Chloe Barnes Advisory Council on Rare Diseases

Report to the Minnesota Legislature

January 1, 2022

Chloe Barnes Advisory Council on Rare Diseases

Report to the Minnesota Legislature for Calendar Year 2021

As required by Minnesota Statute 137.68

Submitted by:

University of Minnesota Board of Regents

Prepared by:

Staff in the Medical School at the University of Minnesota

Report Preparation Costs:

Per the requirements set forth in Minnesota Statue 3.197, the cost to prepare this report was $300.00

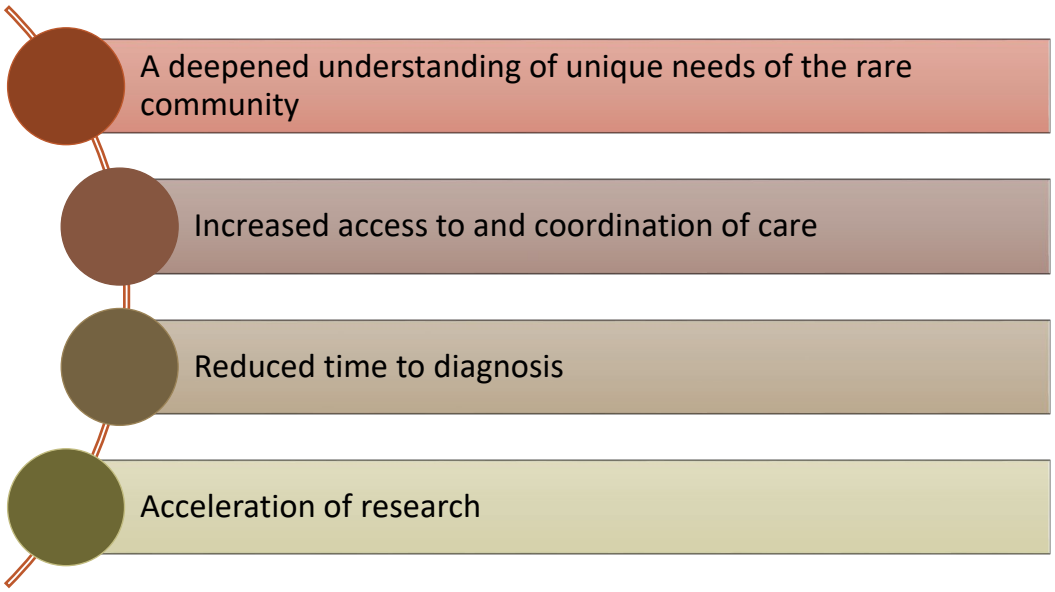
Background

In 2019, the Minnesota State Legislature passed a bill for the creation of the Chloe Barnes Advisory Council on Rare Diseases with oversight provided by University of Minnesota Board of Regents. The Council is housed in the University of Minnesota Medical School and chaired by Jakub Tolar, MD, PhD, Dean of the Medical School and Vice President for Clinical Affairs.

Council members represent the following institutions: ALS Association, Be the Match Foundation, CentraCare, Children's Minnesota Hospital, Gillette Children's Specialty Healthcare, Hennepin County Medical Center, Mayo Clinic, M Health-Fairview, Minnesota Department of Health, Minnesota State Legislature (House and Senate), National Foundation for Ectodermal Dysplasia, Sickle Cell Foundation of Minnesota, and the University of Minnesota.

The Orphan Drug Act in 1983 defined a rare disease as "a disease or condition that affects less than 200,000 people in the United States." According to the National Institutes of Health (NIH), there are approximately 7,000 rare diseases affecting between 25 and 30 million Americans. This equates to 1 in 10 Americans living with a rare disease, which equals between 25-30 million or roughly 8-10% of the population. While each individual rare disease community may be small, collectively, they represent a significant portion of the population.[1] Their etiologies are diverse, but the barriers they face when seeking care are common. Abbey Meyers, founder of the National Organization for Rare Disorders (NORD) said, "Families affected by rare diseases represent a medically disenfranchised population that falls through the cracks of every healthcare system in the world."

The Chloe Barnes Advisory Council on Rare Diseases (the Council) is a cross-sector, multi-institutional collaborative endeavor that seeks to address the gaps in care present in this patient population. Its ***vision*** is a Minnesota where every citizen living with a rare disease has access to a timely diagnosis, expert/coordinated care, as well as individualized treatment, management, and support throughout the lifespan. Its ***mission*** is to provide advice on research, diagnosis, treatment, and education related to rare diseases.[2] The Council has executed on its strategic plan, articulated under the following pillars:



2021 Year in Review

The Council executes its mission through workgroups organized around four pillars (see diagram on previous page). In addition to ensuring that the Council addresses the duties enumerated in the enacting legislation, work group leads meet regularly to identify areas of cross collaboration and report these activities back to the full Council at quarterly meetings. An executive subcommittee as well as operating procedures guide governance practices. Workgroup highlights are:

Barriers to Care	Cost	Coordination of Care	Acceleration of Research
• Completion of nationwide patient survey, largest to date in the USA • Completion of statewide clinician survey • Completion of various educational materials related to the surveys directed at medical commuity, patient community, and policy makers • Mentorship of PhD candidate Samad Qureshi's dissertation project focused on applying bioinformatics to improve the identification of rare disease diagnosis	• Collaboration with barriers workgroup for patient and provider survey content and identification of top themes • Mentorship of PhD candidate and Masters student at the University of Minnesota School of Public Health to produce a cost impact study in the state of Minnesota • Engagement with a payor focused on adressing process barriers such as delayed diagnosis and poor care coordination for rare disease patients with the goal of understanding and potentially extending identified best practices	• Collaboration with barriers workgroup for patient and provider survey content and identification of top themes • Development of registry of clinicians able to support transition of care for rare patients across multiple healthcare institutions • Collaboration with MDH and Gillette Childrens to address transition of care and host a roundtable in 2022	• Collaboration with barriers workgroup for patient and provider survey content and identification of top themes • Collaboration with the Clinical and Translational Science Inistitute at the University of Minnesota on a data capture project that would allow researchers to identify which research endeavors relate to rare diseases • Ongoing work on a white paper entitled "*Nurturing nascent advocacy and research groups along the trajectory toward clinical trial readiness: recommendations from the Chloe Barnes Rare Disease Advisory Council* "

Progress through Collaboration: The Council recognizes that creating collaborations across and outside of Minnesota is vital to improving care for geographically diverse rare disease populations diffused throughout every health care system in the state. Over 2021, the Council both continued existing partnerships and expanded its collaborations with many different organizations to accomplish its mission with the following results:

- *St Cloud State University and Oregon State University* - completion of an IRB-approved statewide patient survey under the direction of Amanda Hemmesch, PhD, Associate Professor of Psychology and Kathleen Bogart, PhD, Associate Professor of Psychology to determine unique barriers to care amongst the rare disease community.
- *Sanford Health CoRDS*- collaborated on an annual summit, participated in a podcast, collaborated on the dissemination of the patient survey. Collaboration that is more formal will be determined in 2022.
- *National Organization for Rare Disorders (NORD)* - Assisted in organizing and co-hosting monthly patient listening sessions for the Minnesota rare disease community during COVID to understand the pandemic's impact on patients. NORD has used the listening sessions to construct responses to COVID impacts at the national level. Led multiple webinars for NORD to assist other states in passing legislation for and operationalizing their state councils.
- *State of Virginia Rare Disease Advisory Council*- Presented on the work of the Minnesota Council at November quarterly meeting to assist the VA Council in operationalizing their Council
- *College of Pharmacy's Center for Orphan Drug Research* - University of Minnesota-Council administrator Erica Barnes and Council member Paul Orchard served on the Center's eighth

annual Rare Disease Day event planning committee. More than 230 University of Minnesota students, staff, and faculty, as well as individuals from health care organizations, patient advocacy groups, and the biomedical industry attended virtually.

- *University of Minnesota "Students for Rare" Club* - In 2021, a group of students launched the first club on campus. The club, co-advised by Council administrator Erica Barnes, hosted numerous events including a campus-wide event featuring a motivational speaker living with a rare disease.

Stakeholder Engagement, Outreach, and Awareness*:* Historically, there has existed few collective initiatives around stakeholder engagement and outreach in the rare disease community as a population. Patient groups and researchers addressing rare diseases that are fragmented geographically and across health care systems. Data and information pertaining to rare diseases are neither centralized nor standardized.[3] Many rare disease patients have no knowledge of other individuals living with their rare disease and most patient advocacy organizations have lesser capacity as compared to more common disease communities. For these reasons, awareness and outreach continued to make up a vital aspect of the Council's collective work to address the community's needs. In 2021, the Council continued its outreach, specifically to the medical community through a provider survey (see "Results and Accomplishments").

Surveys

In 2021, the Council made strides towards its first pillar of a deepened understanding of the needs of the rare disease community with the completion of one statewide and one nationwide survey.

The Council's patient access survey is the largest survey of the rare disease community to date in the United States and the results have been submitted for publication to the international journal *Orphanet*.

