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PRIMARY CARE FOR RARE DISEASE PATIENTS:

EXPLORING SERVICES RECEIVED, HEALTHCARE PROVIDERS INVOLVED, AND PATIENT SATISFACTION

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Abstract

Rare disease is a significant healthcare burden, affecting an estimated 25-30 million U.S. citizens. This equates to 1 in 10 Americans, over half of which are children. It has been suggested that primary care can significantly aid the medical management of rare disease patients' unique conditions, but data on the relationship between rare disease and primary care is limited. Current knowledge consists of physician opinion pieces and analyses of medical records, but it lacks patient-reported data and perspectives. For the first time, 282 U.S. patients with rare diseases were surveyed regarding which primary care services they were receiving and which healthcare professionals were involved. Our study was limited to metabolic disorders listed by the National Institute of Health Genetic and Rare Diseases (GARD) Information Center and excluded conditions with a life expectancy under 12 years. The anonymous 12-question online survey was sent to patient organizations and advocacy groups associated with these conditions. Overall, respondents reported receiving the majority of their primary care from primary care physicians (PCPs), including family physicians, internists, and pediatricians, rather than specialists. The majority (86%) of respondents reported being satisfied with their primary care. Factors like distance from a major urban area, gender, and age did not significantly correlate to patient satisfaction. Our study suggests that despite their generalist training, PCPs play an important role for rare disease patients. It is therefore important that PCPs be prepared to care for these patients. Future research into health insurance, rare disease subtypes, and the training PCPs receive regarding rare disease would further our ability to address this healthcare burden.

Keywords: primary care, rare disease, primary care physicians, patient satisfaction

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Introduction

Despite their name, rare diseases are a common occurrence in healthcare. The Orphan Drug Act of 1983 defines a rare disease as a condition affecting fewer than 200,000 people in the United States. While it is rare to have one of these diseases, the National Institute of Health estimates there may be as many as 7,000 different types of rare disease, affecting 25-30 million Americans today ("FAQs About Rare Diseases", 2017). This equates to 1 in 10, or 10%, of the U.S. population. The burden of rare disease has only recently been recognized as a major healthcare problem in the U.S. and other countries, and those affected experience many challenges when seeking their unique medical care. These typically concern delayed diagnosis, suboptimal care, and inability to afford disease-specific medications (Stoller, 2018). Subsequently, families face emotional burdens as they navigate what is known as the "diagnostic odyssey" as well as life after receiving a diagnosis. Individuals with rare diseases must often see various specialists multiple times a year. While the need for specialist care is well understood, the extent to which rare disease patients value their primary care providers (PCPs) is uncertain.

For many, PCPs play a vital role in one's healthcare team–acting as a coordinator of care, a continuous monitor of health data, and a trusted provider. To understand the role of primary care in the management of rare disease, it is important to consider primary care's main purpose and the unique issues faced by patients with rare disease. The American Academy of Family Physicians (AAFP, 2019) defines primary care in the following way:

> Primary care includes health promotion, disease prevention, health maintenance, counseling, patient education, diagnosis and treatment of acute and chronic illnesses in a variety of health care settings. [It] is performed and managed by a personal physician often collaborating with other health professionals… Primary care provides patient

> advocacy in the health care system to accomplish cost-effective care by coordination of health care services. Primary care promotes effective communication with patients and encourages the role of the patient as a partner in health care.

As such, primary care can play a critical role in a patient's medical management and long term wellbeing. Primary care physicians (PCPs) act as advocates and points of contact through every concern–small and large. For the purposes of our research, we follow the AAFP definition for primary care and PCPs. This includes any specialist in family medicine, internal medicine or pediatrics "who provides definitive care to the undifferentiated patient at the point of first contact, and takes continuing responsibility for providing the patient's comprehensive care" (AAFP, 2019).

Literature on primary care is written primarily by physicians, and it generally emphasizes the aspects of primary care that set it apart from other specialties. These include *trust, support, advocacy, coordination of care,* and *a patient-centered approach* (Dudding-Byth, 2015). PCPs manage referrals to and communication with specialists, influencing the whole of a patient's medical management and ensuring *coordination of care* (Dimatteo, 1995). While literature from the patient perspective is limited, it suggests that patients also view these themes as benefits unique to primary care. Hruby, Pantilat, and Lo (2001) surveyed patients and caregivers regarding the involvement of their PCP in an inpatient setting. They found "about 50% of respondents believed that a PCP (rather than a separate hospital physician) should inform a patient of a serious diagnosis or discuss choices between medical and surgical management" (Hruby, Pantilat, & Lo 2001). The reasoning behind this sentiment was broken down with questions about patient-physician relationships. When asked whether or not they agreed with the statement: "It is important to me that my doctor *know me as a person,"* 88% did, and 73%

reported "I have *more trust* in a doctor I've known a long time than one I've just met" (Hruby, Pantilat, & Lo 2001). While this was a small study, the responses strongly echo the beliefs about primary care seen in the physician-written literature. Physicians and patients both recognize the patient-centric, trustworthy, and supportive services that primary care provides, and patients in this study show it is something they desire in their medical care. It follows that the goals and training of PCPs are well-suited to address the needs of those in the rare disease space.

Patients with rare disease and their families face challenges in their healthcare as unique as their conditions. They can be medical, financial, and emotional in nature, and they "largely regard being diagnosed, receiving optimal care, and affording disease-specific medications" (Stoller, 2018). From the provider perspective, "challenges… include gaining knowledge and experience in caring for such patients, and the availability of local experts and of expert guidelines" (Stoller, 2018). As can be imagined, experts with enough understanding to treat rare diseases are few and far between. As of the time of writing this paper, the National Organization for Rare Disorders (NORD), has only published 16 physician guides for rare diseases (NORD, 2018). That is 16 out of the more than 7,000 identified conditions.

From the patient perspective, this lack of knowledge contributes to a frustrating, costly, and lengthy process known as the diagnostic odyssey. The diagnostic odyssey is well-established with surveys from around the world finding some rare disease patients may wait between 5 and 30 years to receive a diagnosis. During which time they may receive improper care and incorrect medical interventions, including surgery and medications (EURORIDS, 2017). This period can have an extreme emotional toll on individuals and their families (Nutt & Limb, 2011).

Even after receiving a diagnosis, many caregivers feel they lack resources and support (Anderson, Elliott, & Zurynski, 2013). For many families, stressors concern children's inability to self-care in the future, discrimination from the rest of society, poor psychological development (e.g. low self-esteem), and personal guilt for having passed the genetic disorder to their children (Weng et al., 2012). Because of their experiences raising a child with a rare disease, some parents choose not to have any more children (Pelentsov, O'Shaughnessy, Laws, & Esterman, 2014). For these families, psychological worries and emotional concerns are a significant aspect of life with rare disease, and the potential for effective primary care to minimize these concerns is currently unstudied.

In conjunction with the other unique issues in rare disease, many families face a significant financial burden to attain proper care. In Europe, surveys indicate up to 25% of families travel to other regions and countries to access specialist care and diagnoses (EURORIDS, 2017). This costs time and money. In Australia, 77% of rare disease survey respondents report receiving some form of financial support through the government (Anderson, Elliott, and Zurynski, 2013). Over half (52%) of these families indicated that the financial assistance they received was inadequate to cover their needs and 73% said that additional income was needed to cover medical expenses (Anderson, Elliott, and Zurynski, 2013). US studies on rare disease financial burdens are limited, and it is not understood how effective primary care could play a role in reducing these costs.

Data regarding the relationship between primary care and rare disease is scarce. In opinion pieces and reviews, physicians cite this dearth of information and issue a call to action for PCPs. Knight and Senior (2006) argue:

> Many patients with rare diseases will present their symptoms first to a GP. They will also attend a GP in between visits to the specialist, they will require diagnosis and treatment of common ailments, and will benefit from the preventive health services offered by general practices. They will require the accessible, relationship-based advocacy and support role that is at the heart of good general practice… A thoughtful, proactive, ongoing response… of a continuing relationship with a GP may reduce many of the negative experiences of patients with rare diseases.

This argument echoes the themes seen in the literature on primary care's general purpose, namely *support, advocacy, coordination of care, patient-centered approach,* and *continuity of care*. They further declare "a systematic, generic primary-care approach to rare disease may reduce problems such as lack of coordinated care, lack of information, [and] delayed diagnosis" (Knight & Senior, 2006). They call for more research into the patient experience and request that primary care rises to meet the needs of this community (Knight & Senior, 2006).

