Characteristics of Rare Disease Advocacy Organizations and the Facilitators and Barriers that Influence Advocacy Capacity

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DISSERTATION

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DEDICATION

To my wonderful two sons, Julius R. and Calvin D. Slimko. You are both the reason I remained convicted and committed to this work through all the sacrifices and compromises we endured together. Thank you for inspiring me and supporting your mother to endure until the finish. In memory of my late father who taught me resilience and passed on me my warrior spirit to advocate for rare disease patients, Marino Lagatuz Jr. this is dedicated to you and my Lagatuz Family for always being there and teaching me to reach my highest potential in anything I pursued. To my mother, Felisa M. Lagatuz, thank you teaching me the value of education and giving me the gift of curiosity. To my loving and trusted partner, Steve, thank you for being my rock on this journey!

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MLS

PREFACE

Rare disease patient advocates are a special group of dynamic, tireless, and selfless parents, volunteers, caregivers, family members and friends. These diseases may be rare, but the threat they pose to testing human will is all too real. Rare disease patients must have the courage and resilience to keep on fighting not only for themselves, but for others and even for generations to come. These patients and their advocates have paved the way for new supporters and have continued to create their legacy while at times beating unfavorable odds, and sometimes just getting plain lucky.

It is important to tell their story: the story of a 24/7 job whose work sometimes goes unnoticed while also dealing with the psychological upheaval of the life of a child, friend, or colleague being on the line. For all the rare disease stakeholders, and all the folks fighting toward new milestones to save or improve the quality of life of RD patients, I hope this work helps explain the enormous effort, energy, and commitment it takes to advocate for the RD community. The term "rare" implies that these patients face very individual circumstances. But the nature of being human is to want what others have, and for RD patients this is true as well – they want to belong, to be cared for, and to know they have hope of a future to enjoy their life and the world around them.

As a mother of a child with a rare disease, I hope this work teaches and creates awareness for others, that while rare disease may be called "rare," what we are after is commonplace to any mom or caregiver for someone with a disease. We wish for hope, comfort, care, peace of mind, and a cure. To those who continue to hope, and to those who continue to fight: *together we are stronger, and we will always be connected by being rare*.

TABLE OF CONTENTS

I. BACKGROUND AND PROBLEM STATEMENT 1

a. BACKGROUND - RARE DISEASES 2

i. U.S. Definition 3

ii. Global Definition 3

iii. Patient Challenges 4

iv. Lack of Basic Knowledge 4

v. The search for a diagnosis AKA “Diagnostic Odyssey” and Implications 4

vi. RD Patients Are Geographically Dispersed Globally 6

vii. Lack of Access to Reliable Epidemiological Data 6

viii. High Cost Implications of Living With an RD 6

ix. High Cost of Therapeutic Treatment 7

b. HISTORY OF RARE DISEASES AND PUBLIC HEALTH 8

i. High Cost of Orphan Drugs to National Healthcare 8

ii. Examples of RD impacting PH 14

iii. Positive Benefits of RD documented toward PH 14

iv. 1983 Orphan Drug Act 16

v. Historical and Public Policy Context Promoting Research & Development for RDs and Orphan Products 16

c. RD ADVOCACY AS AN IMPORTANT PUBLIC HEALTH STRATEGY 18

i. RD Alignment with Public Health Framework 18

ii. Advocacy as an Important Public Health Strategy 18

iii. RD Advocacy vs. Traditional Patient Advocacy 18

iv. Goals of RD Advocacy Organizations 19

d. PROBLEM STATEMENT AND RESEARCH QUESTIONS 20

e. LEADERSHIP IMPLICATIONS AND RELEVANCE 21

i. Leadership Implications 21

ii. PH Leaders Can Challenge the Status Quo 21

iii. Identification of Adaptive Challenges with RDAOs 22

iv. Importance of Perspectives: Looking at Balcony and Field of Play 22

TABLE OF CONTENTS (continued)

v. Relevance and Significance of the Study 22
II. CONCEPTUAL AND ANALYTICAL FRAMEWORKS 24
a. LITERATURE REVIEW 24
i. Role of Advocacy in Non-Profit Organizations 24
ii. Lack of Formal or Accepted Definition for Advocacy 24
iii. Multiple Definitions of Advocacy in Literature 25
iv. Public Health Advocacy Definition 26
v. Rare Disease Advocacy Remains Undefined and Undocumented 26
b. THE ADVOCACY AND POLICY CHANGE COMPOSITE LOGIC MODEL 26
i. Capacity-Building 28
ii. Engagement and Outreach 28
iii. Increasing Advocacy Capacity 29
iv. Improved Services and Systems 29
c. CONCEPTUAL FRAMEWORK 29
i. Public Health Frameworks and Model 29
ii. Essential Services of Public Health 30
iii. The Core Functions of Public Health 31
1. Assessment 31
2. Policy Development 31
3. Assurance 32
iv. Conceptual Framework 33
III. STUDY DESIGN, DATA, AND METHODS 42
a. Analytical Approach 42
i. Research Rationale 43
ii. Study Setting 43
iii. Study Selection 44
b. DATA SOURCES AND COLLECTION 45
i. Document Review 45
ii. Interviews 46

TABLE OF CONTENTS (continued)

iii. Data Analysis Plan 50
iv. Codebook 50
v. Validity and Reliability Considerations 52
IV. RESULTS 54
a. OVERVIEW OF THE CHAPTER 54
b. RESEARCH DESIGN: PHASE I: SEMI-STRUCTURED INTERVIEWS 54
i. Phase I: Semi-Structured Interviews 55
1. Analysis of Data and Determining of Coding Scheme: Code Development 55
2. Memo Development and Use of Memos 57
ii. Final Determination of Results 57
c. RESEARCH DESIGN: PHASE II: FACILITATED SESSION WITH GLOBAL GENES 59
i. Facilitated Discussion with Global Genes 59
ii. Characteristics of Study Sample 60
iii. Member Criteria of Rare Alliance Foundation Members 64
iv. Recruitment of Volunteers to Participate in Dissertation Research 65
d. SUMMARY OF PHASE I FINDINGS 66
i. Research Question 1: What are the organizational capacity factors that influence RD advocacy? 69
ii. Factors and sub-factors that Influence RD Advocacy Factors and sub-factors that Influence RD Advocacy 69
1. Factor 1 - Organizational Infrastructure, Including Themes and Sub-Factors 70
2. Factor 2 – Organizational Capacity - Funding 78
3. Summary of Factors 3, 4, and 5: Organizational Capacity - Genesis, Prioritization Process, and Role/Title 85
iii. Sub-research Question 1: What are the characteristics of rare disease organizations that undertake advocacy activities? 95
1. Characteristic 1 - Engagement and Outreach 95
2. Characteristic 2 - Gaining Value from Other People 109
iv. Sub-Research Question 2: How have these characteristics facilitated or acted as barriers to advocacy? 117
1. Barriers to Organizational Capacity 119

TABLE OF CONTENTS (continued)

2. Barriers to Advocacy Capacity 129
3. Facilitators to RD Advocacy 144
V: DISCUSSION 156
a. INTRODUCTION 156
b. CONCEPTUAL FRAMEWORK 156
c. RDAO LEADERSHIP 157
d. KEY FINDINGS 158
1. Various types of engagement of people are critical to advancing RD advocacy 158
2. The practice of RD advocacy is not a linear process. 161
3. Barriers and Facilitators to RD Advocacy that were identified also aligned with Organizational Capacity Factors 163
e. IMPLICATIONS FOR THE PUBLIC HEALTH PROFESSIONALS 167
f. NEW CONCEPTUAL MODEL: RECOGNIZING THE MULTIPLE DIMENSIONS OF RD ADVOCACY TO MAXIMIZE ORGANIZATIONAL CAPACITY 168
g. STUDY STRENGTHS AND LIMITATIONS 174
h. CONCLUSIONS 176
i. RECOMMENDATIONS FOR FUTURE STUDIES 177
APPENDICES 178
Appendix 1: Research Protocol 178
Appendix 2: IRB Exemption Letter 184
Appendix 3. Measurement Table 187
Appendix 4: Codebook 192
Appendix 5: Oral Informed Consent Script 200
Appendix 6: Recruitment Email 201
Appendix 7: Facebook Notice for Recruitment 202
Appendix 8: Semi-Structured Interview Guide- Rare Disease Advocacy 203
BIBLIOGRAPHY 205
VITA 211

LIST OF TABLES

TABLE I TOP EIGHT HIGHEST PRICED ORPHAN DRUGS IN THE US, 2016 12

TABLE II DATA MANAGEMENT OF RDAO SEMI-STRUCTURED INTERVIEWS.....49

TABLE III CHARACTERISTICS OF STUDY SAMPLE 63-64

TABLE IV SUMMARY OF ORGANIZATIONAL CAPACITY FACTORS AND THEMES.....67

TABLE V SUMMARY OF CHARACTERISTICS OF RDAOS 68

TABLE VI SUMMARY OF ORGANIZATIONAL FACILITATORS AND BARRIERS TO RD ADVOCACY 68

TABLE VII TOTAL NUMBER OF ORGANIZATIONAL RESPONSES AND NUMBER OF CODES FOR ORGANIZATIONAL INFRASTRUCTURE 70

TABLE VIII TOTAL NUMBER OF ORGANIZATIONAL RESPONSES AND NUMBER OF CODES FOR FUNDING.....78

TABLE IX FUNDING THEMES RELATED TO FACTORS THAT INFLUENCE ADVOCACY 79

TABLE X APPROACH TO PRIORITIZATION PROCESS, ROLE/TITLE OF INTERVIEWER AND GENESIS OF RDAO AND SUMMARY OF CODES PER ORGANIZATION 85

TABLE XI CHARACTERISTICS OF RDAOS – ENGAGEMENT & OUTREACH, NUMBER OF CODES PER ORGANIZATION 96

TABLE XII SUMMARY OF THEMES FOR CHARACTERISTICS OF RDAOS-ENGAGEMENT & OUTREACH.....96

TABLE XIII SUMMARY OF CODES FOR NETWORKING, THIRD-PARTY TECHNICAL SUPPORT, PARTNERING AND MENTORING 110

TABLE XIV SUMMARY OF CODES PER ORGANIZATION FOR BARRIERS TO ACHIEVING ADVOCACY CODE 118

TABLE XV SUMMARY OF THEMES RELATED TO BARRIERS TO ACHIEVING ORGANIZATIONAL CAPACITY.....119

LIST OF TABLES (CONTINUED)

TABLE XVI BARRIERS TO ADVOCACY CAPACITY THEMES AND NUMBER OF REPORTING ORGANIZATIONS AND FREQUENCY OF QUOTATIONS.....129

TABLE XVII FACILITATORS TO RD ADVOCACY THEMES 145

TABLE XVIII FACILITATORS TO ADVOCACY SUMMARY OF CODES.....146

LIST OF FIGURES

Figure 1 Relative number of orphan drugs in development in 2016, by therapeutic category....9

Figure 2 Worldwide orphan drug sales and share of the prescription drug market.....11

Figure 3 Advocacy and policy change composite logic model 27

Figure 4 The 10 essential services of public health and core functions of public health 30

Figure 5 Conceptual framework of building RD advocacy capacity and impacts 33

Figure 6 Purposive sampling thru Global Genes Foundation Alliance membership.....45

Figure 7 Phase I: Semi-structured interview analysis steps 58

Figure 8 Phase II: Facilitated discussion with Global Genes patient engagement team summary.....59

Figure 9 Map of main office locations designating different capacity levels of the RDAOs 66

Figure 10 Benefit of social platforms to helping Orphan Drug research and development.....161

Figure 11 Conceptual Model of Rare Disease Advocacy.....170

Figure 12 Rare Disease Advocacy Capacity Roadmap and RDAO Intake Data.....174

ABBREVIATIONS

CAGR	Compound and Annual Growth Rate
DRPH	Doctor of Public Health
GG	Global Genes
NIH	National Institutes of Health
NORD	National Organization for Rare Diseases
PH	Public Health
RDLA	Rare Disease Legislative Action
RD(s)	Rare Disease(s)
RDAO(s)	Rare Disease Advocacy Organization

SUMMARY

Rare disease patient advocacy organizations (RDAOs) enable individuals with mutual interests to bring their collective resources together, sharing knowledge so they can influence key stakeholders who are advancing work on behalf of that specific Rare Disease (RD) or family of RDs (Terry SF, 2001); (Dunkle M, 2010). Most RD patients are children seeking a cure for diseases that are often first overlooked, mis-diagnosed, and sometimes ignored by primary caregivers (Genes, 2018). Once a diagnosis is made, RD patients often don't have the luxury of time to seek treatment and cures due to rapid disease progression, time it took to obtain the correct diagnosis, therapy time commitments, and lack of access to experts to evaluate their rare disease.

Advocacy for RD can significantly reduce the time it takes for patients and their advocates to 1) gain access to RD experts, 2) obtain a correct diagnosis and 3) find effective treatments. Capacity building, defined as the ability of nonprofit organizations to fulfill their missions in an effective manner (Twombly, 2001) is critical for the lifespan of a nonprofit organization. Even more important are advocacy efforts done on behalf of the organization to advance its overall mission. We see this every day in marketing campaigns for the Red Cross, ASPCA, and other organizations that are great at getting their message out. They not only advocate for their own goals, but show people the important work that they're doing, building the capacity for more work to be done. Advocacy on behalf of RD patients exists but is not documented much in the literature. Only 5% of these diseases have treatments (Genes, 2018), yet patient advocacy organizations can play critical roles in positively influencing clinical research, drug development, and regulations by championing funding, increased awareness, and creating

relationships among key stakeholders including experts, drug developers, and biotech companies in order to advocate for change, speed up research, or foster tools and resources important to RD patients (Davio, 2018). While individual rare diseases are low in prevalence, RDs as whole community represent 10% of the US population (America, 2018) Which equates to an estimated 350 million patients worldwide and approximately 30 million patients in the United States (Griggs, 2009); (America, 2018).

RDs have a significant impact on various public health issues such as healthcare costs, access and affordability of therapies, quality of life issues and the application of RD knowledge to other diseases around the globe that speak to the unique needs of these patients (Valdez, 2016); (Eurodis, 2005). More information and research are vital for the broader rare disease community, for all its stakeholders, patients, and families to better understand their environment, how to effectively advocate for patients, and to increase understanding for advocates and caregivers to foster better support, care, and connections (Eurodis, 2005).

The ability for RDAOs as nonprofit 501c3 organizations to prepare, anticipate, mobilize, and execute advocacy activities is critical to facilitate public health approaches to addressing rare disease including prevention messages, early screening and testing, and surveillance. This study examined the organizational capacity factors that influence RD advocacy, identified barriers and facilitators to RD advocacy, and described the characteristics of RDAOs who undertake advocacy work. Improved understanding and awareness of these factors may aid in guidance of RDAO advocacy strategies and foster improved engagement among the many stakeholders involved in RD advocacy.

This study had two aims. One aim was to describe and create a greater understanding of the characteristics of capacity and capacity-building strategies used for advocacy efforts in rare

disease organizations who advocate on behalf of patients and families that have a single rare disease. The second aim was to identify facilitators and barriers to undertaking advocacy activities. This study may provide information that could further develop capacity-building programs.

To better understand the characteristics of rare disease advocacy capacity, this study used a qualitative case study research design to allow for greater examination of characteristics of these types of organizations, along with identifying their facilitators and barriers to conducting rare disease advocacy. Global Genes, based in CA was selected as a non-profit umbrella organization that supports the global rare disease advocacy community and focuses on building capacity within RD organizations. Global Genes Foundation Alliance Members who were actively engaged, and had dedicated advocacy managers, were selected using purposeful sampling among 30 RDAOs.

The methods comprised a two-phase approach. Phase I included semi-structured interviews to gain a deeper understanding and context to how and why these organizations function as an RD advocacy organization. Phase II included a facilitated discussion where the researcher presented de-identified findings to a group of no more than 15 Global Genes advocacy staff members.

a. Study Findings

1. Various types of engagement of people are critical to advancing RD advocacy

The greatest number of codes in the entire study were *engagement and outreach* and *gaining value from other people*. Engagement and outreach were defined as connecting with people who have similar RD or stakeholders of the RD in order to inform, educate, or connect within a specific RD community. Gaining value from other people for RD advocacy in ways related to

mentoring, networking, partnerships and using third party technical support. Taken together, this represents the important role people serve both internally and externally to RD advocacy. This is important for several reasons. First, with the lack of basic resources available to the RDAOs and individual RDs, advancing advocacy activities is best done with and through people (Dunkle M, 2010). In the absence of research, experts, funding, therapy, or a cure, advocacy that requires multiple steps is advanced through people. By partnering, leveraging mentors or experts, building networks, and seeking greater exposure or support from other people or experts, organizations acquired deeper learning, connections and problem solving.

Second, this study also shed light on the importance of telling the RD story in order to increase awareness and engagement amongst key people who could advance RD advocacy work. These findings may help GG in their future planning and placing an emphasis on their planning efforts as it relates to how RDAOs can best achieve their greatest potential through more opportunities to engage with people, drive advocacy activities and advocate for patients with RDs.

2. The practice of RD advocacy is not a linear process.

This study provided an opportunity to document how RDAOs in this community support and build their own organizations. A key finding was that the process for RD advocacy is not linear. RDAOs were run mostly by family members who may have been parents, or a relative who was also juggling several responsibilities. The unique role that leaders in RDAOs have is trying to run a 501c3 nonprofit organization while dealing with the emotional, physical, and social demands of caring for their family member with an RD. This process is not linear in that advancement in one area of RD advocacy leads to the next advancement that is closer to obtaining a cure. For example, and RD advocate may advance scientific research with a drug

company but have to juggle managing and administering a non-profit organization at the same time. While all that is taking place, they may have to go home to care for their family or child with RD. Yet, despite all this, RDAOs continue to advance their work and try to keep moving forward.

One major example of the complexity of pursuing advocacy work, often from scratch, while caring for a family member with an RD, was understanding the RDAOs prioritization processes – the process by which they define and tackle advocacy goals. While many described a more formal process, what was really happening in their RDAO was a different story evidenced by further descriptions of additional extraneous workplans that were not identified in approved strategic plans. In fact, RDAO representatives described handling multiple priorities that could change and veer away from their original plan. This leads to the organization recognizing important roles and bouncing between various dimensions that address the needs of the individual with RD, organizational capacity, advocacy capacity, and the ability to connect all dimensions to the overall world of RDs.

3. Barriers and Facilitators to RD Advocacy that were identified also aligned with Organizational Capacity Factors

The organizational capacity factors represented ways that RDAOs can get their advocacy work done. The barriers that were identified such as fundraising, prioritization process, diagnosis with an RD, and lack of RD resources were mentioned conversely as the important factors that positively advance RD advocacy. This implied that these barriers that can also serve an organizational capacity factors if they are not lacking.

A main facilitator for advancing RD advocacy is starting a 501c3 nonprofit organization. This finding also aligns with the genesis findings in the organizational capacity factors. The challenge of building a community was also identified as a barrier, yet this important characteristic describes RDAOs who were undertaking RD advocacy work. This is another alignment of where this factor is lacking, when it is present, serves as a positive facilitator to RD advocacy.

4. The power of an RD advocate in RDAOs

Interviewees with the RDAOs in this study left an immediate impression by the way they talked about their work with conviction describing how they never gave up, and always sought the help and support of others. Key characteristics of these advocates emerged as having traits of compassion, genuineness, and resiliency. These are identified and described as core elements for RD advocates. Most likely RD advocates are personally connected to the RD, and their passion, commitment, and belief in this work compels them to be driven and dedicated to their work because it is part of their family.

a. Implications for Public Health Professionals

RDAOs are an important consideration in public health and can easily get dismissed due to its categorical name as rare diseases. RD may have low numbers of people per disease, but overall, RDs represent 25-30 million people globally. Understanding more about how RDAOs interact and build organizational capacity to advance advocacy is helpful not only to umbrella advocacy organizations such as GG, but also to the various stakeholders affiliated with supporting rare diseases. PH professionals can help translate the important contributions of RDAOs to PH as it relates to PH policy, legislation, research, and advocacy. Furthermore, PH professionals can play an important role in advocating for greater recognition of RDAOs as a

minority population that deserves and requires health equity relative to health care, access, affordability and greater PH surveillance practices to further understand RDAOs role in PH in the future.

b. Key Conclusions

Greater awareness and understanding how RDAOs build organizational capacity are important not only for RD communities but for all the stakeholders, actors and people who play supportive and lead roles. This study's examination of the factors that contributed to organizational capacity can help organizations, researchers, governmental officials, pharmaceutical and biotech companies, RDAOs, and others identify priorities for investigating organizational capacity vulnerabilities and possible strategies to improve advocacy practices in the face of working in RD advocacy that oftentimes has sparse and lacks unavailable resources.

RDAOs are an important population to continue studying and supporting due to their impact and contributions to public health, and to also address future RD impacts to public health such as access to health care, costs, and healthy equity related issues. They have the potential to greatly influence healthcare and will continue to need care, dedicated experts, understanding, and support. RDAOs should be considered in health policy and legislation to remain an important part of the future public health agendas.

c. Recommendations for Future Studies

As in conducting action-based research, this study represents a segment of time. Furthering this work to continue extend the study to include an examination of the proposed model and application to an organization like Global Genes would be worth exploring to test feasibility of the revised conceptual model and proposed individual intake data approach to see if feasible, useful and beneficial to the organization.

Potential Future Research Questions for Continuing Action Based Research:

1. Is organizational capacity improved when applying the revised conceptual framework? Why or why not?
2. Does applying an individual advocacy workplan and strategy effective for RDAOs? What do they gain, or what do the miss by applying this approach?
3. How has individualized support enhance or deter from GG advocacy strategies?

I. BACKGROUND AND PROBLEM STATEMENT

Rare diseases impact an estimated 1 in 10 Americans, or 30 million people, and 350 million people worldwide are living with a rare disease (Field MJ, 2010). Of the RD population, 50% are children, where a third will die before the age of five (Genes, 2018). Yet, in 2000, less than $20B was spent on drugs to treat RDs globally, and in 2020, that amount is projected to increase to $176B, representing approximately 19% of the world's total drug expense (America, 2018). These figures illustrate the incredible growth of the RD community. The majority of these individuals lack a single treatment option and have limited access to resources. Powerful patient advocacy and advocacy groups are a major approach to obtaining more funding for research, brokering key collaborations, and activating communities to conduct "gain success" for these patients (Stoller, 2018). However, because RD organizations have limited staff and resources, little is known about RD advocacy approaches and building capacity. An opportunity exists to explore advocacy capacity to inform how RD organizations can improve their advocacy approaches.

Rare diseases are often misunderstood and misdiagnosed, as they affect a very small subset of the population. Primary care doctors must evaluate symptoms with their current knowledge. Also causing confusion is the fact that RDs are often framed as diseases that affect only a small part of the population, lack published literature, and are not well documented or well understood. They are thus perceived not to impact public health due to their low incidence and prevalence – but I will show why this is not true and how RD advocacy can turn the tide (Field MJ, 2010).

One example of a disease that started out unknown and has significantly impacted public health is AIDS (Field MJ, 2010); (CDC, 2010). As the infection spread and health care providers

had the ability to improve diagnostic capabilities as well as data collection systems, researchers developed effective treatments that reduced mortality despite not developing a cure. This once-known rare disease rose to approximately 470,000 people by 2007 and the number of individuals infected with HIV exceed 1.1M (CDC, 2010) (Field MJ B. T.). It no longer remains a RD, but is now a more prominent, widely-known disease. With greater awareness, patient advocacy, funding and research, AIDS treatment, awareness, education and quality of life has greatly improved.

The examination of RDs on a broader platform such as public health, has a greater population impact. RDs affect 30 million Americans, or 1 in 10 people, and 350 million people worldwide (Eurodis, 2009) (America, 2018). According to Global Genes, a rare disease advocacy non-profit organization, if all people with RDs lived in one country, it would be represent the world's third most populated country. An estimated half of the people affected by RDs are children, and RDs affect more people than AIDS and cancer cumulatively (Genes, 2018). Yet, 95% of RDs do not have a single FDA approved drug treatment and no cures are available, and almost half of RDs do not have a specific disease foundation or supporting organization dedicated to researching their disease (Genes, 2018). Examining RDs at the organizational level and determining how RDAOs can increase capacity will inform research into better and more fruitful advocacy to provide sound healthcare, fund research, and treatment, therapy, and cure development. Collectively, RDs have similar health issues like many other Americans, including physical disabilities, access to affordable and quality care, learning disabilities, and needs for sound childhood health and overall development.

a. BACKGROUND - RARE DISEASES

This section provides background and context to RDs including recognized definitions from US and global perspectives. Rare diseases are a complex topic because they require connecting

many different underdeveloped or hard-to-find sources. An outline of challenges is provided that describes the multi-faceted challenges associated with living with or caring for someone with a RD. Additionally, these challenges can influence the viewpoint of advocacy needs for the patient, the clinicians, and investigators. Identifying these important perspectives collectively is important when addressing the challenges RD advocates may face (Stoller, 2018) (WHO, 2013).

i. U.S. Definition

In the United States, rare disease is defined as a condition that affects fewer than 200,000 people. Congress created this definition through in the Orphan Drug Act of 1983 (FDA, 1983). Rare diseases were simultaneously referred to as orphan diseases as a result of the lack of interest by drug companies to adopt them develop treatments. This definition was needed to establish which conditions would qualify for the new incentive programs provided in the Orphan Drug Act of 1983 (Field MJ, 2010; FDA, 1983). In the United States, not many rare diseases are tracked when a person is diagnosed. Additionally, this also includes some infectious diseases, birth defects, and cancers, as well as the diseases on state newborn screening tests. Since most rare diseases are not tracked, it make is challenging to effectively know the precise prevalence and incidence of people with rare diseases (Field MJ, 2010).

ii. Global Definition

According to the World Health Organization, there are between 5,000 and 8,000 rare diseases. The total number of Americans living with a rare disease is estimated between 25-30 million, 30 million in Europe, and 400 million worldwide. An approximation is that 1 out of 15 persons worldwide could be affected by a RD. RDs are chronic diseases and can be life threatening (WHO, 2013) (Eurodis, 2005).

iii. Patient Challenges

Despite positive developments over the last decade or so, the burden of rare disease continues to exist for a variety of reasons. RDs have basic and varying challenges compared to diseases with prominent prevalence such as asthma or heart disease. This is most appaprent at the clinical development stage when rare disease poses complications to completion of a study (WHO, 2013).

iv. Lack of Basic Knowledge

Compared to more common diseases, basic knowledge about RDs is lacking, such as cause of the disease, pathophysiology, and natural progression of the disease. Epidemiological data is either scarce or not available in many cases. The limited data in the literature or general practice guidelines of health care practitioners significantly hinders the ability to both diagnose and treat RDs. The availability of public funding for basic foundational research into the disease process remains needed both globally and nationally (WHO, 2013).

v. The search for a diagnosis AKA "Diagnostic Odyssey" and Implications

Due to the dearth of basic knowledge, an RD patient often endures a long journey in order to gain a fundamental piece of knowledge, a confirmed diagnosis of their RD, so that they may begin moving forward with care and treatment. A "diagnostic odyssey" is a term often used with RD patients, , that describes their upfront challenge in navigating the health system to seek a diagnosis. With an RD, gaining access to information that is readily available and part of everyday medical practice is not common. Thus, the odyssey describes how an RD patient may enter, leave, and return to the healthcare system just to find a diagnosis – a potentially multiyear long process for some patients (Evans, 2018).

EURODIS, a non-governmental organization that represents European RD patient groups, conducted the biggest study regarding time-to-diagnosis. It's been a decade since they surveyed 6,000 patients in 17 countries who were affected by one of eight RDs. Results showed that approximately 25% of patients waited from 5-30 years to gain a diagnosis for their condition and two-fifths (Shire, 2013) were misdiagnosed within their duration of a diagnostic odyssey (Eurodis, 2005). A more recent study conducted by Shire Pharmaceuticals in 2013 (Shire, 2013) uncovered that US patients endured an average of 7.6 years and UK patients 5.6 years until diagnosis. The study noted that patients surveyed typically saw up to eight doctors and were given two to three misdiagnoses along their journey. In another survey completed by Engage Health on behalf of GG, an average time of diagnosis to be 4.8 years was witnessed, while the longest time reported was 20 years (Chan, 2017)(Engel PA., 2013).

Several implications arise for RD patients who get lost in their diagnostic odyssey. Lacking a diagnosis can lead to feelings of anxiety, frustration, and stress. Moreover, misdiagnoses can also result in wrong or inappropriate treatments, or living on false hope that their answer has arrived, erroneously ending the patient's need for continued learning. (Eurodis, 2009).

Beyond emotional implications, lacking a diagnosis also puts patients in precautionary situations such as prolonging or postponing having children due to not knowing what potential risks may lie ahead. Other patients may embark on "medical pilgrimages" to reach specialist centers (Dharssi, 2017). For example, 2% of respondents to the EURODIS survey in 2009 found that some patients travelled to a different country to get an accurate diagnosis (Eurodis, 2009). Enduring many out-of-pocket costs may also add financial stress on patients and their families (Anderson, 2013) (Eurodis, 2009).

Patients who get lost in the system in pursuit of the right treatment, expert answers, and ultimately a diagnosis, may cause governments to absorb undue healthcare costs. If on the wrong path or experiencing misdiagnosis, the healthcare costs related to tests, procedures, treatments, referrals and other healthcare spending can impact the healthcare system tremendously (Chan, 2017).

vi. RD Patients Are Geographically Dispersed Globally

RD patients do not live in big clusters or pockets throughout the globe. As a consequence, medical expertise for each of these diseases is scarce and dispersed. Fragmented disease knowledge drives the need for critical investments in fundamental research that go hand-in-hand with investments in dedicated infrastructure and international networks (biobanks, registries, networks of expertise) (WHO, 2013).

vii. Lack of Access to Reliable Epidemiological Data

Due to low prevalence and incidence, an internationally recognized rare disease classification system that can generate reliable epidemiological data does not exist. A sound classification system with public access would allow for further research into natural history and causes of RDs. Furthermore, safety and clinical effectiveness of therapies and quality of care would also be available to examine (WHO, 2013).

viii. High Cost Implications of Living With an RD

The pursuit of a cure, therapy, or basic to in-depth information can also pose a major financial burden for individuals and families. In a recent survey called the Rare Disease Impact Report (Shire, 2013), conducted in the US and UK, healthcare professionals and payers were asked about their perspectives on the impact of rare disease. Almost every one of the 20 payers included in the survey reported that treatment for an RD is relatively more expensive and costs are rising more quickly compared to common diseases or disorders. For example, the average

cost for a person with hemophilia, a rare blood disorder, was ~$131K per year, which totals ~ $8.7M across their lifetime. People with spinal muscular atrophy had average. annual costs of ~$79K. In a study that looked at people with Duchenne muscular dystrophy (DMD), and their caregivers, medical costs rose 16 times as the disease progressed ($4,420 at Stage 1compared to $68,958 at Stage 5)) (Ryder S., 2017) (Review, 2017). As RDs continue to progress, so will the cost implications to support and treat these patients.

Other costs associated with living with a rare disease include direct healthcare costs, direct non-healthcare costs, direct non-healthcare informal costs, and indirect costs.

ix. High Cost of Therapeutic Treatment

It is important to understand the population impact of RDs in society because oftentimes, they are expensive to diagnose, treat, and support. In addition to thinking about direct and indirect costs, even treating a smaller population who does not have a common disease, is noteworthy for discussion. While patients and caregivers may opt to celebrate an emerging drug or approved therapy, this joy may feel bittersweet. The costs for some RD treatments may carry a steep price tag and high risk. For example, the FDA approved a drug, Nusinersen, for spinal muscular atrophy treatment, and 5 days later, the pharmaceutical company, Biogen, announced that each dose would cost $125K. Patients need six doses in the first year, and three per year after that, accumulating costs of $750K per patient in year one, and $375K annually thereafter (HBR, A Gordon Smith). With at least 10,000 SMA patients in the U.S., if just this segment were treated with Nusinersen, the total cost in the first year would be $3.8B and annual cost thereafter would be $1.9B. In addition, this annual figure does not include administration costs. In fact, the total cost would be much larger since this population of SMA patients may live longer, resulting in the need for ongoing and increased treatments over time. This drug made

headlines and gained criticism over the set drug price and the inability for most patients to afford treatments. Within the bigger picture, cost issues present a threat to the entire U.S. health care system, creating an impasse for patients to pay the skyrocketing cumulative costs of therapies for rare diseases (Review, 2017). This threat creates a greater need for increasing our knowledge about rare diseases and understanding the overall implications of public health and RDs.

b. HISTORY OF RARE DISEASES AND PUBLIC HEALTH

As noted, RDs, considered collectively, have a population health impact on quality of life and costs. A population health approach is needed. In public health, its core functions include assessment, advocacy and assurance. See Figure 4. There is a history of this taking place for RDs and public health. One core public health function that is less well understood in RD is advocacy. RD advocacy would greatly benefit from considering a population approach rather than the single or individual disease level. Little is known about organizational capacity for this specific type of RD advocacy.

i. High Cost of Orphan Drugs to National Healthcare

Drugs for orphan diseases can make a significant difference in the life of RD patients, but oftentimes, are paired with a very high cost (Houlton, 2018). An article written in 2018 by S. Houlton entitled, “Orphan Drugs - The High Cost of Hope” provides insight into orphan drugs’ impact on the U.K. National Health System (NHS) budget as they come onto the market. NHS in England deals with over 1 million patients every 36 hours covering antenatal screening, routine screenings, treatments for long term conditions, transplants, emergency treatment and end of life care (NHS, 2016).

Figure 1 (McConnell, 2004) shows the relative number of 2016 orphan drugs in development by therapeutic category. Some of these very high cost drugs on the market are developed to address RDs that exist in small numbers of the population. Orphan drugs can

significantly benefit RD patients, but the cost implications for the NHS are quite significant (Houlton, 2018).

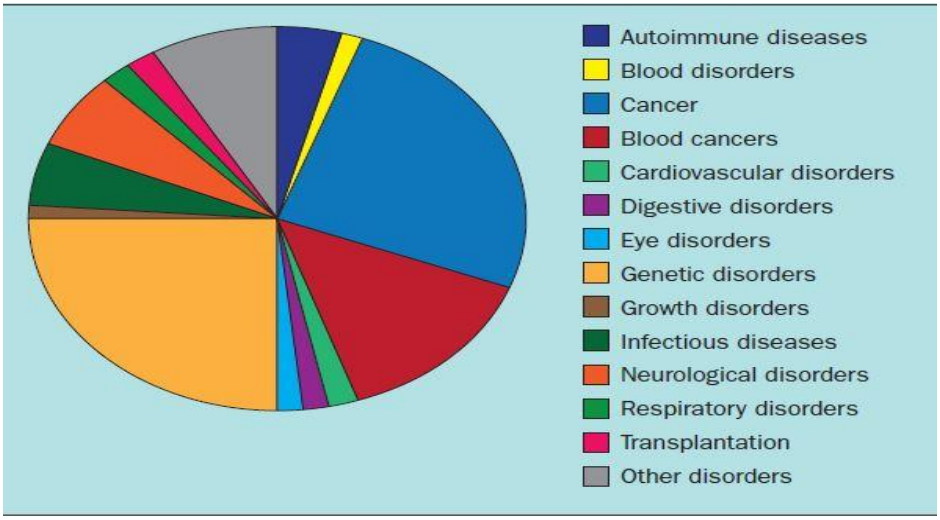


Figure 1. **Relative number of orphan drugs in development in 2016, by therapeutic category** (PhRMA, 2016).

While RDs have their own challenges, developing treatments for this population is also a major, complex challenge. The lack of knowledge, information, and experts often leads to taking a shot in the dark to see what therapies may help. Having a small number of patients means having a small number available to participate in clinical trials. Given the upfront need for solid information to guide drug development, pharmaceutical and biotech companies must also to secure profits from drug sales and address their investors with confidence. Thus, the return on investment for such a small population is not always an attractive revenue for companies and investors.

The premise and rationale behind the concept of the Orphan Drug Act (FDA, 1983). is that it encourages or incentivizes the financial dollars in rare disease research by providing a product an extended seven years of market exclusivity, including tax credits and other incentives

for investing money on this type of research. In order to leverage these benefits, the orphan disease must meet the definition of a rare disease or affect fewer than 200,000 Americans (FDA, 1983).

Legislation in Europe did not emerge until 2000 when the "orphan medicinal product" category was introduced. This category broadened the U.S. definition of rare disease and also included some tropical diseases.

Overlap exists for some drugs that were not intended for RDs, mostly in the areas of cancer, which can also treat a form of the disease that is classified as rare. The designation of "orphan drug" incentivizes pharmaceutical companies to conduct clinical trials in support of more unusual cancers, despite running into issues with recruitment of enough patients.

The rare disease market for potential drugs is anticipated to grow at a steady rate. According to the 2017 Orphan Drug Report from EvaluatePharma, new orphan designations in Europe hit a peak of 208 in 2016; a decade earlier, in 2006, this figure was 82. Unsurprisingly, sales have been rising too, as the chart in Figure 2 shows.

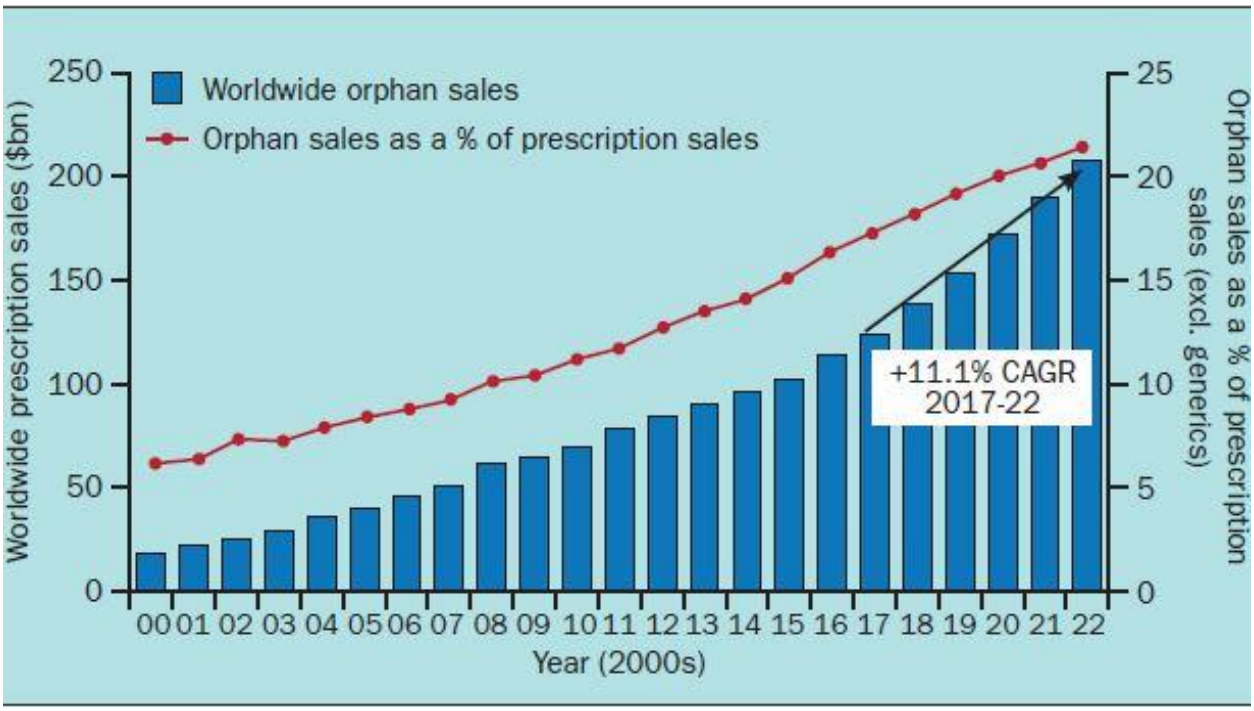


Figure 2. **Worldwide orphan drug sales and share of the prescription drug market (2000–2022) (**Evaluate**,** 2017**)**.

