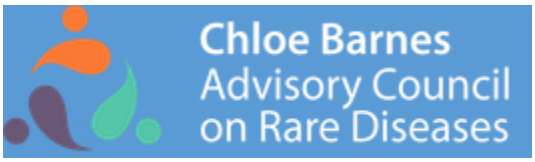
Rare Disease Patient Healthcare Access Survey 2020-2021: Preliminary Qualitative Findings for Barriers and Facilitators



Research Report Prepared by Amanda Hemmesch, Ph.D., & Kathleen Bogart, Ph.D.

This report was prepared for the Chloe Barnes Advisory Council on Rare Diseases by:

Amanda Hemmesch, Ph.D., Professor of Psychology & Co-Director of the SCSU Survey Center, St. Cloud State University
Kathleen Bogart, Ph.D., Associate Professor of Psychology & Director of the Disability and Social Interaction Lab, Oregon State University

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Patient Health Care Access Survey 2020-2021Survey Report

We conducted an online survey of Minnesota and US residents to learn more health care access and quality of life for adults with rare disease and parents of children with rare disease. This report is a summary of the methods of study design and data collection, and preliminary qualitative results.

Executive Summary

This online study included 1,128 participants, including adults with rare disease and parents/relatives/caregivers to individuals with rare disease. Quantitative analyses showed few differences between Minnesotans in this study and the larger US sample, so these groups were not separated for qualitative analyses. Data was analyzed for the full sample to provide a picture of the status of health care access for individuals with rare disease in the US.

Approximately 70% of participants identified as an adult living with a rare disease, 22% were a parent of a child with a rare disease, and approximately 8% were relatives or caregivers to someone with a rare disease. Participants reported that 80% of the individuals with the rare disease were adults and approximately 20% were children. There were 344 different rare diseases represented. Most participants reported only one rare disease (88%), though participants reported having up to 5 rare diseases.

Qualitative Findings: Barriers and Facilitators for Diagnosis, Management, and Treatment of RD

Participants were asked to share their three most significant barriers and facilitators to diagnosis, management or treatment of their RD. Barriers were designed to capture experiences that have felt difficult or frustrating; facilitators were designed to capture experiences that felt positive or helpful.

Two overarching themes emerged when analyzing this data. When considering barriers, participants often responded by describing **lack or absence of** factors they perceived would be helpful to their experiences with RD. When considering facilitators, participants often responded that they were **finally** able to find what they thought was necessary, implying a need for persistence regarding their RD.

The ten subthemes that were identified are (in no particular order): doctors and healthcare providers; diagnosis; medications and treatments; insurance and other costs; social support; knowledge, understanding, awareness, and education; research; psychosocial factors; accessibility and accommodations; and nothing (i.e., a lack of identified barriers or facilitators). These subthemes cut across barriers and facilitators, such that each subtheme was identified as a barrier in some instances and a facilitator in others.

Methods

We cooperated with the Chloe Barnes Advisory Council on Rare Diseases and their Barriers to Care committee to conduct a study of health care access and experiences living with a rare disease. See Appendix I for the questionnaire.

Data was collected with an Internet survey. Data was cleaned and analyzed using standard statistical and qualitative methods with the software packages SPSS (IBM), Microsoft Excel, and Qualtrics. This report outlines the study design, data collection methods, and preliminary qualitative results.

This study was conducted from February 2020 – February 2021. The study design and questionnaire were developed February – September 2020. Data was collected October 2020 – February 2021. Data cleaning and analysis was conducted in March – April 2021 for the quantitative portion of the

study. Qualitative data cleaning and analysis began in May 2021 and is ongoing. This study was reviewed by the St. Cloud State University, Oregon State University, and University of Minnesota Medical School institutional review boards.

This study was conducted during the COVID-19 pandemic. Participants were instructed to answer according to their experiences before the COVID-19 pandemic (except when responding to standardized measures, like the PROMIS scales), and that there was a separate question near the end of the survey to ask about whether their experiences were different during COVID-19.

Study Design and Data Collection

An Internet survey offers many advantages. Internet surveys tend to be perceived as more accessible and more convenient by participants, and can reach participants who are not easily reachable through telephone or mail surveys. Our communications culture has changed, and continues to change, very rapidly. Today there are substantial portions of the general population whose primary means of communication is electronic, and who very infrequently use, and often actively avoid using, (voice) telephone communication. Similar to landline telephones of the past, nearly everyone in our society has access to Internet communications, and so this method offers the potential for reaching a very large swath of the population of individuals with rare diagnoses. Additionally, data collection by way of an Internet survey is often, although not always, cheaper than traditional telephone or mail surveys.

However, Internet surveys present a particular set of challenges for survey researchers, and for those using the information generated through survey studies. As there is no comprehensive list of Internet or email users, and no way to randomly generate Internet contacts such as email addresses, internet surveys require generating contacts through other means. For this study, we used targeted recruiting through health care providers, rare disease support organizations, members and affiliates of the Chloe Barnes Advisory Council on Rare Diseases, Facebook and online posts, and snowball sampling.

Protecting Participants' Privacy and Data Security

There were no identifying features on the questionnaires that would indicate a particular household or individual. The software system used for data collection, Qualtrics, collects IP addresses of the device on which the survey is taken, as well as rough geographic location information. This information, like an email address, could be used to identify an individual respondent. Therefore, we removed IP addresses and all location data from the dataset before analysis, leaving only an anonymous dataset. All information is stored on password protected computers and password protected institutional cloud storage systems.

Participants were limited to those age 18 or older who were currently living with a rare disease, or were parents, relatives, or caregivers to someone currently living with a rare disease.

Online Data Collection

Data was collated by the Qualtrics software, and then exported as necessary as a csv file to use with other analysis software packages.

Multiple responses from the same IP address were not allowed for this study. The prevention of multiple responses was a concern because online surveys can enable motivated participants to skew

the data by submitting multiple surveys in an attempt to give their opinions more weight. Data corresponding to the same IP address were checked before IP addresses were removed from the dataset to ensure that no duplicate entries were included per IP address (the first fully completed submission or most complete of multiple submissions was retained and any other submissions were removed).

Data Cleaning and Preparation

The dataset was cleaned by removing any data from the testing phase of the project (16 cases), responses from individuals who did not agree to take the survey (32 cases; see Appendix I for implied consent language), and removing those who replied to the age demographic question with an age under 18 years old (4 cases). Additionally, 25 cases from duplicate IP addresses were removed from the dataset. During preliminary cleaning, 116 cases were also removed because they reported that the person with the rare disease recovered or died. After preliminary cleaning, the dataset included information from 1,445 participants.

Participants were asked to identify their rare disease(s) (see Appendix 2). Two trained undergraduate research assistants checked self-reported diagnoses against the list of rare diseases maintained by NORD's Genetic and Rare Disorders Information Center (https://rarediseases.info.nih.gov) to determine if they met the criteria to be considered rare diseases. Coding-related discrepancies and questions were resolved by KB. 317 participants' data were removed from the dataset because they did not self-report at least one rare disease.

This resulted in a final dataset that included 1,128 participants with verified rare diagnoses. Of this participants, 868 (77%) completed the entire survey; 260 (23%) have some missing data. As a result of missing data, sample sizes vary across analyses.

Data Cleaning and Preparation

We used a thematic approach for qualitative data analysis (Braun & Clark, 2006). This approach to qualitative data analysis involves multiple steps. Thematic analysis is relatively flexible and can be used to identify themes in qualitative data whether or not existing theories can be used as a lens for interpreting data. Themes identify a pattern of responses within the data that captures something important about participant responses to the topic. The goal is to identify themes that capture the richness (in terms of breadth or depth) of the data. Because little theory exists that identifies potential barriers or facilitators for healthcare access in rare disease, we used a bottom-up approach to data analysis. For each open-ended item, a team of three coders (two undergraduate students and AH) completed the following steps: data familiarization, generating initial codes, searching for themes, reviewing themes, and defining themes. Due to changing availability, additional undergraduate coders were recruited to replace those who were no long able to participate in this project. During data familiarization, coders independently read through participants' responses at least twice. When generating initial codes, coders took notes about items that seemed to capture something important about the data. After each individual coder identified their own initial codes, coders met weekly to discuss their notes and identify themes that were agreed to capture important information in the data. When searching for, reviewing, and defining themes, coders met regularly to discuss how to organize and define codes. Coders decided that this data included both overarching themes and smaller subthemes that described specific barriers and facilitators. All of the themes

described in this report were finalized when coders achieved consensus (i.e., all three coders agreed on the theme and its definition).

Preliminary Qualitative Results

Barriers and Facilitators for Diagnosis, Management, and Treatment of RD

Participants were asked to share their three most significant barriers and facilitators to diagnosis, management or treatment of their RD. Barriers were designed to capture experiences that have felt difficult or frustrating; facilitators were designed to capture experiences that felt positive or helpful.

Coders identified themes for barriers (e.g., challenges) and facilitators (e.g., aides) independently, then reviewed each set of themes together to identify themes that applied to both barriers and facilitators. Coders then identified subthemes that summarized responses to the barriers and facilitators items that provided more detail about participants' responses.