The Council's clinician survey was designed to increase our understanding of the needs of the community from a healthcare provider perspective as well as gather information on what support a clinician may need to improve care for their patients with a rare disease. Outcomes and themes from the surveys will serve as the basis for identifying priority work efforts in 2022. Summaries of both surveys can be found in the Appendix.

Recommendations

As mandated in law, a significant function of the Council is to provide advice to state agencies. In addition to numerous communications with various state agencies, the following are examples of how the Council provided advice and/or recommendations:

- In collaboration and consultation with NORD and partially in response to a rare disease patient contacting the Council, the Council provided guidance to the Governor's office on adding rare diseases to the COVID vaccination prioritization list.
    - Result: The rare disease community was added to the prioritization list. The Council received a note of appreciation from a patient:

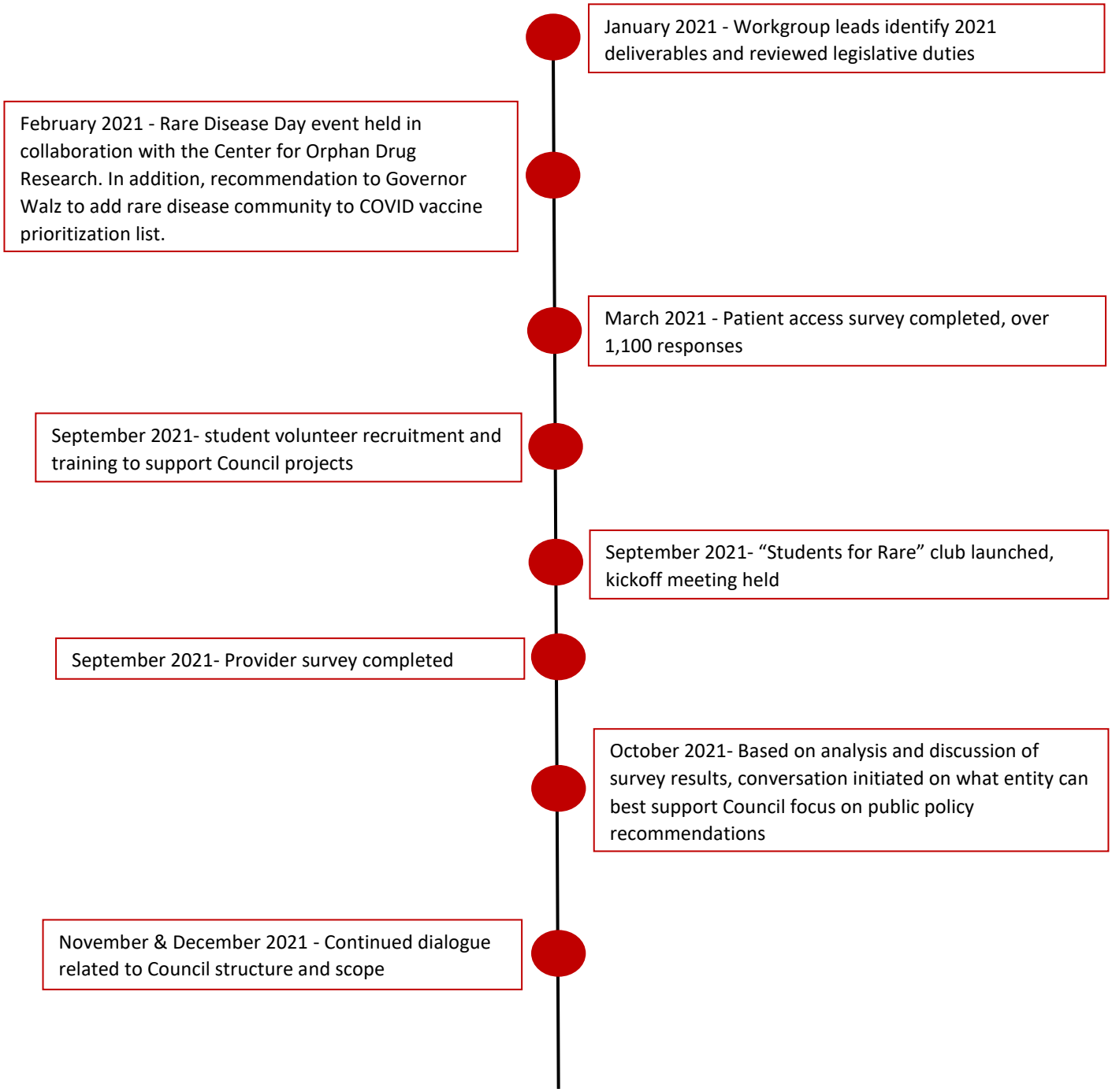
        "Thanks so much for writing back so quickly! This is amazing!! It looks like it has been added to Phase 1b – tier 2 People with rare conditions or disabilities that put them at higher risk…. This would be such a relief!! Please do share my message and gratitude to you and the Council. Merci infiniment!!"

- In collaboration and consultation with NORD, the Council provided a statement to Opioid Prescribing Workgroup (a DHS established advisory body to forward the Opioid Prescribing Improvement Program) on the unique pain management challenges in the rare disease community.
    - Result: The Opioid Workgroup requested a list of rare disease non-profits so that they could communicate with rare disease patients to learn more about the unique needs of the community firsthand and ensure that their recommendations were inclusive of the community,

Mentorship and education of the next generation

The Council forged and expanded a number of relationships at the University of Minnesota with the goal of better equipping future professionals to provide care to the rare disease community. In addition to two PhD candidates from the School of Public Health and Bioinformatics departments respectively, the Council recruited and trained one master's student and five undergraduates to assist on these research projects. Because of a collaboration with an undergraduate, a "Grand Challenge" class now focuses on rare diseases. The Council assisted in establishing a student run Rare Disease Club.

2021 Council Highlights



Future direction of the Council

A core value of the Rare Disease Advisory Council is patient-centered care, as articulated in Pillar One. The Council believes that the people who are impacted most should drive all actions: the patients and their families. All quarterly meetings open with a "mission moment" at which time patients or caregivers with direct experience of a rare disease diagnosis were asked to tell their story and answer questions from Council members. This grounded the Council in its mission (see appendix for all mission moment presenters). Following the completion of the patient survey, the Council determined that, based on reported results, influencing policies that can improve the lives of the rare disease community should be a core component of Council activities. As such, the Council has engaged in ongoing discussion around what organizational entity might best support addressing policy related to rare disease.

Appendix

1. Comparison table of statute with actions taken in 2021
2. Council inaugural work plan
3. Council member bios
4. Patient survey results
5. Provider survey results
6. Mission moment presenters

Citations

1 https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases

2 Council work plan

3 Valdez R, Ouyang L, Bolen J. Public Health and Rare Diseases: Oxymoron No More. Prev Chronic Dis 2016;13:150491. DOI: http://dx.doi.org/10.5888/pcd13.150491.

Appendix Item 1: Statute and Council Actions in 2021

Statue	Council Action
Identify **best practices** for rare disease care implemented in other states, at the national level, and at the international level, that will improve rare disease care in the state and seek opportunities to collaborate with similar organizations in other states and countries.	• Numerous collaborations with CoRDS program at Sanford Health throughout 2021, more formal collaborations planned for 2022 • Completed "Show Me ECHO" (Missouri telementoring program) training to familiarize the Council with activities being undertaken in other states to provide further training in patient care to physicians • Numerous collaborations with National Organization for Rare Disorders (NORD) to both support and learn from implementation of Advisory Councils in other states • Presentation to Virginia Rare Disease Advisory Council to foster collaboration and communication
Work with the state's medical schools, the state's schools of public health, and hospitals in the state that provide care to persons diagnosed with a rare disease to develop **resources** or **recommendations** relating to quality of and access to treatment and services in the state for persons with a rare disease	• Multi health system clinician contact registry launched, ongoing recruitment activities being completed • Council administrator is a staff advisor for "Students for Rare" (MN chapter of NORD) , first ever rare disease club in the state of MN • Mentorship of PhD candidate in University of Minnesota Bioinformatics Program, five undergraduate volunteers also engaged in research project • Mentorship of PhD candidate in University of Minnesota School of Public Health's Health Policy program with the goal of creating a white paper related to cost impacts and recommendations for reducing costs
a **list of existing, publicly accessible resources** on research, diagnosis, treatment, and education relating to rare diseases	• Physician contact registry • List of non-profits in the state of MN that care for rare diseases created and made available to several interested organizations
identify problems faced by patients with a rare disease when changing health plans	• Inclusion of questions on both patient and clinician survey asking participants to identify barriers related to health plans
recommend how to remove obstacles faced by these patients to finding a new health plan and how to improve the ease and speed of finding a new health plan that meets the needs of patients with a rare disease	