Since the publication of Knight's and Senior's work, two studies have addressed the prevalence of rare disease within primary care. Phillips (2009) quantitatively analyzed patient records from family physicians (FPs) in the US. He found that family physicians were almost always the first to identify the rare disease problem and often established the diagnosis. They provided care for the problem more than half of the time and continued care in 76% of patients. (Phillips, 2009). This small study supports the idea that PCPs can and do provide necessary care for those with rare disease. It did not measure quality or outcome of care nor did it assess patient perspective.

Jo et al. (2019) performed a larger, cross sectional study using a national dataset to estimate the prevalence of rare disease in primary care in the US. They found that "among

outpatient visits to PCPs, rare diseases account for 1.6% of the visits" (Jo et al., 2019). While seemingly small, 93% of patients with rare diseases were established patients. These patients often visited their PCP with a chronic problem related to their condition, and "they had a significantly higher total number of chronic diseases compared with patients without rare diseases" (Jo et al., 2019). Through their analysis, they found that patients with rare diseases were 52% more likely to be referred to another provider (Jo et al., 2019). This is the largest study of rare disease in primary care to date. It does not state whether the referrals to other providers are for a consult or for a shared care relationship between primary and specialty care. The authors declare "better understanding of the overall management of patients with rare diseases managed solely outside of primary care would help to improve the care for these patients" (Jo et al., 2019). While research has begun to understand the role of primary care in rare disease, it is not yet known how patients are receiving their care overall. Neither this study nor the others on primary care and rare disease fully capture patients' feelings toward primary care.

In summary, the existing literature has begun to describe how primary care contributes to the health management of rare disease patients, but it lacks their perspective. Our knowledge of rare disease care is missing the voice of the patients. In our survey, we assess patient demographics, opinions, services received, healthcare providers involved, and satisfaction regarding their primary care.

Materials and Methods

Recruitment

In order to prevent confounding and create a more homologous cohort, we restricted our study to rare metabolic conditions. We randomized the National Institute of Health's (NIH) list

of these conditions and selected the first 206 conditions (National Institute of Health, n.d.). Eligibility criteria required that conditions have an average life expectancy of at least twelve years. To recruit participants, we reached out to the patient organizations and social media groups associated with each condition and requested they share our survey with their members. For participants, eligibility criteria required they live in the United States.

Data Collection

A 12-question survey was created via SurveyMonkey. The Sarah Lawrence College Institutional Review Board declared our survey exempt from full review. The board followed the requirements of the Common Rule, which states surveys that do not identify participants are exempt. This survey was open from December 22, 2019 to February 2, 2020. Questions sought information about demographics, distance from a major urban area, which primary care services patients were receiving, which healthcare professionals were providing said services, and patient satisfaction. Types of questions on this survey included categorical (multiple choice), numerical, and free response. Information collected from surveys was stored on password-protected SurveyMonkey and Google Drive accounts. Interested participants provided informed consent at the start of the survey, and we did not compensate participants for completing the survey.

Data Analysis:

We analyzed quantitative data using the statistical analysis tools in Microsoft Excel. Quantitative data is presented in the form of counts, percentages of the total, and median. Chi-squared tests were performed to determine if there were any differences by categorical variables. To further assess correlation between categorical and continuous variables, we ran logistic regressions.

For free response questions, analysis was performed using inductive coding. All three investigators read open-ended responses and identified major themes. After discussion, major themes were compiled to generate a single list of codes. Responses were read again and meaningful units were identified and coded with the relevant code. Responses could fall into multiple categories and were cross-referenced with the *Primary Care Services Received and Healthcare Providers Involved* chart responses (Table 1). Themes appearing in at least 5% of responses were considered common enough to be described in detail.

Results

A total of 282 people responded to the survey. We excluded 48 individuals from our data analysis that did not live in the U.S. and three who did not provide an answer for this question (5.5% of the total responses). Of the remaining 231 U.S. respondents, a few skipped one to three individual questions on the survey; however, we considered all of these surveys to be complete since they answered the majority of questions and reached the final question (#12). We included all of these responses in the data analysis.

Demographic Information

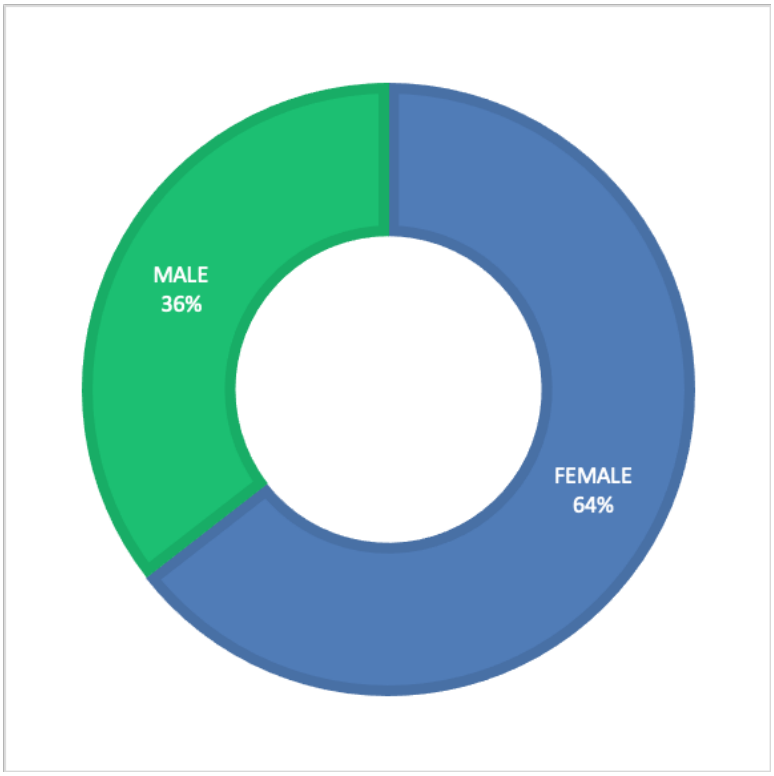
In the first part of our survey, participants answered questions regarding demographic information. This included their relationship to the person with the rare metabolic condition, the individual's gender, the individual's age, the state in which they lived, and the distance between their home and a major city. We also asked respondents to acknowledge the statement, "If you are not the individual with the rare disease, please answer the following as if he or she was filling out the survey." Most confirmed this (n=209, 89.5%), but 22 (10.5%) skipped the question.

The majority of respondents filled out the survey on behalf of their child (n=134, 58.0%) or family member (n=18, 7.79%) with a rare condition. 79 (34.2%) self-reported. Two hundred

and twenty-eight respondents reported the gender of the individual with the rare condition. Most were females (n=147, 64.5%), and 81 were males (35.5%; see Figure 1). Two hundred and thirty respondents gave the individual's age. These individuals ranged from under one year old to 88 years old. The mean age was 24.8, and the median was 17. Most (n=122, 53%) were under the age of 20, and 79 (34.3%) were under the age of 10 (Figure 2). Responses represented 45 states with the majority coming from California (n=20, 8.66%), New York (n=20, 8.66%), and Texas (n=12, 5.2%). Most individuals reported living within 30 miles from a major city (n=157, 68.0%) with 34 (14.7%) living between 30-60 miles away, and 40 (17.3%) living over 60 miles away.