When considering barriers, participants often responded by describing lack or absence of factors they perceived would be helpful to their experiences living with RD. When considering facilitators, participants often responded that they were finally able to find what they needed, implying a need for persistence regarding their RD. The ten subthemes that were identified are (in no particular order): doctors and healthcare providers; diagnosis; medications and treatments; insurance and other costs; social support; knowledge, understanding, awareness, and education; research; psychosocial factors; accessibility and accommodations; and nothing (i.e., a lack of identified barriers or facilitators).

Barriers Theme: "Lack"

The primary theme that was identified when reviewing participants' responses about barriers to successful diagnosis, treatment, or management of their RD was "**lack**." Participants wrote about a lack of many types of support that they believed would be helpful to their RD experience. This sense of lack or absence was pervasive across the barriers identified. For example, many participants wrote about how the absence of a doctor/healthcare provider knowledgeable in RD hindered their ability to get an accurate diagnosis and pursue treatments to manage symptoms. Many participants also wrote about the lack of a cure for their RDs, and sometimes a lack of (effective) treatments to manage symptoms, too. A lack of knowledge and awareness about their specific RD and RDs in general was also identified as a barrier. Participants seemed to be wanting for more to help them manage their RD. Participants wrote about the need to persist and to advocate for themselves as they tried to manage their RD. "Lack" was associated with a variety of negative participant responses, including frustration, anxiety, depression, stress, and stigma.

Facilitators Theme: "Finally"

The primary theme identified when participants considered facilitators was "**finally**." Participants wrote about the persistence needed to manage their RD and the healthcare system. For example, many participants who identified doctors and other healthcare providers as facilitators wrote about how they had to search to find the right doctor/provider, one who didn't dismiss their concerns and who seemed knowledgeable about their RD. Consistent with other RD research, participants often reported diagnostic odysseys as barriers; however, participants also highlighted the importance of an accurate diagnosis as a facilitator for managing their RDs. Relatedly, participants wrote about their

experiences seeking treatments to manage their RD symptoms that felt like trial-and-error, and expressed relief when they were able to receive a medication or treatment that improved their quality of life. Participants identified personal attributes as important in their RD experience, life optimism, persistence, and a positive outlook. As a complement to internal factors, participants highlighted the importance of social support from various sources (e.g., family, friends, and online support communities like Facebook groups) as important for their ability to manage their RDs. Although many different facilitators were identified, many participants expressed relief upon finally finding the resources that were important to their RD experience. In addition to relief, facilitators seemed to bring feelings of validation and the opportunity for greater inclusion and improved quality of life.

Subtheme: Doctors and Healthcare Providers

One subtheme we identified in the qualitative responses about both barriers and facilitators was the critical role that doctors and other healthcare providers play in the experience of people with rare disease (RD). Participants reported that doctors/healthcare providers were both barriers and facilitators for managing their RDs. For example, doctors and other health care providers were seen as barriers when they dismissed participants' concerns about symptoms and when they lacked knowledge about RDs. Doctors and other providers were sometimes perceived as not listening, caring, or knowing. Participants also reported experiences of medical bias as barriers, including perceptions that their gender, race, age, or being overweight or obese prevented them from receiving quality care from doctors and other providers. Some participants reported barriers that could potentially qualify as medical gaslighting, a term used to describe doctor/provider behaviors that downplay, question, or dismiss patients' concerns. For example, some participants felt that doctors and other providers were too quick to assume that mental health issues were responsible for their symptoms; as a result, those providers did not search for other explanations for participants' distressing symptoms.

However, doctors were seen as facilitators when participants believed that they listened to their concerns and took them seriously, were willing to search for causes of symptoms, and when they had knowledge/expertise about RD generally and about participants' specific rare diagnoses. Participants often described needing persistence to find the right doctor(s) or provider(s). Participants seemed grateful to find doctors and healthcare providers who were willing to work with them to help them diagnose and manage their RD.

Subtheme: Diagnosis

Another subtheme that we identified in the data was importance of diagnosis. Participants reported that lack of diagnosis and misdiagnosis were significant barriers living with RD. The identification of diagnosis as a significant barrier is consistent with quantitative findings from this patient survey, which found that many participants experienced delays and some experienced misdiagnosis before receiving a correct RD diagnosis.

Receiving an accurate diagnosis was identified as an important facilitator, as that often opened the door to being taken seriously by doctors/providers and pursuing medications or treatments to manage RD symptoms. Diagnosis also gave participants the information needed to start learning about their RD symptoms and management, and to start seeking RD-specific social support.

Subtheme: Medications and Treatments

Many participants also identified symptoms as a potential barrier for managing their RD and for more general engagement with work, school, and other domains important for quality of life. Participants identified a variety of symptoms as barriers, including pain and fatigue, as well as mobility, sleep, sensory, neurological, and communication issues (among others). Some participants were aware that there was no cure for their RD and were disappointed/frustrated in a lack of other options for managing their RD. As a result, many participants mentioned medications and treatments when considering barriers and facilitators.

Medications were perceived as barriers when they did not exist to treat participants' conditions, work consistently, or maintain efficacy over time, and when they were classified as controlled substances. Participants also reported that lack of treatment options (including FDA-approved treatments) was a significant barrier. The inability to effectively manage their symptoms has potentially important effects on overall quality of life; participants who identified medications or treatments as barriers often wrote about how they impacted their ability to fully participate in activities that were important to them, like work, school, and social relationships.

Other participants reported that medications and treatments were important facilitators. Medications, treatments, and medical services (e.g., physical, speech, and occupational therapy) allowed some participants to manage their symptoms to limit their impact on quality of life. Some participants reported needing to try multiple medications or treatments before finding one(s) that was effective. Some participants also reported seeking non-medical treatments to manage their RD, like diet and exercise. The details of managing symptoms via medications or treatments varied widely across participants, but these experiences shared a common theme of appreciation for maintaining or improving quality of life.

Subtheme: Insurance and Other Costs

Consistent with quantitative findings, participants wrote about insurance and costs when considering significant barriers and facilitators.

Participants identified difficulty getting health insurance approval for tests, treatments, services, and specialist appointments as a barrier. Outside of (or perhaps related to) insurance as a barrier, participants also identified the cost of tests, medications, and treatments, and their finances more generally as barriers.

Other participants identified health insurance as a facilitator for getting tests, treatments, services, and specialist appointments. In addition to health insurance, participants also reported that other programs and waivers designed to reduce the financial impact of RD were facilitators.

Subtheme: Social Support

Another subtheme identified in participants' responses was the importance of social support.

Participants didn't specifically mention a lack of social support as a barrier, but they described not being believed, taken seriously, or understood by others (e.g., family, friends, colleagues), which could represent a lack of social support. Participants also identified isolation and a lack of inclusion in work, education, and social settings as barriers, which are related to a lack of support. There has been much research connecting isolation and a lack of social support to poor physical and mental health outcomes, which provides important context for interpreting the experiences that participants shared when considering barriers. A few participants mentioned that they felt that it was difficult to

diagnose/manage/treat their RD because so few people share their diagnosis, which was both isolating and made navigating healthcare more difficult.

Social support was mentioned more directly and frequently when participants considered facilitators. Participants identified a variety of formal and informal sources of social support. Regarding informal support, participants reported receiving social support from spouses/partners, family, friends, siblings, extended family, peers, and their communities. Participants also reported receiving social support through Facebook and other social media platforms and websites. These online platforms helped participants connect to others who share their RD. Many participants sought online support, especially when they perceived that the healthcare system failed them. Online support provided participants with more information about how to pursue a diagnosis and how to manage both RD symptoms and the interpersonal aspects of having an RD. Participants also identified more formal sources of social support as facilitators to their experience living with RD, including from local, national, and international support/advocacy groups/foundations, conferences, and support groups. In additional to social connection and support, these formal sources of support often also provided knowledge and empowerment.

Subtheme: Knowledge, Understanding, Awareness, and Education

Knowledge, understanding, awareness, and education were commonly cited when participants considered both barriers and facilitators.

As a barrier, participant responses identified lack of knowledge, understanding, awareness, and/or education as challenges both when interacting with doctors and health care providers and more generally. Many participants wrote about the difficulties they experienced when doctors were not familiar with their RD and with finding quality information about their RD.

As a facilitator, knowledge, understanding, awareness, and/or education were identified as helpful when participants were able to find knowledgeable doctors and health care providers, and when participants were able to learn more about their RD. Participants searched for information about potential causes of their RDs, the progression of their RDs, and medications, treatments, services, or cures that could improve quality of life. The information that participants sought ranged from informal online information and lived experiences to more formal sources like research papers and results from trials.

Subtheme: Psychosocial Factors

Participants identified psychosocial factors as significant barriers and facilitators. When considering barriers, participants reported that social processes like stigma and discrimination had significant effects on their experiences with RD. Additionally, some participants identified personal responses to their RD or healthcare access, such as self-doubt, self-stigma, anger, depression, anxiety, and stress, as barriers. These negative feelings and experiences could potentially complicate participants' attempts to manage their RDs.

