

Chloe Barnes Advisory Council on Rare Diseases

Report to the Minnesota Legislature

Calendar Year 2020

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As required by Minnesota Statute 137.68 which went into effect on July 1, 2019.

Submitted by:

Advisory Council Chair, Jakub Tolar, MD, PhD.
Dean of the Medical School
Vice President for Clinical Affairs

Prepared by:

The report was prepared by Erica Barnes, Advisory Council Administrator.

Report Preparation Costs:

Per the requirements set forth in Minnesota Statute 3.197, the cost to prepare this report was \$328.14.

Background

In 2019, the Minnesota State legislature passed, and the governor signed into law, a bill for the creation of the Chloe Barnes Advisory Council on Rare Diseases with oversight provided by the University of Minnesota Board of Regents. The Board of Regents charged the Medical School and Dean Jakub Tolar with the implementation and oversight of the council.

Council members represent the following institutions: ALS Association, Be the Match, Catalyst Medical Clinic, Children’s Minnesota Hospital, Gillette Children’s Hospital, Hennepin County Medical Center, Mayo Clinic, MHeath/Fairview, Minnesota Department of Health, National Foundation for Ectodermal Dysplasia and the Sickle Cell Foundation of Minnesota.

The FDA defines a rare disease as a condition that affects fewer than 200,000 people in the US. There may be as many as 7,000 rare diseases and the total number of Americans living with a rare disease is estimated at between 25-30 million or roughly 8-10% of the population. While each individual community may be small, collectively they represent a significant portion of the population¹. Their etiologies are diverse, but the barriers they face when seeking care are common. Abbey Meyers, founder of the National Organization for Rare Disorders 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 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 diseases². Over the 2020 calendar year, the Council created its strategic plan, articulated under the following pillars:



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

² Council work plan

Results and accomplishments

Work group activities: In 2020 the Council executed on its mission through the creation of four work groups. Throughout the 2020 year, these work groups focused on Pillar I through their particular lenses, following the principle that “if you cannot measure it you cannot improve it”. Through information-gathering strategies focused on deepening the understanding of the rare community *as a population*, the workgroups laid the groundwork for 2021. Below are work group accomplishments for 2020:

Progress through Collaboration:

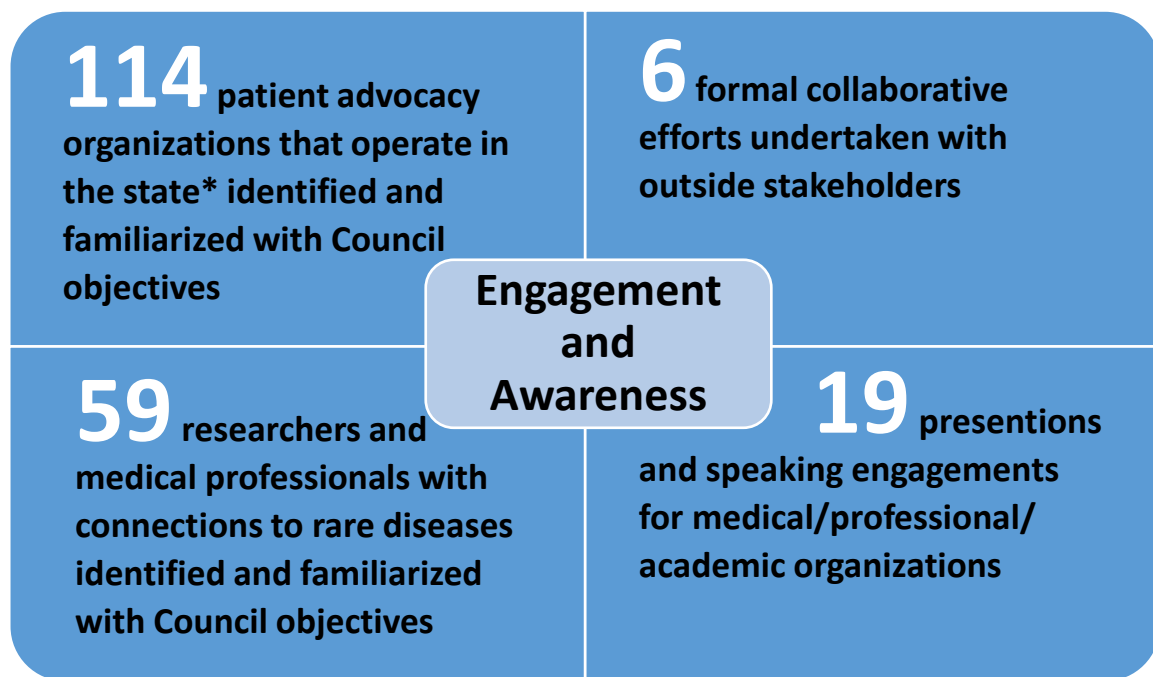
Barriers to Care	Cost	Coordination of Care	Acceleration of Research
<ul style="list-style-type: none"> launched and actively recruiting for state-wide patient survey (see appendix for survey methodology) finalized state-wide provider survey provided input for 3rd place NIH "Rare is not Rare" submission 	<ul style="list-style-type: none"> provided input to barriers work group for patient and provider survey content hosted focus group for researchers and advocates to identify unique costs in research hosted a number of subject matter experts to identify unique costs to rare disease care 	<ul style="list-style-type: none"> provided survey design and content input to barriers work group for patient and provider survey content created contact registry of clinicians able to support transition of care for rare patients engaged with 17 division heads and specialists to discuss improvements to transitioning patients with rare diseases from pediatric to adult care 	<ul style="list-style-type: none"> explored opportunities for collaboration with Sanford Health CoRDS program participated in Sanford Health "CoRDS cast" podcast episode