Figure 1

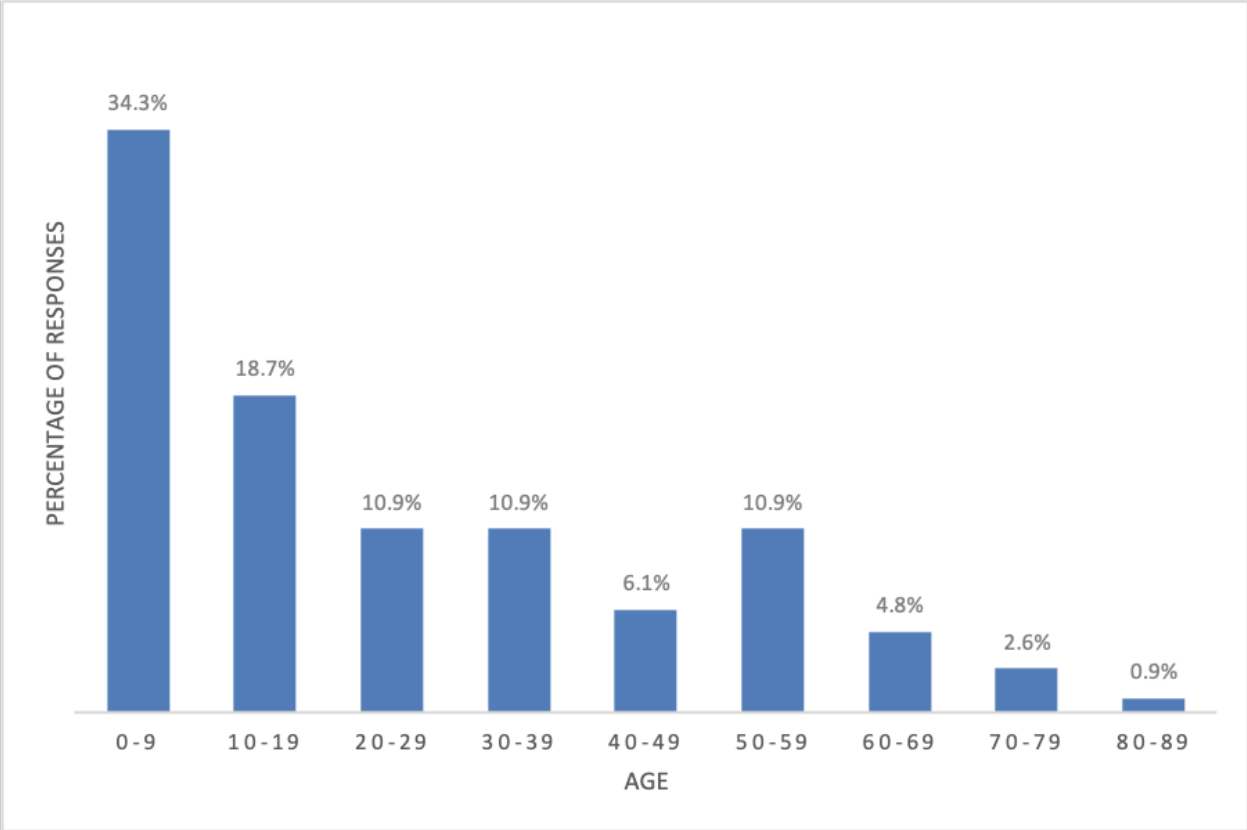
Gender of the Individual with the Rare Condition



Note: Results from 228 individuals; responses left blank (n=3) are not pictured.

Figure 2

Age of the Individual with the Rare Condition



Note: Results from 230 individuals; responses left blank (n=1) are not pictured.

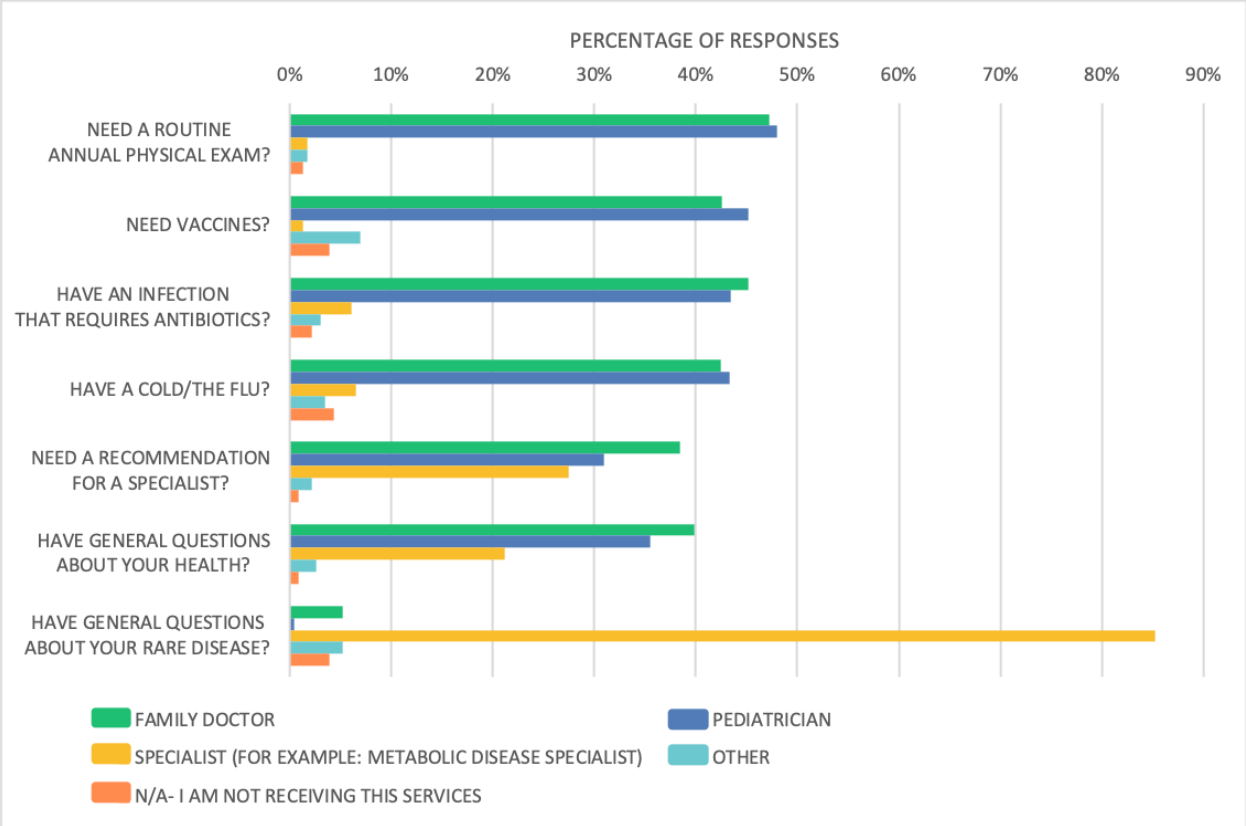
Primary Care Services Received and Healthcare Providers Involved

Participants were asked to select which healthcare provider they go to, if any, for various primary care services. The prompt "Who do you go to when you…" was followed by the following scenarios: "need a routine annual physical, need vaccines, have an infection that requires antibiotics, have a cold/the flu, need a recommendation for a specialist, have general questions about your health, have questions about your rare disease?". They were given the following options for healthcare providers: family doctor or other adult primary care doctor (internist); pediatrician; specialist (for example: metabolic disease specialist); other; and N/A–I am not receiving this service. Responses are visualized in Figure 3 and given as percentage of

the total in Table 1. Overall, respondents reported receiving the majority of their primary care services from PCPs (represented as green and dark blue bars in Fig. 3). For questions regarding their rare disease, 197 (85.3%) reported receiving this service from specialists, and 12 respondents (5.2%) reported receiving information on their rare disease from a source other than the healthcare providers listed. In each of the primary care categories, a small percentage of respondents reported they were not receiving the listed service. This was most notable for treatment of a cold/the flu, which 10 individuals (4.3%) reported they do not receive.

Figure 3

Primary Care Services Received and Healthcare Providers Involved



Caption: Bar chart depicting survey respondents' selections for healthcare professionals in response to the prompt "Who do you go to when you…"