When considering facilitators, participants identified psychological factors as helpful for their experience living with RD. For example, participants reported that their perseverance/persistence, self-advocacy, outlook, positive attitude, and a sense of empowerment were helpful for navigating their RD. Some participants mentioned faith and religion as facilitators, too. Social support was also commonly mentioned as a facilitator (see above for more detail). Participants often attributed their

success to managing their RD and navigating the healthcare system to these positive personal characteristics.

Subtheme: Research

An additional subtheme that was identified in the data was the importance of research. Participants reported that lack of research, and the subsequent dearth of knowledge, about RD and their treatments or cures was a barrier to their ability to successfully manage their RD.

Research about RD and treatments/cure was identified as a significant facilitator. Participants were interested in learning more about and participating in clinical trials and other studies that could improve understanding of their RD and potential treatments.

Subtheme: Accessibility and Accommodations

A final subtheme that was identified across barriers and facilitators related to accessibility and accommodations. Participants reported a few ways that they encountered accessibility as a barrier. Participants reported experiencing problems with the legal aspects of accessibility, including challenges getting appropriate Individualized Education Programs (IEPs) in education settings and accommodations protected under the Americans with Disabilities Act (ADA) in work and other settings. Participants also identified accessibility issues around transportation/travel for medical appointments, which can be more difficult when participants experience symptoms that interfere with sensory and mobility systems (i.e., a lack of affordable, accessible public transportation; poorly designed clinics). Finally, some participants wrote about the long distances that they are required to travel to access specialists, special centers, and treatments as accessibility issues.

Participants also reported positive/successful experiences with accessibility. When considering facilitators, some participants pointed to IEPs and reasonable accommodations provided by the ADA (e.g., disability parking) as important facilitators. Participants also identified programs such as unemployment insurance, and Social Security Disability Insurance as facilitators that helped them to manage their RD. Additionally, participants experienced sensory and mobility aides as facilitators that helped them to manage their RD symptoms. Finally, some participants wrote that apps, electronic medical records (especially those that allow asynchronous communication, like messaging with doctors/providers), and telehealth improved their experience living with RD. All of these facilitators improved participants' inclusion in important domains of their lives, including collaborating with doctors and other healthcare providers about managing their RDs.

Subtheme: Nothing/None

A subgroup of participants did not respond to the open-ended questions asking about barriers or facilitators, or responded with answers like 'none' or 'nothing'. Some of these participants may not have experienced barriers or facilitators for their RD diagnosis, management, or treatment, and some may have opted not to answer the open-ended questions. When comparing these preliminary qualitative results with those from the quantitative portion of this study, it is not surprising that some participants did not report barriers or facilitators. Quantitative results showed that between one-third and one-half of participants reported that common barriers to healthcare access (e.g., travel distance, lack of childcare, insurance) were never barriers for them, and many participants reported accessing a variety of healthcare-related doctors/providers, treatments, and services.

Changes to Healthcare Access due to the COVID-19 Pandemic

Qualitative responses to these open-ended question are still being analyzed.

One Single Item to Improve Healthcare Experience

Qualitative responses to these open-ended question are still being analyzed.

APPENDIX I: QUESTIONNAIRE

Q1

Rare Disease Health Care Access Survey

This survey is a partnership between the Chloe Barnes Advisory Council on Rare Disease established at the University of Minnesota, St. Cloud State University, and Oregon State University

Purpose: You are being asked to take part in a research study. The purpose of this research study is to examine the experience of seeking and accessing health care for rare diseases, including unique barriers that individuals living with rare disease may experience.

Activities: The study activities include completing a survey that is adapted from established questionnaires. Your participation in this study will last about 30 minutes.

Risks: Although we have steps in place to maintain your confidentiality, there is almost always a chance of breach of confidentiality in web-based research. Risks associated with internet research may include: the existence of data on backups or server logs beyond the time frame of the research project; interception, corruption, loss, or destruction of data, or that data may arrive late, be incomplete, or contain viruses.

Benefit: We do not know if you will benefit from being in this study, but completing the survey may help you gain knowledge and insight into your experiences with your rare disorder. By understanding challenges that people with rare disease face, there may be programs/offerings put in place to address those challenges.

Payment: You will not be paid for participating in this survey.

Confidentiality: The information you provide during this research study will be kept confidential to the extent permitted by law. Research records will be stored securely for the use by the research team. If the results of this project are published, your identity will not be made public.

Voluntary: Participation in this study is voluntary. You are free to skip any questions that you would prefer not to answer. You are free to withdraw at any time without penalty. If you choose to withdraw from this project before it ends, the researchers may keep information collected about you and this information may be included in study reports.

Study contacts: Erica Barnes, Executive Director, Chloe Barnes Advisory Council on Rare Disease, demo0050@umn.edu, 612-626-8932; Amanda Hemmesch, Associate Professor, St. Cloud State University, arhemmesch@stcloudstate.edu, 320-308-3215.

If you would like a paper copy of the survey to complete and mail to the study team, please reach out to Erica Barnes at the contact information included above.

By clicking on the 'Agree' button below, you indicate that you've read this information, any questions you had have been answered, and you agree to take part in this study.

o I agree to take part in this study (1)

o I do NOT want to take part in this study (2)

End of Block: Consent

Start of Block: Demographics

Q2 Thank you for agreeing to take our survey! We will ask you a series of questions about your/your family member's background, rare disease, the way you/they obtain care and information about your disease, and your/their socioemotional concerns.

We are interested in understanding your experience accessing care BEFORE the COVID-19 pandemic. There will be a question near the end of the survey to ask about your experience related to COVID-19 where you can share how things may have changed or stayed the same.

For the purpose of this survey: A caregiver is a person who (usually without being paid) provides ongoing care or assistance to another person living with a rare disease. Caregivers are often family members of the person living with a rare disease. A paid support worker is a person who is paid to provide ongoing care or assistance to another person living with a rare disease.
In this survey, we use the phrases “the person with the rare disease” or "you/your family member" to refer to the person living with a rare disease. If you are a paid support worker, please interpret this as the person you provide care for.

Q3 Is the person answering this survey:
o A person living with a rare disease (1)
o A relative/caregiver of a person with a rare disease (2)
o A parent of a child with a rare disease (3)
o A paid support worker (4)
o Other (please describe) (5) _____

Q4 Is the person answering this survey aged 18 years or older?
o Yes (1)
o No - This survey should be completed by an adult at least 18 years old (2)

Skip To: End of Survey If Is the person answering this survey aged 18 years or older? = No - This survey should be completed by an adult at least 18 years old

Q5 Is the person answering the survey:
o Female (1)
o Male (2)
o Something else (please describe) (4) _____

Q6 Has the person with the rare disease recovered from the disease or died?
o Yes - This survey should be completed by someone with current experience with rare disease (1)
o No (2)

Display This Question:
If Has the person with the rare disease recovered from the disease or died? = Yes - This survey should be completed by someone with current experience with rare disease

Q7 Thank you for your willingness to contribute to a deeper understanding of the rare disease community. While this survey is limited to individuals currently living with a rare disease, your input is valuable. If you would like to be involved in the work of the Rare Disease Advisory Council, please contact Erica Barnes at demo0050@umn.edu.

Skip To: End of Survey If Thank you for your willingness to contribute to a deeper understanding of the rare disease commun... Is Displayed

Q8 Is the person with the rare disease a child (under 18) or an adult (18 or older)?
o Child (1)
o Adult (2)

Q9 What year was the person with the rare disease born?
▼ 1930 or before (1) ... 2021 (92)

Q10 What is the race/ethnicity of the person with the rare disease? Select all that apply.
▢ American Indian or Alaska native (1)
▢ Asian (2)
▢ Black or African American (3)
▢ Hispanic or Latino/a (4)
▢ Middle Eastern (5)
▢ Mixed race (6)
▢ Native Hawaiian or Pacific Islander (7)
▢ White or Caucasian (8)
▢ Other (9) _____
▢ Rather not say (10)
▢ Don't know (12)

Q11 Is the person with the rare disease:
o Female (1)
o Male (2)
o Something else (please describe) (4) _____

Q12 What is the marital/partnership status of the person with the rare disease?
o Single (never married or partnered) (1)
o Dating/in a relationship (2)
o Married or partnered (3)
o Divorced or separated (4)
o Widowed (5)
o Don't know (6)
o Not applicable (7)

Q13 What is the highest level of education obtained by the person with the rare disease?
- o Some high school or less (1)
- o High school diploma or GED (2)
- o Some college (3)
- o Associates or technical degree (4)
- o Bachelor's degree (5)
- o Some graduate school (6)
- o Graduate degree (7)
- o Don't know (8)
- o Not applicable (9)

Q14 What was the approximate household income (USD) in 2019, before taxes, of the person with the rare disease?
- o Under $20,000 (1)
- o $20,000-39,000 (2)
- o $40,000-59,000 (3)
- o $60,000-79,000 (4)
- o $80,000-99,999 (5)
- o Over $100,000 (6)
- o Rather not say (7)
- o Don't know (8)
- o Not applicable (9)

Q15 The person with the rare disease lives in a:
- o Large city (>500,000 residents) (1)
- o Medium city (100,000-500,000 residents) (2)
- o Small town or city ((3)
- o Rural area (outside of a town or city) (4)
- o Don't know (5)

Q106 What state does the person with the rare disease live in?
▼ Alabama (1) ... Prefer not to say (52)

Skip To: Q17 If What state does the person with the rare disease live in? != Minnesota

Q16
What county does the person with the rare disease live in?