There are several reasons why analysts attribute future growth for the rare disease orphan drug market. In fact, one analyst sees growth beyond the traditional prescription market, noting, "It is definitely growing at a much faster rate than the overall prescription market," says the report's author, EvaluatePharma Senior Analyst Andreas Hadjivasiliou. Hadjjvasiliou also mentions there has been an historical interest in orphan drugs by both small and big pharmaceutical companies, and that early innovators in this space have already proved it is a viable business model. Interest and profitability encourage innovators to be drawn to doing work in orphan drugs.

Hadjivasiliou provides other reasons for growth: he notes that the number of drug approvals is on the rise and drugs that treat larger indications are also being awarded orphan designations. Additionally, personalized medicine supports research into new orphan drugs. He

identifies that as therapies become more and more individualized by targeting specific gene mutations, the science will allow for researching orphan drugs that target a smaller population.

The 2017 EvaluatePharma analysis demonstrates that the smaller the population treated by a drug, the higher the price will likely be. Hadjivasiliou recognized what this means for RD patients and families, noting that, "A lot of these people are in a life-or-death situation, so there is a very strong incentive to provide the drug, whatever the cost."

Table I shows the prices of the eight highest-priced orphan drugs, according to EvaluatePharma's 2017 report. The data is presented in U.S. dollars, whereas U.K. prices are typically lower. The differential in prices is usually lower for orphan drugs (Houlton, 2018).

Brand name	INN	2016 cost PPPY,* USA, in $	Product type	Therapeutic area
Soliris	eculizumab	599,842	antibody	paroxysmal nocturnal haemoglobinuria, atypical haemolytic uraemic syndrome
Naglazyme	galsulfase	492,213	enzyme replacement therapy	mucopolysaccharidosis VI
Eloctate (Elocta in UK)	anti-haemophilic factor (recombinant) Fc fusion protein	459,332	recombinant protein	recombinant factor VIII to treat haemophilia A
Cerezyme	imiglucerase	457,930	enzyme replacement therapy	Type I Gaucher disease
Gazyva (Gazyvaro in UK)	obinutuzumab	449,984	antibody	follicular lymphoma and chronic lymphocytic leukaemia
Cinryze	C1 esterase inhibitor	416,029	blood product	hereditary angioedema
NovoSeven	eptacog alfa	369,245	recombinant protein	recombinant factor VIIa to treat haemophilia A and B
Fabrazyme	agalsidase beta	361,057	enzyme replacement therapy	Fabry disease
*PPPY: per patient per year				

TABLE I. TOP EIGHT HIGHEST PRICED ORPHAN DRUGS IN THE US, 2016
(Evaluate, 2017); (Houlton, 2018).

The very high costs of orphan drugs can have a major impact on a healthy national budget, such as in the U.K. Hadjivasiliou notes that "If you look at the absolute numbers, these

drugs aren't necessarily causing the same budget impact as some of the other drugs that are prescribed more widely to a larger number of people, but [payers] are now looking closely at these drugs and asking if they demonstrate enough benefit to the patient to justify the price. Prior to this examination, such questions may not have been raised. As people start to ask more and more questions, orphan drug costs do not remain under the radar anymore (Houlton, 2018)."

There are different costs associated with the approval of orphan treatments as they translate to less money for people receiving other kinds of services within the NHS, and less money for the salaries of NHS workers. Orphan drugs are a challenge to correctly budget for, because even though the cost-per-patient price is much higher, the impact on the budget as revenue is low; orphan drugs lower, as orphan drugs represent a small portion of the overall NHS budget (Houlton, 2018).

Professor Wailoo Upadhyaya, an expert quoted in the 2017 report, stated that some may argue that the NHS should be prepared to pay more for rare drugs as compared to something intended for more common diseases. RD patients are easily identifiable as there may be less than 10 in the total population. Professor Wailoo pointed out the dilemma of an argument regarding why others should be ready to pay more, which is to provide equal chances of being treated which typically affect children with severe conditions. Many arguments are not really directly tied to being rare but could equally apply to severe but common conditions (Houlton, 2018). Upadhyaya personally stated that patients who equally pay into the systems are afforded the chance to hope for treatment by encouraging innovation by pharma companies in a fair and just manner (Houlton, 2018).

ii. Examples of RD impacting PH

Looking beyond these unique yet high -riced therapies for RDs, there have been also been some positive benefits documented toward public health. At its core, public health is an organized effort by society to keep its members healthy and to prevent disease and disabilities.

iii. Positive Benefits of RD documented toward PH

There are some notable successes with a proven population benefit to public health that range from rare disease screening, approaches for prevention, as well as policy or legislation. Some of these examples include routine surveillance and screening that can identify larger trends (e.g. autism may be an example). Screening can lead to solutions to improve quality of life or complete prevention (Valdez, 2016). Secondly, public health agencies (state level) have commitments to support long-term case management, and some treatments for children with rare diseases exist i.e. congenital hypothyroidism, sickle cell disease, and phenylketonuria (Valdez, 2016). Lastly, work done by rare disease focused organizations may also help eliminate disease, i.e. March of Dimes (Valdez, 2016). Here I outline a few examples of the public health benefits for applied rare disease interventions.

1. Newborn Screening

Implementation of newborn screening exists to ameliorate or prevent adverse metabolic and developmental consequences within children born with rare conditions that have treatments (Valdez R. O., 2016). The National Institute of Child Health and Human Development (NICHD) has been involved in newborn screening efforts since the 1960s. An example of the Institute's earliest research successes was validation of the mass screening test developed by Dr. Robert Guthrie for the metabolic disorder phenylketonuria (PKU) (Alexander, 2003) (Health, 2017).

For more than half a century, newborn screening for selected metabolic and other rare genetic conditions has been a major public health program in the U.S. and many countries

around the world. These programs have arisen because of the availability of life or disability saving interventions coupled with the ability to perform timely diagnoses and treatments early in life. The number of screened conditions has increased steadily over time with more than 30 disorders now recommended by the Advisory Committee on Heritable Disorders in Newborns and Children.

2. Mandatory Folic Acid Fortification

Mandatory folic acid fortification of enriched cereal grain contributed to the reduction of neural tube defects. The CDC's *Morbidity and Mortality Weekly Report* published a study examining how neural tube defects (NTDs) have been prevented since the U.S. Food and Drug Administration mandated that all enriched grain products need to be fortified with folic acid. Researchers uncovered that with the addition and use of folic acid fortification, this resulted in approximately 1,300 babies born annually without an NTD, who may have been affected otherwise (CDC, 2017).

3. Increased Life Expectancy of Cystic Fibrosis Patients

Children with cystic fibrosis in the 1960s had an average life expectancy of approximately 10 years of age. Today, there is still a search for a cure, but targeted treatments have increased the average life expectancy to nearly 50 years (Hurley MN, 2014) (Field MJ B. T.).

4. Decreased Prevalence of Tay-Sachs

Prevalence of Tay-Sachs, a severe genetic disease, has been drastically reduced among the Ashkenazi Jewish population through population screening and strategies. TSD was the first genetic condition where community-based screening for carrier detection was applied. Living with TSD can serve as an example of the benefits of providing public education, carrier testing,

and reproductive counseling to support avoiding a childhood disease that was fatal. The last 28 years provides strong evidence that such efforts can successfully decrease the prevalence of a disease (Kaplan, 2009). Screening Programs for Tay-Sachs have decreased incidence by 90% in high-risk populations in various countries (Rozenberg, 2001).

iv. 1983 Orphan Drug Act

The U.S. Congress passed the Orphan Drug Act in 1983 (FDA, 1983) and it was enacted in the same year to spur the development of drugs for orphan diseases. During the same year, drug therapies for RDs diseases hardly developed. Thirty years later, much more development occurred as an increased segment of industry research and development (R&D) (Herder, 2017) Regulatory drug approvals for diseases affecting fewer than 200,000 persons in the U. S., followed (Herder, 2017).

These examples demonstrate the results that the application of a public health approach to rare diseases can have (Valdez, 2016). Clearly, these are just a few examples of how and public health approaches support rare disease prevention, community education, public policy, population-based health and management, and legislation changes that positively affect populations.

v. Historical and Public Policy Context Promoting Research & Development for RDs and Orphan Products

The connection between public health and RDs began with this first event, noted in 1964. According to the IOM Report on Rare Diseases (Field MJ, 2010) the Committee of the Public Health Service reviewed the impact of changes from the 1962 changes to the requirements for drug approval from regarding the commercial availability of unpatentable drugs and drugs for rare diseases (Field MJ, 2010). Fast-forward to Congressional hearings in the early 1980s, and it focused public attention on RDs while establishing the foundation for passage of the Orphan

Drug Act. Signed into law in 1983, this legislation marked the first significant public commitment of any nation to promote the development of drugs for people with rare diseases. The start of approvals for orphan drugs came that same year the law was passed.

Several active advocates who wanted to push forward legislation came together and formed the National Organization for Rare Disorders (NORD). This organization served and still serves as an umbrella organization for groups supporting patients and families affected by rare conditions. NORD advocates for the identification, treatment, and cure of rare disorders through programs of education, advocacy (Dunkle M, 2010) (NORD, 2018).

Helping to address challenges within the rare disease community are rare disease patient advocacy groups. According to RDCRN (RDRN, 18), Patient Advocacy Groups (PAGs) are organizations that market the needs and priorities of patients. Some of these patient needs could involve supporting research for a specific disease, providing awareness of a disease, and informing the community about a disease. Most patient advocacy groups support one or several diseases (RDRN, 18). Advocacy groups play an important role in cross-collaboration among other public and private groups such as government agencies, commercial companies, academic institutions and investigators.

The following section provides additional content regarding public policy and legislation related to rare diseases in the U.S. and internationally. For the purposes of this study, the focus was to examine and learn more about rare disease advocacy organizations and how they achieve advocacy objectives, rather than to focus solely on policy. Policy is one aspect of advocacy and is currently better documented than broader advocacy efforts. This study aims in part to fill the dearth of literature on RDAO advocacy as a whole.

c. RD ADVOCACY AS AN IMPORTANT PUBLIC HEALTH STRATEGY

i. RD Alignment with Public Health Framework

The work of RD Advocacy aligns with the three core functions of public health: assurance, assessment and public policy (Medicine, 1988). To support these functions, it is important that public health research builds the knowledge base and identifies strategies to achieve health promotion and disease reduction. Advocacy efforts leverage these research findings to push for new public policies that improve health outcomes.

ii. Advocacy as an Important Public Health Strategy

Advocacy is a vital component of public health practice and vital to carrying out all three core functions. Advocacy within non-profit patient organizations for rare diseases play a unique role given the lack of strong funding sources, including both human and financial resources (Dunkle M, 2010) (Pinto, 2016).

RD advocacy groups historically played an essential role in the process that includes an integrated national strategy to accelerate research and product development in rare diseases (Field MJ, 2010). Effective advocacy is recognized by the IOM report to identify the issues that are of importance to the constituent community, and then to advocate policies and programs that address those issues (Field MJ, 2010). The early initiatives of the Cystic Fibrosis Foundation and the Committee to Combat Huntington's Disease helped start an increasing number of patient advocacy groups to get actively involved in rare disease related research. These advocacy groups served in ways that helped create innovative models for funding and organizing research and product development (Field MJ, 2010).

iii. RD Advocacy vs. Traditional Patient Advocacy

The work of rare disease advocacy organizations likely differs from that of other traditional advocacy organizations. Rare disease advocates must also work in a complex, elastic,

and often unchartered environment (Field MJ, 2010) Their cause is rare and may be less relatable than more common diseases and others may perceive common diseases to have a greater return on investment. Additionally, they must remain highly responsive to opportunities that may allow for movement and improvement.

Traditional approaches to advocacy may not apply or may be adapted to increase awareness and interest. For example, a greater emphasis on the use of personal stories and passion-driven results may be needed by rare disease advocacy organizations. Geographical differences may exist in funding, again allowing rare disease organizations to tap into the local story or issues more effectively. Rare disease advocacy organizations also may be more likely to evolve, such as the March of Dimes, which has been effective for over 80 years (March of Dimes, 2018) or dissolve when they are successful in accomplishing the original goals.

iv. Goals of RD Advocacy Organizations

The goals of RD patient advocacy groups vary, depending in part due to the state of the science within various rare disease states, and may also depend on other factors including the number of affected individuals, the interests and skills of organizational founders and leaders, and the success of fundraising strategies. If researchers haven't identified the genetic or other cause of a condition, or delineated how the disease develops, a group may focus its grants and other activities on closing these gaps in knowledge.

An estimated 50% of rare diseases do not have a rare disease specific foundation (Genes, 2018). Advocacy amongst rare disease organizations, its challenges, and successes are important to understand and document to connect sound and relevant public health approaches in the future. The role of advocacy groups is an increasingly important support system to patients with rare diseases. Often, these groups are started by the patients or family members themselves.

They may lack the skills in operating a 501c3 organization, let alone in seeking advanced training or skills to driving.

As one author noted, he believes that independent advocacy by individual groups dilutes the potential political influence (Reid JE, 2001).. However, in aggregate, these rare disease groups could serve as a strong voice to frame the design and reimbursement of health services, research, and social policy (Chang, 2007). In other words, advocating for rare diseases, one disease at a time, versus advocating on behalf of the entire RD community may prove to be more powerful.

d. PROBLEM STATEMENT AND RESEARCH QUESTIONS

Little examination or analysis has been done to look more closely at how to build the internal capacity of non-profit rare disease patient organizations. Analysis has been focused almost exclusively on staff skills to conduct the work, as opposed to broader concepts that define the critically necessary leadership, management and operations to make an effective advocacy organization (Endowment, 2014). Not much is document regarding the application of advocacy as a key strategy in rare disease organizations, how it is defined, what activities are considered facilitative or challenging advocacy activities, as well as the perception of its role in achieving outcomes.

This study has two primary objectives: 1) to understand the characteristics of capacity and capacity building strategies for rare disease advocacy organizations, and 2) to identify factors that influence advocacy capacity and capacity building activities for rare disease organizations.

The main research question for this study is: What are the factors that influence advocacy capacity for rare disease organizations?

Sub-research questions are:

1) What are the characteristics of rare disease organizations that undertake advocacy activities?

2) How have these factors facilitated or presented as barriers to the organization's ability to conduct advocacy on behalf of rare disease patients?

e. LEADERSHIP IMPLICATIONS AND RELEVANCE

i. Leadership Implications

As companies face their own tough challenges to survive in today's competitive global marketplace, the work of leadership is needed not just to provide solutions to their employees, but to forge ahead by asking tough questions to survive. In applying this thinking regarding the work of leadership, Heifetz and Laurie identified the adaptive challenges that must be addressed. These adaptive challenges are not black and white and present themselves as systemic problems without clear-cut answers. However, solving these challenges does not rest on the shoulders of the leader alone. Rather, solving these challenges requires the adaptive work of asking tough questions and leaning on the experience and collective intelligence of people working on the front lines. (Heifetz RA, 1997).

Further leadership implications include challenging traditionally-held notions of "the way we do business," while inviting opportunities to draw out issues and discuss patients' realities, even if harsh or difficult to address. This study's objective was to listen and learn directly from RD advocacy leaders to better understand their work in the trenches at a professional and organizational level.

ii. Public Health Leaders Can Challenge the Status Quo

In conducting and pursuing this research as a leader, the opportunity existed to challenge the ways RDs have historically been perceived and to link the ideas that a disease with low

numbers can in fact have a population-based impact when considered collectively. This conceptual change reframes RDs in the context of a broader conversation: moving RD advocates from working on one particular disease at a time to making change for 30 million global patients. By connecting this work to public health, it may shed light on RD patients while simultaneously uncovering difficult challenges. Additional challenges include issues such as affordability and accessibility of healthcare to a small sub-population, preserving or improving quality of life, economics, resources, and uncovering other barriers that affect the RD community.

iii. Identification of Adaptive Challenges with RDAOs

Rather than providing public health practitioners with a list of solutions, this work aims to work directly with the folks that conduct RD advocacy within various non-profit RD organizations. This approach identified specific adaptive challenges in order to directly support the RD and PH communities that they both impact.

iv. Importance of Perspectives: Looking at Balcony and Field of Play

As Heifetz and Laurie also point out, getting on the balcony is just as important as knowing what is going on in the field of play. Working within this analogy, this research takes into account the perspective of the organizational level while stepping on the balcony to understand the implications from a broader perch, the relationship of RDs to public health. In doing this, the work of leadership is addressed by moving back and forth between the action and the balcony. It creates opportunities to identify emerging patterns that identify the ability to mobilize people to do adaptive work.

v. Relevance and Significance of the Study

The purpose of this work is to define the characteristics of rare disease advocacy organizations, to better inform the rare disease community, and bring awareness to key

stakeholders. This study may provide information that could further develop capacity-building programs and inform advocacy strategies for rare disease advocacy organizations globally.

This work is worth doing because of the very nature of rare diseases. When a patient is first diagnosed, it can feel isolating and helpless, yet, if we can describe rare disease advocacy, we can begin to inform the rare disease advocacy community and document prior and ongoing efforts to initiate, execute, and claim success in advocacy efforts. In addition, by documenting and discussing the challenges, this study will inform the community of anticipated obstacles, and solutions, , so as not to lose the precious time that rare disease patients lack. Even more important, this research will provide important skills and training that advance advocacy work, to anticipate needs in the search for a cure or treatment.

II. CONCEPTUAL AND ANALYTICAL FRAMEWORKS

a. LITERATURE REVIEW

For the literature review, I employed a systematic approach using search tools such as PubMed, Mendeley, UIC Library Services and Google Scholar to identify and summarize information related to the research questions, conceptual framework, and descriptions and summaries related to rare disease advocacy from grey and peer-reviewed literature. For grey literature, references reviewed included conference reports, conference summaries, rare disease advocacy toolkits, and organizational reports. White papers, technical reviews, and policy reports were also used. Publicly available information from the Internet and rare disease advocacy-specific websites were also used to review information about the specific rare disease, and other organization-related information.

i. Role of Advocacy in Non-Profit Organizations

Patient advocacy within the non-profit world is not a new concept. In fact, advocacy is the reason most nonprofit organizations are created (McConnell). McConnell states that the desire to elicit a change in a human life or in the lives of a community serves one of the major compelling reasons for creating an organization. Individuals can and do advocate, but organizations do it better. Organizations can garner resources and target their energy toward a goal. This allows for increasing the chances of making change and improving lives on a large scale. Organizations, through advocacy, can change entire social systems and even cultures to improve the lives of one person or millions (McConnell, 2004). We can draw a connection here between the power of advocacy groups versus individuals, and the power of advocating for all RDs versus one RD. Scale matters.

ii. Lack of Formal or Accepted Definition for Advocacy

Upon starting this research, it was important to gain a base understanding of what is really meant by using the term “advocacy.” There is not a standard accepted definition of the

word “advocacy” by authoritative bodies as nonprofit advocacy can be defined in multiple ways. McConnell lists the words “advocacy,” “lobbying,” and “public policy,” noting that for those outside Washington D.C., these words conjure up mysterious concepts that suggest shady practices, special interest, influence peddling, and back room deals. Often, in different articles, these terms are loosely defined and used interchangeably (McConnell, 2004).

iii. Multiple Definitions of Advocacy in Literature

This research examines research from the organizational perspective of the nonprofit world. Many non-profit organizations that provide human services regularly advocate on behalf of individual clients. More often, however, the term “advocacy” as it relates to non-profits, refers to summative of many definitions rather than individual advocacy (Kimberlin, 2010). For the purposes of this research, one popular definition of “advocacy” within the non-profit field is detailed by Jenkins, who stated that advocacy is “any attempt to influence the decisions of an institutional elite on behalf of a collective interest (Kimberlin, 2010) (Jenkins 1987).” Other experts have offered definitions that focus on the conflict naturally as a part of advocacy, suggesting that advocacy organizations declare public interest claims either promoting or fighting against social change. Further, if the public claims are implemented, these would conflict with the social, cultural, political or economic interests or values of other constituencies and groups (Kimberlin, Andrews and Edwards). Other researchers call for speciftying self-interested organizational advocacy from progressive advocacy which is defined as advocacy that aims to look at underlying structural and unequal power. This approach applies strategies that meaningfully engage constituents more broadly with the process of conducting advocacy (Kimberlin, Donaldson). For the purposes of this research, this work is related to specific advocacy for patients with a rare disease.

iv. Public Health Advocacy Definition

A gap in the literature exists in defining rare disease advocacy. This research aims to address this gap. Additionally, little information is published regarding where rare disease work resides within public health work and little is published or made known by practitioners. Advocacy is a key public health function and core strategy for RD networks/organizations to undertake their work. Even within the review of this area, advocacy has multiple definitions throughout the literature. Public health advocacy refers to advocacy that is intended to reduce death or disability in groups of people (overall or from a specific cause) and that is not only used clinical settings. This type of advocacy uses information and resources to reduce the occurrence or severity of public health problems (Christoffel, 2000).

v. Rare Disease Advocacy Remains Undefined and Undocumented

Yet, there remains a gap in describing and defining rare disease advocacy altogether. It is also important for public health practitioners to know more about this gap as we begin to increase our understanding of its impact on public health as greater evidence and research emerges.

b. THE ADVOCACY AND POLICY CHANGE COMPOSITE LOGIC MODEL

The Composite Logic Model (CLM) was developed as an advocacy evaluation (Coffman, 2018). The CLM also serves as a foundational tool for strategy development and planning by providing an overall picture of how various advocacy tactics connect to interim outcomes that can set the stage for policy change. The CLM addresses different inputs and outputs of public policy and advocacy by providing the user with a method to review and ask the overall question of, "W*hat kinds of outcomes can or should be measured, other than achievement of a public policy goal?"*

It is recommended that users choose the components (inputs, activities, outcomes, policy goals, and impacts) most connected to their current work (Coffman J, 2015). Thus, the CLM

was adapted for purposes of this research related to rare disease advocacy capacity to review the various components of what is relevant to RD advocacy capacity. As advocates ask and answered questions, it provided the ability to map out key areas of advocacy and guide the draft for the researcher code book. As indicated by the CLM authors, the purpose of this tool and exercise used together is to be an iterative process and collection of various options (Institute, 2018).

See Figure 3 to review what information was identified as inputs which were identified as capacity-building and preparation/planning. Activities/tactics included "policy and politics" and "communications and outreach." Interim outcomes included were "advocacy," "capacity," and "policy." The impacts were a mix of what was already provided within this logic model and aligned with the literature review.

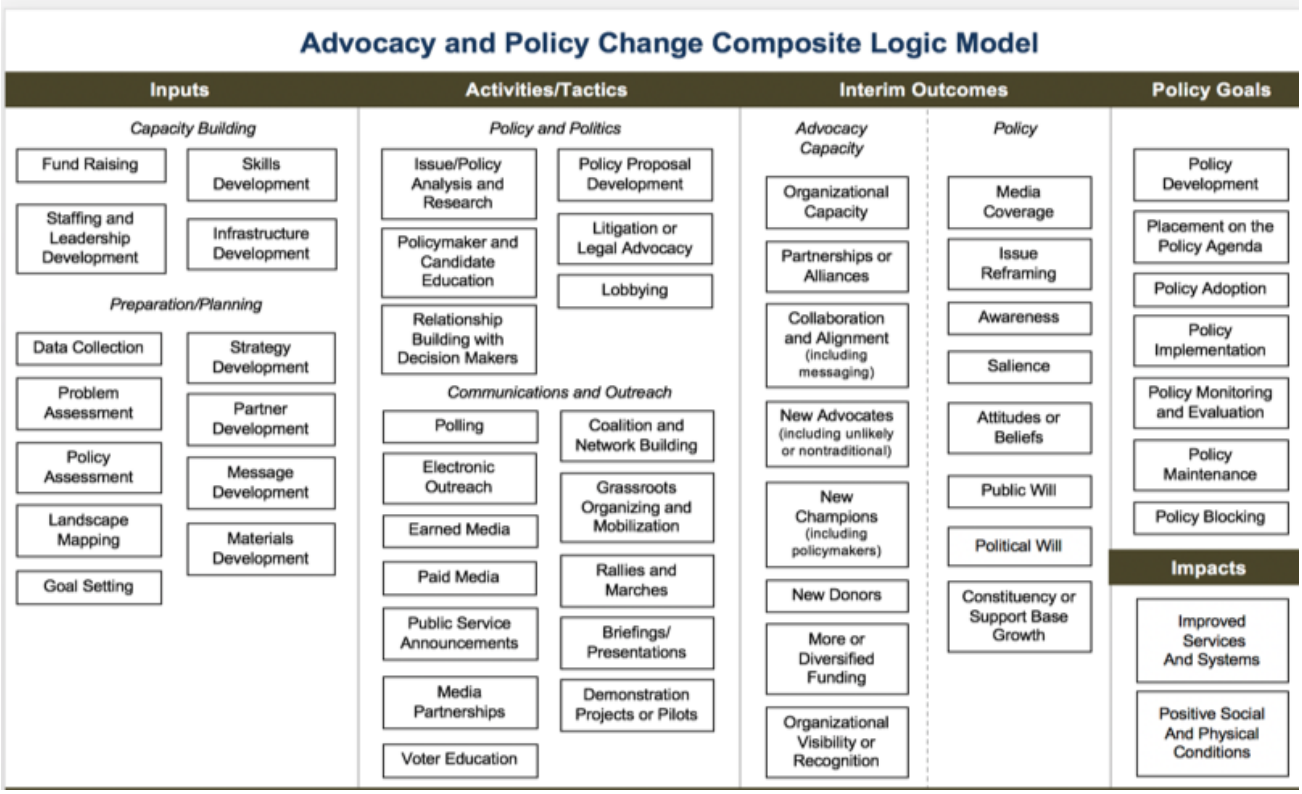


Figure 3. Advocacy and policy change composite logic model

i. Capacity-Building

Capacity-building is defined as the ability of non-profit organizations to fulfill their missions in an effective manner. Non-profits are typically small in size and have limited resources, especially when compared to the enormous challenges they face and the critical issues they aim to address (Twombly, 2001). Administration, finance, human resources, and facilities are among the areas enhanced by capacity-building activities (Twombly, 2001).

ii. Engagement and Outreach

An organization can have a critical mission, solid leadership, and enough resources, but its impact may be limited if not recognized by the community (Institute U. , 2001). Outreach is an important part related to strengthening and growing a mission and its impact. Outreach can be in various forms such as marketing and public relations, community education and advocacy, collaborations, alliances, partnerships, networking and much more (Institute U. , 2001). According to the Amherst W. Wilder Foundation (2000), "For capacity approaches to truly achieve their potential, attention must be given to the web of connections affecting all the persons, organizations, groups, and communities involved." This strategy is part of building social capital and is also good management practice (Twombly, 2001).

Outreach is way for building a foundation of support. The more people who recognize the organization and its work, the greater the opportunity there is to attract folks to the organization and its work, and to attract stakeholders who might push the work forward, such as board members, staff, volunteers, clients, or supporters (Twombly, 2001) (Institute U. , 2001).

Conceptually, organizational outputs and outcomes are the results of many and summative exchanges of the vision and mission, leadership, resources and outreach (Institute U. , 2001). All these factors work in conjunction to creative effective outputs and outcomes driving

the model and helping to mold the endpoints or intended product. However, the endpoints and outcomes provide a feedback loop to the other parts of the model and can improve or decrease their availability and capacity (Twombly, 2001) (Institute U. , 2001).

iii. Increasing Advocacy Capacity

Advocacy capacity is related to overall organizational capacity. Organizations that have sufficient infrastructure, effective staff skills, foundational knowledge, established communication strategies and adequate staffing address taking on any new initiative, advocacy included, much more feasible (City, 2018). Ultimately, investing in advocacy capacity is about investing in people. Advocacy success is not dependent upon unlimited financial resources, high powered political networks, or state-of-the-art communication tools. Advocacy requires people and their passions, armed with the knowledge and skills to leverage their stories for policy change (City, 2018).

iv. Improved Services and Systems

The effectiveness of a non-profit organization depends on demonstrating that their products and services are making a difference to society. In order to show that they are effectively using their resources, there is a need to measure and evaluate the very products and services offered. Funders and investors seek updates on how well a program is running and what it has achieved (Twombly, 2001).

c. CONCEPTUAL FRAMEWORK

i. Public Health Frameworks and Model

To better understand the context of advocacy within public health, it was important to understand the essential services of public health to see how they fit together or connect. There are 10 essential public health services (CDC, 2018) that are fundamental to public health.

Defining them here helps us to understand their interplay with advocacy and specifically, rare disease advocacy.

ii. Essential Services of Public Health

The Core Public Health Functions Steering Committee created the framework for the Essential Services in 1994 (IOM, 1988) (CDC, The Public Health System & the 10 Essential Public Health Services, 2018). The committee was comprised of experts from the U.S. Public Health Service agencies including other major public health organizations. The Public Health wheel in Figure 4 below shows the three core functions on the periphery of the wheel, which are labeled as "assessment," "policy," "development," and "assurance," with key areas listed within the wheel of essential services (CDC, The Public Health System & the 10 Essential Public Health Services, 2018).

Figure 4. The 10 essential services of public health and core functions of public health

iii. The Core Functions of Public Health

Figure 4 shows how the essential health services align with the three core functions of public health, which are "assessment," "policy development," and "assurance" (CDC, CDC Environmental Health Services, 2011; IOM, 1988). The functions of government in public health are described followed by their key strategies as listed by the CDC (CDC, CDC Environmental Health Services, 2011).

Assessment

The wheel begins with assessment. This area includes every activity related to the concept of diagnosing a community, including activities such as surveillance, analyzing why problems occur, collecting and interpreting data, performing research, all the way to the evaluation of outcomes (IOM, 1988). Public health essential services that fall under assessment include:

1. Monitor environmental and health status to identify and solve community environmental health problems
2. Diagnose and investigate environmental health problems and health hazards in the community

Policy Development

According to the IOM Future of Public Health report, policy formulation happens as a result of connecting and working within wide range of public and private organizations and individuals. This process is how a society makes decisions about issues, chooses goals and the appropriate ways to achieve them, handles opposing viewpoints about what needs to be done, and secures resources. An important example provided within this report, related to governmental policy development, includes leadership and advocacy. This is where advocacy plays a key role (IOM, 1988). Public health essential services that fall under policy development include:

3. Inform, educate, and empower people about environmental health issues

4. Mobilize community partnerships and actions to identify and solve environmental health problems
5. Develop policies and plans that support individual and community environmental health efforts

Assurance

The third core function, assurance, is about providing necessary services to achieve goals, either by stimulating the private sector to act, by mandating it, or by directly providing services. It is in this realm where the function in public health seeks to ensure implementation of legislative mandates including statutory responsibilities (IOM, 1988). Public health essential services that fall under assurance include:

6. Enforce laws and regulations that protect environmental health and ensure safety
7. Link people to needed environmental health services and assure the provision of environmental health services when otherwise unavailable
8. Assure a competent environmental health workforce
9. Evaluate effectiveness, accessibility, and quality of personal and population-based environmental health services
10. Research for new insights and innovative solutions to environmental health problems

This PH framework allows us to fit RD advocacy within the traditional definition of PH. The same report that introduced this framework stated that the mission of public health is the "fulfillment of society's interest in assuring conditions in which people can be healthy (Valdez, 2016) (IOM, 1988)." In 2016, Dr. R. Valdez, an epidemiologist at the CDC, wrote an essay asserting the PH approach has limited ability to effect primary prevention. Yet, wider application of a PH approach could greatly benefit RD patients and their families (Valdez, 2016). This framework offers the opportunity to look at RDs with a PH lens.

iv. Conceptual Framework

There were several considerations adapted from the Logic Model that informed the Conceptual Model. Figure 5 represents the building of RD advocacy capacity and its impacts as informed by the literature and adapted from the Policy and Advocacy Composite Model.

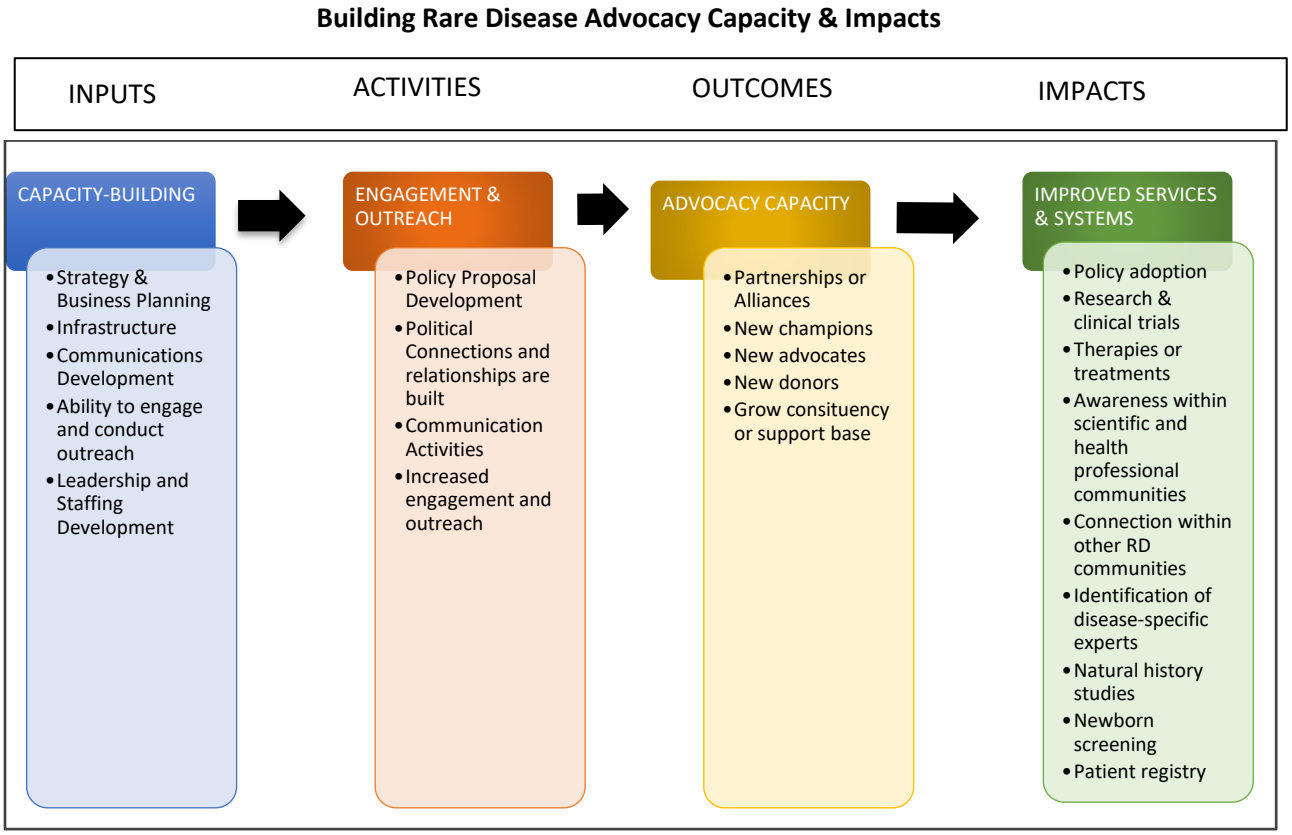


Figure 5: Conceptual framework of building RD advocacy capacity and impacts

1. Inputs—Capacity-Building

According to the National Council of Nonprofits, capacity building is not just about the capacity of the non-profit in the present day, but also about the non-profit's ability to deliver its mission effectively for today and tomorrow's success. It is an investment in the effectiveness and

future sustainability of a nonprofit. Notably, the organization states that when capacity-building is successful, it strengthens a non-profit's ability to reach its mission over time, thus enhancing the non-profit's ability to have a positive impact on lives and communities. The act of building capacity involves many types of activities designed to improve and enhance the way the nonprofit can achieve its mission and sustain itself over time (Profits, 2018).

a. Strategy and Business Planning

The National Council of Nonprofits also states that strategic planning processes identify strategies that will enable a non-profit to advance its mission. This planning involves continually looking to the future and should be a continuous process. In ideal situations, it allows the staff and board to engage in a planning process where they review and become committed to measurable goals, approve priorities for implementation, and agree to revisit strategies on an ongoing basis to address relevancy, changes and prioritization (Profits, 2018).

b. Infrastructure

Capacity-building is important as it allows critical infrastructure, or the way the organization is built, to support and shape charitable work into positive social good. This includes setting up the equipment, systems, and other nuts-and-bolts supports needed to advocate (Profits, 2018). Communications development refers to the creation of various materials to convey information in an oral or written manner. Moreover, advocacy can be done on a shoestring budget but in order to do it on a national or larger scale, the organization must be willing to commit resources to it. Funding and sources of revenue are vital to non-profits' existence (Stalker, 2011).

c. Engagement and Outreach

An important activity for RDAOs is information-sharing through public and private education, which can include conferences, workshops, and meetings where networking can occur (JG, 2013). These different ways to engage refer to connecting with people who have a similar rare disease or stakeholders of the RD to network, inform, educate, or connect within a specific RD community (Coffman, 2018).

d. Leadership and staffing development-related concepts that involve hiring or developing

An organization is as good as its people. Researcher Dr. Ben Asher described leadership and staffing development as two related concepts, "leadership development is the essential condition to achieve virtually all of an organization's objectives. If an organization is developing a dynamic group of leaders at all levels, its problems lessen in proportion to their numbers, strength and distribution. Staff development by definition follows from understandings and actions by leaders within an organization. An organization's leadership development strategy largely determines the extent of effectiveness of its staff development (Asher, 2015-2018)."

2. Activities—Engagement and Outreach

a. Policy Proposal Development

Certain policies can provide a framework, a way to be recognized or positioned, that supports advocacy efforts. "Policy proposal development" refers to a document that details the rationale for and proposed approach to the investigation of a specific policy problem (Coffman, 2018). It is commonly produced in response to an open or targeted call for proposals. In submitting a policy proposal, it is usually in competition with other proposals with the objective to receive funding for a project, fellowship, or post graduate opportunity (Advocacy, 2018) .

RDAOs can benefit from using this approach to compete for resources that are otherwise not available to them.

b. Political Connection and Relationships are Built

Advocacy efforts are difficult to execute with a minimal number of people behind it. It helps to interact with the policymakers or others who have authority to execute upon the issue and create change (Coffman, 2018). This is especially important for RDAOs that may lack prior skills or training in this area; leveraging key relationships can provide support for the work on behalf of their organization.