Table 1

Primary Care Services Received and Healthcare Providers Involved

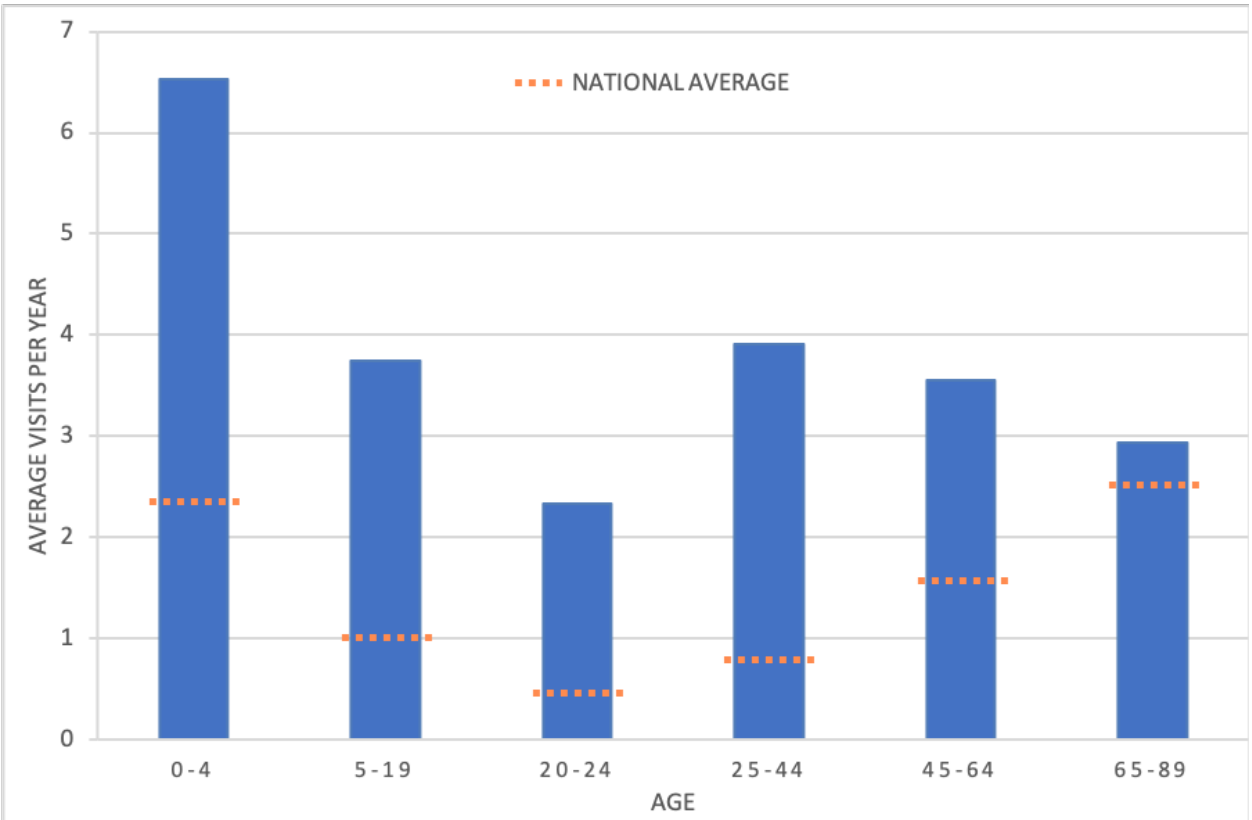
Healthcare Service Worker	FAMILY DOCTOR OR OTHER ADULT PRIMARY CARE DOCTOR (INTERNIST)	PEDIATRICIAN	SPECIALIST (FOR EXAMPLE: METABOLIC DISEASE SPECIALIST)	OTHER	N/A- I AM NOT RECEIVING THIS SERVICE	TOTAL
need a routine annual physical exam?	47.19% 109	48.05% 111	1.73% 4	1.73% 4	1.30% 3	231
need vaccines?	42.61% 98	45.22% 104	1.30% 3	6.96% 16	3.91% 9	230
have an infection that requires antibiotics?	45.22% 104	43.48% 100	6.09% 14	3.04% 7	2.17% 5	230
have a cold/the flu?	42.42% 98	43.29% 100	6.49% 15	3.46% 8	4.33% 10	231
need a recommendation for a specialist?	38.43% 88	31.00% 71	27.51% 63	2.18% 5	0.87% 2	229
have general questions about your health?	39.83% 92	35.50% 82	21.21% 49	2.60% 6	0.87% 2	231
have questions about your rare disease?	5.19% 12	0.43% 1	85.28% 197	5.19% 12	3.90% 9	231

Frequency of Visits for Primary Care Services

Following the question about primary care services and healthcare providers involved, 226 respondents answered the question "Using the situations above as examples of primary care, how many times a year do you visit a doctor for primary care?" For all respondents, the number of visits ranged from 0 to 25, the mean number of visits per year was 4.1, and the median was 3. In Figure 4, blue bars show the average number of visits for primary care services reported by our rare disease respondents. These are parsed into age categories that align with national data from the 2014 Medical Expenditure Panel Survey (MEPS), shown as orange dashed lines (Petterson et al., 2014). Of note, the data from the 2014 MEPS reflects mean numbers of visits to PCPs, which differs slightly from our question.

Figure 4

Annual Visits for Primary Care Services



Note: Results from 226 individuals; responses left blank (n=5) are not pictured. Age categories correspond to data from the 2014 Medical Expenditure Panel Survey (MEPS; Petterson et al., 2014). Blue bars represent results from survey respondents with rare disease. Dashed orange lines show the national average for annual number of visits to PCPs.

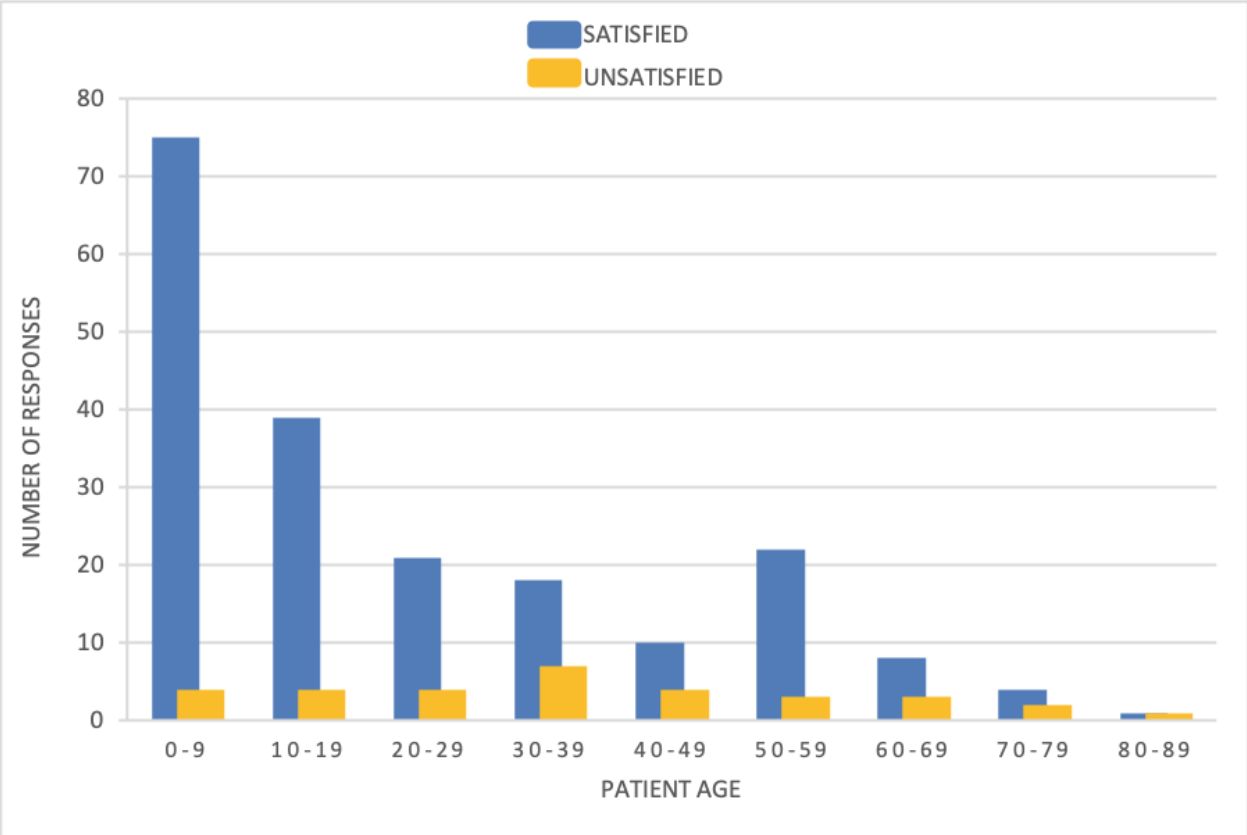
Patient Satisfaction

For the question "Do you feel your primary care needs are being met?", the majority of respondents selected "Yes" (n=199, 86.2%), and 32 (13.9%) selected "No". Neither gender nor distance from a major city were significantly correlated with patient satisfaction (χ^2: $p>.05$ for both analyses). When we assessed for correlation via logistic regression, age was also a poor predictor of patient satisfaction (R-Squared=0.03). Figure 5 shows a histogram of age versus number of respondents. Blue and yellow bars represent patients who feel satisfied and

unsatisfied with their primary care, respectively. The figure shows the significantly larger number of responses and uneven age distribution of the satisfied category. Because of the class imbalance between the two patient satisfaction categories, it is difficult to assess the correlation between age and patient satisfaction.