Identify best practices to ensure health care providers are adequately informed of the most effective strategies for recognizing and treating rare diseases	• Engagement with the Patient Centered Medical Home staff at MDH to determine educational needs of providers • Engagement with multiple medical associations with a goal of creating educational content related to rare diseases for dissemination by the associations
Advise, consult, and cooperate with the Department of Health, the Advisory Committee on Heritable and Congenital Disorders, and other agencies of state government in developing information and programs for the public and the health care community relating to diagnosis, treatment, and awareness of rare diseases.	• In collaboration and consultation with NORD, provided guidance on adding rare diseases to the COVID prioritization list • In collaboration and consultation with NORD, provided a statement to Opioid workgroup on the unique pain management challenges in the rare disease community • Ongoing collaboration with a Minnesota-based patient advocacy organization (Legacy of Angels Foundation) to assist them in making a recommendation to the Minnesota Committee on Heritable and Congenital Disorders for adding Krabbe disease to the Minnesota newborn screening panel • Ongoing collaboration with MDH and Gillette Children's to participate in a roundtable discussion focused on transition of care for pediatric patients with rare diseases
The advisory council shall collect additional topic areas for study and evaluation from the public. In order for the advisory council to study and evaluate a topic, the advisory council must approve the topic for study and evaluation.	• Developed public comment tracker • Administration of rare disease patient access survey

Appendix Item 2: Council Inaugural Work Plan

From the Chair

Rare diseases, when taken as a group, are anything but rare. By committing your time to work together to improve Minnesota's response, you have shown your understanding of the devastating impact rare diseases have on patients, their families, their communities, and our state. This Chloe Barnes Advisory Council on Rare Diseases presents a remarkable opportunity. Over the months to come, we will be working to describe the obstacles that the rare disease community faces and to develop innovative solutions and new approaches to overcome them.

Here are the initial ideas we will work by. We agree to:

- Put aside what we think we know so that we can approach systemic problems in a novel way.
- Focus on identified goals and outcomes.
- Propose actionable, practical, and achievable results.
- Adhere to measurable and meaningful metrics of success.

I am deeply grateful for your commitment to this challenge and to this Council. I look forward to working together to improve the lives of people who are impacted by rare diseases.

Vision

The Chloe Barnes Advisory Council on Rare Diseases envisions a state where every Minnesota Citizen living with a rare disease has access to a timely diagnosis, expert/ coordinated care, as well as individualized treatment, management, and support throughout the lifespan

Mission

The mission of the Minnesota Rare Disease Advisory Council is to provide advice on research, diagnosis, treatment, and education related to rare diseases

Pillars of Focus

1. Deepened Understanding of the Needs of the Rare Community
   The Council will acquire a comprehensive understanding of the systemic barriers across patient groups unique to the rare community
2. Increase Access to and Coordination of Care
   The Council will develop recommendations and resources to improve access to and coordination of care for rare disease patients
3. Identify Strategies for Reducing Time to Diagnosis
   The Council will advise consult and cooperate with multiple institutions in the state to develop information and programs that increase awareness for diagnosis and treatment of rare diseases
4. Encourage the Acceleration of Research
   The Council will foster the increase of rare disease research through awareness and collaboration

Pillar 1: Deepened Understanding

What is our Aspiration?

The Council will have a deep and comprehensive understanding of the systemic barriers across patient groups unique to the rare community

Why is it Important?

Due to the historic lack of collective consciousness around rare diseases as a class, there are many gaps in knowledge related to rare disease impact and care. For example, the incidence and prevalence of the majority of rare diseases in Minnesota are unknown. Additionally, there is no quantitative information on how many providers with expertise in rare disease care are practicing the state of Minnesota. These are just two examples. The Council ascribes to the axiom that what you cannot measure you cannot improve.

Concrete Goals

- Gather Minnesota specific baseline data on important metrics (i.e. prevalence/incidence, possible population disparities, cost burden of rare diseases)
- Identify most the common barriers to care across rare disease populations in Minnesota across the lifespan
- Directly and regularly communicate with rare patient communities to collect input on additional topics for consideration
- Collaborate with various disease-specific and public health organizations around the country to identify best practices in other states and internationally

Pillar 2: Increased Access, Coordination

What is our Aspiration?

The Council will develop recommendations and resources to improve access to and coordination of care for rare disease patients

Why is it Important?

Among the medical community, there are concerns that, for the rare disease population, adult specialists may not exist for a significant group of these patient populations for various reasons. Additionally, the costs of delivery of services/treatments for rare disease patients is a growing concern.

Concrete Goals

- Engage the state's medical schools, schools of public health, and hospitals to develop centralized, publicly accessible resources on diagnosis, treatment, and education relating to rare disease
- Identify problems faced by patients when there is a change in health plans and make recommendations on removing these obstacles to finding a new health plan
- Create a list of existing resources on research, diagnosis, treatment, and education for rare diseases
- Apply knowledge from other disease care models (i.e. more well understood rare disease populations) to improve coordination of care

- Provide healthcare providers with information on best practices so that they are informed of the best strategies for recognizing and treating rare diseases

Pillar 3: Reduced Time to Diagnosis

What is our Aspiration?

The Council will advise, consult, and cooperate with multiple institutions in the State to develop information and programs that increase awareness for diagnosis and treatment of rare diseases

Why is it Important?

The average wait time to a diagnosis for a rare disease patient is 6-7 years. Additionally, a rare disease patient is misdiagnosed 2-3 times. This delay in diagnosis creates inefficiencies in the system and significantly negatively affects the patient's quality of life and health.

Concrete Goals

- Provide resources for primary care providers so that they are adequately informed of the most effective strategies for recognizing and treating rare diseases
- Identify current technological tools to assist general practitioners and primary care providers with effective care management of rare disease patients
- Apply advances in technology more comprehensively to the diagnosis of rare diseases (next generation sequencing)
- Advise, consult and cooperate with state agencies to develop information and programs for the public and healthcare community to increase awareness and improve diagnosis and treatment for rare diseases

Pillar 4: Acceleration of Research

What is our Aspiration?

The Council will foster the increase of rare disease research through awareness and collaboration

Why is it Important?

Currently, only roughly 5% of the over 7,000 rare disease patient populations have an FDA approved treatment for their specific disease.

Concrete Goals

- Nurture the growth and organizational health of rare patient communities (i.e. provide basic information to emerging rare communities relative to characteristics of a mature patient community)
- Facilitate connections between researchers and patients, rare disease experts and providers, and among diverse rare disease patient organizations
- Explore approaches to novel clinical trial design

Appendix Item 3: Council Member Bios

Jakub Tolar, MD, Ph.D.
Position: Council Chair
University of Minnesota

Education:
Medical School, Charles University, Prague, Czech Republic
Residency in Pediatrics, University of Minnesota Medical School
Fellowship in Pediatrics and Hematology/Oncology and Blood and Marrow Transplantation, University of Minnesota Medical School
PhD, Molecular, Cellular, Developmental Biology and Genetics, University of Minnesota

Further Information:
Jakub Tolar, MD, PhD is the Dean of the University of Minnesota Medical School and a Distinguished McKnight Professor in the Department of Pediatrics, Blood and Marrow Transplantation. He is also the Vice President for Clinical Affairs at the University of Minnesota, Board Chair for University of Minnesota Physicians and co-leader of M Health Fairview, the Joint Clinical Enterprise between the University of Minnesota Medical School, University of Minnesota Physicians and Fairview Health Services.

An internationally recognized physician and researcher, Dr. Tolar is known for his care of patients with recessive dystrophic epidermolysis bullosa. His research focuses on using hematopoietic stem cell transplant as a treatment for rare genetic disorders. Originally, from the Czech Republic, Dr. Tolar received his medical education (MD) in Prague at Charles University. In 1992, he came to the University of Minnesota, where he completed his PhD in Molecular, Cellular & Developmental Biology and Genetics.

Erica Barnes, MA ccc-SLP
Position: Rare Disease Advisory Council Administrator/Community Outreach
University of Minnesota

Education:
MA in Speech and Language Pathology, University of Minnesota
BA in Humanities, writing intensive, Maranatha Baptist University

Further Information:
Erica has spent the last 10 years in advocacy and policy for the rare disease community. In addition to founding Chloe's Fight Rare Disease Foundation, she was the first MN State Ambassador for the Rare Action Network powered by NORD (National Organization for Rare Disorders). She was the recipient of the 2020 Rare Impact Award. Erica and her family have traveled extensively and in her free time, she enjoys cooking recipes from the countries she has lived in/visited. She also spends a fair amount of time wresting items away from her dog for whom the world is a giant chew toy.

Tim Schacker, MD
Position: Ex-Officio Appointee
University of Minnesota

Education:
Medical School, University of Minnesota, Minneapolis, MN
Internal Medicine Residency, Oregon Health Sciences University, Portland, OR
Infectious Disease Fellowship, University of Washington, Seattle, WA

Further Information:
Dr. Schacker is interested in how HIV causes immune suppression and why antiretrovirals do not fully restore immunity. His group focuses on inflammatory damage in lymphatic tissues; the principal site of HIV infection that results in fibrosis of the lymphatic structures required maintaining a normal population of CD4 cells. They are testing novel therapies to prevent and/or reverse this process and slow T cell depletion in HIV and improve their reconstitution when antiretroviral is begun. He is also the principal investigator of a federally funded program of projects designed to determine barriers to HIV eradication. In addition, Dr. Schacker has established a collaboration with the Joint Clinical Research Center in Kampala, Uganda to study how constant exposure to common infections like tuberculosis, malaria, and helminthic infections affect rates of HIV transmission and progression.