3. Communication Activities

Connecting with various audiences is important to ensure an organization can maximize its visibility and advocacy efforts. Communication activities includes different ways that materials are disseminated to various audiences in a way that transforms language from research to policy in short, clear messages for targeted audiences (O'Connell, 2018). From a marketing angle, nonprofits conduct communication campaigns that gain public trust, have credibility, work on inspiring issues that grab attention and have a strong record (Communications, 2009).

4. Increased Engagement and Outreach

The RD community has small numbers and are not easily identified or found. Greater awareness is a benefit not only to the RD community, but to its efforts, so they do not go unnoticed. By providing greater facilitation of ways to connect, inform, and educate key stakeholders, constituents, and decision makers, the organization is able to aim for great capacity of its mission (Coffman, 2018).

5. Outcomes—Advocacy Capacity

a. Partnerships or Alliances

While not many resources exist for most RDs, mutually beneficial relationships with other organizations or individuals who support an advocacy strategy can help advance the organization's work (Coffman, 2018). Efficiency is improved by finding others who can strengthen initiatives by t providing needed resources for smaller RDAO with minimal resources.

b. New Champions (including policymakers)

When an organization is working minimal resources and needs to find more members, stakeholders, or funders, the connection with some high-profile individuals who adopt an issue and publicly advocate for it, supports growth by serving as another high visibility resource and voice for the cause (Coffman, 2018). New champions may garner positive attention to help advance an organization's advocacy strategy by aligning or connecting with the community.

c. New Advocates (including unlikely or nontraditional)

Growing its member base is always important for an RDAO. Gaining new advocates who may be volunteers, staff, or previously unengaged individuals who act in support of an issue provide even greater support for the organization (Coffman, 2018). Whether it's providing greater resource, knowledge, networks, or reach, new advocates can increase the power of the organization to carry out its plan (JG, 2013).

d. New Donors

Non-profit revenue streams are always tight and depend on external funding. New public or private funders or individuals who contribute funds or other resources keep programs, staff and organizational priorities sustainable (Coffman, 2018); (JG, 2013).

e. Constituency or Support Base Growth

Advocacy efforts are best effected when a non-profit grows its support base or constituency. This increase in the number of individuals who can be counted on for sustained advocacy or action on an issue is critical to short and long term success of the RDAO (Coffman, 2018); (JG, 2013) (McConnell, 2004).

6. Impacts—Improved Services & Systems

a. Policy adoption

When policy becomes law, non-profit organizations experience success on behalf of their members. The successful passing of a policy proposal through an ordinance, ballot measure, legislation, or legal agreement can provide reflection on the process in ways to operate more efficiently, gain recognition they may have historically lacked, or open new opportunities that were not previously available to a community like RDAOs (Coffman, 2018).

b. Research and Clinical trials

RD patients need clinical trials for researchers, patients, and caregivers to understand their disease and potential reaction to orphan drugs and therapies. To aid in improving study outcomes, sponsors typically leverage experiences and knowledge of actual patients and caregivers in the process of trial design. By doing this, drug makers can gain valuable insights as to the specific and unique experiences of the RD patient (Trials, 2016).

c. Therapies or Treatments

Rare disease patients require therapies and treatments just like any other common disease. Patients can receive care for their condition in various methods like physical care, pharmaceutical interventions, or mental, physical, or emotional approaches (Innonet, 2018) (Lopes, 2018).

d. Scientific and Health Professional Awareness

It is important that RD patients and their family members or caregivers are aware of who can be involved in their care and research. This includes gaining more knowledge about the RD condition and ongoing or planned research, which can include clinical trials, support services, disease-specific information, available databases that have open access, as well as other organization that can offer support for RDs (Office of Rare Disease Research, 2018).

e. Connection to other RD communities

It's important for RD patients, families, and patient organizations to connect with other communities of the same or similar rare disease. Rare disease communities can feel disconnected, isolated, alone, or ignored. But RD communities can step up and help patients and their caregivers feel less isolated, more educated, and more engaged in health outcomes (Pharma Voice, 2017); (JG, 2013).

f. Identify Disease-Specific Experts

The NIH GARD acknowledges that many patients want to meet or know about health care professionals or researchers who have specific knowledge about their RD. When a condition is rare, it is hard to find someone who has seen a multitude of cases. There is not an existing list of RD experts available, but there are ways to identify healthcare professionals who have a unique experience with a particular condition (GARD, 2017).

g. Natural History Studies

According to the National Cancer Institute, the definition of a natural history study refers to a study that follows a group of people over time who have, or are at risk of developing, a specific medical condition or disease. A natural history study collects health information to

understand how the medical condition or disease develops and how to treat it (Institute N. C., 2018).

h. Newborn Screening

Newborn screening at birth can identify conditions that can affect a child's long-term health or survival, according to the CDC (CDC, Newborn Screening Portal, 2016). This refers to routine screening of millions of babies in the U.S. by taking some blood from the newborn's heel that allowed for the testing for specific genetic, endocrine, and metabolic disorders (Valdez R. O., 2016). The purpose of newborn screening is for early detection, diagnosis, and intervention with the hope to prevent death or disability of the newborn (JG, 2013).

i. Patient Registry

A patient registry refers to an organized collection of uniform types of data that use observational study methods to evaluate specific outcomes for a population defined by a disease, condition, or exposure (Innonet, 2018) (Gliklich RD, 2014). RDAOs support creation of patient registries in order to apply the information to their RD population when in most instances, prior data is lacking (JG, 2013).

j. Summary

In summary, a multitude of factors were taken into consideration when examining rare disease advocacy capacity, public health approaches, and critical factors associated with policy and advocacy in general. Very little is published and available regarding fundamentally describing what it means to conduct advocacy for rare disease patients or communities and describe current factors associated with organizational capacity. Some of what is published demonstrates how rare disease patient advocacy organizations have been involved in patient-related clinical trials, design, or implementation to support therapies or drugs for certain diseases.

These factors outline RD community needs to build capacity in both organizational and advocacy work.

III. STUDY DESIGN, DATA, AND METHODS

a. ANALYTICAL APPROACH

To better understand the characteristics of rare disease advocacy capacity, this study used a qualitative case study research design. Global Genes was selected as an organization that supports the global rare disease advocacy community and focuses on building capacity within RD organizations. Global Genes Foundation Alliance Members who were actively engaged with advocacy managers were selected using purposeful sampling among 30 RDAOs. According to Yin (2003), a case study design should be considered when the focus of the study is to answer "how and why" questions, when the researcher cannot manipulate behavior of participants in the study, and want to cover contextual conditions because the researcher believed they were relevant phenomenon under study (Baxter P, 2008).

This study had two purposes. One purpose was to describe and create a greater understanding of the characteristics of capacity and capacity-building strategies used for advocacy efforts in rare disease organizations who advocate on behalf of patients and families that have a single rare disease. The second purpose was to identify facilitators and barriers to undertaking advocacy activities. This study may provide information that could further develop capacity-building programs.

The methods comprised a two-phase approach. Phase I included semi-structured interviews. Semi-structured interviews allow researchers to gain a deeper understanding of a context (Maxwell 2013). Phase II included a facilitated discussion where the researcher presented de-identified findings to a group of no more than 15 Global Genes advocacy staff members.

Working with a rare disease advocacy organization like Global Genes allowed for taking cues, questions, puzzles, and problems, from the perceptions of practitioners in mainly local

practice contexts (Kathryn Herr, 2015); ((Agyris, 1991). Working in phases allowed for addressing the research questions in real time, while changes occurred within the setting, situation, and researcher (Kathryn Herr, 2015).

i. Research Rationale

For this study, the main sources of data were collected from qualitative and semi-structured interviews, documents, and publicly available information from rare disease organizational websites to help answer research questions. Phase II was included for member checking with Global Genes and data presentation.

At the start of this research, research questions were initially identified, from reviewing available third parties in a U.S. focused search, and Global Genes was one of a few potential organizations identified. They were selected because of their rare disease advocacy work and because they had established training and capacity-building programs for the rare disease communities. Upon building a rapport with Global Genes, and identifying areas of mutual interest, Global Genes was also interested to review findings as it related their own capacity-building for RDAOs with data collection. The rare disease community does not benefit from the support that more commonly recognized diseases have, thus there is less awareness, less advocacy, less capacity, and therefore an unequal distribution of aid and dollars for research, etc. One of the most steadfast values of the RD community and public health practice is the importance of every human being to society, yet more attention and funding is typically reserved for diseases with greater understanding, knowledge, and visibility.

ii. Study Setting

This study was conducted via voluntary telephone interviews, with interviewees spanning across the contiguous U.S. International addresses and contacts were excluded. From a contact list of over 500 names, the list was organized to delete duplicate names and international

addresses. As of May 23, 2017, a recommended list was provided that was reviewed by the advocacy managers. A recommended list comprised of 30 organizations and their executive directors and public websites was available. After volunteer recruitment via a GG webinar with Foundation Alliance members, group emails, and individual follows-ups, the purpose sample was comprised of five members from Low Capacity groups, four from Medium Capacity groups and six from High Capacity groups, who responded to volunteer during the course of the study. See Figure 6, which shows how the purpose sample was determined.

iii. Study Selection

The selection criteria aimed at a purposive sample that would maximize the opportunities for the researcher to learn more about various levels of advocacy capacity. Global Genes advocacy managers selected 30 organizations based on self-defined criteria as existing criteria were not previously determined. The selection process looked at the following characteristics, based on interactions with the sample:

- Age of organization;
- Amount of organizational funding;
- Ability to garner funding in order to advance therapeutic or scientific advancement;
- Number of Staff; and, whether they were,
- Engaged and willing to work with Global Genes or had a positive history of working with advocacy staff.

Data and information were compiled by Global Genes advocacy staff with suggestions per their assessment in lieu of existing criteria to reference.

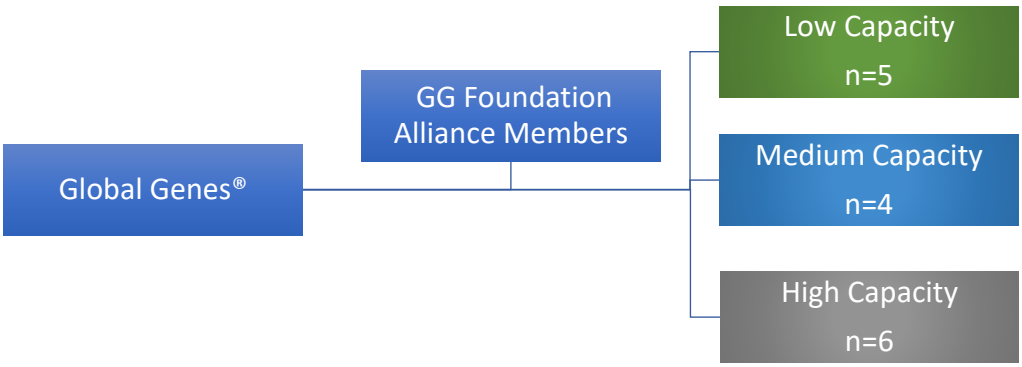


Figure 6: Purposive sampling thru Global Genes Foundation Alliance membership

1. b. Data Sources, Data Collection, and Management

The main sources for this study were collected from semi-structured qualitative interviews, documents, and publicly available information to help answer the research questions. Measurement tables aligned with the conceptual framework were used to aid the data collection processes (Appendix 1).

b. DATA SOURCES AND COLLECTION

i. Document review

There were two phases involved in document review. Phase I included gathering publicly available information before the semi-structured interview to better understand the rare disease and the organization. Phase II included revisiting the publicly available website to confirm information gathered from the semi-structured interviews and fill any gaps from the recordings, such as references to a mission statement available online but not cited during the interview. Other research materials included a literature search and review of websites, and other RDAO related information when publicly available. Data was analyzed for major emergent themes and compared to themes and constructs found in the literature. These reviews provided information that may not have been observable and provided new meaning to develop empirical knowledge (Patton, 2015).

ii. Interviews

There were two phases of interviews conducted for this research. Phase I included semi-structured interviews. Semi-structured interviews allow researchers to gain a deeper understanding of a context (Maxwell 2013). Phase II included a facilitated discussion where the researcher presented de-identified findings to a group of no more than 15 Global Genes advocacy staff members.

1. Interview Selection Criteria

It was anticipated that a total of up to 30 organizations with no more than two organizational representatives would be interviewed, with each interview lasting approximately 60 minutes. An initial list of RDAOs was obtained by reviewing the Global Genes Foundation Alliance Membership list. Global Genes is a global advocacy non-profit organization that supports RD advocacy through its Foundation Alliance Membership. For the purpose of this study, an organization was considered if it met the GG Foundation Alliance Criteria. RDAOs that were engaged at all levels with GG, including newly started 501c3 organizations, and well-established organizations were considered in the purposive sample.

Contact information was obtained from the GG Advocacy Manager with the provided contact name listed as the Foundation Alliance. In the case where multiple individuals were listed, advocacy managers chose names of individuals engaged with GG. The advocacy managers convened to review the list and sort names according to their definitions of low, medium, and high advocacy capacity without prior criteria to reference.

Organizations that were chosen were based the following criteria:

- **High Advocacy Capacity:** Up to 10 disease-specific rare disease organizations that are highly involved with advocacy. High advocacy was defined as organizations who had paid staff, volunteers, funded resources, and over 15 years of age.

- **Medium Advocacy Capacity**: Up to 10 disease-specific rare disease organizations that are somewhat involved with advocacy. Medium advocacy was defined as organizations who had some paid staff, volunteers, minimally funded resources and over 10 years of age.
- **Low Advocacy Capacity:** Up to 10 disease-specific rare disease organizations that are least involved with advocacy. Low advocacy was defined as organizations who had a minimal number of paid staff and volunteers, few funded resources, and not more than 5 years of age.

2. Interview Guide

A semi-structured interview guide was used during the interviews to address the different factors of interest, including facilitators and barriers to conducting rare disease advocacy (Appendix 2). The interview guide reflected the research questions and conceptual framework but allowed for opportunities to include other questions based on input from the interviewee.

The interview guide was pilot tested in February 2017 with a rare disease advocacy executive director who has done rare disease advocacy for over 30 years. The pilot interview allowed for adjustments to address level of understanding, flow of the interview, clarity, anticipated length of time, as well as appropriate probes to address research questions.

3. Interview Procedures

A list of potential rare disease organizations was identified by the advocacy managers at Global Genes. The list included organizations they assessed as low, medium, and high capacity. They were recruited through direct email lists, posts on Facebook, and a request for volunteers during a Foundation Alliance webinar (Appendix 3). Each organization was contacted via email, and follow-up emails were sent at least two to three times. Upon agreement to participate via

email or telephone, informed consent was given orally at the start of the telephone interview, with confirmation of their approval to proceed and understanding of the terms (Appendix 4).

All interviews were completed by telephone. Consent to participate in the 60-minute interview was given over the telephone and agreement was provided that no identifiers would be used in the final report and recorded interviews would be destroyed upon completion of receiving transcriptions. All interviews were recorded from a smart phone via rev.com services and transcribed through the same service. The interviews lasted an average of 60 minutes.

4. Summary

The research protocol aim was to conduct 30-60 semi-structured interviews with 30 organizations (1-2 per organization). Out of the 30 organizations invited to volunteer for the study, a total of 15 agreed to be part of the study. Most of the organizations had one person available who could be interviewed who was working on rare disease advocacy or was the main contact person within the Foundation Alliance of Global Genes. With a lack of already-designated criteria for each organization, advocacy managers developed their best drafts at various levels of organizations deemed, low, medium, or high capacity.

5. Memos

Memos were written after each interview session, as well as after coding the data in ATLAS.ti, to document researcher reflections throughout the data collection process, after reviewing and reading transcriptions and during the analysis process. Memos included contextual insights, observations, potential outliers and notable mentions related to a particular theme or interviewee's comments.

The memos were an integral part of the analysis process, as a point of reflection, and used for comparison or contrast in information throughout the analysis process. Memo notes were typed in a personal computer and used for additional reference and interpretation of interviews.

6. Data Management

A table was developed that listed the various sources of information, de-identified names and organizational information, and included tracked information when the interview was scheduled, recorded and deleted.

De-Identification	Source	Advocacy Capacity	Date of Semi-Structured Interview
A1	Semi-Structured Interview	Medium	8/10/17
A2	Semi-Structured Interview	Medium	10/30/17
A3	Semi-Structured Interview	Medium	9/7/17
A4	Semi-Structured Interview	Low	9/7/17
A5	Semi-Structured Interview	High	9/27/17
A6	Semi-Structured Interview	Low	10/27/17
A7	Semi-Structured Interview	High	9/29/17
A8	Semi-Structured Interview	Low	10/29/17
A9	Semi-Structured Interview	High	8/11/17
A10	Semi-Structured Interview	Medium	9/11/17
A11	Semi-Structured Interview	High	9/5/17
A12	Semi-Structured Interview	Low	9/13/17
A13	Semi-Structured Interview	High	9/15/17
A14	Semi-Structured Interview	Low	8/11/17
A15	Semi-Structured Interview	High	9/15/17

TABLE II. DATA MANAGEMENT OF RDAO SEMI-STRUCTURED INTERVIEWS

Verbatim transcriptions were provided via a paid transcription service called Rev.com. After transcriptions were reviewed and analyzed the files were deleted. Data regarding verbal-informed consent and memos were saved and securely stored electronically on a password-protected computer. The interview transcriptions and memos were uploaded into ATLAS.ti 8.0 for backup storage and analysis.

iii. Data Analysis Plan

The data analysis was completed via numerous steps. The analysis for the qualitative data and the document reviews included reviewing and organizing the data by noting major themes and patterns, developing summaries and descriptions, and creating parent and child codes for the various themes uncovered. After this first phase, the data was developed to show the relationships and outline the results.

1. Document Review Analysis

The initial review of completed transcripts (n=15) was used to manually code, then create a visual diagram of each.

There was minimal physical document review analysis. If information that may have been important for a code was left out of interviews and further context was needed, use of publicly available data on the internet was used and included in documentation. The initial documents were coded using a codebook aligned with the research questions (Appendix 5).

2. Semi-Structured Interviews Analysis

iv. Codebook

The first step was to develop an initial codebook using themes based on the conceptual model and from the literature (Appendix 6). New and emerging codes were added after reviewing the transcripts and getting feedback from a second coder to edit definitions set forth in the codebook. See Appendix for final codebook used in analysis process.

1. Coding protocol

A coding protocol was developed to ensure reliability of the interview analysis. This included a review of the *a priori* codes and working with another coder to review two transcripts for accuracy and clear usage of the codebook definitions. Prior to any interviews that were completed, both coders agreed to code at the full paragraph level (Appendix 2).

2. Inter-Coder Reliability

A master's prepared second coder and researcher with experience in qualitative research was consulted. Both the primary researcher and second coder learned a new version of ATLAS.ti, version 8.0. Both researchers agreed to review two transcripts and codes to manually test the codebook and protocol. The goal was to test the *a priori* codes, determine accuracy of coding, and assess consistency between both coders to determine if changes to the protocol were needed. Feedback from the coder assisted the researcher to further refine and define code definitions and noted examples in the transcripts. Some codes were added as a result of consultation to further determine support for answering the research question. The second coder added comments to the codebook and in the ATLAS.ti 8.0 margins compare coding side-by -side and physically see where discrepancies or other interpretations occurred. Both researcher and coder discussed the proposed edits and they were included as part of the final version of the codebook.

3. Memos

The protocol was to memo during or after a process and note if context was needed for a sentence of a section. Memoing was also used if the coders identified a potential new code (Miles and Huberman, 1994). Upon completion of each semi-structured interview, the researcher

created a memo, along with writing memos while coding and cleaning transcripts. The researcher also created memos at the end of all interviews.

4. Codes and Sub-codes

After the first review cycle of the transcripts, the data was categorized into larger codes, or parent codes, first manually and then using ATLAS.ti. This assisted in identifying relationships, categories, or themes within the rare disease organizations. The types of organizations interviewed were grouped into low, medium, and high capacity categories.

v. Validity and Reliability Considerations

Yin (1989) and Levy (1988) stated that qualitative research should consider construct validity, internal validity and reliability to assess the conclusions being drawn. To mitigate validity, triangulation and member checking was used. Triangulation is important for credibility and is enhanced when multiple sources of information are incorporated. In this case, it included websites, literature, interviewees and document reviews. As Stake and Stringer note, the inclusion of perspectives from diverse sources enables the researcher to clarify meaning by identifying ways the phenomena are being perceived (Stake, 2005) (Stringer, 2014).

Member-checking provides members with opportunities to review raw data, analyses, and reports derived from research procedures and enables them to verify that the research adequately represents their perspectives and experiences (Stringer, 2014). Member checking was part of the focus group work identified as part of Phase II of the research. The principal investigator presented a final report with key findings to a focus group to confirm accuracy and facilitate two-way feedback. The summary of this focus group and key themes are outlined in the Chapter IV.

To ensure reliability a formal study protocol was developed for replication for data collection of the 16 RDAOs (Yin, 1994). All documents and interviews were reviewed a

minimum of two times, while a second coder aided for consistency and design of the protocol and provision of inter-coder reliability.

IV. RESULTS

a. OVERVIEW OF THE CHAPTER

Chapter 4 presents the dissertation study results from a qualitative analysis of semi-structured interviews (n=15) with Rare Disease Advocacy Organizations (RDAOs). This chapter contains an overview of the research questions, description of the analysis process, and the results presented by each research question, including emerging themes.

b. RESEARCH DESIGN: PHASE I: SEMI-STRUCTURED INTERVIEWS

This research was designed with two phases. The first phase included semi-structured interviews with analysis of data and results. Phase II included a facilitated discussion with Global Genes, an organization focused on connecting and empowering the rare disease community, to assess findings, determine accuracy and gain feedback. There were three research questions for this study, divided into one main research question and two sub-research questions.

The main research question was, **"**What *are the organizational capacity factors that influence RD advocacy?*"

There were two sub-research questions in Phase I:

1.) *What are the characteristics of rare disease organizations that undertake advocacy activities?*

2.) How have these characteristics facilitated or acted as barriers to the organization's ability to conduct advocacy on behalf of rare disease patients?

Semi-structured interviews and fact checking data from transcripts was sourced from publicly available RDAO websites. Overarching themes that evolved from the research data provided insight into how RDAOs build organizational and advocacy capacity, including how their advocacy approaches were best leveraged to build their RD community, or address facilitators and barriers to their work.

i. Phase I: Semi-Structured Interviews

Semi-structured interview transcripts were analyzed using the steps presented in Figure 7. Content and thematic analyses were based on findings using ATLAS ti.8.0 software. Throughout the analysis phase, a few codes emerged and after discussion and feedback from other researchers, emerging codes were added. An updated codebook was finalized to code findings using ATLAS ti.8.0 software. See Appendix 4.

1. Analysis of Data and Determining of Coding Scheme: Code Development

A physical cut and sorting of responses was used upon the completion of each semi-structured interview. This involved using large poster boards with color coded notes arranged by each question and the responses. This visual exercise provided the opportunity for pawing (HR., 2019), an ocular scan or eyeballing for patterns and themes based on clusters of colored notes and frequency of responses. This process allowed for researchers to identify where responses were not provided, less developed, and to observe where colored patterns of notes had the greatest frequency of responses per interview and compare across all interviews. This process aided in the creation of assigned codes that described the cluster of responses. After all codes were laid out on a poster board by various colors, groupings of responses were created that resulted in 11 “child” codes termed: age of organization, role/title of organization, genesis, organizational infrastructure, prioritization process, engagement and outreach, mentoring, networking, partnerships, barriers and facilitators. Each child code was then grouped under a broader descriptive header attached to four parent codes termed: organizational capacity, connecting with constituents and stakeholders, gaining value from other people, and identifying and addressing barriers. Together these coding strategies was used to describe data and seek relationships. It allowed for organizing the data in superordinate and subordinate concepts

(Qualitative Data, Analysis and Design) to display the relationships. This worked served for the basis of code book development and the final code book. See Appendix 4.

Coding Process using ATLAS ti. 8.0

After the code book was initially developed, data was then downloaded into ATLAS ti. 8.0 for coding using this software program. All 15 transcripts were prepared by reviewing text, correcting errors in transcription and removing any identifying information. Coding was then completed in Atlas ti 8.0 using the code book.

Coder Reliability

An experienced qualitative researcher with a background in coding and using Atlas ti. 8.0 was used to review drafts of codes, transcript preparation, code definitions, overall codebook, protocol and to test transcripts before PI began coding in software program. The researchers selected two interview transcripts to manually test the codebook and protocol. The purpose was to test the *a priori* codes, determine what was pertinent to keep for research, assess the consistency between both coders and determine if documents needed to be updated. Refinements were addressed in the software program as it had the ability for both researchers to code simultaneously and track changes or comments. This resulted in some enhancements to the process such as ensuring that the software was coding a contiguous section of a paragraph to gain all the sentences related to the topic. The code list was revised, and definitions were updated based on coder feedback and experience coding documents. This provided greater clarity for definitions and discussions on each code for feasibility of use. An emerging code was identified and added through this process. That code was called *genesis,* which referred to the reason why the organization started as an RDAO and how it began.

2. Memo Development and Use of Memos

The process of hand-writing memos was conducted throughout the analysis phase to identify new learnings, divergence, patterned responses, concepts, ideas, or synergies. Memos were taken after each interview was completed, during the coding process, reviewed with the second coder, and throughout the analysis phase, informed codes, identification of themes, and organization of results.

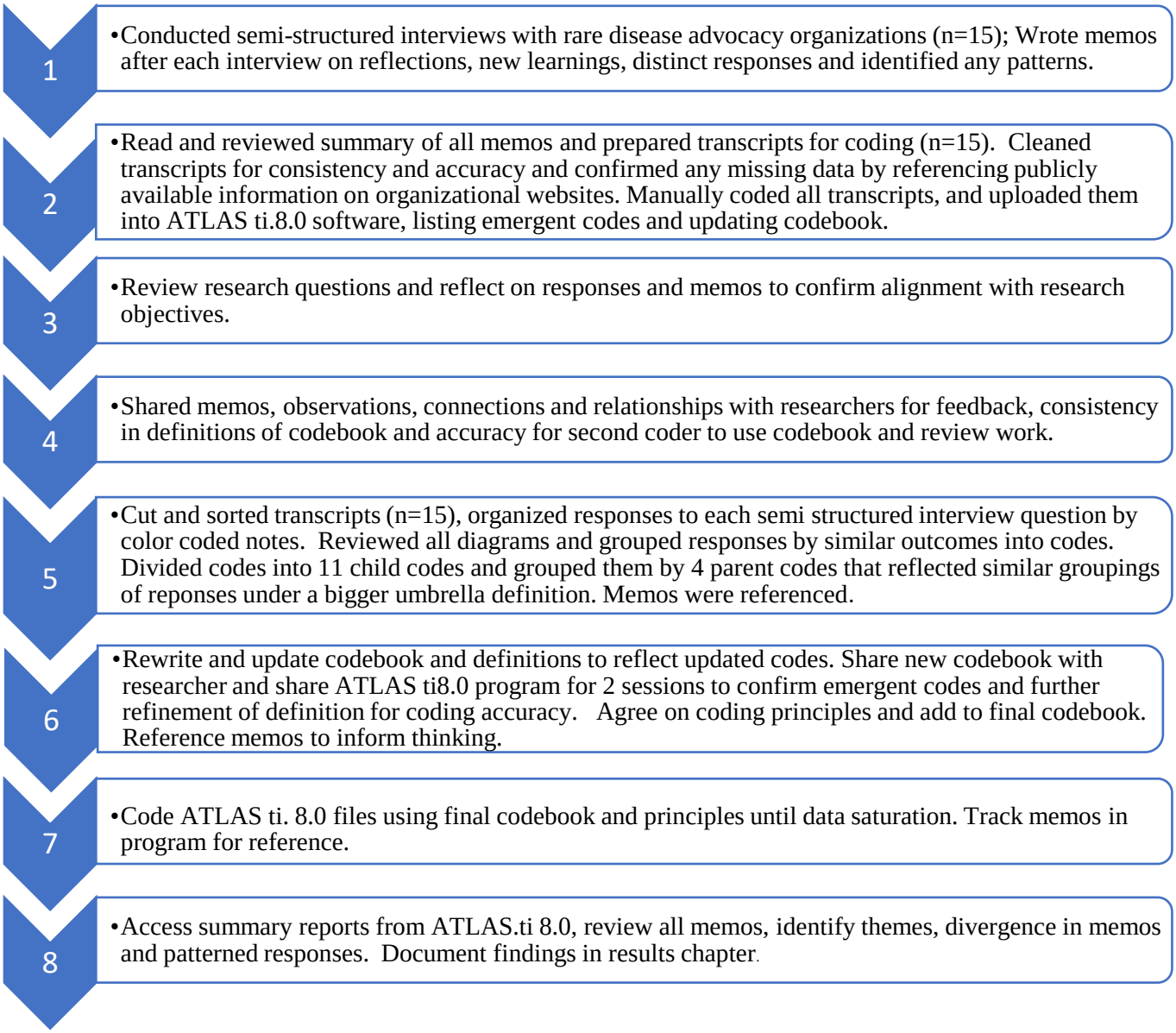
Themes were determined by highest frequency of data within a code that had similarities or patterned responses within the same cluster of data across multiple RDAOs. Coding was stopped upon reaching saturation (Qualitative Data, Analysis and Design). Saturation occurred where new data and their sorting started to confirm categories, themes, and conclusions already reached.

ii. Final Determination of Results

In order to answer the research questions, results are presented in order of density or relative to higher number of codes vs. the codes with lower numbers. Results from lower number of codes were reported due to the fact that RD is still a developing area of research. Data about RDAOs and their organizational capacity can help inform this community and contribute to this area of advocacy research.

Outliers were included in memos for consideration and some were mentioned in the results section as noteworthy to include in the final write up.

Figure 7. Phase I - Semi-structured interview analysis steps



c. RESEARCH DESIGN: PHASE II: FACILITATED SESSION WITH GLOBAL GENES

This research was designed with two phases. The objective for Phase II was to report results back to Global Genes (GG) to confirm that findings aligned with work that is happening in the field of RD advocacy, identify any gaps, and determine if the data was useful to inform their strategic planning. Data for Phase II included the summary of results related to each research question from Phase I and key questions identified by GG. See Figure 8.

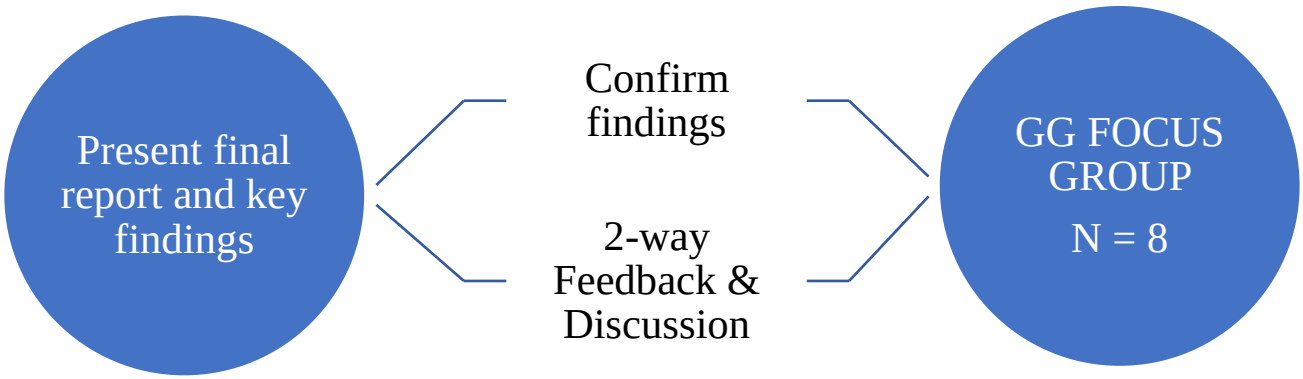


Figure 8. Phase II facilitated discussion with global genes patient engagement team summary

i. Facilitated Discussion with Global Genes

The results of the study were presented to GG via remote access from personal computer to address the findings related to the research questions. Overall, GG feedback agreed the findings were aligned with what they are seeing in the field of RD advocacy. They verbally noted that all participants were nodding their heads in agreement throughout the presentation and had no additional changes or feedback. Eight patient engagement managers attended the presentation, and all agreed the findings helped them validate their past and existing RD advocacy work and appreciated seeing it laid out in the results section. They stated they will reference the results of this study to inform their current strategic planning as it relates to key

trainings, addressing RDAOs earlier in the process, and being prepared ahead of time to recognize barriers to RD advocacy, especially for onboarding new members. The findings related to the virtual organizations provided additional thinking for GG regarding best practices in delivering information and considering webinars to improve accessibility of information beyond in person meetings. Also, GG provided feedback that it was a good reminder knowing RDAO advocacy managers hold multiple roles and responsibilities including parenting and caring for an RD child or family member while leading their own RDAO. One of the patient engagement managers also noted they agreed with the Phase I results that RDs are an integral component of public health and recognized that RDs help inform a broader community about common public health needs such as access to care, health education, and/or support for other RD patients.

A review of the organizational capacity criteria compared with the actual results was also of interest for GG. There was a bit of surprise in that despite some were younger in age, they may have had a higher amount of services or fundraising compared to the higher capacity organizations. The comparison of these discrepancies particularly where there were financial numbers involved was of interest. For example, one organization was high capacity, yet had only 3 people on staff but conversely raised over $300M in RD research. Another organization was less than 1 year old, with no staff, yet had a clinical trial in the works for >1M. Another organization was identified as low capacity but had a viral fundraising program and raised $2 million dollars but also had volunteer staff only.

ii. Characteristics of Study Sample

Global Genes is a global RDAO, based in Aliso Viejo, CA, and was one of four rare disease umbrella organizations contacted to collaborate for this study. After reviewing and contacting other rare disease advocacy groups, GG agreed to provide minor guidance for this

study, acknowledging they are connected to many of the global and U.S. based RDAOs via their RARE Foundation Alliance membership. The RARE Foundation Alliance is a program within GG that is focused on working with RDAOs who become members of their global RD advocacy network. Other RD umbrella organizations fell outside the scope of this research and/or were reluctant to partner due to competing priorities, or lack of time or human resources.

Purposive Sample Using Organizational Capacity Criteria

For the purpose of this study, GG created organizational and advocacy capacity levels designated as low, medium and high capacity with broad based criterion based on 30 RDAOs that were currently part of their network. A purposive sample (n=30), with a goal of 10 from each ranking, was chosen from an identified pool of active Global Genes RARE Foundation Alliance members. GG patient engagement managers helped identify and stratify the purposive sample as potential RDAOs who would be willing to participate in research. The RARE Foundation Alliance is a coalition of over 500 RDAOs within the GG network. The goal of the alliance is to exchange best practices, share lessons learned and drive better outcomes for the entire rare disease community (Global Genes RARE Foundation Alliance , n.d.).

Description of Newly Created Organizational Capacity Criteria and Characteristics of Study Sample

The organizational capacity criteria were based on feedback and definitions created by GG patient engagement managers whereas low capacity organizations (n=5) were described as being a newly formed 501c3 nonprofit organization less than 5 years old, that provided a limited scope of services, obtained or held limited funding resources and likely had limited to zero paid staff. High capacity organizations (n=6) were described as a 501c3 organizations, older than 10 years of age, providing a higher scope of services, held higher funding resources and had paid staff comparative to the other low and medium capacity organizations. Medium capacity

organizations (n=3) were described as somewhere in the middle of both low and high, with some staff who were salaried, some scope of services, and over 5 years of age.

Description of Study Sample

Global Genes provided a pre-screened list of RARE Foundation Alliance Members, along with their self-defined organizational and advocacy capacity criteria created for the purpose of this study. This comprised of advocacy capacity levels for each RDAO, of low (n=10), medium (n=10) and high (n=10) capacity organizations. Role and titles for all contacts were listed as Executive Director (n=30), including publicly available websites (n=30) and email addresses of key contacts (n=30). See Table III.

Despite the use of organizational capacity criteria at the start of the study, it turned out to be unrealistic and did not apply to this study. The criteria served as an unhelpful distinction of various RDAOs whereas similarities existed for most of the organizations in terms of type and number of staff, having advisory boards, and the amount and type of services, which was not clearly distinguishable by capacity level. Thus, results are not presented using the criteria.

At the end of the study, organizations that participated were spread across the U.S. with 6 in the low capacity category, 4 medium capacity, and 5 high capacity organizations. The ages of the 15 organizations ranged from being less than 1 year old to over 65 years old. See Table III for overall summary of description of the study sample.

TABLE III: CHARACTERISTICS OF STUDY SAMPLE

	CRITERIA	RELATION TO RD IND.	RD AFFECTS	TITLE	ROLE	TYPE OF STAFF	TYPE OF BOARD (S)	OFFICE TYPE	AGE OF ORG.
1	LOW	Father/ Mother	Child	President, Founder	Run the business operation of it; fundraising, wife is in a volunteer capacity works w/ industry & engages with scientists	Volunteer	SAB	Virtual and home office	2017
2	LOW	Father, Mother	Child	President, Executive Director	Co-Founder and Executive Director	Husband and wife do 90% of work, 3 other people help on occasion;100% volunteer	N/A	Home Office	2013
3	LOW	Mother	Child	President Founder,	Run day to day	All volunteers; no paid staff	SAB	Home Office	2017
4	LOW	Employee	Child	Executive Director	Run day to day	Volunteer	BOD MAB SAB	Virtual	2017
5	LOW	Grandma	Child	President, Founder	Run day to day	0 staff, all family-husband,2 sons, and 1 daughter in law	BOD	Home Office	2015
6	LOW	Self	Child	On Board of Directors	Writer & social media coordinator	1 staff is PT virtual assistant	SAB	Virtual	2017
7	MED	Mother	Child	Co-Founder, Exec Director	Run day to day	Volunteer	None	Home Office	2011
8	MED	Father	Child	Co-Founder and President	Run day to day	5 board members; no staff; mainly volunteers	SAB	Virtual	2009
9	MED	Employee	Child	Development Manager	In charge of all fundraising	6 FT; 5 PT; volunteer Scientific Director	N/A	Office HQ	1995

10	HI	Mother	Child	Founder, CEO	N.A	11 paid staff	SAB	HQ	2002
	HI	Self	Child	N/A	N/A		BOD SAB		1995
11	HI	Employee	Child	Retired President and CEO	Retired	600 paid staff 74 Chapters	BOD MAB SAB Audit committee Compensation Committee	HQ	1955
12	HI	Employee	Child	VP Policy and Outreach	Started out PT, then FT > 4 yrs.	12-15 paid staff	Board of Directors	HQ and Wash DC Office	2006
13	HI	Employee	Child	Dir Program & Services	In charge of programs and advocacy training and services		MAB SAB Board of Trustees Advocacy Comm. Finance Comm	HQ 9 regional offices	1986
14	HI	Mother	Child	Executive Director	Run day to day	3 staff; volunteer	Board of Directors	Office HQ	1988

iii. Member Criteria of Rare Alliance Foundation Members

All of the selected RDAO organizations met GG eligibility criteria to receive member benefits. RARE Foundation Alliance members are eligible if they are a support group, a U.S. foundation with 501c3 status, can provide access to information via a website or active Facebook page, and provide RD information or support in one or more of the following areas (Global Genes RARE Foundation Alliance , n.d.):

- Patient/caregiver support, healthcare provider education, public awareness and advocacy and/or research.
- Website or Facebook pages that focus on a specific patient or family's journey are not eligible for member benefits.
- Exclusive access for networking and sharing includes quarterly Foundation Alliance webinars to share rare industry updates, initiatives, resources and expert speakers, private Facebook for foundation leadership discussions, members only website, technology support for one patient/family-focused educational content webinar, and discount to their RARE Patient Advocacy Summit.
- Members have the exclusive opportunity to participate in the RARE Patient Grant program, gain a promotional opportunity for the organization's foundation, and one-on-one GG support that include contact with the GG advocacy team to help answer questions and connect rare advocates to partner resources (Global Genes RARE Foundation Alliance , n.d.).

iv. Recruitment of Volunteers to Participate in Dissertation Research

Recruitment efforts began with a webinar presentation by the PI to RARE Foundation Alliance participants, including a follow-up direct email from GG patient engagement managers as well as the PI, seeking volunteers for the study. See Appendices 5-7. Thirty organizations agreed to participate and 15 completed participation in this dissertation study. See Figure 9, which provides the Map of Main Office Locations designating different capacity levels of the RDAOs in this study. Geographically, the 15 organizations and various advocacy capacity levels were spread throughout the United States.