Figure 5

Patient Satisfaction by Age



Note: Results from 230 individuals; responses left blank (n=1) are not pictured.

Open-Ended Responses and Patient Preferences

In open-ended text boxes, we invited participants to comment on which type of healthcare professional provided their primary care services. A total of 212 participants commented. The responses were coded by theme (see Materials and Methods). We did not further describe 138 (53.3%) responses, which either did not answer the question (e.g.

respondents wrote “N/A”) or whose chart reflected they were receiving the majority of their primary care services from PCPs without providing further explanation. Common themes in the remaining 121 (46.7%) answers are listed in Figure 6. The most common reason cited for choosing to see a specific provider was distance (n=32, 26.4%). Notable quotes included:

- “My primary is in my hometown. The specialist is a 45 minute drive. She has taken an interest in GA1, even though it is not her specialty, and is in contact with the metabolic clinic any time it is warranted.”
- “Our pediatrician is fairly local and familiar with my journey with this condition, and keeps in communication with [the] metabolic team when needed.”
- “The pediatrician is very close. If we think a particular problem is rare disease related we call CHOP and ask questions. I wish there were a better way to get comprehensive care.”

Similarly, 22 respondents (18.2%) cited other logistical reasons, including insurance, hospital organization, or scheduling. Notable quotes included:

- “I see my metabolic doctor every 6 months so I use him as my primary.”
- “Only reason is because they are closer and I can get an [appointment] immediately. I cannot see my genetics [doctor] unless it is scheduled way in advance.”
- “Most of the services listed were related to general health. Our metabolic [doctors] won't give us the flu vaccine, for example (we've tried, because our kid is more comfortable with the metabolic team because she [sees] them more often). Generally, physicians keep to their own–metabolics treats metabolics, [pediatrics] treats all else.”

Twenty-two responses (18.2%) explained that they were receiving some or all of their primary care services from a specialist. Notable quotes include:

- "My pediatrician is the specialist when it comes to general care and vaccines. However, our metabolic physician is aware of when I get vaccines, [approves] any medicines before I take them, etc. the two doctor['s] offices are in communication with each other, too, so that each can do their job better when it comes to my condition."
- "Genetic doctor who is a pediatric metabolic specialist. She understands my needs better and the possible adverse issues to certain drugs and procedures for people like me. Our previous primary care [doctor], [didn't] care to learn or understand the rare disease part of me. My genetic [doctor] understands me as a whole, and the condition I have, the primary care did not or care to take time to learn. My geneticists [oversees referrals] to other specialist[s] as well [so] that [I] can benefit… when in need of one."

Nineteen responses (15.7%) described a desire for PCP-provided primary care with some mentioning themes like *comprehensive care*. Notable quotes include:

- "We see the pediatrician for most things so that we have one provider that has a full understanding of her overall health. The specialists' offices are less likely to pay attention to anything outside of their speciality."
- "We have so many specialists our PCP helps tie everything together."
- "Not everything is related to the rare disease - so a primary care [doctor] is necessary."

- "I visit my primary care doctor because he has been with me since birth and understands me as a person. He is not an expert in metabolics but is an expert when it comes to me and how I deal with my disease. My pediatrician consults with my specialists and approves needs such as physical therapy."
- "I have not found any other doctor, including a specialist, that has been able to help alleviate or improve my symptoms. Even the director of a major university hospital department near me was unable to provide any care for me and referred me back to my primary care doctor. My primary care doctor has been the only doctor who has made it his mission to discover what is going on in my body and to help me the best he can."

Only 13 respondents (10.7%) stated their PCP had insufficient knowledge of their rare disease. One respondent wrote:

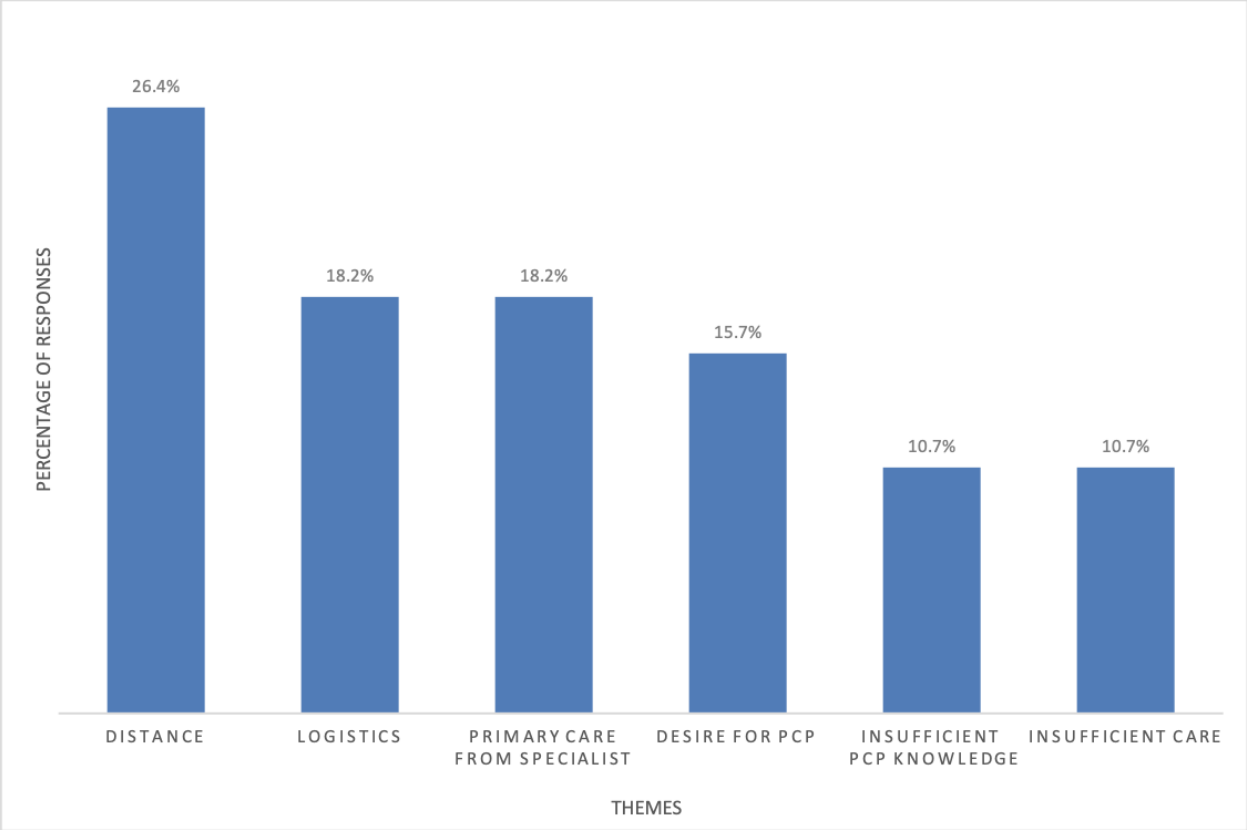
- "PCP wasn't comfortable caring for him. We now go to an internal specialist who has taken on the role of PCP."

Also, only 13 respondents (10.7%) stated they were receiving insufficient specialty care, primary care, or both. Notable quotes included:

- "I have [nowhere] else to go. No one seems to understand my genetic needs. Some care is better than none."
- "I seriously lack the assistance of a medical geneticist routinely, and a primary care doctor who has even minimal understanding of my disease and the medications I take, and those medications that are seriously contraindicated in my case. I am TERRIFIED in the event of an emergency."

Figure 6

Open-Ended Response Themes



Note: Results from 121 individuals; responses left blank or categorized as "No further explanation given" (n=138, 53.3%) are not pictured. Full titles of each category are as follows: Distance; Logistics (Insurance, Hospital Organization, or Scheduling); At Least Some Primary Care Services From Specialist; Desire for a PCP; Insufficient PCP Knowledge of Rare Disease; and Insufficient Primary Care, Specialty Care, or Both.