▼ Aitken (1) ... Refused (125)

Q17 Does the person with the rare disease:
(Select all that apply)
- ▢ Work full-time (1)
- ▢ Work part-time (2)
- ▢ Volunteer (3)

- ☐ Attend school/college/university (4)
- ☐ Not work because you/they are not able (5)
- ☐ Not work because you/they choose not to (6)
- ☐ Not work because you/they are unable to find job (7)
- ☐ Don't know (8)
- ☐ Not applicable (9)

End of Block: Demographics

Start of Block: RD questions

Q18 Next, we'll ask questions about your/your family member's rare disease(s).

A disorder/disease is generally considered rare if it affects fewer than 200,000 individuals in the United States. A list of rare diseases can be found here. Rare disorders are discovered and prevalence estimates change frequently, so even if your disorder does not appear in the list, you may write it in below.

Q19 How many rare diseases do you/your family member have?
- o 1 (1)
- o more than 1 (please write number here) (2) _____
- o Don't know (3)

Q20 What is the name of your/your family member's rare disease(s)?
_____

End of Block: RD questions

Start of Block: Health follow-up questions/diagnosis

Q21 Select the statement below that best describes the time course of your/your family member's rare disease(s).
- o It is stable; not expected to change very much over time (1)
- o It is progressive; expected to get more severe over time (2)
- o It is episodic; I have periods of time when I am stable or get better followed by periods when I get worse. (3)
- o It is improving; it is expected to get better over time. (4)
- o Don't know (5)

Q22 How long have you/your family member experienced symptoms of the rare disease(s)? If you have more than one rare disease, respond about the one you have had the longest.
- o 0-6 months (1)
- o 7-11 months (2)
- o 1-3 years (3)
- o 4-6 years (4)
- o 7-9 years (5)

o 10+ years (6)
o Don't know (7)

Q23 How long after first seeking medical help did it take for you/your family member to get a confirmed diagnosis? If you have more than one rare disease, respond about the one you have had the longest.
o 0-6 months (1)
o 7-11 months (2)
o 1-3 years (3)
o 4-6 years (4)
o 7-9 years (5)
o 10+ years (6)
o Still undiagnosed (7)
o Don't know (8)

Q24 Have you/your family member received a diagnosis of this condition confirmed by a doctor(s)?
o Yes, I/they have a confirmed diagnosis (1)
o No, I/they have an unconfirmed diagnosis (2)
o No, I/they do not have a diagnosis (3)
o Don’t know (4)

Q25 How many doctors have you/your family member seen to get a confirmed diagnosis? If you/your family member have not received a confirmed diagnosis, how many doctors have you seen so far in your attempt to get a diagnosis?
o 1 (1)
o 2-3 (8)
o 4-5 (2)
o 6-10 (3)
o 11-15 (4)
o More than 15 (5)
o Don't know (6)
o Not applicable (7)

Q26 How long ago was the confirmed diagnosis made?
o Less than a year ago. Please indicate how many months ago. (1) _____
o More than a year ago. Please indicate how many years ago. (2) _____
o Don't know (3)
o Not applicable (4)

Q28 Who diagnosed the person with the rare disease’s rare condition? Select all that apply.
▢ General practitioner/primary care doctor (1)
▢ Allied health professionals (e.g., optician, physical therapist, podiatrist, speech therapist, nutritionist, occupational therapist) (2)
▢ Local medical specialists in Minnesota (e.g., specialist doctors). Please describe (e.g., geneticist, neurologist, allergist) (3) _____

▢ Regional medical specialists in the Midwest (e.g., specialist doctors). Please describe (e.g., geneticist, neurologist, allergist) (4) _____
▢ National medical specialists outside the Midwest (e.g., specialist doctors). Please describe (e.g., geneticist, neurologist, allergist) (5) _____
▢ Hospital emergency department (6)
▢ Hospital outpatient services/clinics (not specialist centers in your/your family member's condition) (7)
▢ Dental services (e.g., dentist, dental hygienist) (8)
▢ Mental health service (e.g., psychiatrist, psychologist, counselor) (9)
▢ Alternative health service (e.g., acupuncturist, naturopath, homeopath, or any other alternative health service) (10)
▢ Other (please describe) (11) _____
▢ Don't know (12)
▢ Not applicable (13)

Q29 Sometimes people with rare disease(s) experience situations related to the diagnosis, management or treatment of their rare disease that have felt difficult or frustrating. These are often called "barriers". Reflecting on your own situation, please describe your MOST SIGNIFICANT barrier that you have experienced. Please be as specific as possible. (If there are none, please note "none"):

_____

Q30 Please describe your second most significant barrier:

_____

Q31 Please describe your third most significant barrier:

_____

Q32 Sometimes people with rare disease(s) experience situations related to the diagnosis, management or treatment of their rare disease that have been positive or helpful. These are often called "facilitators". Reflecting on your own situation, please describe the MOST HELPFUL support that you have experienced. Please be as specific as possible. (If there are none, please note "none"):

_____

Q33 Please describe your second most significant facilitator:

_____

Q34 Please describe your third most significant facilitator:

_____

Q35 Please rate the provider first seen at the onset of symptoms, and also the provider who made the diagnosis. If the provider seen first made the diagnosis, only answer the questions for Provider who made diagnosis.

How would you rate their...

	Provider seen first					Provider who made diagnosis				
	Poor (1)	Fair (2)	Neutral (3)	Good (4)	Excellent (5)	Don't know/Not applicable (6)	Poor (1)	Fair (2)	Neutral (3)	Good (4)	Excellent (5)	Don't know/Not applicable (6)
Knowledge of rare disease? (1)	o	o	o	o	o	o	o	o	o	o	o	o
Willingness to ask other local physicians (from Minnesota) to help make a diagnosis? (2)	o	o	o	o	o	o	o	o	o	o	o	o
Willingness to ask regional or national (in or outside the Midwest) experts to help make a diagnosis? (3)	o	o	o	o	o	o	o	o	o	o	o	o
Willingness to research different diseases to make a diagnosis? (4)	o	o	o	o	o	o	o	o	o	o	o	o
Willingness to investigate the cause of symptoms? (5)	o	o	o	o	o	o	o	o	o	o	o	o

Q36 Looking back, at the time of confirmed diagnosis, do you think you/your family member were provided enough information on your/their condition?

o Yes (1)
o No (2)
o Don't know (3)

Q37 Did you/your family member understand all of the information you were given about your/their condition(s) at the time of diagnosis?

o Yes (1)
o No. What information didn't you understand? (Please describe) (2) _____
o Don't know (3)

Q38 What other comments do you have about you/your family member being diagnosed with a rare condition? What information was helpful/unhelpful? Was there information that you wish you had received but didn't?

_____
_____
_____
_____
_____

End of Block: Health follow-up questions/diagnosis

Start of Block: Insurance

Q39 Thank you. Next are some questions about health insurance.

Q40 What kind, if any, of health insurance or health care coverage does the person with the rare disease have? Select all that apply.

- ▢ Private insurance (1)
- ▢ Healthcare marketplace insurance (ACA or Obamacare) (2)
- ▢ Dental insurance (3)
- ▢ Medicare (for US residents) (4)
- ▢ Medicare supplement (for US residents) (5)
- ▢ Medicaid (for US residents) (6)
- ▢ Medicaid waiver (7)
- ▢ Military related health care including VA health care, TriCare, and CHAMP-VA (8)
- ▢ Other (please describe) (9) _____
- ▢ The person with the rare disease does NOT have health care coverage (10)
- ▢ Don't know (11)

Q41 You/your family member paid approximately _____ (USD) out-of-pocket (of your/their own money) in the 2019 calendar year for health claims (physician visit co-pays, deductibles, co-insurance, prescriptions, TEFRA):

- o $0-$499 (1)
- o $500-$999 (2)
- o $1,000-$1,499 (3)
- o $1,500-$1,999 (4)
- o $2,000-$2,999 (5)
- o Over $3,000 (6)
- o Don't know (7)

Q42 Have you/your family member ever been denied or delayed any of the following because pre-approval from health insurance was required?