Lisa Schimmenti, MD
Position: Ex- Officio Appointee
Mayo Clinic

Education:
Fellow Pediatric Critical Care, Yale Affiliated Hospitals Program, Yale University School of Medicine
Fellow Pediatric Critical Care, Harbor-UCLA Medical Center
Residency, Harbor-UCLA Medical Center

Further Information:
As a pediatrician and clinical geneticist, Dr. Schimmenti specializes in the care of children with rare and undiagnosed disease with a focus on conditions characterized by hearing and vision loss. She is an integral member of the Genomic Odyssey Board Team in the Center for Individualized Medicine at the Mayo Clinic. Her laboratory program focuses on hearing and vision disorders using both zebrafish and mouse models to understand the mechanism of disease and to identify genes of unknown significance and their role of human hearing and vision loss, with a long-term goal of identifying novel therapies. She has been the PI or co-PI of two R01s and a March of Dimes award in the areas of vision and hearing. We are currently conducting experiments to identify drugs that would ameliorate hearing phenotypes in both zebrafish and mouse models of Usher Syndrome Type 1 and performing experiments to assess the phenotypic consequences of engineered pathogenic variants other zebrafish models of human hearing loss that include PAX2, MYO7A, EYA4, GJB2, NF2, SLC26A4, and ERCC6. Her laboratory is committed to maintaining a record of, and providing training in rigorous and unbiased experimental design, methodology, analysis, interpretation, and reporting of results

Nicole Brown, MSN, PHN
Positon: Ex-Officio Appointee
Minnesota Department of Health

Education:
Master of Science - MS, Pediatric Nurse
Bachelor of Arts – BA

Further Information:
Nicole Brown, MSN, RN, PHN, CPNP-PC is the Children and Youth with Special Health Needs (CYSHN) Section Manager at the Minnesota Department of Health and the state's Title V CYSHN Director. She provides leadership, collaboration, and aligns strategic priorities to create an effective system of care designed to meet the needs of CYSHN and their families throughout Minnesota. Nicole's experience working as a pediatric nurse practitioner at a community-based clinic and as a public health professional at both the local and state level has fueled her passion to create optimal health and quality of life for all CYSHN and their families. Nicole received her Master's Degree in Nursing from the University of Minnesota. She served as the National Association of Pediatric Nurse Practitioners' liaison to the American Academy of Pediatrics Task Force on Improving the Effectiveness of Newborn Hearing Screening, Diagnosis, and Intervention. Nicole is the parent of two youth who is deaf and was identified through newborn screening.

Rep. Kelly Morrison, MD
Positon: Legislative Appointee
Minnesota House of Representatives

Education:
Medical school, Case Western Reserve Medical School
Residency, Obstetrics and Gynecology, Northwestern University Mcgraw Medical Center
Internships, Northwestern University Mcgraw Medical Center Residencies

Further Information:
Rep. Kelly Morrison is serving her second term in the Minnesota House of Representatives, representing House District 33B, which includes the cities of Deephaven, Shorewood, Tonka Bay, Minnetonka Beach, Mound, Chanhassen, Excelsior, Spring Park, Orono and Greenwood. She is an Assistant Majority Leader and serves on the following committees: Health Finance and Policy; Preventive Health; Early Childhood Education Finance and Policy and Environment and Natural Resources Finance and Policy. Rep. Morrison has been a practicing OBGYN physician for almost two decades in Minneapolis. She lives in Deephaven with her husband John and three children. In her free time, she enjoys reading, being outdoors, and spending time in the BWCA with her family.

Rep Tony Albright- Legislative Appointee
Positon: Legislative Appointee
Minnesota House of Representatives

Education:
B.S., business administration, Minnesota State University, Moorhead

Further Information:
State Representative Tony Albright was first elected in 2012 to represent the people of District 55B. Tony has a B.S. in Business Administration and works in the financial services and investment advisory industry. He and his wife, Marianne, have three children. Tony is active in his community of Prior Lake and throughout Scott County, serving on the Prior Lake and Jordan Chambers of Commerce and volunteering with his church. At the legislature, Albright is the Minority Lead on the Human Services Finance and Policy Committee. He also serves on the Higher Education Finance and Policy and the Ways and Means Committees. Tony is a member of the University of Minnesota's Chloe Barnes Advisory Council on Rare Diseases and the House GOP liaison to the Workers' Compensation Advisory Council

Sen. Matt Klein, MD
Positon: Legislative Appointee
Minnesota Senate

Education:
B.S., University of Wisconsin, Madison
M.D., Mayo Medical School
Residency & Chief Residency, Internal Medicine, Hennepin County Medical Center

Further Information:
Matt Klein is a member of the Minnesota State Senate, representing District 52. He assumed office in 2017. His current term ends on January 2, 2023. Klein ran for re-election to the Minnesota State Senate to represent District 52.

Sen. Julia Coleman
Positon: Legislative Appointee
Minnesota Senate

Education:
BA, University of Minnesota - Twin Cities

Further Information:
My husband Jacob and I live in Chanhassen, where he serves as a firefighter. Being a mother is what drives me to step up to the plate and fight for what I believe in, in order to give my sons, and all our children, the best future possible. By fighting to put power back into the hands of parents when it comes to their child's education, protecting our constitutional rights, changing how we address healthcare

costs, opposing a gas tax increase, and holding bureaucracies – like the Department of Human Services – accountable for how they spend our hard-earned tax dollars, I believe we can start to make that future possible.

David Tilstra, MD
Positon: Hospital Administrative Appointee
CentraCare

Education:
BA, Northwestern College
MA, Iowa State University
MD, University of Iowa College of Medicine

Further Information:
Dr. Tilstra is Community Transformation Officer of CentraCare, an integrated healthcare system based in St. Cloud, MN, since January of 2019. His current role focuses on working in the community to change the local infrastructures to build a healthy environment. Dr. Tilstra served as the President of CentraCare Clinic from 2012 to 2019. He was active in managing physician engagement, strategic thinking, system thinking, recruitment, quality improvement, patient satisfaction and safety, and physician relationship activities. Before that, he was the Medical Director of CentraCare Clinic from 2003 to 2012, emphasizing ambulatory clinical improvement, patient experience, and community engagement. Dr Tilstra practices part-time as a Medical Geneticist for CentraCare Clinic since 1995. He obtained a B.A. in chemistry from Northwestern College of Orange City in 1983, followed by a year in a biochemistry graduate program at Iowa State University. He attended medical school at the University of Iowa College of Medicine completed in 1998, followed by a pediatric residency at the University of Iowa Hospitals and Clinics completed in 1991. Dr. Tilstra completed a Fellowship in Medical Genetics at the University of Washington in Seattle in 1994. He is board certified by the American Board of Medical Genetics. He completed an MBA with an emphasis in healthcare administration from the University of Colorado – Denver in 2016.

Barbara Joers
Positon: Hospital Administrative Appointee
Gillette Children's Specialty Healthcare

Education
BA, hospital administration from Marymount University in Arlington, Virginia
MA in health services management and policy from George Washington University in Washington, D.C.

Further Information:
Barbara Joers became Gillette's chief executive officer in October 2013. In her tenure at Gillette, Joers has implemented a focused and strategic approach to system improvement that cultivates Gillette's expertise in core service areas and emphasizes collaboration to improve care for their patients. Under her leadership, the system has emphasized mastering quality and safety, use of data to guide decisions, and continuous improvement in efficiency and stewardship of resources.

Karl Nelsen, PA-C MS
Position: Patient Advocate Appointee

Education:
Augsburg College, Minneapolis, MN Board Certified Physician

Further Information:
Mr. Nelson is Vice-President of the Board of Directors for the National Foundation for Ectodermal Dysplasias. Has been a member of the NFED family since he was 13 years old. He and his wife, Nancy, live in Minnesota with their children, Andrew, Abby and Samantha. Karl and Samantha are both affected by ectodermal dysplasia. He is a physician assistant in interventional radiology at Fairview Southdale Hospital and Fairview Ridges Hospital, Patient advocate for Ectodermal Dysplasia.

Rae Blaylark, Patient Advocate Appointee
Position: Patient Advocate Appointee

Further Information:
Rae Blaylark is a passionate and genuine person who is committed to continuous learning and improvement through a credible and patient approach that creates consistent and dependable results. She has outstanding communication skills, possessing the ability to connect with those she encounters, both personally and professionally. She seeks to help others find their hidden strengths, their values and the necessary skills in order to become productive members in whatever they are called to do.

Arthur Beisang, MD
Position: Physician Appointee
Gillette Children's Specialty Healthcare

Education:
Medical School, St. George's University School of Medicine, Grenada, West Indies
Residency, Pediatrics, University of Minnesota, Minneapolis
Fellowship: University of Minnesota, Minneapolis

Further Information:
Dr. Beisang is a general pediatrician with experience as chief of staff/ vice president of medical affairs. Previously he was director of product development for Bioplasty Incorporated where he developed and patented several medical devices and brought them into commercial production. Also 35 years of intermittent medical device development. Currently Co-Director of the Rett and Rett Related Disorders clinic and Aerodigestive clinic at Gillette.