Discussion

Primary Care for Rare Disease Patients and the Healthcare Providers Involved

To our knowledge, our survey is the first to ask individuals with rare disease how they receive their primary care. Our data shows that, for the primary care services assessed, the overwhelming majority are provided by PCPs. For physical exams, vaccines, infections requiring antibiotics, and treatment for the cold/flu, 85-95% of respondents reported going to PCPs (Table 1). While PCPs provided the majority of services, specialists also played a

significant role in two primary care services: 27.5% of respondents said they see specialists when they need a recommendation for a new specialist (n=63), and 21.2% see specialists for general questions about their health (n=49). Similarly, 18.2% of open-ended responses mentioned seeing a specialist for at least some primary care services (n=22; Fig. 6). Our data corresponds to previous studies of PCP office records, which show that rare disease patients commonly visit PCPs (Phillips, 2009; Jo et al., 2019). Our results also show the complex nature of rare disease care–while PCPs can and do provide most of their primary care services, rare disease patients require additional assistance from specialists. This demands increased coordination from the healthcare teams and from the patients themselves in order to ensure proper primary care. *Coordination of care,* a prominent theme in physician-written literature, is therefore an evident need for the primary care of rare disease patients. It is one way PCPs can lessen the burden of rare disease.

In combination with our chart data (Fig. 3 and Table 1), our open-ended response analysis further suggests patients with rare disease recognize the aforementioned importance, purpose, and philosophy of primary care. Of respondents that explained which providers they prefer to visit, 15.7% (n=19) mentioned a desire to have a PCP providing them primary care. Some cited the ongoing relationship (in some cases since birth) that they shared with their PCP, reflecting the theme of *continuity of care*. Multiple stated a variation of *"my PCP understands me,"* reflecting the themes of *support, advocacy,* and *patient-centered approach.* Our preliminary exploration shows that rare disease patients see the same benefits of primary care that physicians do.

Unsurprisingly, 85.3% of respondents report seeing specialists when they have questions specifically about their rare disease (n=197; Table 1). Thirteen respondents (10.7%) stated they

felt their PCP had insufficient knowledge of their rare disease, and close to one third of patients even require specialists for referrals to other specialists (Fig. 3; Fig. 6; Table 1). As discussed previously, NORD physician guides exist for only 16 of the over 7,000 identified rare diseases (NORD, 2018). This equates to 0.23% of the known rare conditions. Knowledge for PCPs regarding rare disease is evidently scarce, and this is cited as an issue in physician-written literature (Knight and Senior, 2006). It follows that PCPs may be better able to serve their rare disease patients if they had more resources or training opportunities. We are unaware if or how PCPs are trained to handle patients with rare diseases, and our data shows a wide variation in how PCPs respond to these patients.

While our chart (Fig. 3 and Table 1) shows PCPs are providing the majority of primary care services for rare disease patients, the open-ended responses reveal a spectrum in PCPs' effort to learn about these conditions (Fig. 6). One of the respondents who felt his PCP had insufficient knowledge explained, "Our previous primary care [doctor], [didn't] care to learn or understand the rare disease part of me." In his chart, he reported seeing a specialist for most of his primary care needs. Another who received most of his primary care from a PCP said, "[My pediatrician] has taken an interest in GA1, even though it is not her specialty, and is in contact with the metabolic clinic any time it is warranted." For this patient, the PCP educated herself and provided *coordination of care* to support her patient. If all PCPs made the effort to educate themselves on their patients' rare conditions, they could significantly improve the overall management and quality of life for these individuals. As Knight and Senior (2006) argue, "General practice as a specialty has the opportunity to develop a generic approach to the common problem of rare diseases."

While the majority of respondents in our survey stated they were satisfied with their primary care, 32 (13.9%) said they were not. Likewise, 10.7% of open-ended responses (n=13) specifically stated they had insufficient primary care, specialty care, or both. Several respondents mentioned seeking information on their condition from other sources, such as the Internet. For example, one woman who sees a PCP for most of her care wrote, "I get other questions answered by looking on the internet or asking friends in similar situations. I get information about my rare disease from specialists, conferences or the internet." While that individual felt satisfied with her primary care and internet resources, other respondents show how devastating it can be to lack proper care. One respondent named the emotion brought on by improper care when she wrote, "I am TERRIFIED in the event of an emergency." Lacking proper surveillance and *coordination of care* from a trusted provider can be scary and even life-threatening.

Our survey shows that a significant portion of people with rare disease may be receiving unsatisfactory care. Like Jo et al. (2019) declare, "better understanding of the overall management of patients with rare diseases managed solely outside of primary care would help to improve the care for these patients." Our data suggests the portion of rare disease patients receiving primary care solely from specialists is very small; however, these patients may provide insight to future researchers that can better inform PCPs.

Distance and Logistics

While distance from a major city did not correlate with patient satisfaction (χ^2: $p>.05$), it was commonly cited as a reason rare disease respondents chose their healthcare providers (n=32, 36.4%). Surveys in Europe have shown that up to a quarter of individuals with rare disease travel to other regions and countries to access specialist care and diagnoses (EURORIDS, 2017).

It is not well understood how often and how far U.S. patients with rare disease are traveling for care, and our results suggest that distance to providers is a significant consideration in this process. PCPs who are prepared to manage their patients' rare conditions could improve their access to proper care and minimize costs associated with travel. To better understand this as a barrier to care, future studies in the U.S. could assess the distance, frequency, and cost of traveling for rare disease care.

Annual Visits to Primary Care for Rare Disease Patients

Our data suggests rare disease patients seek primary care services more times per year than the average U.S. patient. Per Petterson et al. (2014), the mean number of annual visits to primary care physicians varies by age in the U.S. As seen in Figure 4, our respondents visited healthcare providers for primary care services more times per year than the national average for each age category. In all groups except the oldest, individuals with rare diseases were receiving primary care services two to three times more than the average American. If a patient with rare disease requires primary care services more than twice as often as the average patient, this is a key way PCPs can contribute to rare disease management. To our knowledge, this is the first study to examine frequency of primary care visits for those with rare disease. This data can inform PCP training guidelines and understanding regarding their role for rare disease patients. S*upport, advocacy, coordination of care, patient-centered approach,* and *continuity of care* are the principles of primary care. As Knight and Senior (2016) argue:

> A systematic, generic primary-care approach to rare disease may reduce problems such as lack of coordinated care, lack of information, delayed diagnosis, and other difficulties encountered by people with rare diseases and their carers. It may enable the [PCP],

inevitably confronted by a person with a rare condition, to approach their care systematically.

Our data supports the ideas that PCPs will "inevitably" have rare disease patients and that they are well poised to advance rare disease management. Based on our data, U.S. rare disease patients understand the purpose of primary care and in fact seek it out more often than others.

Limitations

Even though we limited our survey to patients with rare metabolic conditions using the NIH's list, it includes a large variety of conditions. These range in severity, quality of life, life expectancy, and body system affected. The wide variance in these factors could influence individuals' primary care needs. While these metabolic conditions vary, our survey does not capture the experiences of those with any other subtype of rare disease. Additionally, we did not collect the rare disease diagnosis of each respondent. Future surveys of rare disease patients could therefore expand to cover more conditions and compare across the subtypes to better understand how diagnosis influences primary care.

In order to capture individuals who could not complete the survey on their own, we allowed parents and other family members to respond for them. We required respondents to acknowledge that they should answer as if they were the person with the rare condition. Twenty-two respondents (10.5%) skipped this question. Because we did not collect identifying information or conduct the surveys in person, it is possible some respondents answered questions with their own information rather than that of the individual with the condition. This could make some of our data inaccurate or skewed, which may explain the uneven gender distribution seen in question #4. It is possible that we have more female respondents (reported as 64.5% of the total)

than males because these are mothers. Additionally, due to our survey format, we are unable to verify whether or not individuals actually have rare disease diagnoses.

Lastly, due to discrepancies in healthcare systems across the globe, we limited our study to U.S. participants. Our data therefore does not represent trends in other countries' rare disease primary care. Additionally, several respondents cited logistical reasons for choosing their healthcare providers, such as health insurance or hospital system. We did not ask about health insurance information; thus, future research on this subject may better our understanding of the financial burdens associated with rare disease diagnoses.

Conclusion

The goal of our study was to understand which primary care services patients with rare diseases were receiving and which healthcare professionals were providing them. Our survey captured the perspective of those with rare disease and confirmed that these individuals receive the majority of their primary care from PCPs. Our study suggests that despite their generalist training, PCPs play an important role for rare disease patients. It is therefore important that PCPs be adequately prepared to handle rare diseases. Future research into health insurance, rare disease subtypes, and PCP training would further our ability to address the burden of those with rare disease.