The Council recognizes that creating collaborations across and outside of the state is vital to improving care for the rare disease populations that are spread out geographically and diffuse throughout every health care system. Over 2020, the Council forged collaborations with many different organizations to accomplish its mission with the following results:

- **St. Cloud State University-** assisted the Council in conducting an IRB approved, state-wide patient survey under the direction of Amanda Hemmesch, PhD, Associate Professor of Psychology to determine unique barriers to care.
- **Oregon State University-** assisted the Council in conducting an IRB approved, state-wide patient survey under the direction of Associate Professor of Psychology Kathleen Bogart, PhD, to determine unique barriers to care. She researches quality of life with rare disorders, has a rare disorder herself and is an advocate.
- **Nighthawk Marketing-** co-created a submission to the NIH/NCATS “Rare is not Rare” challenge which won third place. The materials created for the competition serves as the basis for a future awareness campaign.
- **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 better understand the pandemic’s impact on patients. NORD has used the listening sessions to craft responses to COVID at the national level. Assisting in disseminating the patient survey.
- **BlueSpire Marketing, Engage Health-** collaborated on the creation of state-wide patient survey.
- **College of Pharmacy's Center for Orphan Drug Research and the Medical School's Stem Cell Institute, University of Minnesota-** Council administrator Erica Barnes and Council member Paul Orchard participated on the Center’s eighth annual Rare Disease Day

event planning committee and presented. 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 the event.

Stakeholder Engagement, Outreach, and Awareness: Historically, collective consciousness around rare diseases as a population and both patient communities and researchers addressing rare diseases have been fragmented across health systems and geography. Data and information are often neither centralized nor standardized³. Many rare disease patients have no knowledge of other individuals living with their rare disease and most patient advocacy organizations have small capacity as compared to more common disease communities. For these reasons, awareness and outreach make up a vital aspect of the Council’s work in order to collectively address the community’s needs. In 2020, the Council laid the groundwork for improving care by compiling databases and centralizing resources in addition to increasing awareness in multiple sectors.



*organizations identified through publicly available databases from the MN Attorney General’s office list of registered charities operating in the state of MN, membership list of MN Council of Non-profits

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

1. Comparison table of statute with actions taken in 2020
2. Chloe Barnes Advisory Council work plan
3. Council Member Bios
4. Patient survey methodology