	Was able to get easily (1)	Experienced delay (2)	Experienced denial (3)	Don't know/Not applicable (4)
Diagnostic test (1)	▢	▢	▢	▢
Medication that is FDA approved or commercially available for your/your family member's condition (2)	▢	▢	▢	▢
Device or medical equipment (3)	▢	▢	▢	▢
Medical service (e.g., occupational therapy) (4)	▢	▢	▢	▢
Investigational treatment (e.g., medication that is NOT FDA approved or commercially available) (5)	▢	▢	▢	▢
Medication that is used for other reasons but not your/your family member's particular condition (e.g., off-label use or for different age groups) (6)	▢	▢	▢	▢

Q43 Has your/your family member's insurance ever denied referral to a medical or dental specialist?

o Yes (please describe) (1) _____
o No (2)
o Don't know (3)

Q44 Has your/your family member's insurer ever denied covering a medical or dental procedure or test due because there is no available treatment for the rare disease?

o Yes (please describe) (1) _____
o No (2)
o Don't know (3)

Q45 Has your/your family member's insurer ever denied you a medical or dental service/procedure due to the rare disease not having a defined "standard of care"?

o Yes (please describe) (1) _____
o No (2)
o Don't know (3)

Q46 Have you/your family member ever been denied seeing a specialist in your rare disease due to that specialist being "out of network"?

o Yes (please describe) (1) _____
o No (2)
o Don't know (3)

End of Block: Insurance

Start of Block: Knowledge and support

Q47 Thank you. Next, we have some questions about the information, support, and care that the person with the rare disease received about their condition.

Q48 Have you/your family member had sufficient information and care for the rare disease from each of the following sources?

	Sufficient information?					Sufficient care?				
	Strongly disagree (1)	Disagree (2)	Neutral (3)	Agree (4)	Strongly agree (5)	Strongly disagree (1)	Disagree (2)	Neutral (3)	Agree (4)	Strongly agree (5)
General practitioner (GP) (1)	o	o	o	o	o	o	o	o	o	o
Medical specialists (2)	o	o	o	o	o	o	o	o	o	o

Allied health professionals (e.g., physical/physio therapist, occupational therapist, speech therapist) (3) o o o o o o o o o o o o o

Dental professional (4) o o o o o o o o o o o o o

Mental health professional (e.g., social worker, psychologist, psychiatrist) (5) o o o o o o o o o o o o o

Patient organizations (e.g., charities) (6) o o o o o o o o o o o o o

Q49 Do you think you now have sufficient knowledge about your/your family member's rare condition(s)? Rate your knowledge from 1 (no knowledge) to 10 (complete knowledge):

o 1 (9)
o 2 (10)
o 3 (11)
o 4 (12)
o 5 (13)
o 6 (14)
o 7 (15)
o 8 (16)
o 9 (17)
o 10 (18)

Skip To: Q51 If Do you think you now have sufficient knowledge about your/your family member's rare condition(s)?... = 10

Q50 If you rated your knowledge as less than 10, specifically, what information do you feel you are missing?

_____

Q51 Do you agree or disagree that you/your family member received sufficient support in the following areas at the time of diagnosis?

	Strongly disagree (1)	Disagree (2)	Neutral (neither agree nor disagree) (3)	Agree (4)	Strongly agree (5)	Don't know (6)	Not applicable (7)
Medical (for example, doctors, nurses) (1)	o	o	o	o	o	o	o
Dental (2)	o	o	o	o	o	o	o
Social (for example, family, friends, church members) (3)	o	o	o	o	o	o	o
Financial (for example, insurance coverage) (4)	o	o	o	o	o	o	o

Psychological (for example, mental health care, counselors) (5) o o o o o o o o o o
Other (please specify) (6) o o o o o o o o

End of Block: Knowledge and support

Start of Block: Specialist centers/care

Q52 A 'specialist center' is a center or clinic that is able to provide expert advice on diagnosis, assessment and treatment of a particular condition or group of conditions. The center may be made up of a team of different specialists, sometimes also including scientists and researchers.

Specialist centers can support patients across your State and/or the United States, not just in their local area.

Q53 Do you know if there is a specialist center for your/your family member's rare disease(s)?
o Yes. If you know the name of the specialist center, and the city, please specify it here: (1) _____
o No (2)
o Don't know (3)

Q54 Do you/your family member access a specialist center for your/their rare disease(s)?
o Yes (1)
o No (2)
o Don't know (3)

Skip To: Q56 If Do you/your family member access a specialist center for your/their rare disease(s)? = Yes

Q55 If you/your family member does not access a specialist center, why not?
o There is not one (1)
o My insurance does not cover it (2)
o I live too far away (3)
o They are not accepting new patients (4)
o It is too long of a wait list to get in (5)
o Other. Please describe: (6) _____
o Don't know (7)

Q56 How many different medical specialists (i.e., specialist doctors) do you/your family member see for your/their rare disease(s)?
o 1-2 (1)
o 3-4 (2)
o 5-6 (3)
o More than 6 (4)
o Don't know (5)

o Not applicable (6)

Q57 What is the furthest distance traveled to access care for the rare disease(s)?
o Less than 10 miles (1)
o 10-29 miles (2)
o 30-59 miles (3)
o 60 or more miles (4)
o Internationally (5)
o Don't know (6)
o Not applicable (7)

Q58 Telehealth means using electronic information and telecommunications technologies like videoconferencing to provide long-distance health care.

Do you use telehealth services to see medical specialists?
o Yes (1)
o No (2)
o Don’t know (3)
o Not applicable/I do not have access to the technology required for telehealth (4)

Q59 Would you be interested in telehealth/ehealth?
o Yes (1)
o No (2)
o Don’t know (3)
o Not applicable/I do not have access to the technology required for telehealth (4)

Q60 Care coordination refers to health care that is provided in a planned way that meets the needs and preferences of the patient. When care is coordinated well, the patient and his or her doctors, nurses, other health care providers, family, and other caregivers all know who is responsible for different parts of the patient’s care, and they communicate with each other so that everyone has the information they need.

A care coordinator is a trained professional who is responsible for coordinating health care services, for example between specialist clinics, hospital staff, GPs, allied health and non-government organizations.

Do you/your family member have a designated care coordinator?
o Yes (1)
o No (2)
o Don't know (3)

Skip To: Q63 If Care coordination refers to health care that is provided in a planned way that meets the needs an... != Yes

Q61 If you have used a care coordinator, rate on a scale from 1 to 10 how helpful it was, with 1 being not at all helpful to 10 being extremely helpful.

o 1 (4)
o 2 (5)
o 3 (6)
o 4 (7)
o 5 (8)
o 6 (9)
o 7 (10)
o 8 (11)
o 9 (12)
o 10 (13)

Q62 If you have used a care coordinator, what worked well and what did not?
_____

Skip To: Q64 If Condition: If you have used care coord... Is Displayed. Skip To: During the past 12 months, did you/yo....

Q63 If you have not used a care coordinator, why not?
_____

Q64 During the past 12 months, did you/your family member visit the following providers for their rare disease(s) and were you/they satisfied with their care?

	Have you seen this type of provider?	If yes, how satisfied were you with their care?				
	Yes (1)	No (2)	Very dissatisfied (1)	Dissatisfied (2)	Neutral (3)	Satisfied (4)	Very satisfied (5)	Don't know/Not applicable (6)
General practitioner (GP) (1)	o	o	o	o	o	o	o	o
Allied health (e.g., physical/physio therapist, occupational therapist, speech therapist) (2)	o	o	o	o	o	o	o	o
Mental health professional (e.g., social worker, psychologist, psychiatrist) (3)	o	o	o	o	o	o	o	o
Dentist (4)	o	o	o	o	o	o	o	o
Specialist doctor (e.g., geneticist, neurologist, allergist) (5)	o	o	o	o	o	o	o	o
Other (please describe) (6)	o	o	o	o	o	o	o	o

Q65 Please describe you/your family member’s experiences and satisfaction with healthcare providers you/your family member visited during the past 12 months.
_____
_____
_____
_____

End of Block: Specialist centers/care

Start of Block: Pediatrics

Q66 Pediatric health service is defined as the specialty of medical science concerned with the physical, mental, and social health of children from birth to young adulthood. Pediatric care encompasses a broad spectrum of health services ranging from preventive health care to the diagnosis and treatment of acute and chronic diseases.

Were you/your family member ever cared for by pediatric health services for your rare disease(s)?
- o Yes (1)
- o No (2)
- o Don't know (3)

Skip To: End of Block If Pediatric health service is defined as the specialty of medical science concerned with the physic... != Yes

Q67 Overall, how satisfied or dissatisfied are you with the care you/your family member receive(d) from pediatric health services?
- o Very dissatisfied (1)
- o Dissatisfied (2)
- o Neither satisfied nor dissatisfied (3)
- o Satisfied (4)
- o Very satisfied (5)
- o Don't know (6)

Q68 Did you/your family member transition from pediatric to adult health services?
- o Yes (1)
- o No (2)
- o Don't know (3)

Skip To: Q73 If Did you/your family member transition from pediatric to adult health services? != Yes

Q69 What was the time period between your/your family member's last visit to pediatric health services and the first visit to adult health services. Please enter the value in weeks (e.g., if one year, write 52 weeks). _____

Q70 Was this time acceptable or unacceptable to you/your family member?
- o Acceptable (1)
- o Unacceptable (2)
- o Don't know (3)

Q71 Did you/your family member experience any problems in the transition from pediatric to adult services?

o Yes. Please describe what the problems were related to: (1) _____

o No (2)

o Don't know (3)

Q72 How would you compare the knowledge of your/your family member's pediatric provider to your/your family member's adult provider?

o They have similar expertise regarding the rare disease (1)

o The pediatric provider has more expertise regarding the rare disease than the adult provider (2)

o The adult provider has more expertise regarding the rare disease than the pediatric provider (3)

o Don't know (4)

Q73 What other comments do you have about your/your family member's experience using pediatric health services, including the transition from pediatric to adult care?