Sheldon Berkowitz, MD, FAAP
Position: Physician Appointee
Children's Minnesota

Education:
Medical School, University of Colorado Health Sciences Center, Aurora, CO
Residency, Ann and Robert Lurie Children's Hospital of Chicago, Northwestern University, Chicago, IL

Further Information:
Dr. Berkowitz has been a Pediatrician for over 35 years and has been in practice at Children's Hospitals and Clinics since 2001. He went into Pediatrics because he enjoys interacting with children and tries to make their visits to the clinic as enjoyable as possible. Dr. Berkowitz enjoys all aspects of primary care and is very interested in expanding the concepts of the Medical Home to all patients. He has a special interest in Bioethics and enjoys writing and lecturing on this subject. Dr. Berkowitz is President-Elect of American Academy of Pediatrics, MN chapter.

Kris Ann Schultz, MD
Position: Physician Appointee
Children's Minnesota

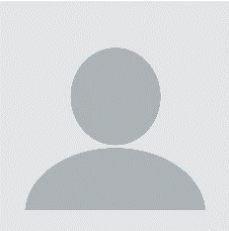
Education:
Medical School, Loyola University Chicago Stritch School of Medicine
Residency, University of Minnesota Medical Center
Fellowship, University of Minnesota Medical Center

Further Information:
Kris Ann P. Schultz, MD, is a pediatric oncologist at Children's Minnesota. She graduated summa cum laude from Drake University (B.A.) and summa cum laude from Loyola University (M.D). She completed her pediatric residency and pediatric hematology/oncology fellowship at the University of Minnesota and received a Master of Science degree in clinical research during the course of her fellowship. Dr. Schultz is the Principal Investigator for the International Pleuropulmonary Blastoma (PPB) and DICER1 Registry and the Principal Investigator and founder of the International Ovarian and Testicular Stromal Tumor Registry. Her current research focuses on development of novel treatments for DICER1-related tumors. She is a member of the Alpha Omega Alpha Honor Medical Society and serves as Co-Director for Cancer and Blood Disorders Research at Children's Minnesota. Dr. Schultz joined the Hematology Oncology program at Children's of Minnesota in 2008 and has particular interest and expertise in the care of children with pleuropulmonary blastoma, ovarian tumors and other rare childhood cancers.

Kerry Hansen, RN
Position: Nurse Appointee
Fairview

Education:
Specialties and Services: Bleeding and Clotting Disorders

Further Information: Kerry Hansen is a nurse clinician at the Center for Bleeding and Clotting Disorders. He also serves as a member of the legislative committee for the Hemophilia Foundation of Minnesota/Dakotas.



Tom Blissenbach, RPh, MS
Position: Pharmacist Appointee
Retired

Education: BS in Pharmacy and MS in Hospital Pharmacy, University of Minnesota

Further Information:
Mr. Blissenbach retired in 2019 as VP of Sales, Marketing and Business Development for Fairview Pharmacy Services, LLC. He received his BS in Pharmacy and MS in Hospital Pharmacy from the University of Minnesota. Tom developed and led numerous ambulatory pharmacy business initiatives at FPS from 2000 to 2019. He was responsible for the sales and marketing group, engagement with pharma around innovative partnerships, research organization supporting drug development trials, and a partnership with U of MN genetics department to support research and treatment for rare diseases and orphan drugs. He has more than 45 years of experience in retail, hospital and alternate site pharmacy. He was a pioneer in the growth of Home Infusion and the evolution of Specialty Pharmacy. His work history includes positions involving multiple-site pharmacy management, sales and marketing, business development and consulting. Throughout his career Tom had consistently been a leader and innovator in the practice and business of pharmacy. Previous employers include HMSS, Caremark and Chronimed. He served in a leadership capacity for numerous local, state and national pharmacy organizations Tom was an inaugural Board Member of the National Association of Specialty Pharmacy (NASP). He was appointed to the Chloe Barnes Advisory Council for Rare Diseases in 2019.

Soraya Beiraghi, DDS, MSD, MS, MSD
Position: Dentist Appointee
University of Minnesota

Professor, Department of Developmental and Surgical Sciences
Director, Division of Pediatric Dentistry

Education: DDS, Indiana University School of Dentistry
MSD (Pediatric Dentistry), Indiana University
MSD (Oral Health and Preventive Science), Indiana University
MS (Clinical Nutrition), University of Kentucky
BS, Biology, University of Kentucky
Certificate, Oral Health Research, Indiana University School of Dentistry

Further Information:
Dr. Beiraghi is Professor and Head of the Division of Pediatric Dentistry at the University of Minnesota, School of Dentistry and has been a Staff member of craniofacial programs for over 25 years at UNMC/BTNRH and UMN. She was the Director of the IADR/AADR Craniofacial Biology Section as well as a member of the Society of Craniofacial Genetics and ASHG. Her primary research focus is in the area of craniofacial anomalies; specifically, she is currently working on genotype-phenotype correlation and

underlying molecular mechanisms. This includes congenital craniofacial syndromes and degenerative musculoskeletal defects, both of which may involve the oral cavity, face, and temporomandibular joint.

2018 Academy for Excellence - Dr. Beiraghi was recognized for her international reputation for care of patients with multiple genetically based craniofacial anomalies and medically compromised patients with significant special health care needs and her ability to demonstrate commitment to excellence in direct patient care, intraprofessional, interprofessional collaborative care, public service and outreach, and innovation in healthcare delivery models.

Amy Gaviglio, MS-CGC
Position: Genetic Counselor Appointee
Center for Disease Control

Public Health Genetics Consultant; CDC, APHL, and Expecting Health
G2S Corporation with the Newborn Screening and Molecular Biology Branch of CDC

Education: Masters of Genetic Counseling, University of Michigan

Further Information:
Amy Gaviglio is a certified genetic counselor and public health genetics consultant who has been working in the Newborn Screening arena for the past 13 years. She is currently a consultant with the Centers for Disease Control and Prevention, Association of Public Health Laboratories (APHL), and Expecting Health. She is co-chair of APHL's New Disorders workgroup, and is a member of several of APHL's workgroups, including Short Term Follow-Up, Legal and Legislative Issues in Newborn Screening, and Health Information Technology. She also serves as Chair of the NBS Expert Panel for the Clinical and Laboratory Standards Institute, Advisor to the Midwest Genetics Network, and is on the Advisory Committee on Heritable Disorders in Newborns and Children's Education and Training workgroup.

Lee A. Jones
Position: Biotech Industry Appointee
Founder, President and CEO of Rebiotix Inc.

Education:
BS chemical engineering, University of Minnesota
Executive Management Program, Carlson School of Business, University of Minnesota

Further Information:
Rebiotix is a clinical stage biotechnology company developing a new class of biologic drugs based on live human-derived microbes. Previously, she was Chief Administrative Officer of the Schulze Diabetes Institute, U of MN and is the former president and chief executive officer of Inlet Medical. Prior to Inlet, she spent 14 years at Medtronic. Currently on the board of Electromed Inc., on the University of Minnesota's Office of Technology Commercialization advisory board, and on the board of MedicalAlley. She is a member of the Sofia Angel Investment Fund and is an advisor to several small companies. She is leading a fast-paced effort to develop a new way of treating disease through Microbiota Restoration Therapy (MRT). The company's first MRT is a biologic drug targeted at recurrent Clostridium difficile infection.

Abigail Miller, MD
Position: Payer Appointee
Chief Medical Officer and Senior Vice President PreferredOne®

Education:
Medical School: University of Minnesota Medical School
Residencies: St. John's Hospital

Further Information:
Prior to her current role, Dr. Miller served as the medical director of inpatient care management and utilization management for Fairview Health Services. She strives to provide excellent care to patients and their families in a compassionate manner.

Paul Orchard, MD
Position: Researcher Appointee
University of Minnesota

Education:
Medical School, Brown University
Pediatrics Residency, University of Wisconsin - Madison
Hematology/Oncology and Bone Marrow Transplantation Fellowship, University of Minnesota

Further Information:
Dr. Orchard is interested in the use of blood and marrow transplantation (BMT) and potentially other cell therapies for inherited metabolic diseases, like Hurler syndrome and adrenoleukodystrophy. He has developed new therapies specifically for this group of patients with the goal of minimizing neurologic deterioration during the transplant process, including the use of antioxidants, reduced intensity transplant regimens and combinations of therapy such as transplant and enzyme replacement. This work also benefits other children-undergoing transplant, particularly in regards to our commitment to minimize the side effects of transplantation.

In addition to his clinical work with patients who have inherited metabolic diseases, Dr. Orchard is also engaged in more basic research studies about determining strategies to enhance the delivery of enzymes to the brain and the peripheral nervous system for patients who lack specific enzymes. In addition, he is interested in testing other types of stem cells that may improve outcomes for patients with inherited diseases, including gene therapy approaches. He also leads research and clinical care for patients with osteopetrosis, an inherited disorder leading to increased density of bone, for which BMT is a treatment option. Dr. Orchard is considered an international expert in this disorder and its treatment.