Item 1: CBAC Statute of Actions Comparison in 2020

Legislation text	Work plan/Council activities
<p>Provide advice on research, diagnosis, treatment, and education related to rare diseases.</p>	<ul style="list-style-type: none"> • 15 presentations by Council administrator to physicians, patient groups, educators, state agencies, students, and industry at conferences and Grand Rounds • Publication of national op-ed piece: <i>How Rare Patients Can Overcome the Barriers to Care</i> https://www.futureofpersonalhealth.com/rare-diseases/how-rare-patients-can-overcome-the-barriers-to-care/#
<p>Developing resources or recommendations relating to quality of and access to treatment and services in the state for persons with a rare disease.</p>	<ul style="list-style-type: none"> • Website created • Work plan created • List of all non-profits operating in the state of Minnesota that serve individuals living with a rare disease compiled, organizations contacted* • Physician contact registry of providers across multiple systems with expertise in caring for individuals with rare diseases compiled
<p>Advising, consulting, and cooperating 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.</p>	<ul style="list-style-type: none"> • Engagement with Dr. Nathan Chomilo, DHS state director of Medicaid • Engagement with Mark McCann, Public Health lab manager for newborn screening in the state of MN • Third place award recipient for submission in NIH/NCATS “Rare is not Rare Challenge”
<p>Identifying problems faced by patients with a rare disease when changing health plans, including recommendations on 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.</p>	<ul style="list-style-type: none"> • Launched state-wide patient survey to identify barriers to care in rare patient communities, including issues related to health plans

<p>Identifying best practices to ensure health care providers are adequately informed of the most effective strategies for recognizing and treating rare diseases.</p>	<p>Work plan Pillar 3, concrete goals: Educate primary care providers so that they are adequately informed of the most effective strategies for recognizing and treating rare diseases</p> <ul style="list-style-type: none"> • Grand Rounds presentation • Provider facing survey launched to determine baseline knowledge of rare diseases in the Minnesota medical community
<p>The advisory council shall collect additional topic areas for study and evaluation from the general public.</p>	<ul style="list-style-type: none"> • Tab on website encouraging the public to contact the Council • Log created and available to Council members that lists all public feedback and requests as well as action taken on public requests

Item 2: CBAC Work Plan 2020

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 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.

Best,
Jakub Tolar, Council Chair

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. Deepen 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 when 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 impacts 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

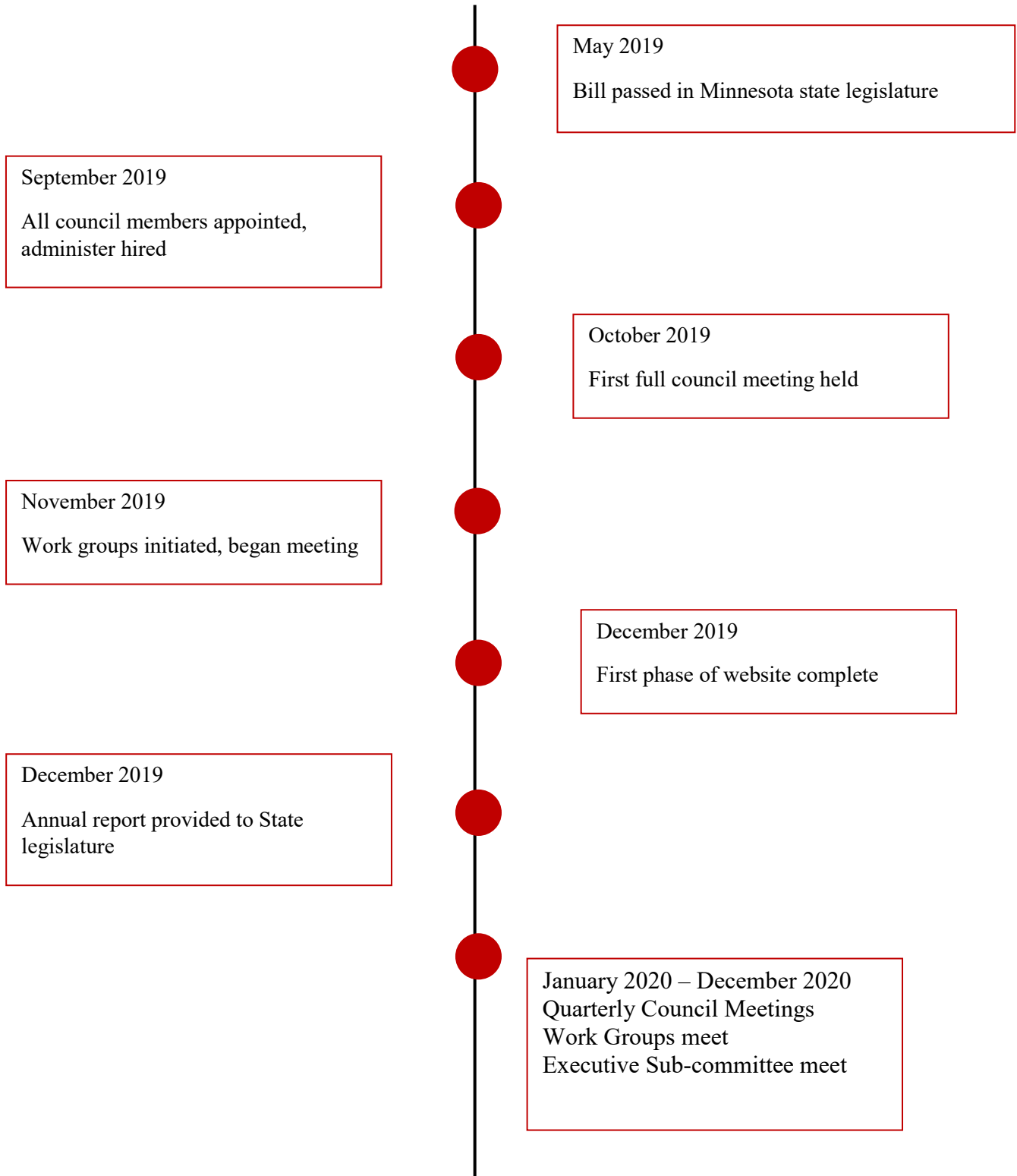
Why is it Important?