_____

End of Block: Pediatrics

Start of Block: Relocation

Q74 Thank you. Next, we have some questions about challenges you/your family member may have had getting care or treatment for the rare disease(s).

Q75 Did any of the following limit you/your family member's ability to get medical or dental care or treatment for your/their rare disease?

	Never (1)	Sometimes (2)	About half the time (3)	Most of the time (4)	Almost always (5)	Don't know (6)
Finances (for example, travel costs, cost of treatment/insurance) (1)	o	o	o	o	o	o
Travel distance (2)	o	o	o	o	o	o
Difficulty getting time off of work (3)	o	o	o	o	o	o
Lack of childcare (4)	o	o	o	o	o	o
Lack of or delay in referrals (5)	o	o	o	o	o	o
Not covered by insurance (6)	o	o	o	o	o	o
Other (please describe) (7)	o	o	o	o	o	o

Q76 Have you/your family member needed to relocate in order to access treatment or clinical trials for their rare disease long-term?

	No, we have not relocated (1)	Temporarily (6 months or less) (2)	7 months to 2 years (3)	2 years or longer (4)	Permanently (5)	Don't know (6)
In-state (1)	o	o	o	o	o	o
Out of state (2)	o	o	o	o	o	o

Skip To: End of Block If Have you/your family member needed to relocate in order to access treatment or clinical trials fo... [No, we have not relocated] (Count) = 2

Q77 If you/your family member relocated because of the rare disease, please describe why:
_____

End of Block: Relocation

Start of Block: Patient Satisfaction Questionnaire Short Form

Q78 Think about your overall experience with healthcare for your/your family member's rare disease(s) when responding to the following statements. If you are a caregiver, please answer these questions in terms of how the person with the rare disease is feeling. If the person with a rare disease is under age 10, please answer based on your experience with them as a caregiver.

	Strongly disagree (1)	Disagree (2)	Neither agree nor disagree (3)	Agree (4)	Strongly agree (5)	Don't know (6)	Not applicable (7)
Healthcare providers are good about explaining the reason for medical tests (1)	o	o	o	o	o	o	o
I think my healthcare providers' office has everything needed to provide complete medical care (2)	o	o	o	o	o	o	o
The medical care I have been receiving is just about perfect (3)	o	o	o	o	o	o	o
Sometimes healthcare providers make me wonder if their diagnosis is correct (4)	o	o	o	o	o	o	o
I feel confident that I can get the medical care I need without being set back financially (5)	o	o	o	o	o	o	o
When I go for medical care, they are careful to check everything when treating and examining me (6)	o	o	o	o	o	o	o
I have to pay for more of my medical care than I can afford (7)	o	o	o	o	o	o	o
I have easy access to the medical specialists I need (8)	o	o	o	o	o	o	o
Where I get medical care, I have to wait too long for emergency treatment (9)	o	o	o	o	o	o	o

Healthcare providers act too businesslike and impersonal toward me (10) o o o o o o o

My healthcare providers treat me in a very friendly and courteous manner (11) o o o o o o o

Those who provide me medical care sometimes hurry too much when they treat me (12) o o o o o o o

Healthcare providers sometimes ignore what I tell them (13) o o o o o o

I have some doubts about the ability of the healthcare providers who treat me (14) o o o o o o o

Healthcare providers usually spend plenty of time with me (15) o o o o o o

I find it hard to get an appointment for medical care right away (16) o o o o o o

I am dissatisfied with some things about the medical care I receive (17) o o o o o o

I am able to get medical care whenever I need it (18) o o o o o o

End of Block: Patient Satisfaction Questionnaire Short Form

Start of Block: PROMIS 29 profile - CHILDREN

Q79 Next, we will ask about your child's overall health and well-being.

Q80	Excellent (1)	Very good (2)	Good (3)	Fair (4)	Poor (5)
In general, would you say your child's health is... (1)	o	o	o	o	o

Q81 In the past 7 days...

	Never (1)	Almost never (2)	Sometimes (3)	Often (4)	Almost always (5)	Don't know (6)	Not applicable (7)
My child could do sports and exercise that other kids his/her age could do (1)	o	o	o	o	o	o	o
My child could get up from the floor (2)	o	o	o	o	o	o	o
My child could walk up stairs without holding onto anything (3)	o	o	o	o	o	o	o
My child has been physically able to do the activities he/she enjoys most (4)	o	o	o	o	o	o	o

Q82 In the past 7 days...

	Never (1)	Almost never (2)	Sometimes (3)	Often (4)	Almost always (5)	Don't know (6)	Not applicable (7)
My child felt like something awful might happen (1)	o	o	o	o	o	o	o

My child felt nervous (2) o o o o o o o

My child felt worried (3) o o o o o o o

My child worried when he/she was at home (4) o o o o o o o o

Q83 In the past 7 days...

	Never (1)	Almost never (2)	Sometimes (3)	Often (4)	Almost always (5)	Don't know (6)	Not applicable (7)
My child felt everything in his/her life went wrong (1)	o	o	o	o	o	o	o
My child felt lonely (2)	o	o	o	o	o	o	o
My child felt sad (3)	o	o	o	o	o	o	o
It was hard for my child to have fun (4)	o	o	o	o	o	o	o

Q84 In the past 7 days...

	Never (1)	Almost never (2)	Sometimes (3)	Often (4)	Almost always (5)	Don't know (6)	Not applicable (7)
Being tired made it hard for my child to keep up with schoolwork (1)	o	o	o	o	o	o	o
My child got tired easily (2)	o	o	o	o	o	o	o
My child was too tired to do sports or exercise (3)	o	o	o	o	o	o	o
My child was too tired to enjoy the things he/she likes to do (4)	o	o	o	o	o	o	o

Q85 In the past 7 days...

	Never (1)	Almost never (2)	Sometimes (3)	Often (4)	Almost always (5)	Don't know (6)	Not applicable (7)
My child felt accepted by other kids his/her age (1)	o	o	o	o	o	o	o
My child was able to count on his/her friends (2)	o	o	o	o	o	o	o
My child and his/her friends helped each other out (3)	o	o	o	o	o	o	o
Other kids wanted to be my child's friend (4)	o	o	o	o	o	o	o

Q86 In the past 7 days...

	Never (1)	Almost never (2)	Sometimes (3)	Often (4)	Almost always (5)	Don't know (6)	Not applicable (7)

My child had trouble sleeping when he/she had pain (1) o o o o o o o o o o

It was hard for my child to pay attention when he/she had pain (2) o o o o o o o o o

It was hard for my child to run when he/she had pain (3) o o o o o o o o o

It was hard for my child to walk one block when he/she had pain (4) o o o o o o o o o

Q87 In the past 7 days...

	1 No pain (1)	2 (2)	3 (3)	4 (4)	5 (5)	6 (6)	7 (7)	8 (8)	9 (9)	10 Worst pain you can think of (10)	Don't know (11)
How bad was your child's pain on average? (1)	o	o	o	o	o	o	o	o	o	o	o

End of Block: PROMIS 29 profile - CHILDREN

Start of Block: PROMIS 29 profile ADULTS

Q88 Next, we will ask about your/your family member’s overall health and well-being. If you are a caregiver, please answer these questions in terms of how the person with the rare disease is feeling.

Q89	Excellent (1)	Very good (2)	Good (3)	Fair (4)	Poor (5)
In general, would you say your health is... (1)	o	o	o	o	o

Q90	Without any difficulty (1)	With a little difficulty (2)	With some difficulty (3)	With much difficulty (4)	Unable to do (5)	Don't know (6)
Are you able to do chores such as vacuuming or yard work? (1)	o	o	o	o	o	o
Are you able to go up and down stairs at a normal pace? (2)	o	o	o	o	o	o
Are you able to go for a walk of at least 15 minutes? (3)	o	o	o	o	o	o
Are you able to run errands and shop? (4)	o	o	o	o	o	o

Q91 In the past 7 days...

	Never (1)	Rarely (2)	Sometimes (3)	Often (4)	Always (5)	Don't know (6)
I felt fearful (1)	o	o	o	o	o	o
I found it hard to focus on anything other than my anxiety (2)	o	o	o	o	o	o

My worries overwhelmed me (3)	o	o	o	o	o	o
I felt uneasy (4)	o	o	o	o	o	o

Q92 In the past 7 days...