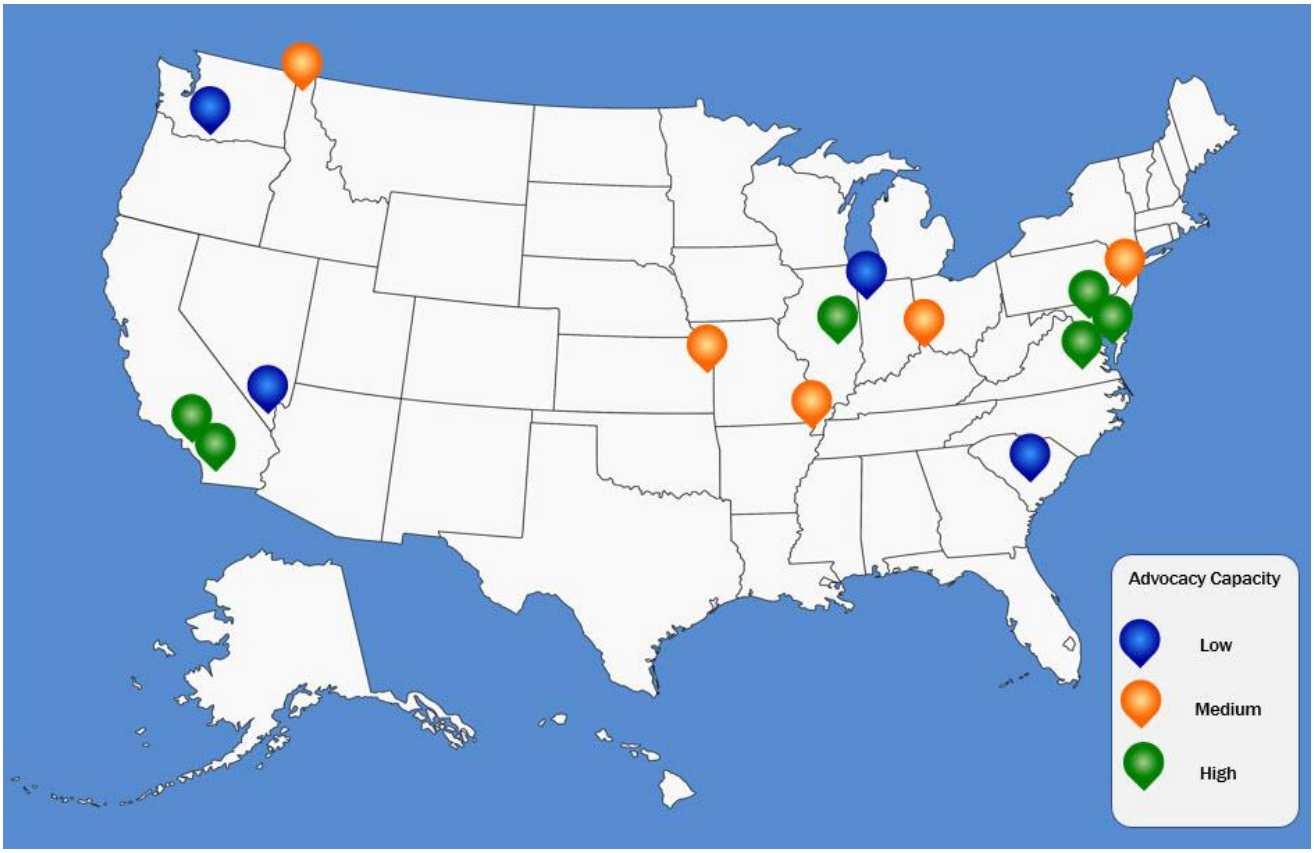


Figure 9. Map of main office locations designating different capacity levels of the RDAOs

d. SUMMARY OF PHASE I FINDINGS

Tables IV-VI provide the summary of results outlined in this chapter. Codes from Phase I of research were used to answer the research questions and identify emerging themes for each question.

Research Question #1:
What are the organizational capacity factors that influence RD advocacy?

Factors	Sub-Factors	# of Codes	Theme(s)
1. Organizational Infrastructure		140	1. Most RD advocates were related to a person or child with the RD
	Age of Organization	(140)	1. Age or organization is based on gaining 501c3 status, but advocacy work may have happened before nonprofit status.
	Type & Number of Staff	(140)	1. RDAOs less than 15 years old operate with lean staff, personnel and rely on volunteers, family for supporting the organization.
	Advisory Boards	(140)	1. Most RDAOs had a Board of Directors, Scientific Advisory Board or Medical Board
	Type of Work Office	(140)	1. Most work out of home office and employees work in a RDAO headquarters office.
2. Funding		108	1. The role and skill for fundraising is challenging yet fundamental to RDAOs 2. The immediate need for RDAOs to fund research.
3. Genesis 4. Role/Title 5. Prioritization Process (Low Number of Codes)		74 40 40	1. RDAOs started as not much was available to support RD. 2. RDAO are led by advocates who have had multiple titles and roles. 3. Many of the RDAOs described a structured process but didn't match current work.

TABLE IV: SUMMARY OF ORGANIZATIONAL CAPACITY FACTORS AND THEMES

Sub Research Question #1:

What are the characteristics of rare disease organizations that undertake advocacy activities?

Characteristics	Sub-Characteristics	# of Code-	Theme(s)	Sub-Theme(s)
1. Engagement & Outreach		210	1. Social media is a way for RDAOs to reach their RD community and others. 2. Software tools support RD advocacy. 3. RD Advocacy has a spectrum of audiences.	1. Facebook is consistently used by RDAOs.
2. Gaining Value from Other People (Low Number of Codes)			1. As RDs lack resources, the value of people as assets to advocacy is important to recognize.	N/A
	Networking	86	1. Networking is an important tool for RDAOs because people can serve as important resources of information, especially when resources are scarce.	**N/A**
	3rd Party Technical	75	1. Technical resources can provide expertise in RD advocacy work related to legislation, policy, and research.	**N/A**
	Partnering	70	1. Identifying partners with shared similar values could help advance RD advocacy work.	**N/A**
	Mentoring	22	1. Having a mentor to support RD advocacy can help provide wisdom, education and prevent mistakes.	N/A

TABLE V: SUMMARY OF CHARACTERISTICS OF RDAOS

Sub Research Question #2:

How have these factors facilitated or presented as barriers to the organization's ability to conduct advocacy on behalf of rare disease patients?

	Sub-Characteristics	# of Codes	Theme(s)
1. Barriers		139	
	Barriers to organizational capacity	(139)	1. Fundraising challenges 2. Lack of prioritization process 3. Need for skilled staff, and true RD advocates
	Barriers to advocacy capacity	(139)	1. Diagnosed with RD 2. Unknown or Lack of RD disease specific information 3. Finding/Identifying greater numbers of RD patients and importance of building a RD community 4. Lack of parent empowerment
2. Facilitators		146	1. Starting a 501c3 for RD advocacy 2. When faced with adversity, RD advocate is driven by personal determination and will to fight 3. Using transferable skills from professional experience and putting it to use to advance RD advocacy

TABLE VI: SUMMARY OF ORGANIZATIONAL FACILITATORS AND BARRIERS TO RD ADVOCACY

i. Research Question 1: What are the organizational capacity factors that influence RD advocacy?

Definition of Organizational Capacity

In order to answer this question, data was organized by the parent code *Organizational Capacity*. In this study, *organizational capacity* was defined as describing the activities designed to improve and enhance the way a nonprofit can achieve its mission and sustain itself over time (Profits, 2018).The factors for this parent code were based on 5 child codes named organizational infrastructure, age of organization, funding, genesis, prioritization process and role/title of interviewee. The results in this section are presented in descending order related to the total number of codes per each child code. Definitions for each code are included in each of their results sections.

Two emerging themes developed in the code entitled *funding* and are described. The eight factors described answer the question: *which factors influence RD advocacy capacity?* When an RDAO can maximize its organizational capacity, advocacy capacity is positively impacted. For example, addressing organizational capacity allows for the organization to improve or enhance its advocacy efforts, funding capacity, and staff in order to have a stronger advocacy presence with a more organized way of conducting advocacy work.

ii. Factors and sub-factors that Influence RD Advocacy Factors and sub-factors that Influence RD Advocacy

Five factors and four sub-factors were found to influence RD advocacy. The five factors are: organizational infrastructure, funding, genesis, role/title of RD advocate, and prioritization process. The four sub-factors related to organizational infrastructure are age of organization, type and number of staff, advisory boards, and type of work office.

Among the organizational capacity factors studied, *organizational infrastructure* had 140 codes (See Table VII), followed by the coded section called *funding,* which had 108 codes. See Table IV. A grouping of factors that had a lesser number of codes were grouped together, which were *prioritization process*, and *role/title*. An emerging code was identified with the second coder and added, which was *genesis*. Together, all of the codes represented factors that helped build advocacy capacity for the RDAO interviewed in this study.

RD Organization	Organizational Infrastructure Code
1	10
2	18
3	7
4	8
5	11
6	15
7	9
8	10
9	6
10	5
11	11
12	9
13	9
14	9
15	3
Total = 15	Total = 140

TABLE VII: TOTAL NUMBER OF ORGANIZATIONAL RESPONSES AND NUMBER OF CODES FOR ORGANIZATIONAL INFRASTRUCTURE

1. Factor 1 - Organizational Infrastructure, Including Themes and Sub-Factors

This data helps answer how organizational infrastructure is an important factor to influencing RD advocacy. For the purposes of this study, *organizational infrastructure* was defined as describing the type of staff and number of staff within an RDAO, inclusion of a scientific or medical advisory board, funding, and description of type of work office. Factors were identified as they aligned with child codes hierarchically related to *organizational capacity* as a code. Each

code in this group was summarized, described and supported by data as factors related to advocacy capacity building.

All 15 RDAOs were RARE Foundation Alliance members at the time of study, assigned with advocacy capacity criteria. These included having a scientific or medical advisory board, 501c3 status and conducting RD advocacy-related work as an organization. The results of this section are best summarized in Table X. The results are organized, and color coded by original organizational and advocacy capacity rank assigned by GG. One theme emerged in this section related to the relationship of the RD advocate to the person or child with the RD.

Organizational Infrastructure Theme 1 – Most RD advocates were related to the person or child with the RD

Most RDs are recognized in childhood, and this research showed that most of the interviewees of this study were related to an RD patient either by being a direct parent, spouse, or immediate family member. Nine out of 15 organizations were directly affected by the RD. Eight respondents were parents. One was a grandmother. Two had the disease themselves and one of these two had the disease and was also a parent. Four respondents were employed by an RDAO.

A few of the organizations described their connection to the RD as parents and said the following, “So back in 2009, first my son and then a few months later my daughter was diagnosed with RD disease, which is a childhood neurodegenerative disease that is progressive and universally fatal.”

Two organizations described how RDs affected them directly.

- “My son was diagnosed with RD in late 2002.”

- "Our daughter, in July of 2013, was diagnosed with RD syndrome at the age of three and a half."

Another organization conveys how their son was the first diagnosis in the world with a certain RD.

- "Sure, so it was founded and started because nothing existed. My son was the first kiddo diagnosed in the world after the disease was discovered."

This data reflects how the RD advocates in most cases may be directly related to the person with the RD and the inherent motivate on of parents or family members to help find a treatment or a cure for their loved ones. Of all the RDAOs interviewed, each was passionate about their job and clearly understood their advocacy role on behalf of the RD patient.

Sub-Factor 1 – Organizational Capacity - Age of Organization

Age of the organization since it started was defined as the year or reference in time when the organization started as a 501c3 nonprofit organization. Results for age of organization when it started is summarized in Table III. All 15 organizations provided the year from which their 501c3 started. Among the ages of the 15 respondents, RDAOs in this study ranged from less than 1 year old to over 64 years in age. This was a key criterion for designation in low, medium, or high capacity levels. The low capacity organizations (n=6) ranged from <1 – 4 years old. Medium capacity organizations (n=3) were >5-6 years old. Organization #9 was designated as medium capacity at the beginning of the study, despite the age from the respondent, of being created in 1995. By definition of criteria, this would have been designated as a high capacity organization.

Sub-Factor 2 - Organizational Infrastructure - Type and Number of Staff

A majority of organizations had a small amount of staff ranging from 2-15 people, regardless of assigned capacity level, with one organization that had over 200 employees. This factor was included as part of the definition for organizational infrastructure. Mentions that described the type of staff or volunteers or if they were paid or not were captured. In general, a majority of the RDAOs had a low number of staff for the amount of work they need to accomplish. Some also depend on themselves to do a lot of the work or may lean on families or volunteers.

According to one of the organizations, she confirmed they have no paid staff and rely on volunteers, saying, “Nope, we're completely 100% a volunteer organization.”

Similarly, another RDAO described how their staff are all volunteer based as well and said, “Right now it's all volunteer-based positions, just due to the lack of financial resources that we have. And it's this repeating cycle of, we want to bring people on full-time so that they can focus on the organization, but to do that, we need money.”

Another organization leans on their board members for support and has no staff as well. Its representative said, “So, we have five board members and no real staff.”

Leaning on themselves and family for support, one organization described their staff by saying, “So, we've got myself and my wife are board members, and then her parents are board members, and her grandfather is our treasurer. Typically, it's my wife and the other mom, my wife’s mom, so they're the ones that typically put that together. And we have some volunteers that help with that too, but we develop all of our own communication.”

The low capacity organizations that were just getting started did a majority of the work themselves with help from family. Other staff that were described as unpaid staff, or served as volunteers. See Table III for detail. The medium capacity organizations were similar to low capacity organizations with mainly no staff and volunteers only. There were only three organizations in this category. High capacity organizations had a different landscape of staff. Three out of the four high capacity organizations had over 10+ paid staff, while one had 600+ paid staff with over 74 chapters of their RDAO.

Sub-Factor 3 - Organizational Infrastructure – Advisory Boards

All 15 of the RDAOs mentioned having a scientific or medical advisory board. This was also included as part of the definition of *organizational infrastructure*. Having an advisory board within the RDAO aligns with GG Foundation Alliance Membership criteria. Eight out of the 15 RDAOs respondents confirmed having a Scientific Advisory Board while two of the organizations had both a medical and scientific advisory board panel. The responses for this section reflect the importance of boards for RDAOs and their contribution to helping with advocacy efforts. The types of boards mentioned are the RDAO Board of Directors, Scientific Advisory Board and/or Medical Advisory Boards that collaborate with the RDAO.

Two of the organizations described their boards as comprised of actual RD patients. One organization said the common thread to his board is all are patients, and said, “Some of them are patient advocates. They are sufferers themselves. We have one person on our board currently who is a patient but was also a nurse when she was working. She’s currently medically retired. But yeah, we do have various backgrounds. Our founder has an IT background. Like I said, we have our board members. But the common thread on all is they are all patients.”

Another organization described a similar situation, where everyone on their board has the same RD and said, “In terms of unpaid, pretty much everybody on the Board either has RD or is a family member of somebody with RD so we all do whatever our skillset is.”

Parents are an important foundation to RDAOs as well, some also serve on boards. As one organization described it, they said, “I’ve got some moms who are on my board, but they’re just to be board members.”

Another important role these boards serve is to help advise or guide the RDAO related to research, organizational priorities and by providing individual and unique skillsets.

As one organization put it, their medical advisory board provides troubleshooting support and guidance on research. Its representative said, “Internally, we have our medical advisory board that has various backgrounds and knowledge of RD. And our medical advisory board we lean on a lot to understand the effects and laws in the medical community. They can help us identify where we’re going to potentially run into issues, and how do we get through different barriers, and they also help us in our research standpoint.”

Another organization had a similar description on how the board helps with the strategic plan but also has a separate committee to help determine solutions for education and planning. They said, “The Board determines the strategic plan and then there’s actually a committee. It’s a mouthful. It’s the Education Advocacy Awareness and Membership Committee that then figures out the details of which venues and which topics we’re going to be educating on.”

Two other organizations described their working board working on similar guidance and how each board member brings different skillsets for the RDAO to benefit from. Their members said: “So, with our Board, each year we sit down and define our strategic priorities and then evaluate what belongs in our strategic priorities, what do we have the resources to

accomplish. Priority setting with the leadership and the Board." And, "Our board of directors is a working board, so we've got somebody that does marketing, and somebody that does this and that and all different skillsets."

Outside of these board descriptions, another RDAO shared the importance of non-U.S. board members. Their board has a non-US member and they are actively looking for other international board members. The respondent said, "We do have one board member in the U.K. The rest of us are here in the United States. Our Scientific Advisory Board is all located in the United States at the moment but we're actively recruiting for somebody outside of the U.S."

Sub-Factor 4 - Organizational Infrastructure - Type of Work Office

Nine out of the 15 organizations held a home office without a physical office space outside their home or conducted their work remotely with volunteers within their membership. This factor was included as part of the definition of organizational infrastructure to better describe how RDAOs' physical office structure exists. Three of the four high-capacity level organizations had a physical office headquarters location. One medium-capacity level organization had a headquarters location in Washington, D.C. where most of their policy work was conducted. They said, *"*The Foundation is located in Pasadena, California. I work out of Arlington, Virginia. As I do policy, I go to Washington D.C."

The remainder of the medium and low-capacity organizations mentioned working from home, remote office location, or doing their work remotely. Some of the description of the home offices are as follows. One of the parents said,

> Yep, it's in New York. I was working out of libraries and also just, yeah, it's at home. We are at home, and I started ... When I went back to work, we gave ourselves a year, because we

figured that we could live maybe for a year on one income, we thought we'll take a year and get this foundation running, functioning, and research supported, and I'll get back to work.

Another explained how their organization has grown from a home-based office in the kitchen to the greater organization they are today, saying,

> The organization had grown really from a small kitchen table group of individuals in a support group like atmosphere to a national organization that has over 54 chapters and affiliates. 43 medical centers that have received a designation of centers of excellence. We have more than 160 support groups. As well as funding research program.

Another organization described their main office as located in their house:

> Well, our main office is right here in our house right now. Right now, yeah. I am the only full-time employee at this time, and that's just as of October of 2016. Prior to that, just the three and a half years prior to that, I was also volunteer and our whole organization was volunteer.

Conducting advocacy work remotely or virtually seems to be more common. A one organization explained, “That's the interesting part. I call ourselves a virtual organization, in that we are registered in Idaho, which is where our founder is located.” Despite where their address is for their 501c3 nonprofit organization, much of the work is done virtually or not physically completed in a single office. Many of the volunteers or staff do their work from their home office and advance it using today’s technology and virtual resources.

The type of work office is an important factor to better understanding how RDAOs get their work done and in what capacity. With a majority of the RDAOs working in a home office, this information also reflects a parent is often the CEO of a nonprofit RDAO, and they often have

few staff and low operating costs. Compared to the higher-level capacity organizations, the older an organization is, the more organizational capacity they have developed, and their advocacy capacity is enhanced as well with having a headquarters office, greater numbers of staff, and support to execute advocacy-related work.

2. Factor 2 – Organizational Capacity - Funding

Funding allows an organization the ability to advance advocacy work in all areas. It is the basis of having the means to get behind work and execute key agendas on behalf of the RD patient. The code *funding* was defined as any mention related to financial needs for RD advocacy. This definition was designed to gain a basic understanding about funding needs, including how finances are advocated for and allocated. Funding is a major component to building organizational capacity and the foundation to the longevity of any nonprofit organization. This section had a total of 108 codes. See Table VIII for more detail about the *funding* code.

RD Organization	Funding
1	19
2	7
3	7
4	4
5	12
6	1
7	8
8	5
9	3
10	4
11	7
12	12
13	11
14	4
15	4
Total = 15	Total = 108

TABLE VIII: TOTAL NUMBER OF ORGANIZATIONAL RESPONSES AND NUMBER OF CODES FOR FUNDING

The Role of Funding in Nonprofit Organizations

Funding in this context refers to the act of fundraising as well as recognizing which key areas require financing to support advocacy initiatives. Two themes were identified that describe priorities related to funding for RD Advocacy. They are detailed below in Table IX.

Factors that Influence Advocacy Capacity Themes	# of RDAO Reporting	Frequency	Data Source
Funding Advocacy Theme 1: The Role and Skill for Fundraising is challenging yet fundamental to RDAOs	15	28	Stakeholder Interviews
Funding Advocacy Theme 2: The Immediate Need for RDAOs to Fund Research	15	21	Stakeholder Interviews

TABLE IX: FUNDING THEMES RELATED TO FACTORS THAT INFLUENCE ADVOCACY

Funding Theme 1: The Role and Skill for Fundraising is challenging yet fundamental to RDAOs

All organizations that were interviewed held a 501c3 nonprofit status. As organizations responded to interview questions, the importance of fundraising was a recurring theme that came up with a frequency of 28 times. The need for fundraising for these RDAOs was mentioned most amongst factors that influenced organizational capacity. Financing work to fund important initiatives allowed the RD advocates to continue searching for more information, therapy, or a cure. The structure of running a nonprofit relies on fundraising. Raising funds is fundamental to all their nonprofit business models in order to survive and operate.

One RDAO mentioned that, “All non-profits are in constant fundraising mode. It's the nature of the beast.” RDAOs see fundraising as an important organizational skill in order to elicit action for advocacy. Turning funds into additional opportunities keeps their journey going.

This concept is also well-described by another organization's advocate who shared her feelings about finances and advocacy. She said,

> My feeling is that if the nonprofit is stronger, then our advocacy will be stronger. By the same token, the fundraising, the ... frankly, the more money that we are ... the better we do with fundraising, then the better we're able to carry out those advocacy activities, the better staff we're able to hire with more ... better skill sets, and all of those things.

Adequate funding provides adequate capacity to accomplish more goals in an improved way versus the actions of minimally funded nonprofits, whose leaders must be constantly focused on fundraising.

Fundraising is often required by RDAOs to address the high cost of orphan drugs and clinical trials due to the low number of available patients and specifically those patients who can be treated with a new therapy or a drug. One organization outlined just how much research trials can cost, and their representative said:

> We created our 501c3 and just started fundraising. The lemonade stands, the galas, all of that stuff. We raised about $250,000 in about six months, which was good but wasn't, obviously as you know in rare disease research, that doesn't move the needle very far. We were fortunate in mid-2014. We had a video made, a pro bono video made, that went viral and raised about $2 million in eight months.

However, a couple of organizations' advocates provided cautionary tales about what happens when RDAOs obtain funds. They conveyed that it has to be used wisely so as not to be wasted, and that the act of fundraising needs to translate into action for the RDAO. Another organization discussed the need for funding and the need to spend those funds to build community. She

explained, “You're gonna run out of money if you don't keep fundraising, and there's always community to build, and you can also squander that.”

Another organization shares the importance of action hand-in-hand with fundraising, whose advocate said,

> For me, it's ... again, I'm in fundraising. It's always going to come down to money. Did we raise more than we did last year? But that ultimately doesn't mean anything unless it turns into action. Advocacy and awareness are the same way for me. This is just me personally, not the foundation. I think we can have all the awareness in the world and all the advocacy in the world, but if it doesn't translate into results for patients, then it doesn't mean anything.

The act of fundraising however, is not a simple task nor a skill that everyone has in today’s world. It entails being disappointed, let down, or shut down by those who may not want to donate or give funds. One organization’s advocate mentioned how fundraising can be a challenging job. He said,

> Fundraising, which is a vast majority of our employees, is a hard, hard business. The young kids today are not used to having people say no to them and when you're calling raising money and people say, ‘No, I can't do that. I don't want to do it.’ Hang up the phone, all of that. They're not used to that.

Another organization’s advocate described the challenge of fundraising, despite quitting his job to work on fundraising full-time, saying, “I started a 501c3, I quit my job and started a 501(c)(3). We decided to go out, and we did a lot of fundraising, and of course that still remains one of the biggest challenges.”

In fact, fundraising also came up as an important skill to build for RD advocates. The same organization mentioned their trainings and ongoing skill-building activities. One organization talked about how they encouraged their staff to learn more and learn, "Fundraising 101, RD Foundation 101, all those kinds of things that you would expect. We encourage people to get advanced certification in fundraising, etc."

Funding Theme 2: The Immediate Need for RDAOS to Fund Research

Much of the literature on RDs discusses ways that RDAOs engage and have been directing RD-related research. Surprisingly, this factor didn't produce the greatest amount of codes. Based on the literature, funding or being directly involved in RD research is what is mostly described in the literature for RDAOs. Every RDAO in this study described the importance of funding for RDs and advocating for funding. Future research proves to be beneficial especially where research is scarce.

When RDAOs begin to learn more about their RD, often the first topic they reference is existing or recognized, published research. Most if not all RDs typically find the research is lacking, and it quickly becomes a priority to fund research in order to gain greater knowledge within an unknown territory.

Every RDAO that was interviewed talked about the importance of funding research for their RD. Six out of the 15 organizations talked about an immediate need to fund research right away as their biggest issue they identified. One RDAO advocate described this immediate need for RDAOs to turn to research like an alcoholic turns to a bottle of alcohol and said, "But we quickly realized, much like an alcoholic turns to a bottle, I turned to research, and we quickly realized that the two tangible things that we could do were to find more kids, grow the prevalence, and raise money."

In discussing the need for funding research, another RDAO advocate confirmed, saying, “I mean, the most immediate need, I think, of any rare disease organization and for any rare disease is money, and money specifically to fund research.”

Another respondent talked about how they concluded their immediate need for research dollars:

> As we started looking into the RD research to try to find some way to help our kids, we started to understand more about the rare disease space and that there's just a lack of resources across the board. Whether it be from NIH funding, funding at the individual non-profit level, number of people in the field, not a lot of incentives for biotech and pharma to get into the space. A lot of challenges. So, we knew there was an immediate need for research dollars.

Beyond noting the immediate need for funding research, other organizations commented on the role and challenges of funding research. The role of research goes beyond building understanding about a rare disease; it also serves as a way to bring credible information to a medical professional, to begin a dialogue and to bridge understanding. One organization’s respondent discussed how having a research-backed position was important to share with various medical professionals and guide research with a pharmaceutical company:

> I would say in the rare disease forum, the biggest issue is establishing a research-backed position, so that you can properly go to various people in the medical industry to say, "You need to look at this." The easiest example I have is, when you're looking at funding from, let's say a pharmaceutical company. If I go to them and say, "Hey, I want to do this research experiment, and here's why," they're going to want you to be able to at least prove in some way that this research makes sense. But the rare diseases that are out there

> have none to very little research. So, you are fighting constantly of, we need to research to do more research, but no one's willing to do the research.

Having funding for research also helps RD advocates move the drug development process along. Research is expensive and takes a long time. One organization's respondent echoed this, saying, "We just really wanted to get research funded and get the drug pipeline going because it takes so long to develop a drug that we just wanted things to get started."

However, there were a couple of organizations who cautioned against using fundraising dollars for research. They provided information on how much they have raised over a 20-year period while still needing to take care of families today. Research funding typically needs to be continual and spent over a long period of time in order to best understand an RD. This organization's respondent said," At the same time, while we're raising that money for research, which we've been very successful for in the past 20 years, so $300 million has gone into RD research at this point, we also want to take care of the families today."

On the other hand, when research funding is completed, the goal is not always achieved. A challenge that one organization noted is that even once funding and goals are met, often times priorities do not line up or match industry priorities or goals. This is a challenge because of the dedicated, time, energy and costs of raising money for research. This organization's advocate outlined this frustration, saying,

> And I think a challenge is a parent and a foundation lead for a disease like RD XYZ syndrome, and our goals and priorities, and how that certainly doesn't match up often with the priorities of industry partners, biotech, researchers, physicians. How could it? They're not in the spot of having children or a child with this disease. I find that's a challenge. I find the information you often get from these different groups is not

consistent, and it can change drastically. You wonder if you're getting good information, who you can trust.

3. Summary of Factors 3, 4, and 5: Organizational Capacity - Genesis, Prioritization Process, and Role/Title

The remaining relevant factors that address advocacy capacity are about prioritization process, role/title, and genesis of the organization. They were grouped together and summarized due to the low number of codes for each factor. See Table X for detail about the number of codes per responding organization.

RD Organization	Genesis	Prioritization Process	Role/Title
1	7	1	4
2	9	7	5
3	2	2	1
4	7	1	7
5	6	5	1
6	5	2	0
7	3	2	3
8	5	2	2
9	6	2	2
10	7	3	1
11	6	6	3
12	3	2	5
13	2	1	3
14	4	3	2
15	2	1	1
Total = 15	Total = 74	Total = 40	Total = 40

TABLE X: APPROACH TO PRIORITIZATION PROCESS, ROLE/TITLE OF INTERVIEWER AND GENESIS OF RDAO AND SUMMARY OF CODES PER ORGANIZATION

Factor 3 - *Genesis*

The *genesis* code was added as an emergent code and identified by the second coder. *Genesis* is defined as the reason why an RDAO was created and how it was created. The *genesis*

code occurred 74 times. At first it was a factor that was only assumed, but there was a recurring commonality amongst all the RDAOs interviewed, and so it was added as an emergent code. The findings contribute to general knowledge about how RDAOs are started and how *genesis* as a factor influences advocacy capacity. It was important not to overlook the reason why the organization was formed and how it got started. Upon diagnosis, the journey of RD advocacy begins, and so do the issues that come with working in this complex space.

All 15 organizations who participated responded with similar feedback as to why the organization was first founded. Ten of the respondents were directly affected by the RD they do advocacy work for. This is no surprise as all 15 organizations described the RD as adversely affecting childhood when the patient first identifies and begins to understand and tackle their disease. Reasons for starting their organizations are related to RD challenges such as lack of research, funding, low number of patients, scarce resources and lack of an advocacy organization. Thus, most of the respondents described how they were proactive and decided to create their own 501c3 organization in order to close a gap or address an immediate challenge that would help them to advocate on behalf of their RD patient community. Respondents described how and why they got started and how it relates to RD advocacy.

In particular these organizations shared their personal parental journey in starting an RDAO.

One of the parents shared her personal connection through her son's diagnosis:

> My son was diagnosed with RD in late 2002. I had a business background in sales and marketing and so I knew that I would want to be actively involved in raising money for research to find a cure. I looked to find another organization to work with, not ever expecting that I would actually start my own non-profit and I really didn't know anything about the non-profit world. I was disappointed with the sense of urgency that I saw was in the existing

organizations, and also, we have just kind of a bias towards working with biotech companies because they found them to move much quicker and being just a lot easier to work with. For those reasons, we decided to start our own non-profit.

Another grandmother shared her story of starting their RDAO and said,

Okay. Basically, our organization was started because we ... due to my grandson's severe hemophilia. Our family is very much a proactive family that plans ahead. So as soon as we received his diagnosis of RD and he was only 11 months old, one of our thoughts was, "Okay, what about an automobile accident?" We went about studying procedures to make sure that if mom and dad were injured and unable to treat him or even speak for him that he would receive his medication. Because in an accident, I don't care if you can put medicine like that or need some extra dough. You make sure that you're covered so that you don't freak out.

Another parent shared her experience of starting their RDAO upon diagnosis, and the rarity of her disease. She said, *"*Our organization started, let's see, we started 11 months ago, because my daughter was diagnosed with a very rare genetic mutation in her gene. And it's a neurodegenerative disease, and the prevalence at diagnosis was under 20 known in literature in the entire world."

Within the RD advocacy space, a lack of resources often exists. Other organizations outlined how the start of their RDAO was a result of being proactive or addressing the gap in available help. These four organizations address the reason for their genesis and discussed how the founding of their organization addresses gaps in RD advocacy and the need to be proactive.

One respondent talked about how they had to do something, and said,

We started out saying, "Oh my gosh. Somebody's got to do something about this. Do people know that this is an issue?" That's kind of what I was on the phone, I was emailing, "Do you realize this issue is this? This needs to be fixed." After six months of nothing, Steve realized that God was saying, "Okay. You need to go do something about this because clearly no one's listening, and someone is going to die."

Another story of *genesis* was provided by one of the employees of an RDAO representative who shared the journey of how their founder was determined to do something about the RD in lieu of available resources. She said,

Our organization was founded by a woman whose husband had our disease, and she decided that she was going to do something about it because at that point back in the 1960s, there was no advocacy. There were no community services. There was no research really being done in the area of our disease. So, she was a very compelling kind of personality. And she used her influence and her dynamic personality to get congress to supply the very first research into neurodegenerative diseases, which includes ours. So, she founded the organization in 1967. And over the last, this is her 50th year of providing services to our community.

Addressing similar gaps in RD advocacy, this organization tells a similar story, whose representative said,

The reason that the organization was started was because Katherine saw that there was an unmet need for awareness and education and advocacy. Ninety percent of people who have RD are not diagnosed. There are effective treatments for RD. That means that the people that don't know that they have it are going without or are under treated when they could be preventing heart disease.

Lastly, another RDAO advocate shared historical context to the foundation of their organization and how at the time only one study was available for reference and there was a low number of cases. She said, “Sure. So, the RD Foundation was founded in 1995 by a mother who, her daughter had just been diagnosed with RD. At the time, there were only eight known RD patients in the country. There was, I believe, one research paper in all of the literature.”

Factor 4 - Prioritization Process

Fourteen out of 15 organizations provided responses about their internal prioritization process. The one organization missing a response did not address it in their interview. The code was named *prioritization process* and referred to the process by which the organization endorses a plan, identifies key advocacy areas, or creates a strategic plan for RD advocacy activities. This section had only 40 codes, (see Table X). However, in summarizing this section as a whole, six out of 14 responded to the question and probes related to how their organization had or prioritized their advocacy work with similar responses. These six organizations shared a similar quality at first, answering the question with a focused, structured description of the way they prioritized work, but in speaking further with each of them, they would move away from the previously mentioned one to two priorities and would continue to add on multiple priorities unconsciously.

An example of this type of response is from this organization’s representative who makes the prioritization process sounds simple and easy, yet many priorities were uncovered, indicating complexity. She said,

> We definitely do have a strategic plan in place. We actually developed it into a graph format. So, as new ideas, components, come to me and come across my desk, it's really easy to make that decision whether it fits in it or not. So, we have the overall awareness which is to

identify families that have neurofibromatosis and then to create awareness with all the programs that we do, and then we have three major buckets on the side of that. And that would be to build the research community, we do that through our advocacy, to fund the research, then to build our RD community, our local community, like we had talked about before, which is through some of our educational meetings, materials, chats, websites. And then the third bucket that we work out of is funding our mission and that is all the different things that we need to do, the events and grants that we need to fill out in order to be able to keep ourselves operational.

Prioritizing did not seem as simple a process reflected in one to three clear priorities for the organization. In general, it was found that the majority of the organizations perceived themselves to be focused yet not realizing they managed multiple, layered priorities. One of the respondents had an important realization that having a strategy or list of priorities may work best on paper but is not what is happening in reality.

The following quotes described similar experiences among RDAOs:

One of the interviewees spelled out how strategic plans may land on paper, but action doesn't always follow. She said, "We do have a strategic plan and advocacy is definitely part of that. Unfortunately, and I think you'll find this is true of a lot of organizations that the strategic plan is more on paper than it is in action."

Another organization described how they are doing multiple projects and didn't have enough time or people to gain support. She said, "I haven't done the research on it. I've got four full-time projects. They're my top priority right now. Yeah and not enough people to delegate them to is the problem."

Others describe an iterative process where priorities may change or require a mix of input to get their priorities in place. There is difficulty in placing one priority over the other in many instances. These organizations described needing a flexible prioritization process with the following descriptions:

And then yeah, some of the things we're doing, we do have to prioritize. Clinical guidelines for RD syndrome. They don't exist. That's a project, so does that come ahead of the caregiver preference study or behind it, or can we do them both at the same time? I think it's ever evolving. We don't have a standard process to say this one's coming first and this one's coming second. It's more we discuss it with the board and make that decision there. Some go on at the same time and sometimes those things change too, as you know. The next board meeting we might say, "You know what? This one has now become more of a focus, for whatever reason, for the landscape."

Another RDAO advocate shares how they balance choosing priorities based on speed of impact to the RDAO. He said,

> It's kind of a mix of how we actually do it. Some of the things that obviously we have to handle, and address are internal for our organization. So, for example, setting up our Google Drive to enable collaboration for the people who are a part of our organization. Does that have a direct impact on the patients that we're working for? No. Is it an important tool and an important initiative for us? Yes. A lot of how I look at prioritizing them is, what is going to have the quickest impact right now? And a lot of that being because we are a startup and we're not as established as other organizations, patients that are looking to us ... If I can give them quicker results, they'll have a lot more faith and a lot more confidence in what we're doing, versus doing certain things that may take years or months, that while it'll have a very large impact, the impact takes a while to be seen.

The remaining eight organizations characterized their plan by describing a structure that included a check-in process with their board (a medical board or scientific advisory board, or both) in order to elicit feedback to identify prioritized areas of work. Depending on their role, they characterized a hierarchy-related process with checks and balances before receiving formal approval. One described this structured approach and said,

> Yeah, we do a business plan with our board, a strategic plan, which goes over a lot of things with the foundation. When we have those discussions with the board we do highlight some of those different areas and prioritize what's the focus. I was just saying it the other day, as much as we'd love to, we can't be everything to everyone and do everything. We do a lot of collaborating with other organizations. There's other RD foundations, not only in the United States but around the world. There's RD Society, here in the United States and also in other countries. We do a lot of collaborating and group calls and things to make sure that certain groups are focusing on certain things, and we're not doing duplicative efforts. That's one thing we do.

Another organization relies on an annual meeting to determine their priorities and said,

> We have a meeting annually of government relations individuals, where they kind of chart what the key issues are for the coming year. And everyone has input into that. As a rare disease, we very rarely get what we want. But the issues that they determine are of importance to the entire membership, do of course affect our members as well.

Similarly, another organization shares their structured approach to prioritization: “So, with our Board, each year we sit down and define our strategic priorities and then evaluate what belongs in our strategic priorities, what do we have the resources to accomplish. Priority setting with the leadership and the Board.”