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

Council Timeline



Item 3: Council Member Bios

Jakub Tolar, MD - Council Chair



Position:

- Dean of Medical School, Vice President for Clinical Affairs
- Distinguished McKnight Professor, Department of Pediatrics
- Professor, Pediatric Blood and Marrow Transplant (BMT) Center
- Microbiology, Immunology and Cancer Biology (MICaB) Ph.D. Graduate Program
- Pediatric Blood and Marrow Transplant Physician

Education:

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

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 is focused 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, Rare Disease Advisory Council Administrator



Position:

- Rare Disease Advisory Council Administrator/Community Outreach

Education:

- MA in Speech and Language Pathology, University of Minnesota (Twin Cities)
- BA in Humanities, writing intensive, Maranatha Baptist University

Further Information:

Erica Barnes is a certified speech-language pathologist and rare disease advocate. She founded the Chloe Barnes Rare Disease Foundation following the loss of her daughter, Chloe, to metachromatic leukodystrophy. She is state ambassador for the Rare Action Network-NORD and lead the effort to pass the legislation that created the MN Rare Disease Advisory Council

Tim Schacker, MD - University of Minnesota Medical School Ex-Officio Appointee



Positions:

- Vice Dean for Research, Medical School
- Director, Program in HIV Medicine
- Director, Clinical Translational Research Services, Clinical and Translational Science Institute (CTSI)
- Professor of Medicine, Division of Infectious Diseases and International Medicine Infectious Disease Specialist

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:

Timothy Schacker, M.D. is a professor of Medicine and Director of the Program in HIV Medicine at the University of Minnesota. He joined the faculty in 1996. Dr. Schacker received his M.D. from the University of Minnesota in 1986 and completed a residency in Internal Medicine at the Oregon Health Sciences University and Infectious Disease Fellowship at the University of Washington in 1993. He then joined the faculty of the University of Minnesota.

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 to maintain 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 - Mayo Medical School Ex- Officio Appointee



Position:

- Chair, Department of Clinical Genomics, Mayo Clinic

Education:

- Medical School: MD - Albert Einstein College of Medicine
- BA - Johns Hopkins University Fellow-University of Minnesota (Twin Cities)
- Research Fellowship-University of Minnesota (Twin Cities)

Education (cont.):

- 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:

For children and families impacted by rare or undiagnosed genetic conditions, special methods for careful clinical observation and documentation joined with sophisticated genomic and functional techniques can provide an answer to a long diagnostic odyssey. Dr. Schimmenti and members of the Center for Individualized Medicine and the Department of Clinical Genomics are studying ways to improve the approach to diagnosis in patients with rare and undiagnosed diseases.