Jackie Foster, MPH, RN, OCN
Position: Non-Profit Organization Appointee
Be The Match

Education:
BA nursing, St. Olaf College University of MN, Twin Cities MPH Public Health

Further Information:
Jackie has been a nurse since 2005, working in several roles including as a blood and marrow transplant staff nurse, an outpatient chemotherapy infusion nurse, and an oncology nurse navigator. Currently, she

leads the teams that help patients find clinical trials and develop education for patients and staff at the National Marrow Donor Program / Be The Match in Minneapolis. Her interests are in population-based interventions to increase access to care and implementing effective strategies to communication complex information to different audiences.

The Council has two vacant positons due to retirement or desire to step away. Those are the Social Work Appointee and Patient Advocate Appointee

Appendix Item 4. Patient Survey Summary

This online study included 1,128 participants, including adults with rare disease and parents/relatives/caregivers to individuals with rare disease. 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 and was also broken down to compare Minnesotans to residents of other states and for comparisons within Minnesotans (participants living in the Minneapolis-St. Paul (MSP) metropolitan area vs Greater Minnesota).

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 (88%) reported only one rare disease though some participants reported having up to 5 rare diseases.

Rare disease characteristics and diagnostic odyssey

Participants reported widely varying times between the onset of their symptoms and their eventual diagnosis: 30% waited 0-6 months, 9% waited 7-11 months, 23% waited 1-3 years, 11% waited 4-6 years, 6% waited 7-9 years, and 16% waited 10 more years for a diagnosis. There was also a lot of variability in how many providers were seen during the diagnosis process: 16% of participants saw only 1 provider, 38% saw 2-3 providers, 24% saw 4-5 providers, 13% saw 6-10 providers, and 7% 11+ providers before being diagnosed. This data suggests that multiple providers are often required to obtain a rare diagnosis in the US. This pattern was similar when comparing Minnesotans to the rest of the sample, and when comparing metropolitan versus Greater Minnesotans.

Health insurance

The majority of participants had at least one type of health insurance. Out of pocket spending varied widely across the sample: 35% reported paying $0-499 last year, 12% reported paying $500-999, 17% reported $1,000-1,999, 10% reported $2,000-2,999, and 26% reported paying over $3,000 in out-of-pocket healthcare expenses last year. Minnesotans were more likely to pay over $3,000 out of pocket than the rest of the sample, but there were no differences across the regions studied in Minnesota. While the majority of participants who needed specialized care often were able to easily receive it through their insurance, investigational treatments and off-label use of medications was more difficult to get approved.

Provider types and satisfaction

Most participants visited a general practitioner (73%) and specialist (81%) in the past year. Other types of providers were less likely seen within the past year: 42% of participants visited an allied health professional (e.g., occupational therapist), 33% visited a mental health professional, 45% visited a dentist, and 29% visited another type of provider (e.g., chiropractor, masseuse). Participants were often most satisfied with their specialists. Findings from the Patient Satisfaction Short Form suggested that participants were generally dissatisfied with their providers. Minnesotans were less satisfied with their providers' communication than other participants were.

Knowledge, Care, and Support

Many participants agreed or strongly agreed that the information they received from different providers was sufficient, although this varied across different types of providers: general practitioners (34%), specialists (69%), allied health professionals (53%), dentists (43%), and mental health professionals (39%), and from patient support organizations (59%).

Regarding the different types of support assessed in this study, 59% of participants agreed or strongly agreed that they received sufficient medical support, 39% agreed/strongly agreed that they received sufficient dental support, 49% agreed/strongly agreed that they received sufficient social support, 45% agreed/strongly agreed that they received sufficient financial support, 31% agreed/strongly agreed that they received sufficient psychological support. Minnesotans (metropolitan and outstate) were similar to participants from other states for most of the knowledge, care, and support variables included in this study.

Specialist Centers, Telehealth, and Care Coordination

The majority of participants (63%) reported that they did not access a specialist center for their rare disease. Even though specialist centers visits are not common, the majority of participants (54%) reported that they have 1-2 different medical specialists to help them manage their rare diseases. Approximately 24% of participants see 3-4 specialists, 21% see 5-6 specialists, and 12% see more than 6 specialists. Minnesotans were more likely to report that they had 6 or more specialists than participants from other states.

Nearly half of participants (47%) reported traveling over 60 miles for their rare disease care. Given the long distances that people may need to travel for care, it is not surprising that 58% of participants reported using telehealth services for appointments with specialists.

Only 12% of participants in the sample said that they have used a care coordinator. Among those who have used a care coordinator, the experience has been rated as helpful.

Minnesotans were more likely to report using telehealth services (67%) than other participants (56%), and Minnesotans were more likely to have a care coordinator (24%) than other participants (9%).

Pediatric Care and Transition to Adult Care

Only 27% of the overall sample for this study reported using pediatric health services. The majority of participants who used pediatric services were either satisfied or very satisfied (78%). Minnesotans were more likely to have had a pediatric provider (46%) than participants from other states (22%).

Barriers to Care and Relocation

Regarding finances, the most common responses were that it never (45%) or sometimes (33%) affected their ability to get care. These were also the most common responses to the items asking about travel distance (never: 52%, sometimes: 29%), difficulty getting time off work (never: 59%, sometimes: 23%), lack of childcare (never: 75%, sometimes: 11%), lack of/delay in getting referrals (never: 49%, sometimes: 32%), and for something not being covered by insurance (never: 47%, sometimes: 33%). In contrast, participants from other states were more likely to report that getting time off work never limits their care (61%) than Minnesotans (53%). Other participants were also more likely to say that childcare never limits their care (76%) than Minnesotans (69%). There was also a difference between Minnesotans and the rest of the sample with regard to insurance coverage being a barrier: 40% of Minnesotans said that this was sometimes an issue compared to 32% of participants in other states. As expected, participants living in outstate Minnesota were more likely to say that travel was a barrier about half of the time (9% vs 3%), most of the time (6% vs 3%), or almost always (7% vs 2%) than those living in the MSP area.

Changes in Care due to COVID-19

Approximately 55% of participants reported that the COVID-19 pandemic has changed their ability to access rare disease care and services. Minnesotans were similar to the overall sample on this item, but

within Minnesota, those living in outstate areas were more likely to report changes than those living in the MSP metropolitan areas.

Quality of life

Compared to population norms of United States residents, adults with rare diseases in Minnesota had significantly worse stigma, sleep disturbance, physical function, pain interference, ability to participate in social roles and activities, fatigue, and anxiety. Adults in Minnesota did not differ from the general population on depression levels. Even when compared to adults with common chronic diseases, adults in Minnesota with rare diseases had significantly worse stigma, physical function, fatigue, depression, and marginally worse anxiety.

Compared to the general American pediatric population, children in MN with rare diseases had significantly worse peer relationships, pain interference, fatigue, depression, mobility, and marginally worse anxiety. Norms for children with common chronic diseases are not available for comparison. Children and adults in states other than MN scored worse than population norms on all domains. Adults outside of MN with rare diseases fared significantly worse than people with common chronic diseases on all domains for which norms were available.

Conclusions

Participants in the United States experienced significant diagnostic delays. The majority of participants, and all of Minnesotans in this sample had health insurance, yet more than one quarter had to pay an unacceptable $3,000+ out of pocket last year. Although most participants did not access a specialist center, the majority see one or two specialists for their rare disease. Participants were dissatisfied with their healthcare providers in general, but were more satisfied with their specialists. Only around half felt their medical, dental, and social support was sufficient, yet less than a third had sufficient psychological support. About half of participants travel more than 60 miles for care and/or access their specialists via telehealth. These healthcare challenges take a notable toll on the quality of life of participants with rare diseases, resulting in generally poorer quality of life compared to the average American and Americans with common chronic diseases

Appendix Item 5: Front line Provider Survey Summary

Survey Development

The Provider Survey was developed collaboratively between the Chloe Barnes Advisory Council on Rare Diseases Provider Survey Working Group and the Research Methodology Consulting Center at the University of Minnesota. The Provider Survey contains 28 questions, which are grouped into the following categories: Knowledge of and Familiarity with Rare Diseases; Comfort with Recognizing Rare Diseases; Barriers to Next Steps when presented with a Rare Disease; Coordinating Care and Transitions from Childhood to Adulthood, and Education, Training, and Resources.

Survey Administration

The Provider Survey was administered from March through August of 2021. The survey was administered online via Qualtrics through an anonymous link sent to providers' email accounts. Front-line providers' email addresses were gathered via a data request made to the Minnesota Board of Medical Practice. The survey was anonymous, except that at the end providers had the opportunity to be contacted about the Rare Disease Provider Registry. No incentive was offered to complete the survey.

Response Rate

In total, 327 front-line providers answered one or more questions, with 219 (67%) completing the entire survey. Of those who took the survey, 11% and 7% agreed to be placed on the pediatric and adult registries respectively, and 14% and 19% agreed to be contacted to learn more about the pediatric and adult registries respectively. Item-level response rates varied from 100% for the first question to 69% for the final question. Of the 327 respondents, seven self-identified as surgeons, and their data was excluded from analyses because they are generally not involved in the identification or diagnosis of diseases and therefore not the target population for this survey.