These factors answer the research question, "What factors influence advocacy capacity?" Addressing these factors related to understanding RDAO prioritization process helps to understand how these organizations attempt to prioritize their organizational and advocacy needs, while remaining nimble and flexible to complex challenges and competing priorities.

Factor 5 - Role/Title of RDAO Representative

Fifteen of the interviewees held some type of leadership role in their organization with a majority of organizational representatives sharing their titles as CEO, President, or Executive Director, while managing the day-to-day of running an RDAO. Regardless of title, those with executive-related titles were decision-makers for advocacy, budgets, and overall management of the organization.

Acknowledging the respondent's role and title was beneficial information to better understand the factors influencing advocacy capacity. Overall, eight of the respondents named themselves as founder, co-founder and President or CEO. This is important to understand that over half of the respondents were responsible for building their own RDAO and also have executive or decision-making responsibilities. See Table III for summary.

Of the 30 organizations initially contacted, 15 RDAOs agreed to voluntarily participate in the study by responding to direct email requests for telephone interviews. Position and titles varied by their direct or indirect connection to the RD, titles, roles, amount and type of staff, type of advisory board, office type, and age of organization.

The advocacy capacity criteria included used a blanket title for all organizations which was Executive Director. However, in speaking to all the individuals it was clear that various titles existed with multiple roles and responsibilities.

Five respondents were not directly affected by an RD through their own family but were employees of organizations that were 10-15 years old or more. With the organizational and advocacy criteria, they fell between medium and high-advocacy capacity levels. Older RDAOs had the budget and staff to have different types of roles due to more established infrastructure and budget. A common theme in this results section is that the respondents who were part of this research described the multiple roles they play in the RDAO as evidenced by the multiple titles they shared. These respondents shared similar experiences.

This RDAO respondent described experiencing organizational changes, yet retained multiple roles, and said, "Yeah. And so, I'm co-founder and president of the organization. And it's gone through a few different iterations. I'm president of the board, yeah."

Another interviewee fully acknowledged she did it all for her RDAO and said playfully, "I'm the founder, the president, executive director, web master, advisor, everything."

One of the long-term employees with an RDAO, talked about her multiple roles through the years, surviving change and said,

> I've been with the organization for, this is the start of my 20th year. I've worn a number of different hats during my time with the organization. I started out as Director of Communications, Marketing and Education. And through the years I've become Director of Programs and Services. Advocacy was added to my role. Communications was taken away. Marketing was taken away.

One of the unique roles and titles tells a different story. This RDAO is led by their entire family and the respondent said, "Yes. I am actually the President and Founder. Technical, and it was our family. It was me and my husband and my son and his wife. They are the parents to the child with RD we advocate for."

iii. Sub-research Question 1: What are the characteristics of rare disease organizations that undertake advocacy activities?

This section presents results that answer sub-research questions 1. There are two main characteristics of RDAOs that undertake RD advocacy work. The two characteristics are (1) engagement and outreach (2) gaining value from other people. Four sub-characteristics of gaining value from other people include (1) networking, (2) third party technical support outreach, (3) partnering and (4) mentoring.

Overall, two parent codes named *Connecting Constituents with Stakeholders* and *Gaining Value from Other People* represent the two characteristics. *Connecting Constituents with Stakeholders* had one child code called *Engagement and Outreach*. *Gaining Value from Other People* had four child codes named *partnering, networking, third party technical support, and mentoring.* These four codes had a low amount of codes per child code and were summarized together in one section. Definitions for each code are provided in each section, respectively, to aid in answering and describing the characteristics of RDAOs.

1. Characteristic 1 - Engagement and Outreach

Greater engagement and outreach are strategic tools critical to RD advocacy work. This was also recognized in the adapted logic model. For the purposes of this research, this parent code was first examined and described by three emerging themes and one sub-theme. This section described RD engagement and outreach and represented the largest number of codes with a total of 210 codes, throughout the entire study. *Engagement and outreach* were defined as connecting with people who have similar RD or stakeholders of the RD in order to inform, educate, or connect within a specific RD community. All 15 organizations responded as to how they engage and provide outreach to key stakeholders. This larger volume of codes aligns with the need for RDAOs to create greater awareness about their RD due to lack of resources and their need to

build and find their RD community and connect with others. The summary of codes is seen in Table XI.

RD Organization	Engagement & Outreach Code
1	31
2	24
3	15
4	16
5	8
6	19
7	8
8	14
9	14
10	12
11	14
12	11
13	9
14	11
15	4
Total = 15	Total = 210

TABLE XI: CHARACTERISTICS OF RDAOS – ENGAGEMENT & OUTREACH, NUMBER OF CODES PER ORGANIZATION

The three emerging themes and subtheme for Engagement and Outreach are listed below in Table XII.

Engagement & Outreach - Themes	# of RDAO Reporting	Frequency	Data Source
Characteristics of RDAOs Theme #1 *Social media is way for RDAOs to reach their RD community, and others*	15	39	Stakeholder Interviews
Subtheme #1 to Social Media *Facebook is consistently used by RDAOs.*	15	28	Stakeholder Interviews
Characteristics of RDAOs Theme #2 *Software Tools support RD Advocacy*	10	12	Stakeholder Interviews

TABLE XII: SUMMARY OF THEMES FOR CHARACTERISTICS OF RDAOS-ENGAGEMENT & OUTREACH

Overall, due to its broad definition, this section overlapped in many ways with the child codes, third party expertise, mentoring, networking and partnering. Working with various types

of people provided an opportunity to also engage and conduct outreach, or communications geared towards a certain audience. A differentiation in the two sections, however, is the emerging themes.

Engagement and Outreach Theme 1 - Social media is a way for RDAOs to reach their RD community and others.

Social media is defined as forms of electronic communication, including websites for social networking and microblogging through which users create online communities to share information, ideas, personal messages, and other content, such as videos (Webster, 2019). In terms of engagement and outreach to advance advocacy work, all of the organizations in this study utilized a form of social media to communicate and connect with their RD community. Facebook, organizational websites, and Twitter were mentioned most often along with cross usage of media. This section outlines how these organizations are reaching out to their communities both internally and externally and the reasons behind their engagement activities. All RDAOs mentioned using Facebook (FB) (n=15). The way they use FB to engage and do outreach is described as a subtheme for social media. It was a commonly-referred-to social media tool and had enough responses to create a subtheme.

Four organizations provided context as to the value of social media for RD advocacy. One organization talked about how their RD members use social media because it's easy, but more so because their RD community may have mobility or other issues. Thus, social media allows the RD member to stay connected from their home or desk. A respondent said,

> So, one of the things about the people that have RD, I think that they are often on social media. Some of them because mobility is an issue and some it's just because it's a very easy way to connect with others that are living with the same thing that you are. So, I

> think it's trying to use social media in order to be able to attract them to the organization in a way that we can help them a little bit better by informing them about things that are going on in their area that they might be interested in attending, like educational meetings or maybe potentially even a walk which is a fundraiser, but it is also a support for families, as well.

Another respondent talked about how social media has helped their organization obtain global reach and identify others with the same disease outside the U.S., saying, "And it also allows us to see the global scope of things. We do have patients from the UK, Australia, I want to say there've been a couple from Africa. I think we also have a few in the Asian continent as well."

Another benefit of social media is the ability to do targeted outreach to those folks who are participating in an event in a certain location. The same respondent said, "Yeah, definitely. I think when we go into a new city for one of our workshops, I think that a big part of how we reach out to them is through social media."

On the contrary, one organization used social media to connect with their members, but based on survey results they conducted, social media was not the best way to find people especially if their community members did not have Internet access. This may be due to costs, cultural issues, affordability, or accessibility-related issues. Therefore, online was not their main choice or source to disseminate information. The respondent said,

> …for us, it was very helpful, our local pediatric cancer group who actually tries to support the families with rare disease. They did a survey it was now two years ago I think of families and again this is Las Vegas and we do have a lot of Hispanic culture here, so

> this does play into the results, but what they found out was that only 68% had any access to the Internet. And of that, less than 30% actually used the Internet on a regular basis. Yeah, and that's why we don't rely on online at all. And while we do disseminate information to the website, but we do mailer…"

Social media remains a powerful tool to conduct and provide RD engagement and outreach. Its value for enabling engagement and outreach is an important characteristic of RDAOs.

Engagement and Outreach Subtheme 1:

Facebook is a specific social media tool for RDAOs to connect with RD networks

Other social media tools were mentioned occasionally, but comparatively respondents went into greater depth about the frequency and multiple uses of Facebook (FB). In many cases, FB was mentioned as being used as a closed or private group page or chat room. The organizations described how they use this social media tool mainly in private or public facing roles. Private FB was used within a tight knit group of accepted users for hosting private conversations and creating opportunities for members to talk about their disease or disease specific activities.

One of the RDAO interviewees said, "Yeah, that's more frequent because we have a private Facebook group for the families that are online. …Where they can talk to each other and vent and share." Other organizations described how they use private FB to connect with their community.

As one RDAO manager described FB, they used it when they first started their work and how it helped them maintain their community-based RD group. He said,

> We have a RD closed Facebook group. Now, they didn't have the non-profit Facebook groups back when we started ours, so we just have a closed Facebook group, we haven't

migrated it because we think we'll lose more people than we'll gain. So, we communicate that way. We have our personal Facebook pages, because both my wife and I have fairly large networks, so we have those.

The importance of connecting to stakeholders and family members in order to share RD experiences while describing how they coped was explained by an RDAO respondent who said,

Our biggest outreach initiative that we have is a private Facebook support group. It consists of sufferers and caretakers of those with RD. And it's just a way for patients to go on and engage with one another, talk about what they're experiencing, seeing if others have experienced it, how did they handle it. I've seen discussions of side effects of different prescriptions that they've been given. Even recommendations of doctors in certain areas, or I've seen people try to find a way to explain what they're experiencing to family members or loved ones who may not necessarily understand but want to. And that's also a medium in which I engage with our network.

The usage of both closed and private FB pages was also discussed by a few organizations. Some of them used both open and closed group accounts. One RDAO had both types of FB pages and explained how they use their various open and closed FB pages. They said they used it for various audiences including the public, the rare form of RD members, volunteers, and lastly an online discussion forum. They said,

But we have a private Facebook group that's really more about fundraising, and advocacy ideas, and things like that. There's a larger group of these RD disorders, RD XY disorders, which RD is one of seven. There's a larger group on Facebook, and that talks more about the stuff, as far as new wheelchairs, or my son has this happened to him, do you have any

medical thoughts or advice and things like that. Our group focuses more on the business side and advocacy side. Although my wife, being a physician and the scientific director, she takes medical questions all the time on Facebook direct, or by phone, or by email. We do all of it. I would say our most organized things are either in group email, Facebook group that we have, and also these calls that we have every two months, which again, are really important.

Similarly, another interviewee explained their uses for multiple audiences using various FB pages and said,

On the social media, we have the public but then we have a closed Facebook page, a couple of closed Facebook pages: One for RD, the rare form, and one for any form of RD. Then another one for our volunteers and then we have an online discussion forum. It's on there through our web page you know with a password and all that and it's by topic. In that, we're kind of nascent.

However, one RDAO cautioned the use of FB due to patient or personal anecdotes that may not be an accurate or best way to self-treat. He warned of the importance and responsibility of needing to respond quickly. He said with caution,

We're on Facebook, we're on Twitter, we're on all of that and we have a staff that monitors that all the time and responds to it. And you have to respond because you have to be very careful about that, because you could have a person go on something like Facebook and say, "Oh, you know I aerosolized," not a good example, it didn't happen this way, "but you can aerosolize ammonia sulfate and I feel better." Well, you got to follow that real quick because that can do damage.

FB is described by the various RDAOs as a specific social media tool that can used for different purposes with various audiences.

Engagement and Outreach Theme 2 – Software helps RDAOs reach and organize outreach materials

Ten organizations mentioned RD-related tools that they take advantage of to keep track of their members and data used to understand more about their RD communities. While the RDAO may have a multitude of work to complete, computer-based programs and software can aid nonprofits to act more efficiently, be better organized, and provide time-saving tools. This is important especially in engagement and outreach as it relates to timely communications, follow up regarding events, or maintaining a key list of donors.

For example, one of the respondents discussed the value of using Google Drive to support RDAO collaboration. They said,

> Some of the things that obviously we have to handle, and address are internal for our organization. So, for example, setting up our Google Drive to enable collaboration for the people who are a part of our organization. Does that have a direct impact on the patients that we're working for? No. Is it an important tool and an important initiative for us? Yes.

Specific Customer Relationship Manager Software was mentioned by a few of the organizations. One of the tools can help schedule social media posts on behalf of the RDAO. This RDAO's respondent said, "But, I think that there is a lot of area for growth here, especially with things like Hootsuite and things where you can kind of schedule the social media."

Another software called *Network for Good* was mentioned that aided in the management of RD stakeholders and donors. The representative described it as,

> We have one but it's more on a ... I forget what they call it now but the contact manager and fundraising type stuff. What we have is a Network for Good software for managing our stakeholders and our donors. Yeah. Although, we haven't had that for very long and it's not something that I personally work on, I think that will be very useful in terms of being able to segment our stakeholders and to be able to figure out where they are and what they need.

Managing outreach or communication is important. Another software-related tool used by RDAOs is Mailer Lite. This organization described its value back to the RDAO for managing engagement and outreach and said,

> Yeah, we actually just launched a CRM that we are utilizing for relationship management, as well as a fundraising pipeline as we begin delving into that aspect. I'm currently in the process of selecting an organization manager. In essence, it's just a project management software that will allow me to individualize all the projects that are out there, develop timelines, and create plans for either a year or two years or three years, depending on what I'm working on. Obviously, Mailer Lite being our mass email communication. We use that for our newsletter. If we see things that come up in the interim between the newsletters, we can use that to communicate that out.

In a different way of using CRM, one organization's respondent described how they use it to identify potential partnerships for future development and to support their advocacy capacity. The comments were,

> The CRM really, from the advocacy side, the way that I utilize it is a little bit different than necessarily how others might. From a leadership standpoint, I use it to identify potential partnerships that I could begin developing. In our current network that we have, I've actually

> identified three or four individuals that I'd like to pursue for various reasons. From an advocacy standpoint, it allows us to see how many patients we're reaching, where are they. It can allow us to begin creating regional focuses. So, if I identify that 25% of my network is on the West Coast of the United States, I can begin developing strategies, and how can I best focus attentions on that 25%? And develop a strategy to help service them on a regional basis.

As opposed to specific software, two organizations offered up information on how they use RD-specific tools such as the patient registry or natural history study to aid in outreach and engagement as well. One organization tracks participation with the organization thru their patient registry. The respondent said, “But our main external things that we're doing that patients will see are things like our patient registry. We have a white paper on the condition that's currently in draft. It's recruiting various people to fill different positions in the organization.”

The other organization brought up the value of their RD-specific natural history study where they tried to capture as much data about the RD as possible regardless of location. They said,

> … we're about to launch a natural history study so, again, the more people you can get contributing their data and their experiences to the natural history, the better, regardless of where they're located in the world because, you know, if you don't have enough RD need, it doesn't matter whether you're in Egypt.

Computer software and CRM are important tools that RDAOs use to aid in their engagement and outreach efforts. One of their characteristics is how they use these tools to drive

membership, have organized communication and keep in contact with key donors, to drive forward their goals.

Engagement and Outreach Theme 3 - Targeted Audiences Identified for RD Advocacy

All of the RDAOs in this study provided an example of who the target is for most of their engagement and outreach efforts. The following results provide examples of the spectrum of various audiences that RDAOs may reach. The different types of audiences listed aligned with the organization's education and awareness goals. The act of identifying and naming the audience seemed to depend on where the RDAO was on their journey and how they defined key objectives for their advocacy work.

One of the organizations persistently called their State Department of Health: "I contacted national organizations and I contacted our State Department of Health every two weeks. An email then a phone call, an email then a phone call. I got no replies from anyone. We are the average Americans, you know."

The medical community is important to the RD community and advocates. Within this group of professionals, RDAOs mentioned physicians, medical advisory boards and the medical industry as important target audiences for them.

Related to physicians, one organization's respondent discussed why they are important to their advocacy efforts:

> They also need a physician to understand the disease and care, and whole healthcare professional team that really understand the disease and know how to push the envelope in terms of improving the care and trying new things, and learning from other diseases, and creating new care guides that are going to keep these kids healthier and enable them to live longer.

Another respondent also described the importance of reaching out to the medical community: “From an advocacy standpoint, the more conversations that are happening in the medical community about RD; that will be when we know we've been successful in our advocating.”

One organization’s own medical advisory board was an important medical field audience. They explained by saying,

> To the advocacy side, I would say the most critical is our medical advisory board. And the way that I look at it is, if I can get a doctor onto my medical advisory board, I can get them engaged and invested into my organization. They are more than willing to go out to medical professionals that they know and tell them, "From your perspective as a neurologist or an infectious diseases doctor or a general practitioner, you need to look at this organization and see what they're communicating, in case you find a patient that is exhibiting these symptoms." And I've found that that type of networking and communication has been way more effective than developing research data and publicizing it in an infographic or something else like that, where yes, it looks good and it may even make sense to one that it may seem like it's worthwhile. But having another person in your realm saying it's important, that puts a lot of weight behind the organization's name.

Another respondent added onto the medical professional list of target audiences, but shared information related to the need to target the medical industry to achieve goals, network and create a cadence of communications to stay in touch.

He said,

> In the immediate, it's a knowledge in the medical industry. It is not uncommon for our patients to go in to a doctor who has never heard of RD, and to be told in that sense, or actually a few have been told formally, that they are "crazy," and that their condition does not exist. Well, we sort of built a pipeline, you know we're always, we've put out a few newsletters, not many. But we communicate with our donors and we've built this donor list over since 2009. And as we communicate, those folks are donating money, and I always say that they expect something to happen out of those dollars. So, we communicate what we're funding, what we're proposed to fund, what we're funding, what the expected outcome is. And then every time we have an event or whatnot, we'll update folks on where projects A, B, and C are. So, we communicate our successes. And then that tends to lead to, okay, these folks are really, once you're achieving goals you set out to do, people are willing to be repeat donors, if that makes sense.

Outside the medical community, reaching out to other communities was also just as important. This included organizations such as other researchers, RDAOs, legislative bodies to ensure that they would demonstrate the growing RD community.

Engaging with the researchers was a priority for one organization who talked about how other RDAOs are just as important to target in order to learn more and continue learning and talking about RDs and said, “I think relationships with our researchers has been very important, as well as heads of other patient advocacy organizations.”

As RDs are an important component of public health and public policy, targeted advocacy work on Capitol Hill is important to reach legislative bodies and try to reach out and educate more about RDs on a broader public health topic.

This respondent said,

> So, I think the one that we use most commonly is when we talk about going to the Hill and asking Congress for funding for RD research. Another way we talk about advocacy is being a strong advocate for your own health care or your child's health care by making sure that they are able to have the information that they need so that they ask the right questions, so that they're getting the proper care. And then, since 50% of our patient population has learning disabilities, we also use advocacy in individual education plans (IEP) and educational meetings.

Another organization shared a similar experience with targeting public policy folks, and why it was important to their RDAO and said, ...we really focused, because we didn't have a lot of money, we really focused on research and public policy. We had a public policy on how to raise money and really focused on our care center program and research.

In terms of forward thinking one of the high capacity organizations talked about another key target for them who was the adult community of their RD. Despite being diagnosed in childhood, today's advances in medicine have increased the age of survival as well as quality of life. It leaves this particular RDAO focused on their adult RD community.

The respondent said,

> Right now, one of the things that they are doing more and more of is trying to reach out to their, especially their adult RD community, because we have more of them now, it's not just a pediatric disease. We've got probably 15,000 patients and they all have different obstacles. Thinking about education, thinking about marriage, thinking about birth control, thinking about all those issues. What we're trying to do is bring them into a community in terms of outreach. We're using technology, we can't bring them all

> together in a room by the way because they have unique infections that we can't allow one patient to interact with another and cross-infect them so that really hampers their ability to do that. But we are taking advantage of some of the new technologies with webinars and different kinds of video conferencing where you can put hundreds of people on at a time. When the person speaks then their face comes up, so you can see who they are.

In summary, RDAOs who focus on advocacy have to target a spectrum of audiences in order to carry out their work and continually engage to keep increasing awareness. This section supports the theme that RDAOs target a range of audiences as a key characteristic amongst RDAOs.

2. Characteristic 2 - Gaining Value from Other People

The RD community is mostly a small, niche community connecting within its own network with others who have the same disease. When connecting with other folks in the RD world, looking beyond only those with the same disease, there is value in networking, working with 3rd party experts, partnering, and mentoring. With the Internet and social media, many RDAOs have been able to find and connect to one another locally or globally and create stronger connections to other RD patients and families.

As resources may also be scarce, there is a value in working with other people within an RD network, reaching out to partner in order to advance priorities faster, or leverage other people's assets by tapping into mentors and teachers, in order to save time and avoid making the same mistakes. Some in the RD space genuinely offer coaching and guide new advocates who have recently joined the RD community at-large. This value gained with and through people is an

important asset to the RD community. When there are zero to low resources, people are of utmost value to an RDAO.

Four child codes as seen in Table XIII describe the different degrees of value that RD advocates gain by working with others. These codes were relatively low numbers with codes from less than 10 organizations. There was not enough data to identify emerging themes per code, but summaries of key insights and learning are worth reporting on.

Gaining Value from Other People

RD Organization	Networking	3rd Party Technical	Partnering	Mentoring
1	15	24	24	7
2	12	2	11	1
3	6	3	2	3
4	9	8	3	0
5	4	1	2	2
6	2	5	3	0
7	3	0	1	1
8	9	3	3	3
9	4	5	4	1
10	8	5	4	0
11	6	7	4	1
12	2	2	2	0
13	2	3	3	1
14	3	5	3	1
15	1	2	1	1
Total = 15	Total = 86	Total = 75	Total =70	Total = 22

TABLE XIII: SUMMARY OF CODES FOR NETWORKING, THIRD-PARTY TECHNICAL SUPPORT, PARTNERING AND MENTORING

To further answer the research question related to characteristics of RDAOs to advance advocacy, this section provides results that reflect how RDAOs gain value from other people and what the respondents shared. Four child codes were used, named *networking*, rd *third-party technical support, partnering* and *mentoring*. Definitions for each code are provided in its results section.

Sub-Characteristic 1 – Networking With Others

Networking internally and externally proved to be invaluable to RD communities. Many benefits were described from respondents. One organization even said the core of their work is about relationships: “Yeah. I would say 50% of what I do for the nonprofit is just relationship management.”

Attending various RD or scientific conferences allowed for the opportunity to meet others, learn, and socialize. One respondent said,

> Well, these families, because they have rare diseases, don't have other groups locally or even nationally to meet other rare families, or others like them. So, we get these families together a few times a year now, just for socialization and for support. Several organizations organize meetings for the sole purpose of the RD community to get together, talk to similar people going through similar issues as well as expand the reach to community that has a common thread.

One respondent also described the value of conferences not only for networking but to stay on top of a quick-changing world. Meeting people from previous advocacy paths is also helpful to gain additional insight to RD advocacy work. He said,

> The list goes on and on. Just talking with them, meeting with them, discussing with them, you get a whole lot of insight. Then even in RDAO there were parents that have come before us that have foundations, and we talked with them and got some insight. Things do change so quickly, though. You really have to stay on top of these conferences and continue to talk to people, because what might've been a good strategy four years ago may not work today.

Networking was also found to be an important tool in working with others to have an informal relationship to gather help, get through obstacles, and weigh in on research challenges. One of the RDAO respondents said,

> For me, I use it more as a networking tool than anything. It's more to connect with different nonprofit leaders, and just talk with them and brainstorm how they're looking at different issues as we go through them. We can bounce ideas off one another, and just see if you throw an idea out there, see if anyone's gone through it and talk with them about it. Talk about the challenges. Just more of an informal brainstorming session than anything. We get through different barriers, and they also help us in our research standpoint.

Outside of connecting with others and the value it provided, one respondent also emphasized that since there aren't many resources available for RDAOs, networking and working with others is a form of survival. When one learns something or gains new knowledge, it is intuitive to help the next RD advocate as one knows how hard the work is. They said,

> It's a pay-it-forward community, or at least it should be. Everything that I've learned, and my expertise now is because someone else has already gone through it. Because we don't have those development resources, we're all teaching each other, and I think we as a community have done really well, at least the online committed community, like the people who show up at the Global Genes and NORD summits and things like that, really have embraced the let's help each other get through this mentality. And then can you talk about what you think too the value of networking is in rare disease community? To getting- Oh, it's required. You can't survive without networking in the rare disease community. Because again, most people start these organizations or advocacy work because they want to save a life, whether it's their own or a family member or in memory

> of a family member. You're not ... you don't go to college for this. There's no rare disease school, and oh, when I get out, I'm going to start this. Everybody's doing it because they have a reason, and nobody studies for it, so you have to learn somewhere and the only way you're going to learn is from people who've done it before because it's such a niche.

An important note from this quote is another reminder that these RDAO advocates do not have a training ground or formal RD school to attend to learn their craft. Yet, the value gained by helping others and giving back is part of being a member of this niche community. The lack of a formal training ground or school of RD advocacy underscores the importance of working and partnering with others within the RD world, because they are who you have to help with advocacy.

Sub-Characteristic 2 - Support from Third Party Technical Support Organizations

The RD advocacy space is comparatively small compared to bigger disease states like cardiovascular disease (CVD) or Type 2 Diabetes. Yet, there were a few recurring third-party technical supports that were mentioned by several of the organizations. Most mentioned Global Genes as an asset to their advocacy work. This aligns with the criteria by GG Foundation Alliance members. *Third-party technical support* was defined as specifically-mentioned RDAOs that respondents worked with or sought out as third-party partners for technical expertise to learn about how to conduct various RD advocacy efforts.

Another third-party technical expert is NORD, an umbrella organization that supports U.S. rare disease organizations. They provide RDAOs with technical webinars, networking, resources, tools and access to all the various RD organizations in the U.S.

Aside from advocacy, another value in working with other experts likes Rare Disease Legislative Advocates (RDLA) is to learn about the U.S. legislation process as it relates to RDs

as well as the ability to advocate on Rare Disease Day. This resource is often mentioned alongside GG as a valuable tool to learning and staying informed about advocacy work in Washington, D.C.

Lastly, most of the RD advocates are resourceful in searching the Internet for a variety of information and support, but one organization specifically said that the third-party expert's greatest value is the constant information flow. She said,

> You know, I haven't attended one of their events, but I think I follow information flow. That's another, if you want to put them in that newsletter that I follow, that I signed up with email list. Another one was Faster Cures and Genetic Alliance, those are ones that I follow. I forgot to mention those previously. So, these, just so I have information flowing in to me.

The value of constant information flow is to stay up to speed on important topics, but also as RD advocates are oftentimes parents and juggling multiple agendas, the flow of information can alleviate having to dig and find information and waste valuable time, and better inform an advocate, making them aware of key RD activities.

Third-party technical support organizations for RDAOs were valued in terms of advocacy training, public policy guidance, legislative trainings, information flow on RD topics and connecting with other organizations with shared values.

Sub-Characteristic 3 – Partnering with Others

Partnering was defined as the benefits gained by working with identified or specifically targeted third parties or rare disease advocacy groups that support the advancement of advocacy needs or goals.

Finding people who share similar values and can help advance advocacy work is a benefit to RD advocates. Partnering helped to form joint workstreams and pool various resources. As such, partnering also offered RDAOs an opportunity to join forces, form alliances, and create greater access to co-funding. Some of these partnerships were less formal while some were described as more formally as an agreement. This section did not have many codes but examples of partnering from RDAOs are provided and the descriptions.

For example, one respondent described an agreement with a partner by saying, "I'm very excited to share, finally, that we have an agreement that was just announced last week with NCATS, NIH, and Pharma Company on a three-way partnership for NCATS to do some work on RD."

The other example was about finding the opportunity to co-fund work together. One respondent described the process in this way: "We were then with them for a few years, by that time our fundraising had grown significantly, and we decided to partner with another sub fund, to form a joint organization."

On a broader research scale, another organization talked about their work with NIH: "We really started building the RD program there. I'd been there a long time. The support of the NIH has been an alliance partner for us, well I came out of NIH. We really started to build the RD program there."

Sub-Characteristic 4 – Mentoring

Mentoring was often associated with being introduced and having access to new people or an existing network of people. The other association with mentoring was about teaching. Most responses address the value of being taught a lesson, a skill, a key learning or new way to think of RD advocacy that the advocacy manager was thankful for. *Mentoring* was defined as having

an external or internal colleague or contact that provides feedback or an opportunity to learn directly about RD advocacy.

Teaching and learning were the two assets described by a couple of RDAOs regarding value from current or past mentors. Some examples of this include calling a scientist to help translate a study, learning from others' mistakes, executive leadership, learning from dynamic RD advocates that one would like to emulate, and gaining valuable advocacy or nonprofit management-related skills to do their job better.

One respondent talked about the value of both networking and mentoring, and said,

> Networking and mentoring are huge, but in order to really fully take advantage of them, I had to be the first person to say I don't know anything, which is true. You have to be very direct and I find that people are receptive when I just cut right to the chase and say, "Hey listen, I don't know anything about this, but I'm in dire need of learning about it. Can you help me?" The answer is always, it's very rare that the answer is no to that. Networking and mentorship again, to me, we have to throw out the conventional strategies of how to go about that. I don't think that you need to spend millions of dollars on a giant gala to get everybody together and to learn the advocacy thing. A lot of advocacy, when we ask to learn, a mentor to advocacy, a lot of that is, "Here's how you throw an event, here's how you do all that." I would prefer to give all that money to research and spend $150 on the backroom of a restaurant where there's one giant big round table and get everybody together to learn. It's mostly about just saying I need help, help me. The conventional form of networking to me is often a waste of time, so it's the people who are receptive to me saying, "Hey, I don't know. Can you drop everything that you're doing and help me?" And the people who do that are Global Genes.

The insight here is that networking and mentoring do not require a high degree of financing and time, just being able to be humble and open to learning is useful.

Another advocate cautioned against RD advocates describing mentoring as a service that is readily available and free. She talked about how trust is important and not everyone is always so free to share their knowledge. While there may also be consultants who can share additional knowledge, RDAOs are already strapped for finances as is and this is not a readily available expense for most. She said, "No one tells you how, mentor tells in between A and B but willing to tell in between, but everyone guards their information. Mentorship is a service. More than a paid profession. Many share information for a price. Will be consultants then charge, and you don't know what it's like."

In summary, these four characteristics of gaining value from other people, are important tools to RDAOs who are focused on advocacy. People provide an important asset to an organization to aid in getting work done, learning, passing on wisdom, or avoiding costly mistakes that RD advocates cannot afford.

iv. Sub-Research Question 2: How have these characteristics facilitated or acted as barriers to advocacy?

Respondents discussed diverse barriers that may affect their organization's ability to conduct their RD advocacy-related work. These results answered the second sub-research question and uncovered two different types of barriers. They are identified as barriers that get in the way of achieving organizational capacity or advocacy capacity. This section reflects what all 15 RDAOs identified as barriers with 139 quotations using the code named *barriers*. See Table XIV. The *barriers* code was defined as obstacles that get in the way of advancing advocacy

initiatives. *Barriers* was ini*tially organized by one parent code called *Identifying and Addressing Barriers,* with two child codes named *Barriers* and *Facilitators*.

RD Organization	Barriers to Achieving Advocacy Code
1	28
2	11
3	5
4	13
5	8
6	12
7	6
8	16
9	6
10	9
11	7
12	7
13	3
14	5
15	3
Total	139

TABLE XIV: SUMMARY OF CODES PER ORGANIZATION FOR BARRIERS TO ACHIEVING ADVOCACY CODE

The initial code provided a way to generally define barriers to achieving advocacy. Five themes emerged associated with barriers to achieving RD advocacy, and three themes emerged related to barriers to achieving organizational capacity. The dichotomy of these barrier groupings answer the research question, "How have factors presented as barriers?"

Barriers listed as themes in this section aligned with some components listed in the adapted Advocacy Capacity Interim Outcomes Model. See Figure 3. Building advocacy capacity is important in RD policy and advocacy work, which has also been identified as an important PH function, as mentioned earlier. The results in this section align with what the literature identifies as key barriers to advocating for people living with a rare disease. For most RDs, there is the

lack of knowledge, and dealing with unknown variables leaves advocates juggling multiple areas continually while trying to make progress against their own advocacy goals.

1. Barriers to Organizational Capacity

Barrier Theme 1: Fundraising Challenges

As result of reviewing the *barriers* code, three themes emerged uncovering and describing barriers to achieving organizational capacity. Themes were determined by multiple reviews of transcripts, coding, then tallying top responses from multiple organizations considering frequency, patterned responses and number of RDAOs who responded. The three barriers are: (1) funding or fundraising, (2) lack of a prioritization process and (3) maximizing staff. Together, these areas are fundamental to building organizational capacity in RDAOs. However, the implications for RD advocacy presented unique challenges to building the organizational capacity needed for RDAOs to achieve their advocacy goals. A summary of the themes for barriers to achieving organizational capacity is seen in Table XV.

Barriers to Organizational Capacity	# of RDAO Reporting	Frequency	Data Source
Barrier #1 Fundraising challenges	9	9	Stakeholder Interviews
Barrier #2 Lack of Prioritization Process	7	15	Stakeholder Interviews
Barrier #3 Need for Skilled Staff and true RD advocates	7	19	Stakeholder Interviews

TABLE XV: SUMMARY OF THEMES RELATED TO BARRIERS TO ACHIEVING ORGANIZATIONAL CAPACITY

Financial resources are not only important for RD patients and their future healthcare needs, but also to building capacity for a nonprofit organization to adequately get work done. Thirteen organizations discussed the need for more funding and fundraising capabilities for RD advocacy. The lack of these activities related to funding hindered organizational capacity. One interviewee said, “Now the barriers, it's always money, quite frankly. Follow the money, is what people say.”

In terms of funding, this section highlights the needs RDAOs have for enabling research, drug development, general nonprofit management, or financing organizational needs. *Fundraising* was defined as the need to secure additional money. RD drug development or therapies do not come at a low cost. In fact, some therapeutic upfront investments may start in the millions of dollars versus hundreds of thousands. A way to alleviate that cost or burden for the RD patient is to raise dollars through an organization such as a nonprofit. As one mother stated, “Right now, I need to raise a couple million dollars for a drug trial.” Fundraising supports building organizational capacity in order to support drug development.

Another respondent discussed the importance of funding to develop access to drugs and the ability to afford their high costs:

> Now one of the most important issues for us is access to drugs. Some of the new drugs that the Foundation helped to develop with XYZ Pharmaceuticals and potentially others are very, very expensive at $300,000 a year. Now these are disease modifying drugs, these are not just your typical therapeutic drugs for treating the symptoms. These are treating the basic defect and will add potentially to some of these patients’ decades of life, but they're about $300,000 a year.

Another respondent identified fundraising as a challenge since the kids in their RD world have to undergo genetic sequencing. They said, "We decided to go out, and we did a lot of fundraising, and of course that still remains one of the biggest challenges, and we realized that we're not going to get anywhere without kids getting sequenced."

Fundraising can support an organization, but a foundation of capital for an organization is helpful. Otherwise, a RDAO is left trying to balance how to make a small amount of funds go a long way. Deciding where to be is described by one organization who outlined this vicious cycle, they said,

> "Right now, it's all volunteer-based positions, just due to the lack of financial resources that we have. And it's this repeating cycle of, we want to bring people on full-time so that they can focus on the organization, but to do that, we need money. So, you go to recruit a fundraiser who would only want to do it full-time and with pay, but you don't have the money to pay them, which is why you're bringing on a fundraiser."

Another organization had similar feelings about the need for money for good staff, but that it has to be built from somewhere before it's good for the organization. RDAOs are always in fundraising mode, and looking to secure funds:

> Yeah. All non-profits are in constant fundraising mode. It's the nature of the beast…Sometimes, build a team, find the right staff, understand the expertise that you need. I think that's a need that somebody can recognize. I mean and obviously there's funding. That's a challenge. Can't do it without the funding.

Another example of this RD conundrum is evidenced by needing research to justify more research.

> However, funding, the easiest example I have is, when you're looking at funding from, let's say a pharmaceutical company. If I go to them and say, "Hey, I want to do this research experiment, and here's why," they're going to want you to be able to at least prove in some way that this research makes sense. But the rare diseases that are out there have none to very little research. So, you are fighting constantly of, we need to research to do more research, but no one's willing to do the research.

Beyond the vicious cycle of fundraising, one respondent brought up how fundraising can take an organization away from their mission and become an even greater challenge. They talked about the challenge of spending the dollars and said, “The biggest barrier, as far as I'm concerned, is spending. That's always the issue because when you're spending so much time raising funds, it's difficult to really dedicate the time to your mission.”

Lastly, one respondent raised the point that a gap exists specifically for RD fundraisers. She implied that fundraising is even more different than fundraising for other diseases:

> There's really not a dedicated place for rare disease fundraisers to go. I have, in my capacity, gone to fundraising conferences and I've gone to rare disease conferences, but fundraising for nonprofits is so different than fundraising for rare diseases, so it's actually ... I'm kind of actually working on putting together just a small, very small group of rare disease fundraisers to be able to learn from each other about best practices in what we're doing and talking about challenges and struggles and that sort of thing, because it's just ... it's so different in this space than it is in any other space.

Barrier Theme 2 – Lack of Prioritization Process

The conceptual model identified the need for *strategic and business planning* defined as planning systematically for how to position and deciding what tactics to use to reach the goal.

While this is a recognized need for capacity building, this process arose as a barrier when organizations were lacking a plan or successful way to prioritize work. This lack of a prioritization process distracted organizations from getting advocacy work done or advanced, and absorbed time which can become an issue due to several competing priorities. This evidence helped answer the research question as to what characteristics served as barriers to organizational capacity. Doing work on behalf RD patients is difficult when priorities are not clear or focused and can take an organization away from its mission.

Most of the organizations' respondents offered up their experiences and difficulties with prioritizing work, despite identifying a process to prioritize work with their third-party experts, such as their board, or scientific advisory board. While they described their priorities, many respondents unknowingly mentioned several areas of work that were not part of their written priorities list but were described as just as important. One respondent noted, "We do have a strategic plan and advocacy is definitely part of that. Unfortunately, and I think you'll find this is true of a lot of organizations that the strategic plan is more on paper than it is in action."

Another advocate described their workload, and how much of it can fall on them as the advocate. Since most of the advocate responsibilities fall on the same person, this leaves a gap in delegating additional work. Lack of funding is means that having additional staff on payroll is not always possible. A respondent described how they address multiple and competing priorities: "There might be. That (training) isn't a top priority right now. I haven't done the research on it. I've got four full-time projects. They're my top priority right now. (Competing priorities) Yeah and not enough people to delegate them to is the problem."

Similarly, another said, *"*Exactly. It's prioritizing. And you're right, it's prioritizing. Sometimes you don't prioritize the right things, and don't realize that these other things, maybe

it's worth taking a stop, and taking a break and saying, "I'm going to get more value from learning some of these additional things."

However, some organizations wrestled with the fact that there are so many priorities, you cannot do them all. One stakeholder said, “You can't make the most of the opportunities that might come your way. That's a really hard thing to do.”