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



Position:

- Supervisor, Newborn and Child Follow-up Unit, Minnesota Department of Health

Education:

- MSN Nursing

Further Information:

Nicole Brown has worked in Minnesota's Maternal and Child Health/Public Health field for over 20 years at both the state and local level. She is a supervisor within the Children and Youth with Special Health Needs program and along with her team, provides long-term follow-up for children identified with Newborn Screening Conditions.

Rep. Alice Mann, MD - Legislative Appointee



Position:

- MN legislator, physician practicing family and emergency medicine

Education:

- Medical School: Johns Hopkins University- Master of Public Health,
- Meharry Medical College - Doctor of Medicine.
- Residency - Mayo Clinic Health System in La Crosse, Wisconsin

Further Information:

In addition to serving in the state legislature Dr. Alice Mann is a family medicine doctor in Lakeville, Minnesota and is affiliated with multiple hospitals in the area, including Linton Hospital and Northfield Hospital and Clinics.

Rep. Tony Albright - Legislative Appointee



Position:

- MN legislator, Assistant Minority Leader

Education:

- B.S., Business Administration, Minnesota State University, Moorhead

Further Information:

Rep. Albright has spent 22 years in the Financial Services/Investment

Sen Matt Klein, MD - Legislative Appointee



Position:

- MN legislator, doctor of internal medicine at HCMC

Education:

- B.S., University of Wisconsin, Madison
- M.D., Mayo Medical School residency and chief residency in internal medicine at Hennepin County Medical Center

Sen. Scott Jensen, MD - Legislative Appointee



Positions:

- MN legislator, President-Catalyst Medical Clinic,
- Clinical Associate Professor - University of MN Family Practice Dept.
- Pro Rehab Medical Director,
- Staff Physician – Ridgeview Medical Center

Education:

- B.A., University of Minnesota
- M.D., University of Minnesota

Karl Nelsen, PA-C MS - Patient Advocate Appointee



Positions:

- PA at Fairview Southdale Hospital
- Fairview Ridges Hospital, Patient advocate for Ectodermal Dysplasia

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's a physician assistant in interventional radiology at a large community-based hospital system.

Rae Blaylark - Patient Advocate Appointee



Position:

- Founder & Executive Director - Sickle Cell Foundation of MN
- Sickle cell caregiver

Further Information:

Ms. Blaylark has served the sickle cell community since 2004 advocating for equitable resources, quality of care, and patient rights. She has served as Sickle Cell Services Coordinator of Children's Hospital of Minnesota and in Training & Education, Donor Recruitment, and Community Engagement for Memorial Blood Centers' Sickle Cell Program. Her fight is personal. Rae's 22 year old son battles sickle cell disease.

Karri LaFond - Patient Advocate Appointee



Position:

- ALS caregiver

Education: N/A

Further Information:

Karri's husband, Michael, lived with ALS for almost two years before passing away in February 2019. Throughout his journey, Karri was a strong advocate for him. Karri believes that her lived experience and the barriers she encountered provides her with vital insights that can promote positive improvements for future families dealing with rare diseases.

Arthur Beisang, MD - Physician Appointee



Position:

- 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.

Sheldon Berkowitz, MD, FAAP - Physician Appointee



Position:

- Clinical Medical Director & Physician Advisor 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 25 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 - Physician Appointee



Position:

- Pediatrician, Hematology and Oncology, 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:

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 and she has a particular interest/expertise in the care of children with pleuropulmonary blastoma, ovarian tumors and other rare childhood cancer.

Kerry Hansen, RN - Nurse Appointee



Position:

- Nurse coordinator, Fairview

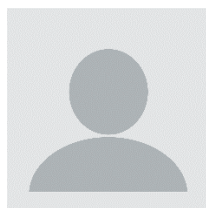
Education:

- 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 - Pharmacist Appointee



Position:

- Consultant for Fairview Pharmacy Services, LLC managing Pharma sponsored outcomes studies and projects

Education:

- BS in Pharmacy, MS in Hospital Pharmacy - University of Minnesota

Further Information:

Mr. Blissenbach is currently a Consultant for Fairview Pharmacy Services, LLC (FPS) managing Pharma sponsored outcomes studies and projects. He developed and led numerous ambulatory pharmacy business units at FPS since 2000. Most recently Mr. Blissenbach led clinical trials in drug development as well as providing direction for a provider led organization that provides diagnosis, care and treatment for children and adults with Lysosomal diseases. Mr. Blissenbach has more than 50 years of experience in hospital and alternate site pharmacy. He was a pioneer in the growth of Home Infusion Services and the evolution of Specialty Pharmacy. His work history includes positions involving multiple-site pharmacy management, sales and marketing, business development, research and treatments for rare diseases. Mr. Blissenbach received his BS in Pharmacy and MS in Hospital Pharmacy from the University of Minnesota. Throughout his career he has consistently been a leader and innovator in the practice and business of pharmacy. Previous employers include HMSS, Caremark, Chronimed and Fairview.