Sample Demographic Characteristics

Of the 320 respondents, 68% (n = 218) provided demographic information. Of the 218 respondents who provided demographic information, one-third (32%) reported being dentists, with primary care physicians (15%) and rare disease specialists (13%) comprising the second and third largest groups, respectively. Approximately 13% of respondents selected "other." Of the "other" responses, specialists who do not manage rare diseases as part of their practice (e.g., dermatologists, psychiatrists), emergency room physicians, and obstetrician-gynecologists comprise the largest groups. As noted above, seven respondents identified as surgeons and their data are excluded from all tables and analyses.

Table 1. Type of Medical Professional

Response Choices	Percent Selecting
Primary care/family practice physician	10%
Pediatrician	6%
Internal medicine physician	5%
Med-peds physician	1%
Physician assistant	5%

Nurse practitioner	1%
Dentist	22%
Specialist who manages rare diseases as part of your practice	9%
Other	9%
Not reported	32%

The type of medical professional variable was re-coded for use in the analyses below. The variable was recoded into four groups: Medical providers (n = 119, 37% of sample), dental providers (n = 70, 22% of sample), rare disease specialists (n = 29, 9% of sample), and respondents who did not report their profession (n = 102, 32% of sample).

Of the 218 respondents who provided demographic information, 63% reported spending most of their time in private or group practices, while 31% reported spending the majority of their time in a hospital based clinic. Five percent of respondents selected "other." The largest "other" responses comprised dental practices, community-based clinics, emergency departments, and single general private practices. The responses are summarized in the table below.

Table 2. Type of Medical Setting

Response Choices	Percent Selecting
Single specialty private practice	19%
Multispeciality group practice	24%
Hospital based outpatient clinic	9%
Hospital based inpatient clinic	13%
Other	5%
Not reported	32%

The final demographic question asked respondents to report the zip code their practice, hospital, or clinic is located. Then, the rural-urban commuting area (RUCA) codes were used to classify each zip code as rural, urban, etc. RUCA codes classify U.S. census tracts using measures of population density, urbanization, and daily commuting and are maintained by the USDA. The most recently available codes are based on the 2010 census tracts. The results show that 85% of respondents work in metropolitan areas, 4% work in micropolitan areas, and 8% work in small towns.

Table 3. Location of Medical Setting

Description of Location	Percent Selecting
Metro: Counties in metro areas of 1 million population or more	61%
Metro: Counties in metro areas of 250,000 to 1 million population	3%

Metro: Counties in metro areas of fewer than 250,000 population	21%
Nonmetro: Urban population of 20,000 or more, adjacent to a metro area	1%
Nonmetro: Urban population of 2,500 to 19,999, adjacent to a metro area	3%
Nonmetro: Urban population of 2,500 to 19,999, not adjacent to a metro area	8%
Other state or country	4%

Note: Thirty-three percent of respondents (n = 108) did not respond to this question.

Results

Knowledge and Familiarity Questions

The first question asked providers, “How would you rate your general knowledge of diagnosis and management of rare diseases?'' All 320 providers responded, with only 13% stating they were very familiar with the facts of diagnosis and management for rare diseases and 31% said they were unfamiliar with the facts of diagnosis and management for rare diseases. These numbers varied however by the type of medical professional with rare disease specialists reporting the highest familiarity and dentists reporting the lowest familiarity.

Table 4. Knowledge of Rare Diseases by Type of Provider

Knowledge of Rare Diseases	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Very familiar	8%	3%	59%	12%	13%
Somewhat familiar	63%	53%	35%	56%	56%
Not familiar	29%	44%	7%	32%	31%
Sample size	119	70	29	102	320

The second question asked providers, “Do you think your knowledge of rare disease diagnosis and management is sufficient to meet the needs of your practice?'' Ninety-six percent of respondents answered the question. The results are presented in Table 5. As can be seen, rare disease specialists were three times more likely to say their knowledge of rare diseases meets the needs of their practice than medical or dental providers.

Table 5. Knowledge of Rare Diseases Meets Needs of Practice by Type of Provider

Knowledge Meets Needs of Practice	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Sufficient	26%	20%	62%	33%	30%

Somewhat sufficient	51%	61%	31%	40%	48%
Insufficient	23%	19%	7%	9%	16%
Not reported	0%	0%	0%	18%	6%
Sample size	119	70	29	102	320

The responses to questions one and two are displayed as a cross-tabulation in Table 6. Of the respondents who report sufficient knowledge of rare disease to meet the needs of their practices, only one-third reported being very familiar with rare diseases, and one-tenth reported no familiarity with rare diseases. This finding suggests some providers do not perceive rare diseases as relevant to their practice. Similarly, of the respondents who reported no familiarity with rare diseases, less than half noted this was insufficient to meet the needs of their practice.

These results varied by type of medical professional. For example, 55% of rare disease specialists reported being very familiar with rare diseases and that their level of knowledge was sufficient to meet the needs of their practice, compared to 5% and 3% of medical and dental providers, respectively. Of note, dental providers were four times more likely than medical providers to rate being not familiar with rare diseases as sufficient to meet the needs of their practice.

Table 6. Knowledge of Rare Diseases vs. Sufficiency of Knowledge

	Knowledge Meets Needs of Practice			
Knowledge of Rare Diseases	Sufficient	Somewhat Sufficient	Insufficient	Not reported
Very familiar	10%	2%	0%	1%
Somewhat familiar	17%	33%	4%	3%
Not familiar	3%	13%	12%	2%

Comfort with Recognizing Rare Diseases

Rare diseases are heterogeneous and can sometimes be quite challenging to diagnose. We were interested in providers' comfort in identifying and initiating a diagnostic workup for patients where common etiologies have been ruled out.

The first question in this section asked providers, “To your knowledge, how often have you encountered a patient/patients with a rare disease in your medical practice?” Eighty-nine percent of respondents answered the question. Overall 8% of providers stated that they had never encountered a patient with a rare disease, and 20% reported frequently encountering patients with a rare disease. Dental providers were over twice as likely as medical providers to report no exposure to a patient with a rare disease. Similarly, medical providers were more than four times more likely than dental providers to report frequent exposure to patients with a rare disease (see Table 7).

Table 7. Exposure to a Patient with a Rare Disease by Type of Provider

Exposure to a Patient with a Rare Disease	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Never	6%	16%	0%	6%	8%
Occasionally	75%	80%	24%	43%	61%
Frequently	19%	4%	76%	15%	20%
Not reported	0%	0%	0%	36%	12%
Sample size	119	70	29	102	320

The second question asked providers, "When presented with a set of symptoms that you have not previously encountered and that do not follow a typical disease course, what are your next step(s)?" Providers had the opportunity to check all responses that applied and to provide any additional next steps not included in the list of response choices. Providers also had the choice to select "does not apply," which 2% selected. The remaining responses are summarized in Table 8 by type of medical provider. Thirteen percent of respondents selected "other." Of the other responses, the most common was to consult or discuss the set of symptoms with a colleague or specialist. Several of the dentists noted that they would call the patient's primary care physician to discuss their concerns or recommend the patient to a medical doctor. A small number of providers noted that they would gather more information about the patient's medical history (including family history).

Table 8. Next Steps When Presented with a Possible Rare Disease by Type of Provider

Next Step	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Wait and see if the disease course presents more diagnostic indicators.	31%	26%	14%	8%	31%
Refer the patient to a specialist.	78%	96%	79%	43%	78%
Order additional testing.	79%	17%	86%	14%	79%
Use online literature search/search engines.	89%	57%	86%	26%	89%
Other	13%	6%	28%	10%	13%
Does not apply	1%	1%	0%	5%	1%

Sample size	119	70	29	102	320

The next question noted that genetic diagnosis is increasingly common and asked providers, "How comfortable are you interpreting results from genetic testing?" Eight-six percent of respondents answered the question. The results are summarized by type of medical professional in Table 9. As can be seen, 71% of dental providers report not being comfortable with interpreting the results of genetic testing, compared to 33% of medical providers.

Table 9. Comfort with Interpreting Genetic Testing by Type of Medical Provider

Comfort with Genetic Testing	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Not at all comfortable	33%	71%	3%	28%	37%
Somewhat comfortable	47%	19%	28%	15%	29%
Comfortable	10%	1%	24%	5%	8%
Very comfortable	7%	1%	41%	4%	8%
I don't know	3%	7%	3%	3%	4%
Not reported	0%	0%	0%	45%	14%
Sample size	119	70	29	102	320

The final question in this section asked providers, "If you receive notification that a patient on your caseload has a positive/abnormal result on a newborn screen, how comfortable are you in coordinating the next steps in confirmatory diagnosis?" Eighty-four percent of respondents answered the question. The results are summarized by type of medical providers in Table 10. Again, differences by type of provider are evident.