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References

AAFP. (2019). AAFP definition of Primary Care. Retrieved from https://www.aafp.org/about/policies/all/primary-care.html#3

Almy, M.D., T. P. (1981). The Role of the Primary Physician in the Health-Care "Industry." *The New England Journal of Medicine*, *304*(4), 225–228. Retrieved from https://search-proquest-com.remote.slc.edu/docview/1870012747?OpenUrlRefId=info:xri/sid:wcdiscovery&accountid=13701

Anderson, M., Elliott, E. J., & Zurynski, Y. A. (2013). Australian families living with rare disease: Experiences of diagnosis, health services use and needs for psychosocial support. *Orphanet Journal of Rare Diseases*, *8*(1). https://doi.org/10.1186/1750-1172-8-22

Aymé, S., Kole, A., & Groft, S. (2008). Empowerment of patients: lessons from the rare diseases community. *The Lancet*, *371*(9629), 2048–2051. https://doi.org/10.1016/S0140-6736(08)60875-2

Baker, J. R., Crudder, S. O., Riske, B., Bias, V., & Forsberg, A. (2005, November). A model for a regional system of care to promote the health and well-being of people with rare chronic genetic disorders. *American Journal of Public Health*. American Public Health Association. https://doi.org/10.2105/AJPH.2004.051318

Bodenheimer, T., Wagner, E. H., & Grumbach, K. (2002). Improving Primary Care for Patients With Chronic Illness. *JAMA*, *288*(14), 1775–1779. Retrieved from www.jama.com

Coumou, H. C. H., & Meijman, F. J. (2006). How do primary care physicians seek answers to clinical questions? A literature review. *Journal of the Medical Library Association : JMLA*, *94*(1), 55–60. Retrieved from http://www.ncbi.nlm.nih.gov/pubmed/16404470

Cuevas, A. G., O'Brien, K., & Saha, S. (2019). Can patient-centered communication reduce the effects of medical mistrust on patients' decision making? *Health Psychology*, *38*(4), 325–333. https://doi.org/http://dx.doi.org/10.1037/hea0000721

Daughtridge, R., & Sloane, P. D. (2002). Physician-patient communication in the primary care office: a systematic review. *The Journal of the American Board of Family Practice / American Board of Family Practice*, *15*(1), 25–38. Retrieved from http://shibboleth.ovid.com/secure/?T=JS&CSC=Y&NEWS=N&PAGE=fulltext&D=emed5&AN=11841136%5Cnhttp://sfx.kcl.ac.uk/kings?sid=OVID:embase&id=pmid:&id=doi:&genre=article&atitle=Physician-patient+communication+in+the+primary+care+office%3A+a+systematic+review&ti

Dellve, L., Samuelsson, L., Tallborn, A., Fasth, A., & Hallberg, L. R.-M. (2006). Stress and well-being among parents of children with rare diseases: A prospective intervention study. *Journal of Advanced Nursing*, *53*(4), 392–402. https://doi.org/10.1111/j.1365-2648.2006.03736.x

Dharssi, S., Wong-Rieger, D., Harold, M., & Terry, S. (2017). Review of 11 national policies for rare diseases in the context of key patient needs. *Orphanet Journal of Rare Diseases*, *12*(63), 1–13. https://doi.org/10.1186/s13023-017-0618-0

Dimatteo, M. R. (1995). The Role of the Physician in the Emerging Health Care Environment. *Western Journal of Medicine*, *168*(5), 328–333. Retrieved from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1304975/pdf/westjmed00332-0038.pdf

Dudding-Byth, T. (2015). A powerful team: The family physician advocating for patients with a rare disease. *Australian Family Physician*, *44*(9), 634–638.

FAQs About Rare Diseases. (2017, November 30). Retrieved from https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

EURORDIS (2017). *EURORDIS SURVEY OF THE DELAY IN DIAGNOSIS WHAT IS A RARE DISEASE? FOR 8 RARE DISEASES IN EUROPE ('EURORDISCARE 2')*. Retrieved from http://www.eurordis.org/sites/default/files/publications/Fact_Sheet_Eurordiscare2.pdf

Fiscella, K., Meldrum, S., Franks, P., Shields, C. G., Duberstein, P., Mcdaniel, S. H., & Epstein, R. M. (2004). *Patient Trust: Is It Related to Patient-Centered Behavior of Primary Care Physicians? JSTOR* (Vol. 42). Retrieved from https://www-jstor-org.remote.slc.edu/stable/pdf/4640855.pdf?refreqid=excelsior%3A595a869d4265126f95dab211a98b86d0

Heritage, J., & Mcarthur, A. (2019). The diagnostic moment: A study in US primary care. *Social Science & Medicine*, *228*, 262–271. https://doi.org/10.1016/j.socscimed.2019.03.022

Hojat, M., Louis, D. Z., & Maxwell, K. (n.d.). A Brief Instrument to Measure Patients' Overall Satisfaction With Primary Care Physicians.

Hruby, M., Pantilat, S. Z., & Lo, B. (2001). How do patients view the role of the primary care physician in inpatient care? *The American Journal of Medicine*, *111*(9B), 21S-25S. Retrieved from http://www.ncbi.nlm.nih.gov/pubmed/11790364

Hummelinck, A., & Pollock, K. (2006). Parents' information needs about the treatment of their chronically ill child: A qualitative study. *Patient Education and Counseling*, *62*(2), 228–234. https://doi.org/10.1016/J.PEC.2005.07.006

I Am A Patient. (n.d.). Retrieved May 3, 2019, from https://www.theabfm.org/patient/i-am-patient

Jo, A., Larson, S., Carek, P., Peabody, M. R., Peterson, L. E., & Mainous, A. G. (2019). Prevalence and practice for rare diseases in primary care: a national cross-sectional study in the USA. *BMJ Open*, *9*(4), e027248. https://doi.org/10.1136/bmjopen-2018-027248

Jones, D. E. J., Sturm, E., & Lohse, A. W. (2018). Access to care in rare liver diseases: New challenges and new opportunities. *Journal of Hepatology*, *68*(3), 577–585. https://doi.org/10.1016/j.jhep.2017.11.004

Klabunde, C. N., Ambs, A., Keating, N. L., He, Y., Doucette, W. R., Tisnado, D., … Kahn, K. L. (2009). The role of primary care physicians in cancer care. *Journal of General Internal Medicine*, *24*(9), 1029–1036. https://doi.org/10.1007/s11606-009-1058-x

Knight, A. W., & Senior, T. P. (2006). The common problem of rare disease in general practice. *Medical Journal of Australia*, *185*(2), 83–83. Retrieved from www.mja.com.

Miller, F. A., Carroll, J. C., Wilson, B. J., Bytautas, J. P., Allanson, J., Cappelli, M., … Saibil, F. (2010). The primary care physician role in cancer genetics: a qualitative study of patient experience. *Oxford Journals: Family Practice*, *27*(5), 563–569. https://doi.org/10.1093/fampra/cmq035

National Institute of Health. *Metabolic disorders*. (n.d.). Retrieved March 8, 2020, from https://rarediseases.info.nih.gov/diseases/diseases-by-category/14/metabolic-disorders

NORD. (2018). Orphan Drugs in the United States. Exclusivity, Pricing and Treated Populations, (December), 22. Retrieved from https://www.iqvia.com/-/media/iqvia/pdfs/institute-reports/orphan-drugs-in-the-united-states-exclusivity-pricing-and-treated-populations.pdf?_=1554370338182

Nutt, S., & Limb, L. (2011). Survey of patients' and families' experiences of rare diseases reinforces calls for a rare disease strategy. *Social Care and Neurodisability*, *2*(4), 195–199. https://doi.org/10.1108/20420911111188443

Orphan Drug Act of 1983, Pub. L. 97-414, 96 Stat. 2049, codified as amended at 21 U.S.C. §§ 360aa-360ee

Ouyang, L., Grosse, S., Raspa, M., & Bailey, D. (2010). Employment impact and financial burden for families of children with fragile X syndrome: Findings from the National Fragile X Survey. *Journal of Intellectual Disability Research*, *54*(10), 918–928. https://doi.org/10.1111/j.1365-2788.2010.01320.x

Pelentsov, L. J., Fielder, A. L., Laws, T. A., & Esterman, A. J. (2016). The supportive care needs of parents with a child with a rare disease: results of an online survey. *BMC Family Practice*, *17*(1), 88. https://doi.org/10.1186/s12875-016-0488-x

Pelentsov, L. J., Laws, T. A., & Esterman, A. J. (2015). The supportive care needs of parents caring for a child with a rare disease: A scoping review. *Disability and Health Journal*, *8*(4), 475–491. https://doi.org/10.1016/J.DHJO.2015.03.009

Pelentsov, L. J., O'Shaughnessy, P., Laws, T. A., & Esterman, A. J. (2014). What are the supportive care needs of parents caring for a child diagnosed with ectodermal dysplasia: A rare genetic disorder? *International Journal of Child Health and Human Development*, *7*(1), 23–29. Retrieved from http://search.ebscohost.com/login.aspx?direct=true&db=psyh&AN=2014-14359-004&site=ehost-live%5Cnlemuel.pelentsov@unisa.edu.au

Petterson S, McNellis R, Klink K, Meyers D, Bazemore A. The State of Primary Care in the United States: A Chartbook of Facts and Statistics. January 2018.