Another organization’s respondent felt similarly while discussing their formal planning:

> Yeah, we do a business plan with our board, a strategic plan, which goes over a lot of things with the foundation. When we have those discussions with the board we do highlight some of those different areas and prioritize what's the focus. I was just saying it the other day, as much as we'd love to, we can't be everything to everyone and do everything.

Another organization lacked a formal process to prioritize their advocacy work and seemed to tackle issues as they came to them. Their respondent detailed how their process was iterative, saying,

> And then yeah, some of the things we're doing, we do have to prioritize. What's the other project you're working on, [Clinical guidelines for RD syndrome.] They don't exist. That's a project, so does that come ahead of the caregiver preference study or behind it, or can we do them both at the same time? I think it's ever evolving. We don't have a standard process to say this one's coming first and this one's coming second. It's more we discuss it with the board and make that decision there. Some go on at the same time and sometimes those things change too, as you know. The next board meeting we might say, "You know what? This one has now become more of a focus, for whatever reason, for the landscape."

RDAOs seem to have the intention to strategically plan and prioritize. Most of these organizations find it difficult to focus on a small amount of priorities. On the flip side, one

organization acknowledged they cannot do it all: be everything to everyone. The data confirmed that there is a lot of work to do for these RDAOs and they simply cannot do it all. These examples of lack of prioritization serve as barriers to achieving greater organizational capacity.

Barrier Theme 3 – The Need for Skilled Staff and True RD Advocates

Many of the organizations responded about their need to optimize or make the best use of each volunteer and staff member in order to build organizational capacity and have success. Finding and keeping good staff and volunteers is important to building successful RDAOs. For these types of organizations, most advocates that were interviewed were a family member, parent, or patient of a person with an RD (n=9). The need to advocate for more RD research, resources, information, or patient needs became the basis to create a 501c3 foundation or organization. Unique to this community is the fact that there aren't many other organizations to lean into that are identical, nor the knowledge or other resources to take advantage of. An emphasis that many organizations shared is when lacking good staff is that building organizational capacity is harder to achieve.

One organization's respondent confirmed the need for capacity and staff. He talked about the importance of having the right people and said,

> Okay, so I think if you're looking at the development of organizations, I think a barrier is to have the capacity, the right staff and the right expertise. It can be, that's a challenge you have to face. You can have great ideas, but if you don't have the right people to implement them, then you can't deliver.

Similarly, another organization mentioned understanding staffing needs: "Sometimes, build a team, find the right staff, understand the expertise that you need. I think that's a need. That's a challenge. Can't do it without the funding."

Leadership and staff development are other key components from the conceptual model. See Figure 3. It is defined as hiring or developing the people to implement an advocacy strategy and establishing a clear understanding of who is doing what. RDAO staff who can be leaders and are empowered to do so will thrive much better than getting lost in a bureaucracy.

This was evident from one organization's respondent who described how they empowered their staff as the decision makers in order to be effective:

> Basically, those decisions regarding what we want to do is basically a staff decision. Now many, many organizations, and I wouldn't say the most effective organizations, they're run by their boards, and the staff is basically just what you say, staff. Every decision has to be run through their board and so they're not very effective in terms of making good decisions and quick decisions. And if every document has to be run through a committee, you can't run effectively. So that's why our board did what they did back in the early 80s and that really helped them to hire good people who really wanted to be leaders and not just followers.

In order to have good people in the organization, some organizations also described key characteristics that RD advocates can possess to get work done. These characteristics are about being well-rounded, results-oriented, and compassionate. When respondents talked about their staff and these characteristics, their voices and thoughtful responses implied that it takes special, genuine people to really back the RD cause and advocacy work. Also, none of the organizations said this type of work is easy. While having good and efficient staff is crucial, RDAOs also described other important characteristics beyond competency.

Working in RD advocacy involves working with different types of people. As one organization's respondent put it, there is a need for a well-rounded personality in order to do this work and be part of a well-rounded organization. He shared his thoughts, saying:

> Then I think the last thing I would say is just the personality to ... Of course, not everybody in the organization needs to have ... I mean, a well-rounded organization has many different personality types, of course, but just the ... You know, the personality to be interested in talking to these people and caring about ... I mean, everybody cares, but in able to…being able to be outside of yourself and have the extroverted ability. That's not natural for everybody to really be able to be the face of the organization, especially in a small organization.

Another stakeholder discussed the rarity of finding true advocates to do this work and take action. Finding good people entails finding rare advocates who truly do the work and work hard to elicit action. He said,

> You know, another barrier that I think, it sort of refers back to when I said being in the rare disease space is indeed rare but finding the true advocates that will take a stand and do something, that is incredibly rare. And now, I don't have a solution to it, but I think finding a way to get more of your base, more of your individual rare disease group inspired and thinking that they can make a difference. You know, they don't have to be scientists. Just getting them to take action, getting them to do something. We've got so many, you know, we have 400 people at our recent family conference. And I just wish there were 10 or 20 of those people that did half of, not in a, I just wish more of those people could step forward and try to make a real difference.

On the other hand, advocacy work requires professionalism and RD advocates need to remember to have compassion. This characteristic for staff is helpful when dealing with people who have serious ailments or are sick every day. One of the respondents described the need to be professional and compassionate:

> That's a good question. I mean, I think something that I try to keep in mind is compassion. It's something that as a professional who's not otherwise connected to rare disease, it's easy for me to forget that the people I work with are sick. Having ... expectations change a little when you ... when somebody you're working with has to go into the hospital for a week, because they have pneumonia, or things like that. We do have some patients on staff and family members on staff, and that kind of helps keep us grounded. At the same time, though, I think a high level of professionalism is necessary in this field. What I mean by that is I think a lot of patient advocacy organizations have a little bit of a ... too much of grassroots feel to them.

Outside of these staff needs and characteristic traits, an important comment was made in this set of codes related to how businesses can think about donating services rather than financial contributions. In the discussion related to having good people one advocate specifically described needing companies to help increase their organizational capacity. She mentioned the opportunity for companies to donate skills or services to nonprofits:

> I think maybe if I could say something to the larger companies, the drug companies, maybe advertising companies, finance companies that can give advice to nonprofits and I don't know if you'll reach all of those, but I think sometimes I know businesses are always asked for donations, donations, donations. Think outside the box and maybe think about how you can donate services to those nonprofits that could be even a lot more help than $25, $50, or $100 at times.

Staff for RDAOs play an integral role in RDAOs and help advance organizational capacity. RD advocacy skills and certain characteristics are worth understanding, as lacking the right kind of skilled staff can serve as a barrier to organizational capacity.

2. Barriers to Advocacy Capacity

The second type of barrier was identified as *barriers to advocacy capacity*. This was defined as obstacles that hinder RD advocacy work. This section reflects five themes related to barriers to advocacy capacity. The themes represent the description of the barrier evidenced from organizational respondent quotes. Five themes emerged from the set of codes named *barriers* developed from the *barrier* code and determined by frequency of responses. See Table XVI for summary.

Barriers to Conducting Advocacy Themes	# of RDAO Reporting	Frequency	Data Source
Barrier to Conducting Advocacy Theme Diagnosed with a RD	15	8	Stakeholder Interviews
Barrier to Conducting Advocacy Theme Unknown or Lack of RD Disease Specific Information	10	28	Stakeholder Interviews
Barrier to Conducting Advocacy Theme RD Life is on the line, without the luxury of time	7	15	Stakeholder Interviews
Barrier to Conducting Advocacy Theme Finding/Identifying greater numbers of RD patients and importance of building a RD community	12	12	Stakeholder Interviews
Barrier to Conducting Advocacy Theme Lack of Patient Empowerment and Parent Empowerment	13	12	Stakeholder Interviews

TABLE XVI: BARRIERS TO ADVOCACY CAPACITY THEMES AND NUMBER OF REPORTING ORGANIZATIONS AND FREQUENCY OF QUOTATIONS

Barriers to Advocacy Capacity: Barrier Theme 1 – Diagnosed With an RD

The first emerging theme for the section on barriers was about the fact of living with an RD, which can be experienced in the form of caring for a child or patient who has an RD or can describe the person who has the RD. This is important, because the characteristics of living with an RD can get in the way of advocacy when prognosis is poor. Ailments of the disease get in the way of advocacy efforts either physically, mentally or emotionally, and childcare is needed while doing advocacy work elsewhere. All of these experiences from having an RD can pose issues for the RD patient or caregiver especially since some RD advocates are patients themselves. By having the disease or caring for someone who has the RD, it becomes a competition to juggle various advocacy priorities on top of getting by day-to-day.

What was overtly mentioned and observed is that most of the RD advocates were affected personally. Nine of the respondents were a close family member of the RD patients, four were employees and one was the RD patient herself. Thus, the summary of this theme captures the barrier to conducting RD advocacy related to the diagnosis as most of the RD advocates are close family members dealing with the disease themselves along with doing advocacy work. Not only were they serving the job of advocacy roles, but also running a nonprofit organization and taking care of all other details related to a family, professional role, and personal agendas all at the same time.

The RD advocate wasn't the only interviewee with an RD, but another RD advocate who was interviewed represented her ultra-rare disease, which is defined as in Europe as affecting fewer than five per 10,000 people or 500 patients per 1 million people. Therefore, she was advocating for herself, her affected children, and those she has connected with worldwide. By having the

diagnosis of an ultra-rare RD, it posed several physical, mental, and emotional challenges for her to also get her advocacy work completed simultaneously.

The barrier to advocacy starts at RD diagnosis. Following diagnosis, barriers are also existent in a variety of ways as dealing with an RD, either as a patient, caregiver, or family member. Several quotes outlined the barriers to advocacy. Upon learning and realizing what RD diagnosis means, it becomes apparent what barriers present itself because of the diagnosis alone. Having a rare disease is not free of the various burdens of disease such as pain, physical, mental or learning disabilities, poor prognosis or dealing with earlier mortality compared to an average healthy person.

Nine stakeholders mentioned the background of their disease and related challenges. Stakeholders in general described the RD disease they are doing patient advocacy work for and provided further context to the severity of the RD based on their current understanding. In addition, stakeholders also described how the RD impacts the patient and, in some cases, themselves. Many of the stakeholders detailed the impact the RD has on quality of life, longevity, as well as its overall effects on health. The parents or advocates described what it's like living with a rare disease with the following comments:

One of the parents describes her reality of diagnosis for her daughter by saying, "My daughter was diagnosed with RD which is a childhood neurodegenerative disease that is progressive and universally fatal."

Another parent shared the same painful realization about kids with the same RD as their own and said, "So, you know I mentioned that kids first start having seizures and over those next two years, they have an incredible drop off of ability as those brain cells start to die." Other parents shared similar RD diagnosis issues related to RD children. They said the following:

- "Heart attacks at age 3 with RD is possible."
- "These RD children who were less than 10 years old, were told they would probably not live to be into their teens, or even on the younger ones, even into nursery school."

Another stakeholder provided information related to the rarity of their child's disease, with very little identified cases in the world and said, "My daughter was diagnosed with an RD. It's a neurogenerative disease, and the prevalence at diagnosis was under 20 in the known literature in the entire world."

Having an RD diagnosis poses immediate challenges depending on the severity and projected prognosis of the RD. These issues can pose as barriers to the advocacy work these RD families or RDAOs take on.

Barrier Theme 2 - RD life is on the line, without luxury of time

Another theme emerged for barriers regarding the concept of time for people with RDs. With the lack of information to adequately act upon addressing the progression of an RD, time is not a luxury because of this uncertainty. There may be uncertainty as to what the diagnosis is, when treatments are most beneficial, or the drug approval process may take a long time, for patients whose RD may progress very quickly. This theme also coincides with the lack of resources theme, as without adequate information, instead of redirecting time to a priority area, that time may be used less effectively to address multiple areas of unknown information. The lack of luxury of time compared to living a regular, healthy life cannot be dismissed for those advocating in the RD space. Results represent seven RDAO responses with a frequency of 15. While one may be busy trying to find out the unknown, or building advocacy capacity, time is of the essence as a life is on the line and improving quality of life depends on work done by the RD advocate.

Patients born with a rare disease have multiple challenges as soon as they become aware that their disease or disorder is not common. This is because there may be little to no resources available to learn more about their disease beyond diagnosis, and their disease may remain unknown, diagnosed or misdiagnosed. The pursuit of this confirmation of a rare disease is often called the *rare disease odyssey* because a patient is often left checking every avenue to determine their disorder, which can take years. Thus, upon awareness of their rare disease, the time spent getting to diagnosis is already a barrier to advocacy, because many times the diagnosis process has taken decades, to only now progress to the next stage of advocating for new needs.

The following respondent quotes illustrate the importance of time to the RD advocate as a person/child with an RD life is on the line, and time is of the essence to find a therapy or cure.

One organization's respondent started her journey because her son needed help, but there wasn't much movement, and because her son's life depended on action, she stated, "I was disappointed with the sense of urgency that I saw was in the existing organizations…" The burden of searching for or finding a cure or therapy falls on the parent, advocate, or caregiver since they are dealing with the RD firsthand. Yet, perhaps in more well-recognized diseases, it may feel like others are already advocates and motivated or know what advocacy work needs to get done.

Another stakeholder who also had an RD discussed the importance of time not only to take care of oneself but others, as well as anticipating future complications of their RD:

> There's a lot of time that they're putting into just keeping their kids going so there's limited time that they have available to volunteer and then, at the adult level, our symptoms can be quite disabling. Also, fatigue is a major symptom. Even adults that are, perhaps, retired, semi-retired like I am, just getting through the day takes up all of our energy…

Some patients are dealing with their RD, while juggling different responsibilities and roles and these may get in the way of their RD advocacy work.

To further describe the time barrier of time, one organization's respondent was a medical doctor reinforced it by saying, "Our disease is time. Everything is time to the parents that are out there today."

The time barrier doesn't leave much room for mistakes or error for RD. With time not always on the RDAO side, it can be a barrier to advocacy capacity.

Barrier Theme 3 - Unknown or Lack of RD-Specific Resources

A rare disease is also greatly associated with dealing with multiple unknown factors. This issue is related to needing to find and identify specific RD-related experts, existing published literature, if available, and having access to medical or clinical resources etc. The lack of many of these resources for RD patients poses issues to getting RD advocacy work done. For example, if there are research needs and one has to advocate for funding, it is difficult to do so when you don't have ample amounts of research to share or emulate. Moreover, the lack of recent or contemporary thinking about RD creates difficulty for advocates in speed of problem solving, especially if the last and only study was conducted several decades prior. It is these kinds of barriers that can stop progress in advocacy efforts or create lengthy delays.

Several stakeholders pointed out how the lack of information added to the uncertainty of being able to learn more about the RD and pointed out gaps that exist where little to virtually no resources are available. The following quotations describe the stakeholders' observations and barriers to living with an RD especially when various type of resources does not exist. These include important advocacy efforts that include resources, support groups, communication outlets, clinical guidelines, treatment, and other advocacy efforts in general. Yet, most

interviewees described the paucity of these efforts or non-existent efforts. These barriers are described below in their comments:

> One of the parents talked about how their pediatric doctors are unaware and won't look for the RD. He said,
>
> > We are really going to impress upon pediatric neurologists that you have to be looking for this. Because if you miss this, it's going to be six or nine months before you see this child again and they're going to have a dramatically worse outcome if you miss the initial diagnosis.

One of the RDAO respondents said there was general difficulty in knowing about the RD condition since it has multiple names. He said, "There's a mixture. It's a lack of education. There is a lack of awareness. But particularly with us is, the same condition is under about five different names." Medical professionals, patients or other healthcare professionals would not know what to look for since the RD information is not easily available.

Other RDAOs describe what this gap in resources meant for their organization. One organization' respondent described this lack of RD knowledge and information: "There's no resources, no support groups, there's no website, there's no contact, nothing. So that left me more motivated, because he was undiagnosed." Another respondent described what it is like to have a child diagnosed with an RD but did not have clinical practice guidelines that existed to treat or address their child. The respondent described this feeling of despair:

> There's not even clinical guidelines. It's everything that's done for these kids is very reactive and not proactive. It's only after a certain complication happened that they cared to do anything. Trying to get physicians to be proactive about their care, because maybe there aren't clinical trials showing this or that is effective. It's just like you feel very neglected, I

suppose, and pushed aside. You feel that your child is pushed aside. It's not intentional, but they simply don't know what to do so they don't do anything.

Another interviewee described how there was nothing available almost 40 years ago, and thus they were lacking historical knowledge and refences and said, "… back in the 1960s, there was no advocacy. There were no community services. There was no research really being done in our disease."

Rare disease patients may be sparse, or geographically dispersed. In addition, there is a lack of available, knowledgeable health care professionals familiar with a certain RD, experts, and specific medical care for RD patients. Other more recognized diseases have greater access to care and experts and may live close to support. In most instances, RD patients do not have the luxury of looking up a readily available in-network healthcare provider who is affiliated with a health center that can treat or provide therapies for RD patients, much less identify a healthcare team within driving distance to be taken care of.

This gap in awareness with physicians also creates a barrier to advocacy. Without understanding from physicians, it is difficult to advance RD care. The following quotes outlined the barriers related to this lack of medical resources or experts that are geographically and conveniently located:

This organization's respondent explains further, saying,

They also need a physician to understand the disease and care, and a whole healthcare professional team that really understands the disease and knows how to push the envelope in terms of improving the care and trying new things, and learning from other diseases, and

creating new care, care guides that are going to keep these kids healthier and enable them to live longer.

Having access to multiple physicians for choice may be something other non-RDs have. This organization's interviewee in particular describes how even finding one physician is a challenge especially when needing to garner interest in working in such a rare area. They explained this by saying,

> We are still working to develop new clinics, and that's something that's based on ... partly in geography, but also partly based on the physicians and the clinicians and the team that's around, because one physician does not a good clinic make, and there are needs in certain parts of the area, or certain parts of the country that can't be met if there isn't a physician that's willing to work on RD or researchers aren't interested in RD in the area. It's a challenge.

Similarly, but in global context, another RDAO respondent talked about how only a dozen or so physicians know how to treat their specific RD and said,

> There's only about a dozen or so specialist doctors in the country and I couldn't even tell you how many worldwide but within the US there's only about a dozen. Correct diagnosis and treatment are the first priorities. Or they may be experienced in another disease, but they have never seen an RD patient with RD.

The lack of resources, healthcare or medical professionals' knowledge, and research serve as major barriers to advancing any advocacy work. The shift or priority for the RD advocate must then be focused on finding or gaining these resources to close the gap.

Barrier Theme 4: Finding/Identifying greater numbers of RD patients and need for building a RD community

Rare diseases are rare because they do not exist in large numbers. Therefore, finding, building or strengthening an RD community, or even identifying patients in greater numbers in order to call it a community, poses an important barrier to advocating for an RD. Communities may be hard to build as RD patients may be dispersed throughout the country or world, and if many are still undiagnosed, they are hard to locate, and if there are very little-known cases, other RD patients with same disease may not exist. Twelve of 15 organizations responded with a frequency of 12.

When little research is available, it makes completing clinical trials difficult due to the low number of volunteers. As one organization emphasized, the process of building community is a benefit when recruiting for RD specific clinical trials, but a barrier when RDAOs are unable to reach the RD community in need. The following quotations from interviewees reflect these challenges from most of the organizations. A respondent said,

> Identifying families that have RD and bringing them into the RD community so that they can continue to communicate with others as we see research progressing. So, identifying them so that as clinical trials become available, through these research dollars that have been spent into RD research work…It's a barrier that we're trying to reach with social media.

This RDAO interviewee discussed similar sentiments and said, "I think one problem that all rare disease organizations face is that we only have so many patients. It makes it challenging, whether it's enrolling in clinical trials or trying to fundraise."

Related to locating RD folks or communities, is the fact that due to low numbers, there is not usually a group of RD patients with the same disease in a short geographical distance. One

RDAO respondent explains by saying, “One of our issues is finding families…Being regional as opposed to a disease specific organization.”

Another missing link for finding or growing communities is the lack of a one-stop place like search engines or the telephone book at one’s fingertips. An organization shares their frustration with this barrier and said, “…to others, it may be small, but being that we really have no direct place to go to find these families, social media has played a huge role in that.”

With the greater reach and use of social media tools, some RDAOs have been able to build their reach by looking outside the U.S. Global reach is possible and another source of information as one organization said,

> …one of the things about a rare disease, which you know, is that you don’t have the numbers so the more numbers you can get, in terms of being heard and in terms of gathering information, the better off you are, so rather than limiting ourselves to 15,000 or so patients within the United States, we encourage people to join us from around the world.

Lastly, one interviewee talked about the difficulty of identifying RD patients and families, if they do not yet have a diagnosis. She said, “It's vital to bring the community together. For us, of course 90% of the people are not diagnosed, so how do you bring together people who don't know they have your disorder?”

Another challenge to building advocacy capacity may be finding and growing an RD community. Many benefits arise from RD communities such as enrollment in clinical trials with more patients, finding people with similar diseases, and having a connection to a similar community of people.

Barrier Theme 5 - Lack of Patient and Parent Empowerment

Another theme that answered the research question identifying barriers to building advocacy capacity was identified amongst the stakeholders who dealt with RD children and understood not only the importance of patient empowerment, but also the need for parent empowerment. Some parents said that they can often be dismissed and are provided a dismal overview of their child's future, with very little hope and positive outlook for the future.

Parent empowerment was described as related to being heard and valued for parent knowledge, closeness to the child with RD, and understanding important observations. The main issue for these parents is having credibility regarding their knowledge about the RD. Parents described these barriers to advocacy in an emotional way. Some felt disheartened, but it didn't seem any felt defeated. Most of the organizations described their reality and how it made them feel. As one parent shared their feeling of being dismissed, he said, "…we get told, basically, that we're not doctors, we're not scientists. So, nothing we say means anything." Another parent of in an RDAO pointed out the empowerment barrier among different stakeholders, and said, "That's a huge barrier and you see it in the various stakeholders in the medical community, not listening to patients."

Other organizations reiterate this frustration of being left off the consideration for important information related to their child with RD. Another parent said, "If you don't involve parents, you're missing so much." One of the mothers of an RD-afflicted child identified a key biomarker related to her son but was still not taken seriously. She shared her frustration by saying, "So, it's very frustrating as a parent to figure out, like hey, you know this is a physical biomarker are the hottest thing in medicine and I had to beg people for an ear because they don't take the parents seriously."

For a patient, or caregiver, this lack of empowerment also exists when dealing with an RD. Feeling unheard, dismissed or unrecognized as a credible voice to the RD space can be frustrating. Some organizations felt that the only voices worth hearing were the scientists or those in the medical community. This gap in recognition of the important role caregivers or parents can play engenders this lack of patient or parent empowerment when advocating for RD patients.

One RDAO interviewee described how being a part of an RDAO is beneficial, yet feels scientists are the only ones with credibility to talk about the RD, by quoting the medical professionals as saying,

> "Oh, we really want to hear from patient advocacy groups and we want to know what you guys think of various things when we decide whether to approve the treatment." But, again, there's more lip service being given than actuality so that's a struggle because, again, the FDA is all scientists and doctors and it's just a mental mindset that only scientists can tell them anything. They're working really hard to get past that but it's still an existing barrier to overcome. It's a barrier that I don't think that they're aware of, you know? Which makes it even harder. It's like they think they're listening, but the mindset is just so ingrained that you only listen to scientists.

Another patient advocate who also has an RD, described how doctors dismiss their feedback and knowledge, and the reality that these doctors may not treat many cases in the lifetime of their practice. She described this reality:

> And their doctors say, "Well there's nothing you can do. It's a middle age onset disease. You're not going to get anything until you're like 30 to 50 years old. And there's nothing you could do anyway because there's no cure." And it's up to the family to have to educate these

primary care providers and local practitioners that there's always something you can do. And that they're really being quite short-sighted. And quite frankly, most primary care providers will see maybe one case of our disease in their medical lifetime.

Other advocates described how their knowledge is brushed off or their condition or reality is pushed aside. One advocate said,

So, they don't know what it is and maybe know a little about it, have completely skewed ideas. But then on the other hand have this concept that, "Oh well there's doctors to take care of that and you guys have medicine," and almost brush you off, for lack of a better word. It's not really covered, or your very, very soft preconceived ideas are detrimental to somebody's life because they're completely wrong.

Another interviewee was describing how disheartened she was with her experience when told of her child's condition:

I think when families get diagnosed with a condition, it's a terminal, degenerative condition that has no treatment, it's such a shock to the natural order of things as we understand it, particularly, I would think, in the U.S. You go to the doctor, you get a diagnosis, and you get a treatment. Maybe the treatment, maybe it's not 100% effective, but there's something for you. You will be treated and cared for in some way. So much of what really the experience is in our disease situation is that families are told, "I'm sorry, your child has this horrible condition and there's nothing out there. Take them home and love them."

Her husband also added how the lack of proactive approaches for parents does not lead to empowerment, further confirming this feeling of being abandoned or pushed aside. He said,

> There's not even clinical guidelines. It's everything that's done for these kids is very reactive and not proactive. It's only after a certain complication happened that they cared to do anything. Trying to get physicians to be proactive about their care, because maybe there aren't clinical trials showing this or that is effective. It's just like you feel very neglected, I suppose, and pushed aside. You feel that your child is pushed aside. It's not intentional, but they simply don't know what to do so they don't do anything. Of course, my wife is a physician, but had she not been a physician and had parents said to us ... Or doctors, I'm sorry. Physicians or the genetics office said to us, "There's nothing you can do. Take your child home. Love them." I understand why a lot of parents do just what the doctors say. They take their child home. They love them. Not that there's anything wrong with that, but it's not exactly putting them in an empowering position to go out and try to make a difference. It's almost like you have to go against the doctor's orders to go out and try to make a difference.

Yet, the lack of patient empowerment has compelled others in RDAOs to ensure the next parent or patient doesn't have to endure the same heartache when dealing with diagnosis. One stakeholder said,

> I also think a lot of the parents out there that are getting diagnosed are still getting that same type of thing that, "Look, there's nothing going to be in time for your child." We know what we're doing. It's all crazy. It's paying it forward so future families don't have to go through ... We haven't even lived through the worst of it with our daughter. But we're likely going to, and once you live through a little bit of this you don't want any parent ever in the future to go through what your child has gone through. That's why we do it.

One other significant comment made in this list of codes is worth noting, and did not warrant multiple or similar responses, but one organization, said, "A barrier is that most don't

understand RD is a business, it's not just a passion. The lack of know how to run a business or nonprofit pose as a barrier to getting advocacy work done."

This observation and quotation are an important area of feedback as the rare disease advocate is on a journey, which has an emotional and connective human element. Yet, the notion that RDs need to be considered as a business is important to understand further, as drug development, seeking a cure, and funding research for example, require basic business skills in order to advance advocacy work to get this work accomplished.

Lastly, another important yet significant note made by one RDAO respondent in this list of codes, is the comment that: "There are many different RD groups within a large RD community. It's important to find common ground that all organizations can agree on, as all organizations want different things." This speaks to the idea of looking at RDs under a greater public health banner, in order to connect different RD organizations closer together to advocate under similar needs to gain greater advocacy impact.

Families, parents, and RDAOs have an important voice to be heard when it comes to understanding, caring for, and observing patients with rare diseases. To best address advocacy capacity, removing the barrier of *lack of parent empowerment* is an important consideration.

3. Facilitators to RD Advocacy

As described in the organizational capacity section in *genesis*, the barriers for RDAOs begin with the knowledge that an RD patient needs help and support. Thus, a facilitator to advocacy is the process of starting one's own 501c3. This idea emerged as a theme and answers the research question as to what facilitates RD advocacy. All 15 organizations provided

responses and over half provided examples of how they started their own nonprofit organization. Three themes emerged by frequency of quotations and similar responses. See Table XVII.

Facilitators to Achieving Advocacy Themes	# of RDAO Reporting	Frequency	Data Source
Facilitator to Advocacy Capacity Theme 1: Starting a 501c3 for RD advocacy	15	8	Stakeholder Interviews
Facilitator to Advocacy Capacity Theme 1: When faced with adversity, RD advocate is driven by personal determination and will to fight	15	8	Stakeholder Interviews
Facilitator to Advocacy Capacity Theme 1: Using Transferable Skills from Professional Experience and putting it to use to advance RD Advocacy	15	6	Stakeholder Interviews

TABLE XVII: FACILITATORS TO RD ADVOCACY THEMES

Facilitator Theme 1- Started own 501c3 for RD advocacy

All RDAOs that were interviewed were 501c3 organizations, representing their journey within different points of time, whether they were less than one year old to over 60 years old in existence. The creation of their nonprofit organization became a catalyst to operationalize their advocacy efforts. All fifteen organizations responded to this set of codes with a frequency of eight. Overall, the *facilitators to advocacy* code had 146 codes. See Table XVIII.

Facilitators to conducting RD Advocacy

RD Organization	Facilitators
1	29
2	24
3	7
4	8
5	8
6	7
7	6
8	14
9	6
10	7
11	8
12	6
13	7
14	8
15	1
Total	146

TABLE XVIII: FACILITATORS TO ADVOCACY SUMMARY OF CODES

One organization's respondent mentioned their initial focus after learning about their child's disease was to start a nonprofit organization:

> From there, my focus was first to become an established 501c3, so going through the paperwork and application process of that. And then from there, it was developing the scope of what our work would be, what I believe to be important, and then bringing in my skill set from the private sector to develop a way for us to bridge the gap between industry and health and nonprofits so that we could have a greater impact in our patients' lives.

The next step for most RD advocates is to start a nonprofit organization in order to address all their RD advocacy needs. It supports a way to raise funds and have an organization that has the capacity to address all needs on behalf of the patient. In the absence of resources, the creation of a nonprofit organization provides a one-stop resource for others and a source of action. Three

organizations described the need to start a nonprofit because nothing existed to support advocacy for their RD.

One respondent said,

> Alright. Our organization was founded by a woman whose husband had our disease, and she decided that she was going to do something about it because at that point back in the 1960s, there was no advocacy. There were no community services. There was no research really being done in the area of our disease. So, she was a very compelling kind of personality. And she used her influence and her dynamic personality to get Congress to supply the very first research into neurodegenerative diseases, which includes ours. So, she founded the organization in 1967. And over the last, this is her 50th year of providing services to our community. The organization had grown really from a small kitchen table group of individuals in a support-group like atmosphere to a national organization that has over 54 chapters and affiliates. 43 medical centers that have received a designation of centers of excellence.

Another organization's advocate was compelled to start their organization as a result of advice she received from a doctor:

> It's a disease that affects women in the prime of their life, so her daughter, I think, was like 22 at the time. So not a baby, like how we usually think of rare diseases. So, she contacted a pulmonologist, and it turned out that this pulmonologist ... Sorry, it's a lung disease. Contacted the pulmonologist and basically said, "What can I do?" He told her that the best thing to do would be to start a foundation. So, the pulmonologist and the mother together started the foundation. That was 22 years ago, and that pulmonologist has continued to this day to be our chief scientific officer and leads our scientific endeavors. Now, 22 years later,

we have ... There are over 2,000 known RD patients in the country, and about 3000 worldwide total. We've raised and invested over $23 million in RD research.

One interviewee said,

> There was one specific project that needed funding, millions of dollars in funding, and was one of the only things out there at the time that would've been a clinical trial that would've gotten up and running and been a chance for my daughter and many other kids. We created our 501c3 and just started fundraising. The lemonade stands, the galas, all of that stuff. We raised about $250,000 in about six months, which was good but wasn't, obviously as you know in rare disease research, that doesn't move the needle very far. We were fortunate in mid-2014. We had a video made, a pro bono video made, that went viral and raised about $2 million in eight months.

Funding advocacy priorities became a reality when the RDAOs created their own foundation or nonprofit organization. The creation of a 501c3 served as a catalyst for some of these RDAOs to grow their organization with staff, medical centers, fundraised dollars, clinical trials, and research.

Facilitator Theme 2: When Faced with Adversity, RD Advocate is Driven by Personal Determination and Will to Fight

The will of the human spirit is an important factor in breaking through obstacles and change. Most of the interviewees described their fighting spirit, advocating for treatments, research, or care they felt was needed for their child or patient. All 15 RDAOs responded with a frequency of 8. There is a determination among many of the RDAOs that status quo is unacceptable and thus advocating for better is the only direction they see. What is even more important in understanding RDAOs is that most cases reflect RD advocates fighting for their child's treatment

or cure. This determination is a unique aspect of this community. As with other patient advocates, most advocates are not the patients or directly affected family member. They may be health care professionals or someone in the medical profession. The majority of the RD advocates interviewed were directly related to the RD patient. Two of the respondents were patients themselves and advocating for their own health and the next generation of RD patients.

One organization described their organizational history and talked about why it was created:

> The RD Foundation was founded in 1955 by a group of parents whose children were, most of them were less than 10 years of age, who basically had been told their children probably would not live to be into their teens, or even on the younger ones, even into nursery school and they just couldn't accept that.

There is a sense of unwillingness to give up or accept a detrimental future for their children. This personality trait is often associated with those on the RD advocacy journey.

Another mother and advocate described her perseverance in convincing scientists for a year that her observation was more than a unique trait for her son, and she said,

> And so, this is something every parent remembers, every new parent I get, I ask them, "Did your child have a full mouth of teeth?" "Oh, they called him a piranha." "Oh, we called him a shark." "Oh, it was the only thing that wasn't developmentally delayed." So, it's clear ... easy biomarker can direct somebody, you know a genetic team to test for this one gene mutation. And it took me a year to convince scientists to scientifically investigate it. And it was "Oh, RD advocate, it's coincidence for those ten kids." And then we'd have 25 kids. "Oh, RD advocate, it's still a coincidence." Then when I had 40 kids, it's like "Maybe, but we looked at the models and it doesn't show anything."

Another RDAO talked about their fighting spirit, and how it defined themselves and their family: Just that if you are going to be advising rare disease organizations, how to get started with advocacy, I would just emphasize pick a topic that your community is passionate about. If you build it, they will come. And much of her understanding. My daughter was fortunate to get a gene therapy clinical trial. She got the low dose, the first safety dose of a clinical trial, which we were very thankful for, of course, but it was a safe dose, and it was the first dose, and it came after she was very progressed in this disease. But we still fight for her, just like we fight for all these children. That's us."

This was said with a sense of pride and great accolades for themselves.

A mother mentioned what it's like to have the drive of a rare disease diagnosis:

Just a couple months after our son's diagnosis, my husband and I both were sitting at our computer at two o'clock in the morning. We both looked at each and said we both like this is what our whole life was prepared to do. In other words, I think the ability to raise money for, you know, it was for profit by working in sales, by having marketing, so understanding how to communicate what it is we're doing and just by having the drive of a rare disease. I think all of that came together, which enabled us to hit the ground running.

Another respondent described how she felt when left with no action. She stepped up and, just kept moving ahead. She said,

I felt that there was emphasis on building the organization and infrastructure. We were just, I guess you could use the word scrappy. We just really cared less about building an organization and establishing infrastructure. We just really wanted to get research funded and get the drug pipeline going because it takes so long to develop a drug that we just wanted

things to get started. We felt like there is a lot of discussion about what to do and not enough action in terms of actually vetting the research, and taking a risk, and putting money into it.

A grandmother advocate described her unstoppable spirit to keep moving forward to get a diagnosis and emergency help for her granddaughter, saying,

> While we are advocating for a change in EMS protocols to protect all individuals with rare disease and chronic illness, we are very active in advocating for women with bleeding disorders because it took us four years and four doctors to get my granddaughter diagnosed and to get emergency help or factor for her. That's because we would not stop.

Lastly one other advocate went back to school to learn about science and biology to better understand his child's condition. He even changed his career and became immersed in everything he could do. He said,

> I went back to school, like you, and learned a little molecular biology, and then ended up getting a job. I work in biotech right now, I'm in the patient engagement program at X company therapeutics, which is a biotech that works on rare neurological disorders. That's what I do.

Many of these advocates proudly display their will to fight through barriers and take risks and chances on behalf of their child. This will to fight is a unique facilitator to staying on the journey and not giving up finding a therapy or cure for an RD patient or child. This may be a facilitator that sets them apart from other advocates, especially knowing the luxury of time does not exist as previously mentioned.

This helps answer the research question as to what facilitators exist for RD advocacy. In summary, the RDAO and their individual advocates play an important part in getting RD advocacy done with a spirit of perseverance, good will, and persistence.

Facilitator Theme 3 - Using Transferable Skills from Professional Experience toward RD Advocacy

RD advocacy is done by parents, volunteers or staff. A result that emerged from this area is how many of the advocates did not formally have training in RD advocacy, but discussed how their individual skills, personality, job experience and learning was a significant part of advancing RD advocacy efforts. 15 RDAOs talked about their skills and frequency for this section was slightly lower than others with only six. However, it was worth noting what RDAO advocates bring personally and professionally in lieu of having an opportunity to get formal training in RD advocacy.

As previously mentioned the RD community is a niche community and there is no curriculum for learning about RD advocacy. These results show what the individual brings to their advocacy work in the ways of their own personality, unique RD experiences, prior or current professional career skills and transferring them to aid in the current advocacy work they lead.

One organization described his current job experience and how he relates it to his advocacy work:

> I work for a distribution company for a large pizza chain. So, a lot of what I deal with in my normal job is bridging the gap between outside vendors into our current strategies and goals of the company. I also work with internal customers at my organization, where I have to bridge the gap between corporate, which is where I'm at, and our different distribution centers located throughout the United States and Canada. So, I'm bringing a lot of that skill set over, where I can work with outside people, be it research scientists or funders or any other third parties and help identify what their goals and strategies are and align them with what our goals and strategies are in our organization.

Several organizations shared how their experience and business skills helped support and facilitate the advocacy work they needed to accomplish in the following quotes. An advocate shared how her sales and marketing skills paid off:

> Sure. My son was diagnosed with RD in late 2002. I had a business background in sales and marketing and so I knew that I would want to be actively involved in raising money for research to find a cure. I looked to find another organization to work with, not ever expecting that I would actually start my own non-profit and I really didn't know anything about the non-profit world. I was disappointed with the sense of urgency that I saw was in the existing organizations, and also, we have just kind of a bias towards working with biotech companies because they found them to move much quicker and being just a lot easier to work with. For those reasons, we decided to start our own non-profit.

Another advocate also shared how owning her own business and consulting company helped her have the skills to do RD advocacy work:

I do business consulting. My background is in accounting and business. I have been a self-employed business owner for 23 years. It's just kind of funny. At 16 years old, I worked for a lawyer, which was one of the best things I ever did. Then I worked at some different accounting offices and then went into my own...created my own business doing business and accounting consulting. It has given me the ability to assess the situation, address the problem, and create a solution. We have greatly taken those business planning skills and just used them in the nonprofit world and advocacy world.

Similarly, a different organization discussed their knowledge in owning a business was applied toward RD advocacy, as she said,

> Because we really jumped into this with both feet and sometimes I'll laugh and say we jumped deep into the pool with concrete blocks tied to our feet. Absolutely although I had the business skills ...my husband and I owned our own construction company, so we were self-employed for years. His mother and father were self-employed owning a construction company before we did and then I had my own separate business. So that was a tremendous help because we knew just basic elbow grease. Just hard work and dogged determination will get you a long way. But to have had mentors to say just give me some idea, tell me some things not to waste my time on, any advice would have been amazing because we literally did not have a clue.