	Never (1)	Rarely (2)	Sometimes (3)	Often (4)	Always (5)	Don't know (6)
I felt worthless (1)	o	o	o	o	o	o
I felt helpless (2)	o	o	o	o	o	o
I felt depressed (3)	o	o	o	o	o	o
I felt hopeless (4)	o	o	o	o	o	o

Q93 In the past 7 days...

	Not at all (1)	A little bit (2)	Somewhat (3)	Quite a bit (4)	Very much (5)	Don't know (6)
I feel fatigued (1)	o	o	o	o	o	o
I have trouble starting things because I am tired (2)	o	o	o	o	o	o
How run-down did you feel on average? (3)	o	o	o	o	o	o
How fatigued were you on average? (4)	o	o	o	o	o	o

Q94 In the past 7 days...

	Very poor (1)	Poor (2)	Fair (3)	Good (4)	Very good (5)	Don't know (6)
My sleep quality was (1)	o	o	o	o	o	o

Q95 In the past 7 days...

	Not at all (1)	A little bit (2)	Somewhat (3)	Quite a bit (4)	Very much (5)	Don't know (6)
My sleep was refreshing (1)	o	o	o	o	o	o
I had a problem with sleep (2)	o	o	o	o	o	o
I had difficulty falling asleep (3)	o	o	o	o	o	o

Q96	Never (1)	Rarely (2)	Sometimes (3)	Often (4)	Always (5)	Don't know (6)
I have trouble doing all of my regular leisure activities with others (1)	o	o	o	o	o	o
I have trouble doing all of the family activities that I want to do (2)	o	o	o	o	o	o
I have trouble doing all of my usual work (include work at home) (3)	o	o	o	o	o	o
I have trouble doing all of the activities with friends that I want to do (4)	o	o	o	o	o	o

Q97 In the past 7 days...	Not at all (1)	A little bit (2)	Somewhat (3)	Quite a bit (4)	Very much (5)	Don't know (6)
How much did pain interfere with your day to day activities? (1)	o	o	o	o	o	o
How much did pain interfere with work around the home? (2)	o	o	o	o	o	o
How much did pain interfere with your ability to participate in social activities? (3)	o	o	o	o	o	o
How much did pain interfere with your household chores? (5)	o	o	o	o	o	o

Q98 In the past 7 days...	1 No pain (1)	2 (2)	3 (3)	4 (4)	5 (5)	6 (6)	7 (7)	8 (8)	9 (9)	10 Worst pain imaginable (10)	Don't know (11)
How would you rate your pain on average? (1)	o	o	o	o	o	o	o	o	o	o	o

End of Block: PROMIS 29 profile

Start of Block: Anticipated stigma scale

Q99 Rate the likelihood that you/your family member will encounter the experiences below in the future because of your rare disease(s).

If you are a caregiver, please answer these questions in terms of how the person with the rare disease is feeling. If the person with a rare disease is under age 10, please answer based on your experience with them as a caregiver.

	Very unlikely (1)	Unlikely (2)	Neither unlikely nor likely (3)	Likely (4)	Very likely (5)	Don't know (6)
A healthcare worker will blame you for not getting better (9)	o	o	o	o	o	o
A healthcare worker will be frustrated with you (10)	o	o	o	o	o	o

A healthcare worker will give you poor care (11) o o o o o o o

A healthcare worker will think that you are a bad patient (12) o o o o o o

End of Block: Anticipated stigma scale

Start of Block: Stigma scale

Q100 Indicate how much you agree or disagree with each of the statements about the way you have felt about your rare disease(s) lately...

If you are a caregiver, please answer these questions in terms of how the person with the rare disease is feeling. If the person with a rare disease is under age 10, please answer based on your experience with them as a caregiver.

	Never (1)	Rarely (2)	Sometimes (3)	Often (4)	Always (5)	Don't know (6)
Because of my rare disorder(s), some people seemed uncomfortable with me. (1)	o	o	o	o	o	o
Because of my rare disorder(s), some people avoided me. (2)	o	o	o	o	o	o
Because of my rare disorder(s), I felt left out of things. (3)	o	o	o	o	o	o
Because of my rare disorder(s), people were unkind to me. (4)	o	o	o	o	o	o
Because of my rare disorder(s), people avoided looking at me. (5)	o	o	o	o	o	o
I felt embarrassed about my rare disorder(s). (6)	o	o	o	o	o	o
I felt embarrassed because of my physical limitations due to my rare disorder(s). (7)	o	o	o	o	o	o
Some people acted as though it was my fault I have my rare disorder(s). (8)	o	o	o	o	o	o

End of Block: Stigma scale

Start of Block: Qualitative

Q101 Thank you! There are only a couple of questions left.

Is there anything that you would like to add about your/your family member’s experience with rare disease that will help the researchers better understand the challenges experienced by patients/families? If so, please share it here:

_____

_____

_____

_____
_____

Q102 Has the COVID-19 pandemic changed your ability to access care and services?
o Yes (1)
o No (2)
o Don't know (3)

Skip To: Q104 If Has the COVID-19 pandemic changed your ability to access care and services? != Yes

Q103 If so, how has COVID-19 changed how you access care and services?
_____
_____
_____
_____
_____

Q104 If there were ONE SINGLE THING that could be provided by the healthcare system to improve your/your families experience with this rare disease, what would it be?
_____
_____
_____
_____
_____

Q105 This is the last question of the survey. Is there anything you would like to add?
_____
_____
_____
_____
_____

End of Block: Qualitative

End of Survey

Appendix 2: Rare Diseases Represented in the Sample

Rare disease name	Count
1q duplications	1
22q13.3 deletion syndrome	1
46,XY,der(7)t(7;9)(q34;p13.1)	1
47 XXX syndrome	1
4q21.1-q21.22	1
ACTG2-related disorders	1
ACTH deficiency	1
Acute intermittent porphyria	1
Acute interstitial pneumonia	2
Acute posterior multifocal placoid pigment epithel	1
Acute respiratory distress syndrome	1
ADCY5-related dyskinesia	1
Addison's disease	14
Adiposis dolorosa	1
Adrenomyeloneuropathy	1
Adult polyglucosan body disease	1
Alagille syndrome	3
Alpha-1 antitrypsin deficiency	1
Alpha-thalassemia x-linked intellectual disability	1
Amelogenesis imperfecta hypomaturation type	1
Amyotrophic lateral sclerosis	4
Angelman syndrome	2
Aniridia	1
Antiphospholipid syndrome	8

Antisynthetase syndrome	1
Aortic valve stenosis	1
Aphasia	1
Aplasia cutis congenita	1
Aplastic anemia	1
Apraxia	3
Arachnoiditis	1
Argininosuccinic aciduria	1
Atypical hemolytic uremic syndrome	3
Autoimmune autonomic ganglionopathy	1
Autoimmune encephalitis	2
Autoimmune hemolytic anemia	2
Autoimmune hepatitis	4
Autoimmune polyglandular syndrome type 2	1
Axenfeld-Rieger syndrome	1
Babesiosis	1
Bardet-Biedl syndrome	1
Becker muscular dystrophy	1
Beckwith-Wiedemann syndrome	1
Behçet disease	9
Beta-Propeller Protein-Associated Neurodegeneratio	1
Biliary atresia	1
Bohring-Opitz syndrome	1
Brain-lung-thyroid syndrome	1
Branchiootorenal syndrome	1

Bronchopulmonary dysplasia	1
Brown-Sequard syndrome	2
Cat eye syndrome	1
Cat scratch disease	1
Catecholaminergic polymorphic ventricular tachycar	1
Cauda equina syndrome	4
Caudal regression sequence	1
Cerebral palsy spastic quadriplegic	1
Cervical dystonia	3
Charcot-Marie-Tooth disease	2
Choanal atresia	1
Chromosome 10 trisomy	1
Chromosome 11q deletion	1
Chromosome 13q duplication	1
Chromosome 17q duplication	1
Chromosome 19p deletion	1
Chromosome 1p deletion	8
Chromosome 20 deletions	1
Chromosome 22q duplication	1
Chronic inflammatory demyelinating polyneuropathy	1
Chronic lymphocytic inflammation with pontine peri	1
Chronic myeloproliferative disorders	1
Chronic recurrent multifocal osteomyelitis	14
Cleidocranial dysplasia	1
Coccidioidomycosis	1

Cogan's syndrome	1
Cold urticaria	2
Common variable immunodeficiency	4
Complex regional pain syndrome	7
Cone-rod dystrophy	4
Congenital adrenal hyperplasia	2
Congenital hydrocephalus	6
Congenital hypothyroidism	1
Congenital laryngeal palsy	1
Congenital myasthenic syndromes	1
Congenital rubella	1
Conotruncal heart malformations	1
Conversion disorder	1
Cornelia de Lange syndrome	9
Corpus callosum agenesis	1
cortical dysplasia	1
Cramp-fasciculation syndrome	1
Craniopharyngioma	1
Cri du chat syndrome	1
Cutis marmorata telangiectatica congenita	1
Cystic fibrosis	11
Cytomegalic inclusion disease	1
Dandy-Walker complex	3
Dentinogenesis imperfecta	1
Dermatitis herpetiformis	1