Phillips, R. L. (2005). Primary care in the United States: problems and possibilities. *BMJ (Clinical Research Ed.)*, *331*(7529), 1400–1402. https://doi.org/10.1136/bmj.331.7529.1400

Phillips, W. R. (2009). Zebras on the Commons: Rare Conditions in Family Practice. *The Journal of the American Board of Family Medicine*, *17*(4), 283–286. https://doi.org/10.3122/jabfm.17.4.283

Pierucci, P., Lenato, G. M., Suppressa, P., Lastella, P., Triggiani, V., Valerio, R., … Sabb, C. (2012). A long diagnostic delay in patients with Hereditary Haemorrhagic Telangiectasia: A questionnaire-based retrospective study. *Orphanet Journal of Rare Diseases*, *7*(1), 33. https://doi.org/10.1186/1750-1172-7-33

Royal Australian College of General Practitioners. (n.d.). *Australian family physician.* Australian College of General Practitioners. Retrieved from https://search.informit.com.au/documentSummary;dn=512019547619809;res=IELHEA

Smith, J., & Sibthorpe, B. (2007). Divisions of general practice in Australia: how do they measure up in the international context? *Australia and New Zealand Health Policy*, *4*, 15. https://doi.org/10.1186/1743-8462-4-15

Stoller, J. K. (2018). The Challenge of Rare Diseases. *Chest*, *153*(6), 1309–1314. https://doi.org/10.1016/j.chest.2017.12.018

Wallenius, E., Möller, K., & Berglund, B. (2015). Everyday Impact of Having a Rare Diagnosis. A Questionnaire Study. *Nordic Journal of Nursing Research*, *29*(3), 13–17. https://doi.org/10.1177/010740830902900304

Weng, H.-J., Niu, D.-M., Turale, S., Tsao, L.-I., Shih, F.-J., Yamamoto-Mitani, N., … Shih, F.-J. (2012). Family caregiver distress with children having rare genetic disorders: a

qualitative study involving Russell-Silver Syndrome in Taiwan. *Journal of Clinical Nursing*, *21*(1–2), 160–169. https://doi.org/10.1111/j.1365-2702.2010.03583.x

Appendix A: Survey Consent

We are asking you to take part in a research study at Sarah Lawrence College. Please read through the following questions and responses that will help you to decide whether or not to participate.
What is the purpose of this study?

- The purpose of this study is to learn how individuals with rare disease receive primary care services.

Why am I being asked to participate?

- You are being asked to participate in this survey because you are an individual with a rare disease OR you are a parent/legal guardian of an individual with a rare disease.

What will I be asked to do?

- You will be asked to complete an anonymous 5 minute survey with questions about routine healthcare services and providers.

Is my participation voluntary?

- Participation is entirely voluntary. You can choose to opt out of the study at any point and doing so will not affect your relationship with Sarah Lawrence College.

Do I have to answer every question?

- You can choose not to answer specific questions without having to justify your choice.

Are there any benefits or risks associated with my participation in this study?

- There are no direct benefits for your participation in the survey.The risks involved with this survey are minimal.

Will I be compensated for my participation?

- No, you will not be compensated.

Will the information I provide be kept confidential?

- The survey does not collect any information (such as your name or date of birth) that could be used to identify you. You will not be identified in any written or oral report of the research study.

If I have any questions or concerns after the study, how can I contact you?

- Please email us at ekahan@gm.slc.edu if you have any other questions. Our faculty advisor, Janelle Villiers, is available at jvilliers@sarahlawrence.eduWho can I contact if I

have questions about my rights as a research participant?The IRB co-chairs Professors Elizabeth Johnston (914-323-6672) and Claire Davis (914-395-2605) at irb@sarahlawrence.edu.Contact information:Researchers: Jasmine Chao, Catherine Mayo, Eliana Kahan Spielmanekahan@gm.slc.edu45 Wrexham Road, Bronxville, NY 10708By clicking "I agree", you are agreeing that you have read the above, understand your rights, and voluntarily agree to participate in this study.

Appendix B: Survey Questions

Title: Research Study on Primary Care for Rare Disease Patients

Question 1: [Survey consent information; See Appendix A]

Question 2: Are you the person with the rare disease or are you filling out this survey on behalf of your child/family member?

- Self
- Child
- Other Family Member

Question 3: If you are not the individual with the rare disease, please answer the following as if he or she was filling out the survey.

- I understand

Question 4: What is your Gender?

- Male
- Female
- Other

Question 5: How old are you in years? (Please use whole numbers-not decimals. For children under 1, please put 0)

- Free response

Question 6: What state do you live in?

- Alabama
- Alaska
- N/A I do not live in the United States
- Arizona
- Arkansas
- American Samoa
- California
- Colorado
- Connecticut
- Delaware
- District of Columbia
- Florida
- Georgia
- Guam
- Hawaii
- Idaho
- Illinois
- Indiana
- Iowa

- Kansas
- Kentucky
- Louisiana
- Maine
- Maryland
- Massachusetts
- Michigan
- Minnesota
- Mississippi
- Missouri
- Montana
- Nebraska
- Nevada
- New Hampshire
- New Jersey
- New Mexico
- New York
- North Carolina
- North Dakota
- Northern Marianas Islands
- Ohio
- Oklahoma
- Oregon
- Pennsylvania
- Rhode Island
- South Carolina
- South Dakota
- Tennessee
- Texas
- Utah
- Vermont
- Virginia
- Virgin Islands
- Washington
- West Virginia
- Wisconsin
- Wyoming

Question 7: How far would you say you live from a major city?

- 0 to 30 miles
- 30 to 60 miles

- 60 + miles

Question 8: For the following primary care services, who do you go to when you

- need a routine annual physical exam?
    - Family doctor or other adult primary care doctor (internist)
    - Pediatrician
    - Specialist (for example: metabolic disease specialist)
    - Other
    - N/A- I am not receiving this service
- need vaccines?
    - Family doctor or other adult primary care doctor (internist)
    - Pediatrician
    - Specialist (for example: metabolic disease specialist)
    - Other
    - N/A- I am not receiving this service
- have an infection that requires antibiotics?
    - Family doctor or other adult primary care doctor (internist)
    - Pediatrician
    - Specialist (for example: metabolic disease specialist)
    - Other
    - N/A- I am not receiving this service
- have a cold/the flu?
    - Family doctor or other adult primary care doctor (internist)
    - Pediatrician
    - Specialist (for example: metabolic disease specialist)
    - Other
    - N/A- I am not receiving this service
- need a recommendation for a specialist?
    - Family doctor or other adult primary care doctor (internist)
    - Pediatrician
    - Specialist (for example: metabolic disease specialist)
    - Other
    - N/A- I am not receiving this service
- have general questions about your health?
    - Family doctor or other adult primary care doctor (internist)
    - Pediatrician
    - Specialist (for example: metabolic disease specialist)
    - Other
    - N/A- I am not receiving this service
- have questions about your rare disease?
    - Family doctor or other adult primary care doctor (internist)

- Pediatrician
- Specialist (for example: metabolic disease specialist)
- Other
- N/A- I am not receiving this service

Question 9: If you selected other for any of the primary care services above please explain.

- Free response

Question 10: Using the situations above as examples of primary care, how many times a year do you visit a doctor for primary care?

- Free response

Question 11: Please explain your reasoning for receiving primary care services from the provider(s) you selected prior.For example, “I see my medical geneticist routinely, so it makes sense to also capture my primary care when I’m there.

- Free response

Question 12: Do you feel your primary care needs are being met?

- Yes
- No