However, one advocate felt that despite all the business background and skills, the specific information on venture philanthropy and rare diseases was not a skill that is encountered with generalized business skills. She mentioned the importance of learning this skill and asking basic questions to continue learning:

> Even with my business background, it's not the business background that tells you about IT, and tech transfer, and even venture philanthropy and things like that. All of those are becoming more and more, in these rare diseases, where foundations, rightly so ... If someone's making big money on this, we want to get a piece of that, so we can put our donor dollars back into more research for our disease. Yeah, all of that is very interesting. I think more training and understanding of those types of things. Again, the way I do it now is I call Sally Smith from RD Organization and say, "Hey, you guys are big into venture philanthropy. Can you tell me how it works?" But I think formal types of trainings would be really helpful.

This last theme for facilitators of advocacy capacity described how RDAO advocates leverage their own unique strengths, training, backgrounds, and skills and apply it toward their RD advocacy work. Their talents are not wasted and are repurposed in areas where they can fully contribute. Personal experiences and working with others to build on these skills is important to gaining greater knowledge of how RDAOs maintain or advance advocacy capacity.

V: DISCUSSION

a. INTRODUCTION

The ability for RDAOs as nonprofit 501c3 organizations to prepare, anticipate, mobilize, and execute advocacy activities is critical to facilitate public health approaches to addressing rare disease including prevention messages, early screening and testing, and surveillance. This study examined the organizational capacity factors that influence RD advocacy, identified barriers and facilitators to RD advocacy, and described the characteristics of RDAOs who undertake advocacy work. Improved understanding and awareness of these factors may aid in guidance of RDAO advocacy strategies and foster improved engagement among the many stakeholders involved in RD advocacy.

Tables IV through VI represent the summary of findings. These findings were supported and confirmed by referencing publicly available information on websites as well as referencing literature to support the findings. This chapter provides a discussion of the study followed by a summary of the key findings, recommendations for an updated Conceptual Model, and application to Global Genes (GG) advocacy work, public health leadership implications, conclusion, and recommendations for future studies.

b. CONCEPTUAL FRAMEWORK

The overarching conceptual framework for this study was based on an adapted logic model by Coffman et al. called the Advocacy and Policy Change Composite Logic Model. This study found some similarities but important differences between the logic model and GG organizations. Similarities included organizational capacity, infrastructure, engagement and outreach, staffing but have differences related to advocacy capacity and improved systems and services. Distinct differences included the lack of inclusion for barriers or facilitators to advocacy work, taking into account the role of the RD advocate, as well as describing the characteristics of RDAOs. The organizational capacity factors suggested by this study

overlapped with two of the inputs highlighted in the Logic Model of Building Organizational Capacity and Impacts. See Figure 5. For example, strategy and business planning and infrastructure were identified but did not include consideration of additional contributors that were described in more detail in this study. Factors related to infrastructure were addressed in more detail by including age of the organization, type and number of staff, advisory boards, and type of work office. Strategy and business planning were described within the prioritization process data, but provided greater details related to current practices, structure of decision making, and examples of priorities for RD advocacy. Although the Advocacy and Policy Change Composite Model outlined important components for determining advocacy and capacity, it lacked considerations of the composition of the organization, the human elements of personal drivers of RD advocacy, and identification of barriers and facilitators along the way.

c. RDAO LEADERSHIP

In this study, RDAOs described an array of factors for organizational capacity related to addressing how to get RD advocacy work accomplished. See Tables IV-VI. All organizations that were interviewed demonstrated these factors, where a greater emphasis from most organizations was related to organizational infrastructure. Overall, findings indicate that a majority of RDAOs are executed by a person who may be related to the RD child or person and in most instances is a parent of a child with a RD. The lead of the RDAO also served multiple roles on behalf the organization in addition to having responsibilities for care of a child with RD, and at home. These same RDAOs are mostly run out of home offices, compared to a few advocates who were employees working in a RDAO headquarters office. Upon becoming a RDAO, most of the organizations main priority is funding research and raising money for RD activities such as clinical trials, research, or drug development. This may be an initial reaction and feel like a natural fit, but understanding the true upfront needs is worth further exploration.

Most of the organizations described a structured, more formal prioritization process related to identifying advocacy goals and projects within their RDAO. But the reality, based on interview data, was that they were managing multiple projects and responsibilities, and their process required them to be flexible, nimble and adaptable to change. The drive to find a cure or therapy for an RD, along with the family-run nature of these RDAOs, may overshadow their need and awareness of building organizational capacity. Formal training in running and maintaining an RDAO lacks and thus, leaves the RDAO to discover and learn along the way of their journey which entails helping their child or others with RDs.

d. KEY FINDINGS

This section summarizes the four key findings of the study which are highlighted in Tables IV-

1. Various types of engagement of people are critical to advancing RD advocacy

The greatest number of codes identified in the study were *engagement and outreach* and *gaining value from other people*. These areas were mentioned the greatest number of times by all 15 organizations interviewed. Engagement and outreach were defined as connecting with people who have similar RD or stakeholders of the RD in order to inform, educate, or connect within a specific RD community. Gaining value from other people for RD advocacy in ways related to mentoring, networking, partnerships and using third party technical support. Taken together, this represents the important role people serve both internally and externally to RD advocacy. This is important for several reasons. First, with the lack of basic resources available to the RDAOs and individual RDs, advancing advocacy activities is best done with and through people. In the absence of research, experts, funding, therapy, or a cure, advocacy that requires multiple steps is advanced through people. By partnering, leveraging mentors or experts, building networks, and seeking greater exposure or support from other people or experts, organizations acquired deeper learning, connections and problem solving.

Second, this study also shed light on the importance of telling the RD story in order to increase awareness and engagement amongst key people who could advance RD advocacy work. In today's global reach that is accomplished through various social media and similar technologies where this community can find and connect with like members, volunteers, and staff through the Internet, or other online tools and social media such as Facebook, Twitter, private chat rooms, or online forums. This was evidenced by several examples given during interviews.

Organizations described n=15 Facebook users as both a public and private platform. Facebook was mentioned so frequently that it was recorded as a subtheme. Key learnings arose related to how Facebook was used by the RDAO for advocacy purposes. One method was to use Facebook publicly, as a platform to identify with others with the same disease, build a community, educate, post events, and build an online community. Other methods included creating additional private or closed Facebook pages to connect with an RD community to talk about more private issues, like care, feelings of isolation, a way to connect often, and to talk about issues patients are experiencing with medicine, diagnosis, care, or trying to find expert medical care. One respondent described followers as sufferers and caretakers of those with a particular RD. Another approach for Facebook that was described was managing multiple pages, separated by roles such as patients, volunteers, or staff, and the public RD community. In all instances, Facebook was updated frequently and cross-referenced with other social media to expand reach and spread content and messages to wider audiences on platforms like Twitter, LinkedIn, or Instagram.

Four of the responses explained the value of social media to their RDAO. These reasons included that social media is easy to use, supports patients who are immobile or have disabilities,

allows for continuity and isn't limited by geographical boundaries to find others with similar interests. Other respondents described how social media is important to identify others with the same disease locally and globally, with the ability to conduct targeted outreach. Providing direct information for events and social gatherings is helpful to directly communicate to the RD community. Dunkel et al from NORD wrote a chapter on RD advocacy, underscoring the importance of knowing target audiences. They support the idea that the essence of effective advocacy is to call out the critical issues identified by the constituent community followed by advocating for policies and programs that address those issues (Dunkle M, 2010).

On the contrary, one organization's respondent mentioned that social media is not always the answer. In her area of providing services, she found that not all potential members have access to the Internet, which may be the result of costs for services, cultural issues, affordability and accessibility related challenges. The need to connect and find support, specifically in RD communities, has led to an increasing number of patients and caregivers turning to social media platforms for its valuable insights on their disease. Some of these known healthcare platforms in the RD community include RAREConnect (Rare Connect, 2019), which is a rare disease discussion group supported by closed access online communities like Inspire (Inspire, 2019) and Smart Patients (Smart Patients, n.d.), Facebook groups, and publicly available disease specific discussion boards. In these various platforms, patients can participate in what this research uncovered: they can share their experiences, important information, provide support, advice or guidance.

Researchers have also recognized the potential for these social platforms in helping orphan drug research and development (Merinopoulou, 2019). See Figure 10.

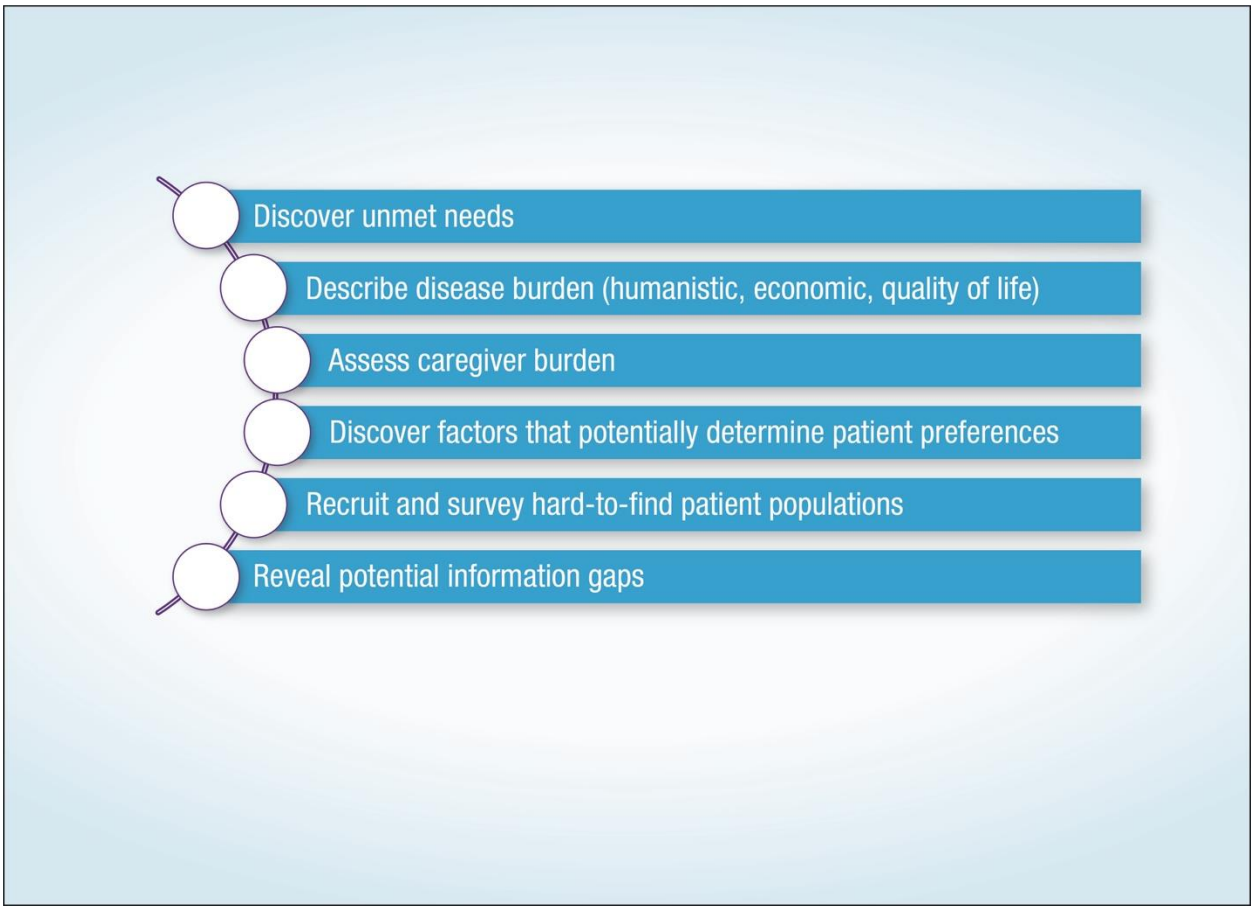


Figure 10: Benefit of social platforms to helping Orphan Drug research and development

These findings may help GG in their future planning and placing an emphasis on their planning efforts as it relates to how RDAOs can best achieve their greatest potential through more opportunities to engage with people, drive advocacy activities and advocate for patients with RDs.

For communities that are hard to build or attract, social media is proving to be an important engagement tool for both researchers and those involved in RD advocacy.

2. The practice of RD advocacy is not a linear process.

This study provided an opportunity to document how RDAOs in this community support and build their own organizations. A key finding was that the process for RD advocacy is not linear.

The Advocacy and Policy Conceptual Model (See Figure 3) suggested that to build an advocacy strategy, one can work from one end of the model to the end, with the end resulting in valuable outcomes for a person with an RD such as a treatment or cure. However, in conducting interviews with the organizations in this study, that was not the case. RDAOs were run mostly by family members who may have been parents, or a relative who was also juggling several responsibilities. The unique role that leaders in RDAOs have is trying to run a 501c3 nonprofit organization while dealing with the emotional, physical, and social demands of caring for their family member with an RD. This process is not linear in that advancement in one area of RD advocacy leads to the next advancement that is closer to obtaining a cure. For example, and RD advocate may advance scientific research with a drug company, but have to juggle managing and administering a non-profit organization at the same time. While all that is taking place, they may have to go home to care for their family or child with RD. Yet, despite all this, RDAOs continue to advance their work and try to keep moving forward.

One major example of the complexity of pursuing advocacy work, often from scratch, while caring for a family member with an RD, was understanding the RDAOs prioritization processes – the process by which they define and tackle advocacy goals. While many described a more formal process, what was really happening in their RDAO was a different story evidenced by further descriptions of additional extraneous workplans that were not identified in approved strategic plans. In fact, RDAO representatives described handling multiple priorities that could change and veer away from their original plan. This leads to the organization recognizing important roles and bouncing between various dimensions that address the needs of the individual with RD, organizational capacity, advocacy capacity, and the ability to connect all dimensions to the overall world of RDs.

3. Barriers and Facilitators to RD Advocacy that were identified also aligned with Organizational Capacity Factors

The organizational capacity factors represented ways that RDAOs can get their advocacy work done. The barriers were identified as fundraising, lack of a prioritization process, and the need for skilled staff and true RD advocates.

Fundraising

Fundraising is fundamental to the existence of RDAOs because most RDAOs are non-profit organizations or foundations. In addition to the advocacy provided by non-profits or foundations, fundraising for RD research and care is just as critical. In a survey conducted by Chat et al, funding was universally viewed as the greatest obstacle to meeting patient needs (Chang, 2007). RD advocacy needs financial backing and depending on the priority of the organization, fundraising may also support clinical trials, recruiting, registries, access to journal articles and other events or activities to keep the organization active. Fundraising was defined as the need to secure additional money. Nine out of 15 organizations mentioned the importance of fundraising. One mother described the need to raise a couple million dollars for a clinical trial. Another organization described the need for access to RD specific drugs, which in some cases can be very expensive. One recent drug has been made available, but at a cost of $300,000 per year. Most of the respondents talked about a vicious cycle of need for funds and fundraising. While budgets and operating costs may be lean, access to capital helps fund part-time or full-time staff. Sometimes dedicated staff is needed to fundraise, but it can be difficult if funds are not immediately available to advance advocacy work.

Lack of Prioritization Process

Almost half of the organizations mentioned another barrier as the lack of a prioritization process, defined in this study as identifying the need for strategic and business planning. While

most described their thoughts of a disciplined and structured prioritization process, in speaking to respondents further, they did not have simple and focused priorities of focus, and many issues were being managed at any given time. One organization admitted to having a strategic plan with advocacy as central to their plan. But admittedly, this organization confessed this plan looks best on paper compared to what is actually taking place. Another reason for this lack of prioritization, is that most of the work is done by the patient advocate. The patient advocate for most organizations was also a parent, a caretaker, a non-profit executive director, and accountant for the organization. One organization described this process as having all responsibilities falling on one person without anyone left to delegate the work to. Another interviewee talked about their want to prioritize but that they may not always prioritize the right things, thus leaving areas of work that may be higher value to be overlooked or put lower on the list of action items. Another RDAO described a more formal planning process, but in their Board meeting, mentioned that they cannot be everywhere, doing everything, and being everything to everyone. Overall, most organizations are stretched thin and constantly finding ways to juggle all their advocacy work in addition to caring for a child or person with RD.

Need for Skilled Staff & True Advocates

Another key barrier that was identified as an obstacle to RD advocacy is the need for skilled staff in RD advocacy and ones who stand to be true advocates. More than half (n=9) of the respondents were advocates that were also a family member, parent, or person with an RD. Over half of the respondents in this study elevated this as a barrier with a frequency of 19. This was the highest number of mentions in the transcripts. As one organization put it, it is about having the right staff with right expertise within the right capacity of the organization. Skilled staff also applied to respondents' comments about the need for greater leadership and staff

development to help empower the staff. One organization had a deliberate focus to allow their staff to make decisions on their own, rather than wait for board approval. This approach led to stronger advocacy engagement and ownership in success.

On the contrary to skilled staff, one RD advocate emphasized the need for professionalism and that advocates need to remember to have compassion. Staff who encounter the RD community oftentimes may work with those who have serious ailments or are sick daily. Sometimes one can be taught to be a successful advocate, yet on the other hand, advocacy also requires one to have empathy, patience, and the ability to recognize the needs of others rather than solely focusing on getting advocacy work accomplished.

Facilitators

Facilitators to advocacy were ways that respondents created their non-profit organization and catalyzed their efforts to operationalize their advocacy agenda. This section had a similar overlap with the Genesis section related to why the organization was started. This section had a total of 146 codes and all 15 organizations provided responses to 3 key facilitators which were 1) starting a 501c3 for RD advocacy, 2) RD advocate driven by personal determination and 3) using transferable skills from professional experiences and applying it to RD advocacy. As such, 15 out of 15 respondents identified a main facilitator for advancing RD advocacy is starting a 501c3 nonprofit organization. This finding also aligns with the genesis findings in the organizational capacity factors. The challenge of building a community was also identified as a barrier, yet this important characteristic describes RDAOs who are in the process of undertaking RD advocacy work. This is another alignment of where the factor is described to be lacking, but when it is present, serves as a positive facilitator to RD advocacy.

Personal Determination

One hundred percent of organizations interviewed also discussed how RD advocates are driven by a personal determination and will to fight. Much of the evidence provided describes the RD advocate as having a fighting spirit, advocating for treatments, research or care needed for the RD child or patient. Many of these advocates also did not accept status quo and seemed to always find ways to push though barriers to help the RD community. This seemed to be relevant particularly because over half of the respondents were also a family member of parents.

This instinct to protect their family and fight for another day was transparent in many voices, and descriptions of how these RDAOs have advanced their agendas. One organization's respondent also had an immense focus to fund research and found herself longing to support a drug pipeline to support her son. She met with anyone who would listen, called anyone who would stay to hear her plea and found out as much information as she could until she was able to set up a clinical trial for her child. Interviewees with the RDAOs in this study left an immediate impression by the way they talked about their work with conviction describing how they never gave up, and always sought the help and support of others. Key characteristics of these advocates emerged as having traits of compassion, genuineness, and resiliency. These are identified and described as core elements for RD advocates. Most likely RD advocates are personally connected to the RD, and their passion, commitment, and belief in this work compels them to be driven and dedicated to their work because it is part of their family.

In an essay by Ayme et al, the authors discuss the notion that patients with RDs and their underlying organizations are among the groups that the most empowered due to their fight for being recognized and better health care. These organizations have paved a way for a new approach to addressing the gap between public research, which did not look at demands and

expectations, and market driven research, which limits research to organizations who are making enough money to provide a justification for private investments. They sum out their paper by saying patient organizations have an active and crucial role in creating research policies and projects on behalf of RDs. Therefore, patients are directly impacting their future (Ayme, 2008)

A study conducted in Australia by Pinto et al, showed similar perceived challenges. Their study showed insufficient funding and consequent limitations of organizational capacity as factors most frequently cited by survey respondents. This was a prominent theme in interviews with RD patient organizations (Pinto D. M., 2016; Valdez R. G., 2016)

e. IMPLICATIONS FOR THE PUBLIC HEALTH PROFESSIONALS

RDAOs are an important consideration in public health and can easily get dismissed due to its categorical name as rare diseases. RD may have low numbers of people per disease, but overall, RDs represent 25-30 million people globally. Understanding more about how RDAOs interact and build organizational capacity to advance advocacy is helpful not only to umbrella advocacy organizations such as GG, but also to the various stakeholders affiliated with supporting rare diseases. PH professionals can help translate the important contributions of RDAOs to PH as it relates to PH policy, legislation, research, and advocacy. Furthermore, PH professionals can play an important role in advocating for greater recognition of RDAOs as a minority population that deserves and requires health equity relative to health care, access, affordability and greater PH surveillance practices to further understand RDAOs role in PH in the future.

Most recently, this public health and rare disease connection was also published in 2016 by Valdez et al. who outline the need for a next generation public health response to RDs (Valdez R. G., 2016). A public health response to rare disease requires a framework to work in a seamless way. A framework was proposed and contained nine elements that can be distilled into

five overarching components: (i) assessment of burden—numbers of affected individuals, health outcomes, quality of life, health-care use, and economic costs; (ii) research on preventable causes and effective treatments; (iii) systems for screening and early identification; (iv) empowerment and education of people with rare diseases, families, and health-care providers; and (v) public policies to promote access to services and treatments for people with rare diseases (Walter, 2016) (Valdez R. G., 2016). The authors stated that the target of this public health framework should be about rare diseases or elements of their sequelae that can be prevented through population interventions. But, in many cases related to several genetic diseases, primary prevention is not always available for rare diseases; thus, secondary prevention is another critical component in the framework.

The public health leaders of tomorrow can recognize the future of this work and understand the connection between public health and RDs.

f. NEW CONCEPTUAL MODEL: RECOGNIZING THE MULTIPLE DIMENSIONS OF RD ADVOCACY TO MAXIMIZE ORGANIZATIONAL CAPACITY

As a result of this study and newly updated Conceptual Model was created based on results from the interviewees. See Figure 11. The first model was adapted to address RD advocacy based on the literature about RDAOs and their roles in advocacy. See Figure 5. It was customized to address RD advocacy specifically whereas the original Advocacy and Policy Composite Logic Model was intended for advocacy and policy work in general or unspecified. See Figure 3.

This study identified some gaps and new dimensions for RD advocacy. A gap in the advocacy and policy conceptual model is recognizing the human element of personal drivers of RD advocacy, and anticipated barriers and facilitators is missing. A model unique to RDs is a

helpful tool to customarily tailor an approach for RD advocates to better understand what's needed to build better advocacy and organizational capacity. Figure 11 represents the revised model.

The first dimension of this new version of the RD Advocacy Conceptual Model is important to recognize the needs of the child with RD and the multiple needs for these children where financial, human, and scientific based resources are scarce. This was evidenced by the data and themes in the genesis section of the results.

Following the individual needs, represents the need to recognize the organizational capacity factors to reach the greatest potential of the organization operating as a 501c3 nonprofit. These factors were identified in Tables IV-VI.

Third, achieving the best ability to conduct RD advocacy is captured in the advocacy capacity dimension. This represents RD specific needs to address facilitators to RD advocacy to continue advancing priorities for the RD. This was informed by the emerging barriers and facilitators in the results section. Also see Tables IV-VI.

The outer dimension or ring also poses as an opportunity for PH education, and advancement for both RD community, its stakeholders and public health leaders to correct the misnomer that RDs affect only a small segment of the population. This is informed by the literature review and background to RD advocacy.

The multiple arrows imply how an advocate within an RDAO may bounce between dimensions in order to get advocacy work completed.

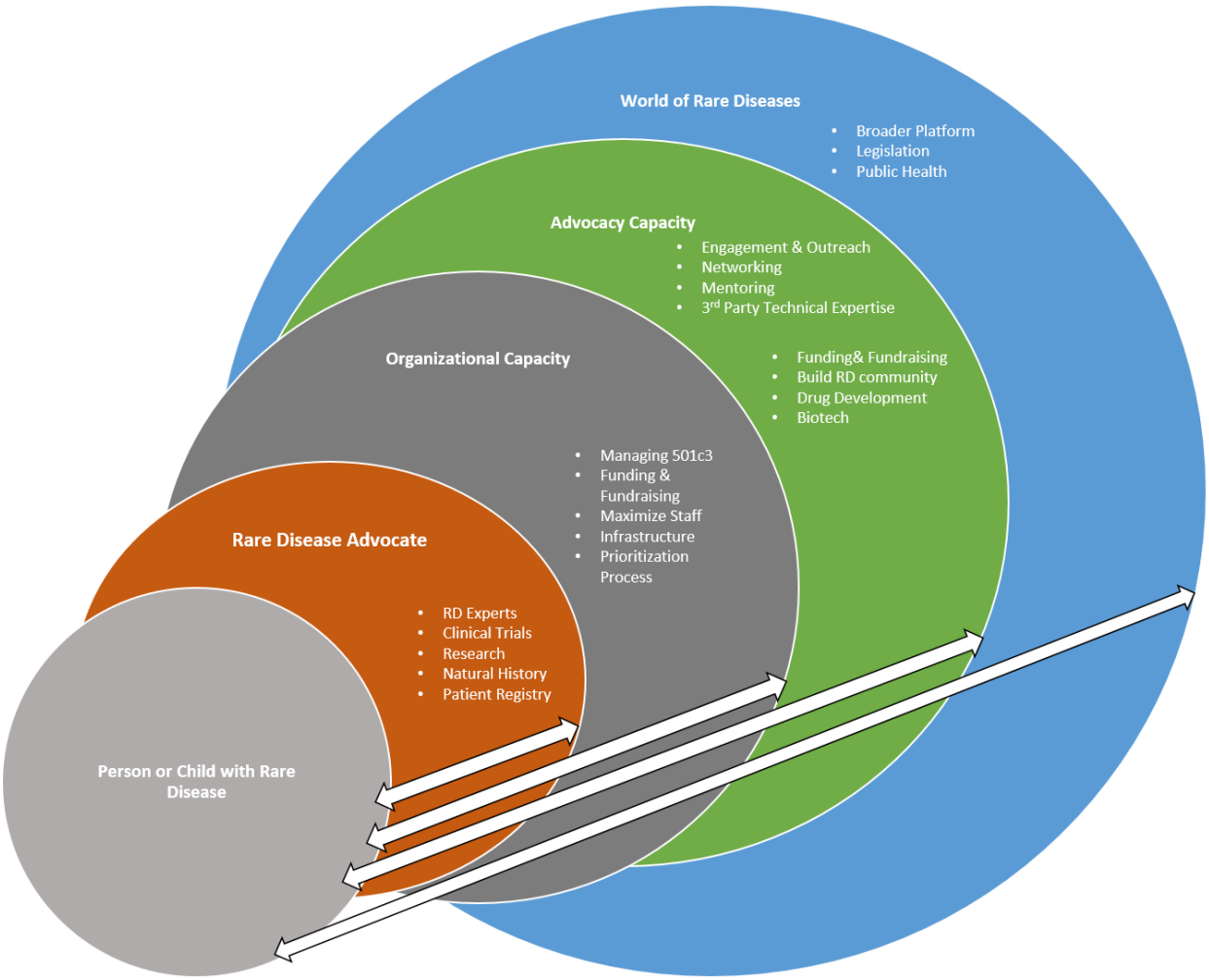


Figure 11. Conceptual Model of Rare Disease Advocacy

Another model that outlines the Stages of RD Advocacy may be helpful to GG and other umbrella RD support organizations in order to directly address organizations. The benefit of this approach is to reach them before they overextend their time and energy on multiple streams of work versus having a targeted, focus RDAO organizational capacity plan.

While applying this thinking to GG strategy, another adaptation of a model was created to consider the RDAO individual goals and their journey to promote customized RDAO support. The individual intake data form represents ways to track, assess and catalog RDAO co-identified immediate and long terms needs. See Figure 12.

The conceptual model was created using evidence from semi-structured interviews in this study. As most of the organization's respondents (n=9) wore multiple hats in terms of serving as a parent or family member, caretaker, executive director of a non-profit organization, or needing to understanding relevant legislation or find more RD patients outside the U.S., this roadmap represents the multiple roles over half the respondents described. Non-parents, or family members were employees working within a non-profit organization, who also described juggling many roles as evidenced by feedback in the barriers code that highlighted the lack of prioritization processes, leaving them to move between several priorities. Their roles also were described in the barrier section. For example, unknown or lack of RD-specific resources, leads advocates to lack references or experts. Therefore, moving between multiple dimensions is required out of the fundamental gaps in rare disease advocacy. This includes how several stakeholders described a lack of basic information, lack of formed communities, RD clinical specific guidelines, or access to experts within a medical system.

Social media emerged as a major characteristic of RDAOs related to engagement and outreach. Two themes were identified explaining how social media is important to RD advocacy as well as a greater usage of a specific social media platform, called Facebook. Social media was described an important tool to connect with others, notify RD communities about events, as well as connect with other RD patients in the U.S. and globally. See Figure 12. As a result of organizations talking about the importance of social media in multiple dimensions of RD advocacy, this skill building and application of knowledge spans across three dimensions, called maximizing organizational capacity, maximizing advocacy capacity and navigating the RD world. As some mentioned, Facebook can be used to communicate directly with staff and volunteers. This supports organizational capacity. Due to the volume of information and reach

that can take place by cross-referencing various social media platforms, this can aid in maximizing advocacy capacity by providing targeted outreach, multiple avenues of communication and ways to address advocacy needs. This can be in the form of posting events related to fundraising, the ability to collect donated money online as well as searching for other patients with the same RD. Lastly, as some organizations mentioned, social media allows RD patients with the ability to find other rare cases that are similar to their own, that may live outside the U.S. For those who do not have many members in the U.S., their RD community is global through the capability of international connections via social media sites. Other members may find each other through today's RD-specific social media, which has contributed to patient empowerment.

This roadmap would be beneficial for new RDAOs introduced to Global Genes. It serves as a guide for discussion with the patient engagement manager by allowing both parties to understand various dimensions of conducting RD advocacy. The RD advocate's status on the roadmap can be captured in intake data, which would include the name of the organization, how long it's been a 501c3, when the intake data was initiated, aspirational goals of the RDAO, along with which educational materials were provided to the RDAO by Global Genes. Lastly, there is an open section for notes to track needs based on the dimension of the RDAO and specific needs to facilitate, strengthen or enhance current advocacy work. This roadmap provides a way for newly introduced RDAOs to understand their current place in RD advocacy as well as understand the various areas of work that may lie ahead. By having aspirational goals, the RDAO can set realistic, timebound and pragmatic goals. As indicated in the Genesis section of coding, when lack of prioritization was identified as a key barrier, the roadmap can help focus in on an area of advocacy.

For the future, it may be helpful for Global Genes to host an introductory session walking new organizations through the roadmap. Additionally, focused seminars covering each dimension would be another way for a participant to learn about a specific dimension and gain the tools and resources to accomplish the RDAO goals.

A list of suggested materials to share with other organizations include:

a.) Manuscript(s) of published research

b.) Global Genes' contact information and website

c.) Draft of RD Advocacy Roadmap

d.) PowerPoint Presentation highlighting findings from this study, and overall study details

e.) Contact information of primary researcher

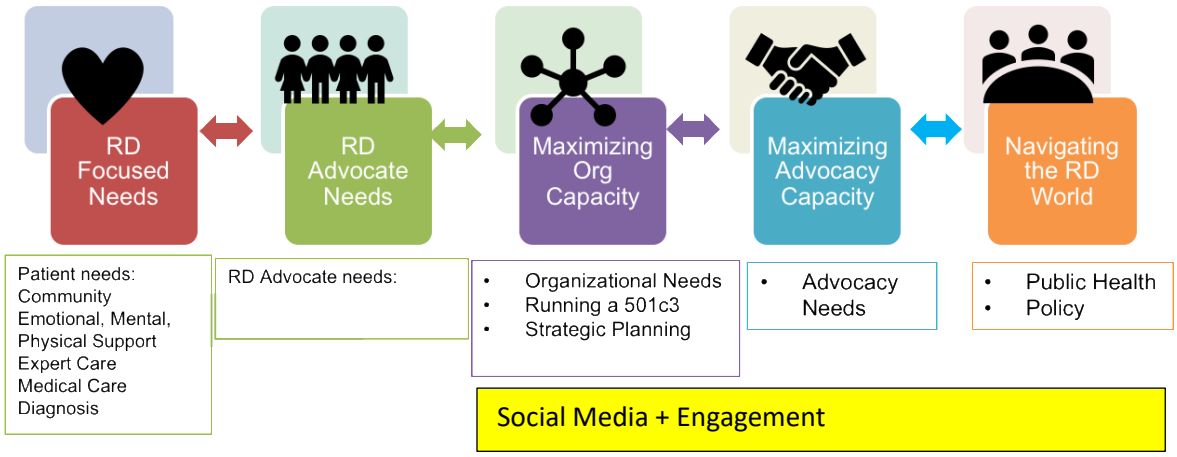


Figure 12. Rare Disease Advocacy Capacity roadmap and RDAO intake data

g. STUDY STRENGTHS AND LIMITATIONS

This study had some limitations. While it provided perspectives from a wide variety of RDAOs, it is small in scope, as a purposive sampling approach was used. Unlike quantitative studies, random selection for study samples is often used to determine who will be selected in the research study population.

Capacity criteria of low, medium, and high advocacy capacity organizations were based on subjective judgement of GG's patient engagement team. It was difficult to measure organizational capacity based on loose criteria for evaluation and cross case comparison. Moreover, it would be difficult to compare with other studies in this same area of study. Another

key consideration in conducting qualitative research, it may be possible that the PI unconsciously influenced results through the way interviews were conducted and analyzed. Additionally, findings may not be generalizable to just the RDAOs in this sample.

Not all RDAOs are uniform or focused solely on one disease or geography. Some of the RDAOs focus for advocacy varies from supporting a specific RD, or a family of RDs. Since most of these organizations may be working from a home office or virtually, geographic location may be difficult assess. Possible variables for consideration in evaluation include domestic versus international differences. With the use of social media and the Internet, and relatively small populations, it may be difficult to distinguish differences based on geographic location, cultural practices, policy implications, current legislation, and how RDs are funded through a national health care structure or other type of mechanism. More research including a broader range of actors, use of mixed methods, and different organizational capacity scales are needed to develop organizational capacity evaluations that can be tracked, measured, and compared across similar studies.

Confirmation bias is another limitation to consider given the researcher's history in working directly in the rare diseases area and personal investment. Confirmation bias, according to R. Nickerson, refers to the unwitting selectivity in the acquisition and use of evidence. Nickerson states that there is evidence to support the view that once a person has taken a position on an issue, one's primary purpose becomes that of defending or justifying that position (Nickerson, 1998). The researcher may have looked for what was wanting to be seen or found. To try and address this, a second coder was used. To test findings, alternative, contrasting or outliers of evidence were identified, and to confirm what was appearing as themes or patterns, member checking was also conducted in Phase II of research.

Construct validity is another concept for consideration in limitations of this study. This refers to the degree to which inferences can legitimately be made in the study compared to theoretical references (Web Center for Social Reserach Methods, 2019). This limitation was addressed through using the peer reviewed and gray literature a-priori to support the constructs explored in this study.

h. CONCLUSIONS

Greater awareness and understanding how RDAOs build organizational capacity are important not only for RD communities but for all the stakeholders, actors and people who play supportive and lead roles. This study's examination of the factors that contributed to organizational capacity can help organizations, researchers, governmental officials, pharmaceutical and biotech companies, RDAOs, and others identify priorities for investigating organizational capacity vulnerabilities and possible strategies to improve advocacy practices in the face of working in RD advocacy that oftentimes has sparse and lacks unavailable resources.

RDAOs are an important population to continue studying and supporting due to their impact and contributions to public health, and to also address future RD impacts to public health such as access to health care, costs, and healthy equity related issues. They have the potential to greatly influence healthcare and will continue to need care, dedicated experts, understanding, and support. RDAOs should be considered in health policy and legislation to remain an important part of the future public health agendas.

As Chang et al concluded in their paper in their editorial in 2007 (Chang, 2007), advocacy as a unified group, of 25 million individuals with rare conditions, would be a powerful and potentially more successful voice. This approach makes sense specifically because the issues of RD patients included the need for universal health care, subsidies or payments for expensive

therapies, drugs or medical devices, and access to specialists throughout the course of the disease.

Further understanding how RDAOs do advocacy, and who is behind the organization, is helpful to gain a greater understanding about the characteristics of these organizations. By also recognizing the barriers and facilitators to achieve RD advocacy, multiple stakeholders, along with the RD community, can understand how to collaborate, support one another and advocate from a public health perspective. This may eventually lead to effective and recognized RD policy, or legislation to further support these RD communities and RDAOs.

i. RECOMMENDATIONS FOR FUTURE STUDIES

This study represents a segment of time. Furthering this work to continue extend the study to include an examination of the proposed model and application to an organization like Global Genes would be worth exploring to test feasibility of the revised conceptual model and proposed individual intake data approach to see if feasible, useful and beneficial to the organization.

In addition, the use of this conceptual model and application to advocacy strategies as it relates to advancing or cultivating rare disease policy or legislation would also be of interest.

Potential Future Research Questions for Continuing Research:

1. Is organizational capacity improved when applying the revised conceptual framework? Why or why not?
2. Does applying an individual advocacy workplan and strategy effective for RDAOs? What do they gain, or what do the miss by applying this approach?
3. How has individualized support enhance or deter from GG advocacy strategies?
4. Did the conceptual model support or accelerate activities that lead to rare disease policy or legislation? Why or why not?

APPENDICES

APPENDIX 1: Research Protocol

"Identifying Characteristics of Rare Disease Advocacy Organizations and the Factors that Influence their Advocacy Capacity"

Research Protocol: Version 1
June 21, 2017

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Background and Significance:

In the United states, a rare disease is defined as a condition that affects fewer than 200,000 people. This definition was created by Congress in the Orphan Drug Act of 1983. Rare diseases became known as orphan diseases because drug companies were not interested in adopting them to develop treatments. This definition was needed to establish which conditions would qualify for the new incentive programs provided in the Orphan Drug Act of 1983[1].

There may be as many as 7,000 rare diseases. The total number of Americans living with a rare disease is estimated between 25-30 million and 30 million in Europe and 400 million worldwide[2]. In the United States, only a few types of rare diseases are tracked when a person is diagnosed. These include certain infectious diseases, birth defects, and cancers. It also includes the diseases on state newborn screening tests. Because most rare diseases are not tracked, it is hard to determine the exact number of rare diseases or how many people are affected[1].

There are compelling reasons to apply a public health approach to rare diseases[3]. The 3 core functions of public health are assurance, assessment and public policy[4]. To support these functions, it is important that public health research builds the knowledge base and identifies strategies to achieve health promotion and disease reduction. Advocacy uses these research findings to create new public policies to improve health outcomes. Advocacy is a critical part of public health practice, and vital to carry out these functions[5]. Moreover, advocacy organizations for rare diseases play a unique role given the lack of strong funding sources[6].

Helping to address challenges within the rare disease community are patient advocacy groups. Advocacy groups play an important role in cross collaboration amongst other public and private groups such as government agencies, commercial companies, academic institutions and investigators. In fact, they are recognized as playing an essential role in the process that includes an integrated national strategy to accelerate research and product development in rare diseases[6]. Effective advocacy is to identify the issues that are of importance to the constituent community, and then to advocate policies and programs that address those issues[5]. The early initiatives of the Cystic Fibrosis Foundation and the Committee to Combat Huntington's Disease helped begin the start of an increasing number of patient advocacy groups getting actively involved in research. The advocacy groups served in ways that helped create innovative models for funding and organizing research and product development[6].