Table 10. Comfort with Abnormal Newborn Screen by Type of Medical Provider

Comfort with Abnormal Newborn Screen	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Not at all comfortable	9%	40%	3%	10%	16%
Somewhat comfortable	29%	6%	3%	8%	15%
Comfortable	19%	3%	17%	1%	9%
Very comfortable	11%	0%	28%	4%	8%
I don't know	2%	6%	0%	1%	2%

Does not apply	30%	46%	48%	27%	34%
Not reported	0%	0%	0%	50%	16%
Sample size	119	70	29	102	320

Barriers to Next Steps when Presented with a Possible Rare Disease

We were also interested in barriers providers have experienced in identifying and initiating a diagnostic workup for patients. As such, we asked providers six questions about potential barriers to diagnosing a rare disease. The response rates ranged from 74-79%. The responses are summarized in Table 11. To examine differences by type of medical professional, the average response (from 1 to 4) for each item is presented by type of medical professional in Table 12 (missing responses are excluded). The results suggest that the type of insurance a patient carries is, relatively speaking, the largest reported barrier providers encounter. No clear pattern of differences emerged between the types of providers.

Table 11. Barriers to Diagnosing Rare Diseases

Question	Never or very rarely	A few times	Many times	Always or almost always	Not reported
How often has the type of insurance your patient carried affected your ability to order diagnostic testing?	25%	27%	20%	6%	21%
How often has the type of insurance your patient carried affected your ability to refer them to specialists?	27%	29%	18%	3%	23%
How often has the type of insurance your patient carried affected your ability to complete the recommended treatment plan?	15%	26%	28%	7%	24%
How often have you referred your patients with rare diseases and difficult diagnostic questions out of Minnesota?	63%	12%	2%	<1%	24%
How often has distance to a specialty center influenced your decision to refer a patient for care?	39%	25%	8%	2%	26%
How often have the productivity expectations of your system impacted your ability to manage care for medically complex patients?	39%	20%	11%	4%	27%

Coordinating Care and Transitions from Childhood to Adulthood

Given the complexity of many rare diseases, care can also be very challenging to coordinate. We were interested in providers' comfort in coordinating care and transitioning from pediatrics to adults for these patients. These questions were restricted to providers who reported they had encountered at least one patient with a rare disease (92% of sample).

The first question asked, “Have you ever had a patient on your caseload with a rare disease that required you to coordinate care across multiple specialties?” Seventy-three percent of respondents answered the question. Approximately 87% of medical providers, 63% of dental providers, and 93% of rare disease specialists reported having one or more patients with a rare disease that required them to coordinate care across multiple specialties. The remaining respondents reported that they had not had a patient with a rare disease that required them to coordinate care.

Then, the providers were asked, “Have you ever experienced difficulty transitioning a pediatric patient with a rare disease to adult care?” Seventy-two percent of respondents answered the question. Overall, 24% of respondents reported experiencing difficulties transiting a patient, including 30% of medical providers, 25% of dental providers, and 69% of rare disease specialists.

The providers who said they had encountered difficulties were asked to identify the type of difficulties. Providers could select all that apply and fill in other options. The results are summarized in Table 12 below. Other responses identified two additional problems: a) a lack of communication between the pediatric and adult systems, and b) that the adult system can be challenging for families to navigate logistically.

Table 12. Difficulties Transitioning Care by Type of Provider

	Type of Provider				
Difficulty	Medical	Dental	Rare Disease Specialist	Not Reported	Total
No adult specialist with expertise was available	59%	53%	85%	100%	30%
No local practitioner willing to take the patient was available.	56%	60%	65%	100%	28%
The patient's insurance presented challenges.	24%	60%	35%	0%	15%
Other	24%	7%	35%	0%	10%
Sample size	34	15	20	2	155

Next, providers were asked, “Have you ever been contacted by a specialist, provider, or family requesting a transfer of care for a patient with a rare disease to your practice that you did not feel you had the ability to care for?” Seventy-three percent of respondents answered the question. The results

are summarized in Table 13 below. Approximately 30% of medical and dental providers and 14% of rare disease specialists reported being contacted to care for a patient with a rare disease that they did not feel they had the ability to care for.

Table 13. Unable to Care for a Rare Disease Patient by Type of Provider

Unable to Care for Rare Disease Patient	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Yes	27%	29%	14%	5%	18%
No, I felt comfortable	36%	12%	79%	5%	23%
No, never contacted	38%	59%	7%	10%	31%
Not reported	0%	0%	0%	80%	28%
Sample size	119	70	29	102	320

Of those who reported not feeling they could care for the patient, 9% said it was because of the type of insurance the patient carried, 77% said the disease was too complex, 19% reported time constraints, and 25% reported other reasons which primarily that the disease was outside the scope of the practice or beyond the knowledge level of the practitioners.

Education, Training, and Resources

We were interested in how well providers' medical training prepared them for rare disease diagnosis and management and if they feel as though you could benefit from additional resources or training related to rare diseases. As such, the first two questions in this section were only asked of providers who reported encountering one or more patients with a rare disease (92% of sample).

First, providers were asked, "To what extent was your formal medical training useful in rare disease diagnosis and management?" Seventy-two percent of respondents answered the question. The responses are summarized in Table 14 below.

Table 14. Medical Training Useful by Type of Provider

Medical Training Useful	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Not very useful	25%	34%	28%	2%	19%
Somewhat useful	51%	49%	31%	12%	35%
Very useful	24%	17%	41%	3%	17%
Not reported	0%	0%	0%	82%	29%
Sample size	112	59	29	59	296

Then, providers were asked, “Do you think your clinical infrastructure is sufficient to support the needs of patients with rare diseases?” Seventy percent of respondents answered the question. The responses are summarized below.

Table 15. Clinical Infrastructure Sufficient by Type of Provider

Clinical Infrastructure Sufficient	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Sufficient	31%	22%	62%	2%	27%
Somewhat sufficient	50%	59%	31%	8%	36%
Insufficient	18%	19%	7%	0%	11%
Not reported	1%	0%	0%	90%	31%
Sample size	112	59	29	59	296

All providers were asked, “What kind of resources are available to you to support you in disease diagnosis and care?” They could check all that apply and were provided an “other” option. The responses are summarized below.

Table 16. Available Resources by Type of Provider

Available Resources	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Abstract services (i.e. Uptodate)	77%	20%	90%	2%	42%
Comprehensive scholastic literature searches	55%	40%	86%	2%	38%
Access to a colleagues to consult with on interpretation of test results	87%	51%	100%	3%	53%
Genetic counselors available to help you with ordering diagnostic tests and interpreting the results	68%	16%	79%	1%	36%
Case managers available to you to help coordinate ongoing care/care coordination support	55%	14%	66%	0%	30%

Other	9%	9%	21%	0%	7%
I don't know of any resources that are available to me.	1%	36%	0%	1%	8%
Sample size	119	70	29	102	320

Then, all providers were asked, “Do you think you could benefit from additional rare disease education or training?” The responses are summarized in Table 17 below. Very few providers stated they would not benefit from additional training; however, most noted that the benefit would depend on the type of training.

Table 17. Benefit from Additional Training by Type of Provider

Benefit from Additional Training	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Yes	43%	25%	35%	3%	24%
Depends on training	49%	63%	62%	2%	38%
No	8%	5%	3%	0%	5%
I don’t know	0%	0%	0%	0%	2%
Not reported	0%	0%	0%	95%	31%
Sample size	119	70	29	102	320

Similarly, providers were asked to, “Select the areas where you could benefit from more information on rare diseases. “They could check all that apply and were given an “other” option. The responses are summarized below.

Table 18. Benefit from More Information on Rare Diseases

Benefit from More Information	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Diagnosis	83%	74%	55%	0%	52%
Treatment and disease management	72%	83%	66%	1%	51%
Research	34%	21%	62%	0%	23%

Patient and family support services	59%	47%	69%	1%	39%
Other	5%	1%	7%	0%	3%
None	5%	4%	3%	0%	3%
Sample size	119	70	29	102	320

Then, providers were asked to, "Select the information sources below that you prefer to use." Again, they could check all that apply and were given an "other" option. The responses are summarized below.

Table 19. Preferred Information Sources

Preferred Information Sources	Type of Provider				
	Medical	Dental	Rare Disease Specialist	Not Reported	Total
Medical journals	69%	57%	93%	1%	47%
Medical associations	29%	26%	38%	0%	20%
CE courses	75%	89%	76%	1%	54%
Patient associations	24%	17%	34%	0%	16%
Online sources	81%	64%	97%	1%	53%
Other	7%	6%	7%	0%	4%
None	0%	0%	0%	0%	0%
Sample size	119	70	29	102	320

Note: CE = continuing education.

Appendix Item 6: Quarterly Meeting Mission Moment Participants

January- Trevor Turner, individual living with Usher Syndrome and Director of Public Policy for the MN Council on Disabilities

April- Sen. Norm Coleman, father and US Senator who lost a child to a rare disease

July- Kari Olavson, mother who lost a child to a rare disease and founder of Brave Souls Photography

October- Tom Kelly, VP of marketing and innovation at UnitedHealth Group, father of a son living with a rare, genetic condition