Dermatomyositis	1
Developmental and Eplileptic Encephalopathy Type 3	1
Developmental delay with variable intellectual imp	1
Diffuse idiopathic pulmonary neuroendocrine cell h	1
Disseminated superficial actinic porokeratosis	1
Dopa-responsive dystonia	1
Dravet syndrome	1
Duane syndrome	1
Duchenne muscular dystrophy	5
DYRK1A-Related Intellectual Disability Syndrome	2
Dystonia	1
DYT-TOR1A	1
Ectodermal dysplasia	16
Ehlers-Danlos Syndrome	45
Ehrlichiosis	1
Empty sella syndrome	1
Eosinophilic gastroenteritis	7
EPDC5-Related Epilepsy	1
Epilepsy juvenile absence	1
Epithelioid sarcoma	1
Erythromelalgia	2
Essential thrombocythemia	1
Factor XI deficiency	1
Familial adenomatous polyposis	5
familial cavernous malformation	1

Familial dysautonomia	1
Familial hypocalciuric hypercalcemia	1
Familial Mediterranean fever	2
Familiar or sporadic hemiplegic migraine	2
Fanconi anemia	2
Florid cystic endosalpingiosis of the uterus	1
Focal cortical dysplasia	1
Fowler's syndrome	1
Fragile X syndrome	1
Friedreich ataxia	7
Frontotemporal dementia	2
Gastroparesis	8
GATAD2B-associated neurodevelopmental disorder	1
Ghose Sachdev Kumar syndrome	1
Gitelman syndrome	1
Glycogen storage disease	19
Gonadal dysgenesis mixed	1
Gordon syndrome	1
Gould Syndrome	1
Gray platelet syndrome	1
Growth hormone deficiency	4
Hantavirus pulmonary syndrome	1
Hemihypertrophy	1
Hemophilia	2
Hereditary amyloidosis	1

Hereditary hemorrhagic telangiectasia	1
Hereditary neuropathy with liability to pressure	1
Hereditary pancreatitis	1
Hereditary sensory and autonomic neuropathy	3
Hereditary Spastic Parapalegia SPG4	3
Herpes zoster oticus	1
Hirschsprung disease	1
HIVEP2-related intellectual disability	1
Holoprosencephaly	5
Homocystinuria	1
hydrocephalus	1
Hyperacusis	20
Hypersensitivity pneumonitis	1
Hypocomplementemic urticarial vasculitis	1
Hypophosphatasia	23
Hypophosphatemic rickets	1
Hypoplastic left heart syndrome	1
Ichthyosis	1
Idiopathic achalasia	1
Idiopathic alveolar hypoventilation syndrome	1
Idiopathic hypersomnia	146
Idiopathic inflammatory myopathy	5
Idiopathic intracranial hypertension	4
Idiopathic pulmonary fibrosis	12
Idiopathic subglottic tracheal stenosis	1

Idiopathic thrombocytopenic purpura	1
IgG4-related disease	1
Immune thrombocytopenia	1
Imperforate anus	1
Inclusion body myopathy	11
Inclusion body myositis	1
Inflammatory myofibroblastic tumor	1
Intellectual disability-severe speech delay	1
Joubert syndrome	1
Juvenile-onset small-fiber polyneuropathy	1
Kabuki syndrome	6
Kartagener syndrome	1
KAT6A syndrome	1
Kawasaki Disease	2
KBG Syndrome	1
Kleine Levin syndrome	1
Klippel Feil syndrome	17
Known recurrent chromosome deletion	1
L1 syndrome	1
Lambert Eaton myasthenic syndrome	1
Landau-Kleffner syndrome	1
Larsen syndrome	1
Leber congenital amaurosis	5
Left ventricular noncompaction	1
Legg-Calve-Perthes disease	1

Leiomyosarcoma	12
Lennox-Gastaut syndrome	2
Leukodystrophy	1
Lichen sclerosus	3
Li-Fraumeni syndrome	1
Limbic encephalitis	1
Limited cutaneous systemic sclerosis	1
Loeys-Dietz syndrome	1
Loin pain hematuria syndrome	1
Lowe oculocerebrorenal syndrome	4
Lymphatic malformations	4
malan overgrowth syndrome	4
Mantle cell lymphoma	1
Maple syrup urine disease	5
Marfanoid hypermobility syndrome	1
Marshall-Smith syndrome	1
Mast cell activation syndrome	12
Median arcuate ligament syndrome	3
Medullary sponge kidney	1
Megacystis microcolon intestinal hypoperistalsis	1
Meier-Gorlin syndrome	1
Mental retardation, autosomal dominant 56	1
Mental retardation, X-linked 99	1
Metachromatic leukodystrophy	2
Microcephaly	2

Misophonia	4
Mitochondrial genetic disorders	5
Mitochondrial myopathy	2
Mixed connective tissue disease	6
Mollaret meningitis	26
Monoclonal gammopathy of undetermined significance	1
Monomelic amyotrophy	1
Mowat-Wilson syndrome	1
Moyamoya disease	1
Mucolipidosis type 4	1
Mucopolysaccharidosis	5
Multiple endocrine neoplasia type 1	4
Multiple system atrophy	5
Muscular dystrophy	3
Myasthenia gravis	6
Mycobacterium Avium Complex infections	1
Myhre syndrome	5
Narcolepsy	56
Neonatal stroke	1
Neuroendocrine tumor	1
Neuromyelitis optica spectrum disorder	1
Neuronal ceroid lipofuscinosis 3	1
Neutral lipid storage disease with myopathy	1
non-radiographic axial spondyloarthritis	1
Noonan syndrome	1

Ocular albinism	1
Oculomotor apraxia Cogan type	1
Optic nerve hypoplasia, familial bilateral	1
Orofaciodigital syndrome 6	1
Osteogenesis imperfecta	8
Ovarian remnant syndrome	1
Pantothenate kinase-associated neurodegeneration	1
Paraneoplastic syndrome	1
Paroxysmal kinesigenic choreoathetosis	1
Pars planitis	1
Partial atrioventricular canal	1
Periventricular heterotopia	1
Periventricular leukomalacia	1
Permanent neonatal diabetes mellitus	1
Pfeiffer syndrome	1
Phenylketonuria	1
Pierpont syndrome	1
Pierre Robin sequence	1
Pitt-Hopkins syndrome	9
Polycythemia vera	1
Polymyalgia rheumatica	1
Porencephaly	1
Potocki-Lupski syndrome	1
Prader-Willi syndrome	1
Precocious puberty	1

Primary biliary cholangitis	36
Primary ciliary dyskinesia	1
Primary orthostatic hypotension	5
Primary sclerosing cholangitis	1
Prolactinoma	1
Propionic acidemia	2
Pseudocholinesterase deficiency	1
Pseudohypoparathyroidism	1
Psoriatic juvenile idiopathic arthritis	1
Pulmonary arterial hypertension	2
Pulmonary artery sarcoma	1
Pyoderma gangrenosum	1
Pyruvate dehydrogenase complex deficiency	1
Recurrent respiratory papillomatosis	11
Relapsing polychondritis	2
Renal agenesis	1
Renal glycosuria	1
Retinitis pigmentosa	2
Rett syndrome	11
Reynolds syndrome	1
Ring chromosome 20	1
Robinow syndrome	3
SAPHO syndrome	1
Schwannomatosis	1
SCN2A related disorders	1

Sensory ataxic neuropathy	1
Short bowel syndrome	1
Soft tissue sarcoma	1
Spasmodic dysphonia	5
Spastic ataxia Charlevoix-Saguenay type	3
Spastic diplegia cerebral palsy	1
Spastic paraparesis	2
Specific antibody deficiency	3
Spina bifida	6
Spinal muscular atrophy	8
Spinocerebellar ataxia	178
Spontaneous coronary artery dissection	1
Sprengel deformity	2
Stevens-Johnson syndrome	1
Stickler syndrome	15
Stiff person syndrome	3
Stiff skin syndrome	1
Sudden sensorineural hearing loss	1
Syringomyelia	2
Systemic capillary leak syndrome	1
Systemic scleroderma	3
T cell immunodeficiency primary	1
T cell lymphoma	1
Takenouchi-Kosaki syndrome	1
TANGO2-Related Metabolic Encephalopathy and Arrhyt	1

Tarlov cysts	3
Tethered cord syndrome	3
Thyroid cancer, medullary	1
Tracheobronchomalacia	2
Transient global amnesia	11
Treacher Collins syndrome	1
Trigeminal neuralgia	4
Ullrich congenital muscular dystrophy	1
Unbalanced translocation chromosome 3p and 6p. col	1
Undifferentiated pleomorphic sarcoma	1
Van der Woude syndrome	1
Visual snow syndrome	1
WAGR syndrome	8
Waldenstrom macroglobulinemia	3
Warfarin syndrome	1
White Sutton syndrome	2
Wieacker syndrome	1
Wiedemann-Steiner syndrome	7
Wolf-Hirschhorn syndrome	7
X-linked adrenoleukodystrophy	2

*Some participants reported multiple rare diseases.