The emphases of advocacy groups vary, depending in part due to the state of the science within various disease areas, and may also depend on other factors which may include the number of affected individuals, the interests and skills of organizational founders and leaders, and the success of fundraising strategies. If researchers haven't identified the genetic or other cause of a condition, or delineated how the disease develops, a group may focus its grants and other activities on closing these gaps in knowledge[7]. Approximately 50% of rare diseases do not have a disease specific foundation[8]. Advocacy amongst rare disease organizations is critical to better understand challenges and successes in order to consider a public health approach in the future. One author believes that independent advocacy by individual groups dilutes the potential political influence. However, in aggregate, these rare disease groups could be a powerful voice to shape the design and reimbursement of health services, research, and social policy[9].

The role of advocacy groups is an increasingly important support to patients with rare diseases. Often, these groups are started by the patients or family members themselves. They may lack the skills in

operating a 501c3 organization, let alone advanced training or skills to drive advocacy or policy. Little examination or analysis has been done to build the internal capacity of non-profit advocacy organizations. Most analysis has been focused almost exclusively on staff skills to carry out work, as opposed to broader concepts that define the critically necessary leadership, management and operations to make an effective advocacy organization[10]. The researcher's purpose for this work is to best describe the characteristics of rare disease advocacy organizations to better inform the rare disease community, and especially to bring awareness to key collaborators that work with rare diseases advocacy groups. After all, advocacy is often achieved through coalitions, each member of which, differs in their objectives[11]. Little is known about the application of advocacy as a key strategy in rare disease organizations, how it is defined, what activities are considered advocacy activities, and the perception of its role in achieving outcomes. Further, by identifying what capacity is needed to undertake advocacy activities, this knowledge may inform best practice for the rare disease advocacy community and ultimately improve advocacy approaches at the population health level.

Purpose

There are two purposes of this study. One is to describe and create a greater understanding of the characteristics of capacity and capacity building strategies used for advocacy efforts in rare disease organizations who advocate on behalf of patients and families who have a single rare disease. The second purpose is to identify facilitators and barriers to undertaking advocacy activities. This study may provide information that could further develop capacity building programs.

Objectives and Evaluation Questions:

This study has two primary objectives: 1.) To understand the characteristics of capacity and capacity building strategies for rare disease advocacy organizations 2.) To identify factors that influence advocacy capacity and capacity building activities for rare disease organizations.

To meet these objectives, there is one main research and 2 primary research questions.

Main Research Question:

What are the factors that influence advocacy capacity for rare disease organizations?

Two Primary Research Questions:

1.) What are the characteristics of rare disease organizations that undertake advocacy activities?

2.) How have these factors facilitated or presented as barriers to the organization's ability to conduct advocacy on behalf of rare disease patients?

Research Design:

The research design is action research, using qualitative methodology.

Data Collection: There are three sources of data that will be collected for this study: A document review of gray and peer-reviewed literature; Semi-structured interviews, and organizational data that is available at the organization-specific rare disease website.

Data Sources:

1. **Document review: Research materials will include a literature search and review of websites, reports, and/or policy briefs that discuss advocacy activities among rare disease advocacy organizations along with any identified resources used in training or skill building. No**

identifiers will be noted. Data will be analyzed for major emergent themes and will be compared to themes and constructs found in the literature. Please note all the information is available in the public domain and can be easily be accessed by conducting a Google search.

2. **Phase I- Semi-structured interviews: Research materials will consist of digital audio recordings of the organizational representatives' verbal response to the semi-structured interviews and verbatim transcriptions of these recordings (electronic and paper copies). Interviews will be conducted by the main researcher. It is anticipated that a total of up to 30 organizations with no more than two organizational representatives may be interviewed, with each interview lasting approximately 60 minutes. Organizations will include the following:**
    - **a.** Up to 10 disease specific rare disease organizations that are highly involved with advocacy.
    - **b.** Up to 10 disease specific rare disease organizations that are somewhat involved with advocacy.
    - **c.** Up to 10 disease specific rare disease organizations that are least involved with advocacy.

3. **Phase II- Facilitated discussion: Researcher will present de-identified findings to a group of no more than 15 Global Genes staff members**.

Recruitment Process and Procedures:
The recruitment process for participating in an interview will follow the same three step process: review, recruit/screen, and assign. All individuals who participate in interviews, as well as other key stakeholders identified by interviewees from their own or other organizations, will be invited to participate in the facilitated discussions. Participation will be voluntary; and no notes will be recorded for individuals who do not provide consent.

1. **Review:** An initial list purposive sample of up to 30 organizations and their contact information will be generated by Global Genes staff who have previous experience and contact with organizational partners within different membership opportunities tied to Global Genes.

2. **Recruit/Screen**:

    a) The researcher will contact the preliminary list of potential study participants and use the recruitment/screening script to guide the conversation. The recruitment script contains screening questions to ensure that eligibility criteria are met and that individuals are offered the correct category for participation. Participants must be age 18 and over, an organizational representative, and who have at least two years of experience in the organization. It is anticipated that all participants will be English-speaking and knowledgeable about rare diseases. Participation is voluntary.
    b) If an individual meets the study eligibility criteria, the researcher will invite them to participate in an interview. In either case, the researcher will provide an overview of the purpose of the study, discuss its voluntary nature and the fact that individuals can withdraw at any time, and that while name, professional role, and entity name will be collected, no other identifying information will be noted on the transcripts. The researcher will also review the consent process in detail based on whether the individual is participating in the interview.

3. **<u>Enrollment and Consent Process</u>: For individuals selected for an interview:** The researcher will ask the individual representative of the organization to select a time when it would be the best time to conduct the phone interview.
    - A convenient date and time for the interview will be scheduled.
    - The researcher will ask their permission to email or mail them the consent in advance and instruct them to read it carefully prior to the date of their interview. The researcher will ask them to send back a signed consent prior to their scheduled interview date and time. Prior to the interview or facilitated discussion, an informed consent will be read to the individual. (Informed consent form)

Instruments: A semi-structured interview guide has been developed in consultation with the literature; all are attached with this IRB application and are listed below:

- Semi-Structured Interview Guide; Rare Disease Advocacy

Participation: Participation efforts include: Telephone and email introductory recruitment/screening scripts; semi-structured interviews to support characterization of rare disease advocacy organizations. The researcher anticipates conducting up to 30 semi-structured interviews that may include up to 60 total organizational representatives (N=2 per organization). A facilitated discussion as part of Phase II is anticipated to include up to 15 Global Genes Advocacy Managers.

Eligibility:
A purposive sample of up to 30 rare disease advocacy organizations that conduct advocacy on behalf of rare disease patients and families in the United States. Only individuals over the age of 18 will be eligible for this study and who have at least two years of experience in the field. It is anticipated that all participants will be an organizational representative working directly in rare disease advocacy and English-speaking.

Risks and Benefits: The risks involved in this study are expected to be minimal. However, participants may be inconvenienced by taking time to participate in an interview. There will be no direct benefits to participants for their involvement in the semi-structured interviews.

Data Management: Precautions to ensure confidentiality in all components of the study include the following: The researcher received training in human subjects' protection at the University of Illinois at Chicago; Only appropriately trained research committee members have access to study data on an as-needed basis; and semi-structured interview transcripts, as well as notes from the facilitated discussion, will be stripped of identified information and stored on a computer that is password protected. Data will not be distributed unless tripped of identifiers.

Data security: Each data source will be assigned an identification number. Datasets in computer files will be stored with all personal identifiers such as name and address removed. A key to the identification of participants will be stored in a secure location remote from the data. Data in computer files will be stored on an access-protected server housed at the School of Public Health. Data will be available by remote access through an encrypted tunnel. Researchers will share data for analysis and data management by using the shared network drive. Data will not be stored or transmitted using external media including USB devices and CD ROMs. All records of data on hard copy will be maintained in a locked room within locked file cabinets in the home office of the researcher.

Data Analysis: Data analysis will be an iterative process and will occur simultaneously with data collection. Interviews transcripts will be de-identified, logged, and stored in encrypted files. Data will be coded using Atlas.ti software; documents will be inventoried and coded; field notes will be transcribed and coded; themes will be identified using within, between, and cross case analysis.

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Appendix 2: IRB Exemption Letter

UNIVERSITY OF ILLINOIS
AT CHICAGO

Office for the Protection of Research Subjects (OPRS)
Office of the Vice Chancellor for Research (MC 672)
203 Administrative Office Building
1737 West Polk Street
Chicago, Illinois 60612-7227

Exemption Granted

July 7, 2017

Michelle Slimko

Public Health

1603 W. Taylor Street

Phone: (773) 517-9756

RE: Research Protocol # 2017-0711
"Identifying Characteristics of Rare Disease Advocacy Organizations and the Factors that Influence their Advocacy Capacity"

Sponsors: None

Dear Michelle Slimko:

Your Claim of Exemption was reviewed on July 7, 2017 and it was determined that your research protocol meets the criteria for exemption as defined in the U. S. Department of Health and Human Services Regulations for the Protection of Human Subjects [(45 CFR 46.101(b)]. You may now begin your research.

Exemption Period:	**July 7, 2017 – July 7, 2020**
Performance Site:	UIC
Subject Population:	Adult (18+ years) subjects only
Number of Subjects:	75

The specific exemption category under 45 CFR 46.101(b) is:

(2) Research involving the use of educational tests (cognitive, diagnostic, aptitude, achievement), survey procedures, interview procedures or observation of public behavior, unless: (i) information obtained is recorded in such a manner that human subjects can be identified, directly or through identifiers linked to the subjects; and (ii) any disclosure of the human subjects' responses outside the research could reasonably place the subjects at risk of criminal or civil liability or be damaging to the subjects' financial standing, employability, or reputation.

You are reminded that investigators whose research involving human subjects is determined to be exempt from the federal regulations for the protection of human subjects still have responsibilities for the ethical conduct of the research under state law and UIC policy. Please be aware of the following UIC policies and responsibilities for investigators:

1. Amendments You are responsible for reporting any amendments to your research protocol that may affect the determination of the exemption and may result in your research no longer being eligible for the exemption that has been granted.

2. Record Keeping You are responsible for maintaining a copy all research related records in a secure location in the event future verification is necessary, at a minimum these documents include: the research protocol, the claim of exemption application, all questionnaires, survey instruments, interview questions and/or data collection instruments associated with this research protocol, recruiting or advertising materials, any consent forms or information sheets given to subjects, or any other pertinent documents.

3. Final Report When you have completed work on your research protocol, you should submit a final report to the Office for Protection of Research Subjects (OPRS).

4. Information for Human Subjects UIC Policy requires investigators to provide information about the research to subjects and to obtain their permission prior to their participating in the research. The information about the research should be presented to subjects as detailed in the research protocol, application and supporting documents.

Please be sure to use your research protocol number (listed above) on any documents or correspondence with the IRB concerning your research protocol.

We wish you the best as you conduct your research. If you have any questions or need further help, please contact me at (312) 355-2908 or the OPRS office at (312) 996-1711.

Sincerely,

Charles W. Hoehne, B.S.
Assistant Director, IRB #7

Office for the Protection of Research Subjects

cc: Paul Brandt-Rauf, Public Health, M/C 923

Christina Welter, Public Health, M/C 923

Appendix 3. Measurement Table

1.) What are the characteristics of rare disease organizations that undertake advocacy activities? 2.) How have these factors facilitated or presented as barriers to the organization's ability to conduct advocacy on behalf of rare disease patients?					
Constructs	**Sub-constructs to explore**	**Description and Factors**	**Data Collection Approach**	**Possible sub-codes**	**Analysis**
Inputs	**Strategic Planning and Business Planning**	Strategy planning is defined as planning systematically for how to position and deciding what tactics to use to reach the goal.	Annual reports Power point presentations, Budgets Goals documents	Strategy document Planning documents Posted on website, Internet Reviewed by Board Developed with Board and share broadly Budget, and funding forecasting documents	
	Infrastructure Development	Defined as setting up the equipment, systems, and other nuts-and-bolts supports needed to advocate.	CapWhiz program, Donor software, Databases Internet access Computers Phone system 800 Line, multi line capability Office related bills/maintenance document	Physical office Computer Phone/phone line Software licenses/agreements Internet Access Interactive webinar capability	

	Engagement and Outreach	Defined as using technologies such as email, websites, blogs, podcasts, and cell phones to reach a large audience and enable fast communications.	Email blast documents Newsletters Social media content Blogs Podcasts Webinars Website content Feedback from targeted audiences	Email Websites Blogs Podcasts Test Messaging Social media Webinars	
	Communications Development	Defined as creating publications, brochures, websites, or other "communications collateral" to deliver advocacy messages.	Communication plans and strategy documents Drafts of advocacy messages documents	Publications Brochures Website content Leave Behind materials Table Tents	
	Staffing and Leadership Development	Defined as hiring or developing the people to implement an advocacy strategy and establishing a clear understanding of who is doing what.	Hiring strategy Development planning documents Meeting minutes, agendas, Proposals for positions Job Descriptions Job postings Pictures of staff with titles	Reporting structures Hired Staff Staff titles and descriptions 501c3 status Designation of time, FTE, vs part time vs volunteer Professional development courses	

			Organizational charts Budget information Staff agendas	Policy analysis training Advocacy training Certifications Continuing education Webinars Leadership Courses	

Constructs	Sub-constructs to explore	Description and Factors	Data Collection Approach	Possible sub-codes	Analysis
Outputs	**Partnerships or Alliances**	Defined as mutually-beneficial relationships with other organizations or individuals who support or participate in an advocacy strategy.	Legal contracts MOUs Programs with collaborative campaigns or work	Support groups Advocacy groups Other similar disease groups List of participants or individuals Collaborative work	
	New Champions	Defined as high-profile individuals who adopt an issue and publicly advocate for it.	Ads PSAs, commercial or other media, Speeches List of new legislators	Celebrity endorsers Federal, state or local leaders Company presidents Sports figures Models Hearings Testimony	
	New Advocates (unlikely or non-traditional)	Defined as individuals who support rare disease that is outside usual RD network.	Databases Lists of new advocates Website content Meeting agendas Partner lists	Annual reports Photos Advocate partners	

	New Donors	Defined as new public or private funders or individuals who contribute funds or other resources for a cause.	Donor data Donor budget information Number of new donors	Public funders list Private funders list Other funders	
	Constituency or Support Base Growth	Defined as increase in the number of individuals who can be counted on for sustained advocacy or action on an issue.	Database of members Number of people who visit Capitol Hill Number of people who wrote their congressman Social Media advocacy efforts	Volunteer activites Newsletters Budgets Attendance records	

Appendix 4: Codebook

Version 5 Final Code Book for Characteristics of RD Advocacy Organizations

Key Principles:

- Code the whole sentence in which a key word or phrase is found. Look at paragraph level and include sentences before and after.
- Sentences can be categorized with more than one code.
- All codes and all sub-codes will be used for the interviews and document reviewed.
- Write memos, or short notes while coding.
- After completing a coding session, write a longer memo.

Construct	Instructions
Age of the organization- This refers to year or reference in time when the organization started as a non-profit organization.	Use for codes for all mentions regarding the year the organization started as a 501c3. • Year started • Started X amount of years ago Examples include: started in 1999 but became a 501c3 in 2000, began 40 years ago
Role and Title of the Interviewee- Use this for mentions from the interviewee about their title and role within the rare disease organization.	Use this collection of codes for all mentions related to titles and roles of the interviewee. • Founder or Co-Founder • President, CEO • Director of Fundraising • Director of Policy, and Advocacy • Board Member • Volunteer Examples include, Board Member and Co-Founder, CEO
Organizational Resources- This refers to the type of staff within the organization, the number of staff, inclusion of a scientific advisory board, or medical advisory board, Board of Directors, an organizational vision and mission, and type of office infrastructure.	Use this collection of codes for all mentions of the organizational design of the advocacy organization and all mentions that describe the makeup or structure of the organization or committees, staff or volunteers. • Staff- paid, FT, or volunteer • Established vision or mission statement • Medical Advisory Board • Board of Directors • Scientific or Medical Advisory Board • Office or Headquarters • Budget Examples include, 5 paid staff, 2 volunteers, Medical Advisory Board of 5 Scientists, and virtual office, work out of home, headquarters, medical advisory board that I all volunteer staff with scientific officer
Prioritization Process- This refers to the process by which the organization takes to endorse a plan, key advocacy areas or strategic plan for advocacy.	Use this collection of codes for all mentions regarding approaches to achieve organizational priorities related to advocacy work. • Established strategic planning process • Based on member feedback • Board decisions • Driven by organizational needs and focus areas.

	Examples include, board decisions, formal business planning process, parent driven feedback, partner with Medical Doctor.
Genesis- This refers to the reason why the organization started as a rare disease advocacy organization and may include how it got started.	Use this code for mentions related to the purpose, or reasons why the advocacy organization was formed. Some examples include: • Child with a rare disease • New opportunity to serve others • Need for RD funding for research • Recommended by third party expert
Funding- This refers to any mention related to needs for money, in the context of supporting any RD advocacy efforts.	Use this collection of codes for all mentions of the need for money to advance advocacy objectives. • Skills • Fund Research • Fund Clinical Trials • Access to Care • National history studies Examples include, fundraising, fund research, find members, fund clinical trials, provide greater access to care.

1.) Organizational Capacity Context

2.) Skills and Training Needs Context

Construct	Instructions
Valuable Skills- This refers to important and recognized competencies by the interviewee that they view as important to achieving RD advocacy goals.	Use this collection of codes for all mentions of skills needed to get advocacy work accomplished, and other skills mentioned in general. • Fundraising • How to manage a non-profit organization • Business and communication skills • Understanding the drug development process

	• Understanding the basics of Rare disease advocacy Examples include fundraising activities, marketing skills, translating science, helping make decisions, how to navigate social media
Wishlist of Trainings- This refers to an ideal list of available skill building activities identified by the interviewee that they feel would help the organization improve its advocacy skills if they had access to such training opportunities.	Use this collection of codes for all mentions of the skills or trainings needed for the organization. • Fundraising • Understanding basic science • Research for RD organizations • How to manage volunteer staff Examples include reading and translating scientific literature, managing nonprofit financials, raising more than a million dollars in one campaign, access to genetic testing or newborn screening

3.) Connecting with Constituents and Stakeholders Context

Construct	Instructions
Engagement and Outreach – This refers to connecting with people who have a similar rare disease or stakeholders of the RD in order to network, inform, educate, or connect within a specific RD community.	Use this set of sub codes with any mention of engagement or outreach activities. • Social Media • Conferences • Mail • Phone Calls • Private Chat Room Examples include, Twitter, Instagram, Facebook, Meetings, donor activities i.e. 5K fundraiser, newsletters, hugs, letters

4.) Gaining Value from Other People Context

Construct	Instructions
Mentoring- This refers to the value of having an external or internal colleague or contact that provides feedback	Use this set of codes with any mention of opportunities involving direct teaching or advisement related to RD advocacy.

or an opportunity to learn directly about RD advocacy.	• Teaching or advising about advocacy • How to effectively work with scientists • Talking to high level executives from biotech company Examples: teacher who taught policy or legislative advocacy, learning how to read a scientific paper directly from a scientist, calling a biotech executive to learn about drug development
Networking- This refers to benefits gained by connecting with people within and external to the RD community that can help advance or support RD advocacy efforts.	Use this set of codes with any mention of benefits gained or lessons learned as a result of interacting with people or organizations internal or external to the RD community that the organization is advocating for. • Introducing children to researchers • Be around smart people • Use web search to contact people • Form a rapport with scientists Examples: meet with other patients with RD in a community, meet other families in RD community, gain insights to previous make mistakes or successes
Partnerships- This refers to benefits gained by working with identified or specifically targeted third parties or rare disease advocacy groups that support the advancement of advocacy needs or goals.	Use this set of codes with any mention formal or informal relationships with third party organizations that advance or benefit the rare disease organizations. • Medical Doctor at university or hospital • Hospital • Health Professional society • Biotech or pharma company • Other foundations or charities Examples: Relationships or formal partnerships with researchers, other healthcare professional societies, heads of patient advocacy groups, medical doctors, hospitals or clinics
Third Party Technical Support- This refers to specifically mentioned rare disease organizations they work with or seek third party partners	Use this set of codes for any mention of a third-party consultant(s) or organization that is leveraged for

specifically for technical expertise to learn about how to conduct various rare disease advocacy efforts.	their trainings, information of consultations to help advance advocacy work. • NORD • Global Genes • Every Life • Genetic Alliance Examples; Global Genes Foundation Alliance, NORD and policy update, Every Life helpful for legislative information.

5.) Perceptions of Advocacy Context

Construct	Instructions
Immediate Advocacy Need(s)- This refers to rare disease advocacy priorities identified that are deemed necessary to address in the near term as critical importance to the organization.	Use this set of codes with any mention of advocacy priorities or needs for the organization that are determined to be addressed at top of action lists. • Funding • Increased Awareness or education • Research • Therapeutic solutions • Access to resources • Finding patients and members Examples: money to support activities, education for healthcare professionals, drug development, working with a lab
Definition of Advocacy This refers to how the organization describes and defines advocacy from the organizational perspective and includes what it may mean from their own individual role and responsibility within the organization.	Use this set of codes with any mention of defining advocacy including explicit comments on how their organization defines advocacy, including what it means from their own role in the organization. • Awareness and Education • Engagement • RD patient empowerment • Support health care professionals that work with RD community • Search for a cure

	• Help with funding Examples include education for affected families, better diagnosis and treatment, go to Capitol Hill to ask for funding
Definition of Success for Advocacy Efforts- This refers to how the organization characterizes success for achieving its advocacy work.	Use this set of codes with any mention of success factors. • Servicing patients • Feedback • Greater constituency or membership • Knowledge of support and care • Improved quality of life Examples include money raised to drive science agenda, how many stories that include impact provided by the organization, better quality of life

6.) Identifying and Addressing Barriers Context

Construct	Instructions
Barriers to achieving advocacy goals- This refers to obstacles that get in the way of advancing advocacy initiatives.	Use this set of codes with any mention of barriers or obstacles related to advocacy work and other mentions in general. • Lack of patients • Not enough funding or money • Lack of experts • Lack of patient credibility • Lack of research Examples include feeling intimidates, caregiving issues, fatigue, disability, challenges with enrolling in clinical trials
Overcoming Barriers- This refers to the approaches made to address obstacles in conducting rare disease advocacy work.	Use this set of codes with any mention of addressing or overcoming any barriers to advocacy. • Host conference • Connect with patients • Create a patient registry • Get further educated • Raise more money

	Examples include send out more mailers, work with other organizations, cultivate friendships, focus on advocacy in every state

Appendix 5: Oral Informed Consent Script
06/21/2017

Thank you for your interest in being involved in this research. My name is Michelle Slimko and I am a Doctoral candidate at the University of Illinois School of Public Health located in Chicago, IL. My email is mslimk2@uic.edu and can be reached at 773-517-9756 on my cell phone. The purpose of this research

is to gain an understanding of the characteristics of rare disease advocacy organizations and better understand how those factors influence advocacy capacity or the ability to conduct advocacy related activities by conducting interviews with those actually doing advocacy at these organizations.

I will conduct an interview with you while on the telephone and record your responses on an audio recorder. Once we complete our interview, that will last no more than 1 hour, I will review the transcripts of our interview. If there is any information that identifies you or your organization, I will strip that information from the transcripts for the purpose of this research to protect your privacy and confidentiality of the research information. You will be coded with a pseudonym to also de-identify your information on the recording. I anticipate this to be research of minimal risk since we will be speaking via telephone and conducting a phone interview, other than being inconvenienced of your time. All participation by you is voluntary and at any point you wish to opt out, I will stop the interview and end the recording.

Do you have any questions for me before we proceed?

Appendix 6: Recruitment Email

Rare Disease Advocacy Research

To: Global Genes Advocacy Managers

Dear Global Genes Advocacy Managers,

We are seeking your help as Rare Disease Advocates who may be interested to participate in a Qualitative Research Study

Are you interested in voluntarily participating in a qualitative research study to better understand what rare disease advocates face and how they overcome these challenges?

Eligible Participants must be:

- At least 18 years old
- Speak English
- Worked in rare disease advocacy for at least one year
- Work at Global Genes as an Advocacy Manager

If you meet this criterion, Michelle would love to include you as part of a facilitated discussion to hear the findings of her research conducted with Foundation Alliance Members via teleconference.

There is no compensation for participating.

If you are interested and wish to help contribute to knowledge about rare disease advocacy, please contact Michelle Slimko at mslimk2@uic.edu

Appendix 7: Facebook Notice for Recruitment

Foundation Alliance Facebook Page Update:

Seeking Rare Disease Advocates interested to participate in a Qualitative Research Study

Are you interested in voluntarily participating in a qualitative research study to better understand what rare disease advocates face and how they overcome these challenges?

Looking for Foundation Alliance Members who are:

- At least 18 years old
- Speak English
- Worked in rare disease advocacy for at least one year

If you meet this criterion, Michelle would love to contact you to participate between late July and August for **no more than one hour of your time** for a scheduled interview via telephone call.

There is no compensation for participating.

If you are interested and wish to help contribute to knowledge about rare disease advocacy, please contact Michelle Slimko, Doctoral student in Public Health Leadership from the University of Illinois at Chicago School of Public Health, at 1603 W. Taylor St. Chicago, IL 60612. Cell phone is 773-517-9756 or email directly at mslimk2@uic.edu.

We appreciate your interest and value your input!

Thank you!

Slimko, Doctoral student in Public Health Leadership from the University of Illinois at Chicago School of Public Health, at 1603 W. Taylor St. Chicago, IL 60612. Cell phone is 773-517-9756 or email directly at mslimk2@uic.edu.

We appreciate your interest and value your input!

Thank you!

Appendix 8: Semi-Structured Interview Guide- Rare Disease Advocacy

Introduction

a. I am interested in gaining a deeper understanding of the characteristics of rare disease advocacy organizations, and in your case, on behalf of <insert specific rare disease>. Can you tell me more about your organization, for example, how it got started, and why it was founded?

    a. What is your role in the organization? How long have you been there?

        Probes: Where is your organization located?

        What is the mission or vison of your organization?

        What year did it begin?

        How is your staff arranged or organized?

        Do they have a separate office space with dedicated staff? Geographic location(s)?

        How would you describe your organizational structure and type of staff positions working on behalf of <insert rare disease>?

    b. Can you tell me more about <insert name of rare disease> and what you think are the immediate advocacy needs for patients and families dealing with <insert rare disease>?

Transition: *Okay, thank you for sharing that information. Let's move on to talking about your organization and the advocacy activities conducted on behalf of patients with <insert rare disease.>*

Facilitators and Barriers of Rare Disease Advocacy

For as long as you have been with your organization, can you tell me about what type of advocacy your organization has been involved with? Can you tell me more about any other historical knowledge regarding advocacy for <insert rare disease> you are aware of that has evolved?

    a. For your organization, what does advocacy mean?

        Probe: What are you trying to achieve via advocacy? (Is it to influence? Is it to invite change? Is it to support a group or individual?)

    b. How does your organization prioritize these advocacy initiatives?

        Probes: What kind of process does your organization undergo when identifying these priorities?

        If you have a strategic plan, how is advocacy a part of your strategic plan?

        Who is involved in this process?

    c. Can you share examples of barriers or challenges you may have faced when conducting advocacy?

        Probes: How did you overcome some of these challenges? Does it still exist?

        Are there any skills or professional development training that may have helped you get through this challenge?

    d. How do you determine if your advocacy efforts are successful? In your own words, what outcome(s) determines success for the organization?

        Probes: Who do you partner, consult, or hire to help you with advocacy?

        What type of activities or advocacy related work do these third parties assist with?

Advocacy Capacity

b. *Transition: Thank you. Now, I'd like to shift our conversation to now learn more about skills and trainings used to undertake the advocacy activities you just described.*

Leadership and Staffing Development

a. Can you tell me what trainings, webinars, or development courses your staff has participated in to learn more about advocacy, or develop other/new skills?

   Probes: How did you learn about these resources?

   Where do you go to get your information for professional development training(s)?

   Do you have a primary resource? What is the best type of training for you and staff? (in person, webinar, short course) and why?

b. Are there trainings you wish you had access to that would benefit your knowledge in conducting advocacy more effectively? If yes, can you tell me what future trainings you may be interested in learning more about?

Engagement & Outreach

a. How do you engage and connect with your organization's members and stakeholders? How frequent is this engagement?

   Probe: What are some of the types of examples of engagement or educational outreach?

   What role does social media play in your advocacy efforts? What social media does your organization use? Can you describe any specific software or computer programs that you use and how you use them?

b. How did you learn about how to use these tools?

   Probes: Do you have a specific resource you use?

   How did you learn about this resource?

   How often do you use these tools?

c. What type of outcomes have these tools provided that is beneficial to your advocacy efforts?

Communications and Development

a. Can you tell me other ways you share information with your key stakeholders beyond electronic media?

b. How do you develop communication materials for your various audiences?

Probes: What are your typical outlets for communication and information sharing?

Who on your staff manages the writing and communications plans?

Partnerships & Alliances

a. In your opinion, which existing partnerships are critical to your advocacy work?

b. Please describe how you developed these partnerships or alliances?

Probes: How were they identified?

Can you tell me what you think the organization gained as result of partnering that helped advance its advocacy agenda?

Conclusion

c. *Thank you so much for sharing your time with me and telling me more about your role and important work your organization is doing on behalf of <insert rare disease>. Is there anything else I haven't asked about that you want to add or share?*

   a. *Do you have any questions for me?*

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VITA

Michelle L. Slimko, MPH, RDN, LDN

Michelle Slimko has been a Licensed, and Registered Dietitian since 2000 and graduated cum laude with B.S. in Nutritional Sciences from Benedictine University and Summa Cum Laude with M.P.H in Community Nutrition from Benedictine University as well. She is currently a doctoral candidate as a Doctorate in Public Health Leadership from the University of Chicago, School of Public Health.

Education

University of Illinois-Chicago, School of Public Health, Chicago, IL- Adult Student, began Fall 2009

Doctor of Public Health Leadership candidate expected Dec. 2018

Benedictine University, Lisle, Illinois

B.S. Nutrition Science (Honors Cum Laude), 1998

Master of Public Health (Honors, Summa Cum Laude), Community Nutrition, 2000

Professional Experiences:

Promoted to SVP, Global Sustainable Nutrition & Research, & Chief of Staff to Global Chief Science Officer, Rosemont, IL Present Role

Senior management role a Chief of Staff with accountability for 40+ R&D and health professional employees, Staff Lead for Dairy Innovation Center Sustainable Nutrition committee comprised of dairy company executives, and strategy and development lead for science agenda in support of our Global Chief Science Officer.

Promoted from VP Strategic Partnerships, Regulatory & Planning May 2013-2017

In this role I represented America's 43,000 dairy farmers ensuring their strategic investments in science and research match strategies that identify, maintain, and create public and private partnerships on their behalf. Responsibilities for relationship management include Joslin Diabetes Center, USDA ARS, NIFA and ERS, Chicago Council on Global Affairs, Foundation for National Institutes of Health, Global Innovation Partnerships for dairy- Taco Bell, McDonald's, Domino's, Pizza Hut, and 8 fluid milk companies, Whey Protein Research Consortium and US Dairy Export Council.

PepsiCo America Foods; Barrington, IL 2011-2013

Principal R&D Nutrition Scientist (Promoted twice from Associate Nutrition Scientist).

- Nutrition Brand Support for Quaker Food and Snacks- claims, substantiation on pack, media and regulatory
- Quaker brand nutritionist for health professional meetings and interface with NGOs
- Conduct nutrition education trainings to R&D, Legal, Marketing, Nutrition, Regulatory
- Managed clinical trial for satiety, claim language with legal, application to portfolio

- Supervise 1 direct report

PepsiCo Global Health Policy; Barrington, IL (reported to VP, Nutrition)

2010-2011

Principal Nutrition Scientist

- Lead community health clinical research initiative and strategy for Performance with Purpose Corp. agenda, Community Commitments
- Assessed and researched potential solutions for food industry to address local Chicago food deserts
- Built business case and potential community research action plan with 2M dollar budget
- Presented findings and scope presentation for proposal to PepsiCo CEO
- Supervised 1 direct report

PepsiCo International Food, Snacks and Beverages; Barrington, IL 2005-2010 (reported to SVP and Dir.)

Sr. Nutrition Scientist

- Managed network of 12 PepsiCo International Nutrition Scientists (PINT), main company point of contact for internal review process and representative for International Food Safety and Nutrition Team (IFSAN) , a network of over 130 scientists, interaction for identification, development and approval of nutrition and product claims, fortification initiatives, advertising claims, PepsiCo International (PI) company policies on safety, nutrition and health & wellness issues for food, snacks and beverages.
- Thorough understanding of intl. nutrition processes and procedures to comply with global legal, food regulatory and corporate policies.

- Executed and managed global communication plans related to PINT and IFSAN. Lead and facilitated global nutrition meetings with stakeholders from IFSAN, including Govt. Affairs, Public Policy, Sustainability, Corporate Communications, Crisis Management, etc.
- Developed and communicated metrics to senior management that measured global nutrition deliverables, database usage, and Internet traffic.
- Lead global alignment within technical and business communities. Served as key ambassador to share best practices globally to enable business solutions and maintain consistency across key business regions.

Key Accomplishments/PepsiCo:

- Built Quaker Snacking Strategy for 2012+, including research and scientific review of literature, leverage key academic opinion leaders and contracted work from scientists for data review and scientific symposium. Company lead for consumer research project in collaboration with IFIC.
- Worked directly with South Side Chicago stakeholders to strategically partner industry with public stakeholders to help address the need to eliminate food deserts.
- Executed strategy behind bi-annual global nutrition scientific meetings to collaborate with global foods, and beverages scientific affairs, safety and regulatory.
- Lead strategy behind creating first ever global Quaker branding guardrails, company wide
- Lead and facilitated 2008 global technical expert meeting on the Nutrition Science of Nuts, Barcelona, Spain, including academic experts, and managing regional budgets.

- Key presenter in South Africa local meeting, representing global nutrition to influence key external stakeholders, Ministry of Health, and other NGOs about PepsiCo Sustainability commitments to H&W.
- Gained global alignment acting as company lead for PI Health &Wellness internal classification for nutrition guidelines. Served as key guide to product development and innovation as nutrition commitments became more public.
- Created and lead first ever global claims, fortification and substantiation approval and review process for PI businesses. Strong negotiation and cross collaboration efforts for PepsiCo Intl. regions to adopt review process and integrate company-wide change. Created a data warehouse for global usage across technical community. Garnered support from PI CEO to fund and endorse this process.
- Completed global tour of major PepsiCo R&D processing plants and technical centers, serving as PepsiCo International nutrition lead via PI Support Center efforts. Exposure and thorough understanding of regional and technical capabilities for foods, snacks and beverages.

Abbott Laboratories; Abbott Park, IL **2001-2005**

Senior Corporate Health Promotion & Wellness Coordinator

- Designed and developed health promotion programs and strategies for broad range of health areas, including wellness education, disease prevention and disease management programs to reach a diverse global workforce. Responsible for managing wellness budget of 60-80K and calculating and analyzing company savings (ROI). Worked with Human Resources, Employee Health, Risk Management, and Corporate Legal to ensure appropriate handling of sensitive issues.

- Established global communication plans and training programs that applied educational and behavioral principles to influence adult learning, behavioral change and promoted a positive corporate culture.
- Provided benefits coordination to Human Resource professionals, Occupational Health Nurses and fitness professionals worldwide for local wellness programs.
- Supervised administrative assistant and 3 graduate students for internship.
- Program manager for Mothers at Work workplace breastfeeding program.
- Assisted director in day to day operations of Executive Health Appraisal Program.

Woodstock Memorial Hospital; Woodstock, IL **2004-2005**

Clinical Dietitian-Registry (Part-time)

Abbott Nutrition International, Scientific & Regulatory Affairs, Abbott Park, IL 2000-2001

Medical Communications Associate, Contractor

- Researched clinical findings for nutritional business development areas.
- Project Manager for key poster presentations and publications for world congresses.
- Coordinated nutritional therapy physician education program for Latin American countries, and wrote, edited and revised Global Slide Kit for scientific content and quality.

St. Joseph Hospital; Chicago, IL **2001-2003**

Clinical Dietitian-Registry (Part-time)

Will County Health Department; Joliet, IL **1999-2000**

Cardiovascular Health Promotion Specialist

Lombard Pharmacy; Lombard, IL **1994-1999**

State of Illinois Licensed Pharmacist Technician

Naperville Community School District 300; Naperville, IL **1998-1999**

Nutrition Educator

Past Memberships

PepsiCo: Women's Inclusion Network, Barrington Community Service, Toastmaster's Club,

Industry Nutrition Advisory Panel AHA

Publications :

Slimko, M. L., Tricarico, J. and Miller, G. D. (2017), Proceedings from the United States Department of Agriculture and National Dairy Council Collaborative Research Planning Meeting Held August 24, 2016. Journal of Food Science, 82: 1513–1515. doi:10.1111/1750-3841.13465.

Healthy Food for a Healthy World- Leveraging Agriculture and Food For Global Nutrition Blog
https://www.thechicagocouncil.org/sites/default/files/GlobalAg-HealthyFood_FINAL.pdf

Partnering to Progress the Future of Agricultural Research Blog

https://www.linkedin.com/pulse/partnering-progress-future-agricultural-research-michelle/

Who do Americans trust when it comes to food? Blog and article

http://archive.constantcontact.com/fs142/1101490782895/archive/1123530809811.html

Slimko ML, Mensah GA. The Role of Diets, Food, and Nutrients in the Prevention and Control of Hypertension and Prehypertension. Cardiology Clinics, 28(4): 665-+.

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http://foodfrontiers.pepsicoblogs.com/2011/02/pepsico-recognizes-american-heart-month-more-work-needs-to-be-done-but-there-is-good-news/# ; Feb 14, 2011.

M. Fleming, M. Lagatuz, MPH, C. Stein, J. Moreschi. Development and Evaluation of Educational Video and Leader Guide. Journal of the American Dietetic Association, Sept. 2000, A-26, Volume 100, #9.

Professional Activities: Society Memberships, Committees, and Leadership Posts

a. Member, Academy Nutrition & Dietetics, since 2000

b. Public Health & Community Nutrition Dietetic Practice Group, since 2017

c. Research Dietetic Practice Group, since 2017

d. Foundation for NIH Metabolic Steering Committee, 2017

e. Student Member, American Public Health Association, since 2016

f. Board of Directors, Neurofibromatosis Network, since 2009

g. Community Volunteer, Community School District 26, Math Education

h. Volunteer Builder, Habitat for Humanity, 2012

i. Benedictine University Nutrition Club

j. Women's Volleyball Team, Captain

Awards:

1. 40 under 40 award from Benedictine University

2. Nutrition Leadership Award, PepsiCo Nutrition Team

3. Chief Science Officer Nutrition Leadership Award, PepsiCo

4. Cum Laude, BS, Benedictine University, 2000

5. Graduated Summa Cum Laude, MPH, Benedictine University, 1998

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