Soraya Beiraghi, DDS, MSD, MS, MSD - Dentist Appointee



Positions:

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

Education: N/A

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 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.

Barbara Joers - Hospital Administrator Appointee



Position:

- CEO, Gillette Children's

Education:

- BA, Hospital Administration - Marymount University in Arlington, Virginia
- MA, Health Services Management and Policy - 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.

Srijoy Mahapatra, MD - Hospital Administrator Appointee



Position:

- Chief Clinical Innovation Officer, M Health

Education:

- B.S., Electrical Engineering, Master of Business Administration, and Certification in Artificial Intelligence - Massachusetts Institute of Technology
- Doctor of Medicine (MD) - Tufts University

Further Information:

Dr. Mahapatra leads system innovation efforts to create and advance clinical programs to help patients live longer, healthier lives. Prior to Fairview, he held several roles at Abbott Labs and St. Jude Medical, where he led worldwide clinical trials in cardiology, structural heart, and electrophysiology. Dr. Mahapatra was an Assistant Professor of Medicine and Biomedical Engineering at the University of Virginia as well as Head Team Cardiologist for NCAA student-athletes. He continues to see patients as a consulting cardiologist to the National Basketball Players Association, NFL, and NHL. He has been involved in several startup companies, holds 11 patents, and has had his work appear in more than 150 publications. Dr. Mahapatra completed

a Cardiology & Electrophysiology fellowship at the Mayo Clinic and advanced training in complex ablation at Haut Levaque in Bordeaux, France, and INCOR in Sao Paulo, Brazil.

Janet Ziegler, MSW, LICSW - Social Work Appointee



Position:

- Clinical Social Worker – Pediatric Blood & Marrow Transplant Program

Education:

- University of St. Thomas/College of St. Catherine (Masters of Social Work) Board Certifications
- MN Board of Social Work - Licensed Independent Clinical Social Worker (LICSW)

Further Information:

Ms. Ziegler has both professional and personal/family experiences with rare diseases and the challenges involved with diagnosis, treatment, and accessing resources. Ms. Ziegler has spent the past 31+ years working with individuals and families dealing with serious medical and mental health conditions. Her interest in rare diseases is related to her subsequent work with patients and families receiving care through the Adult and Pediatric Blood & Marrow Transplant Center at the University of Minnesota Medical Center/ University of Minnesota Masonic Children's Hospital over the past 20+ years. Recognizing that rare illnesses can impact every realm of a person's life Ms. Ziegler's work has focused on addressing the psychosocial needs of patients and families. She provides counseling, education, advocacy and community resource coordination to patients and their caregivers. In addition she is involved with: education & mentoring of other health care team professionals; process improvement projects; working with professional organizations around best practices for social workers & advocating for patient needs; development of patient/family educational materials; and collaboration with community resource organizations.

Ms. Ziegler is passionate about learning from, working with, and advocating for the needs of families living with rare diseases. She recognizes that rare diseases impact every member of society and every community; therefore, it is critical to address related concerns on both the individual and macro levels.

Amy Gaviglio, MS-CGC - Genetic Counselor Appointee



Position:

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

Education:

- Master's - University of Michigan

Further Information:

Amy Gaviglio is a certified genetic counselor and has been employed by the Minnesota Department of Health, Newborn Screening Program for the past 12 years. Amy oversaw follow-up for the program and provided guidance for informatics, education, ethical, and policy related initiatives. She is currently co-chair of APHL's New Disorder workgroup, and is a member of APHL's Short Term Follow-Up and Legal and Legislative Issues in Newborn Screening workgroups. She also serves as vice-chair of the NBS Expert Panel for the Clinical and Laboratory Standards Institute, is on APHL's Newborn Screening and Genetics in Public Health Committee, and the Advisory Committee on Heritable Disorders in Newborns and Children's Education and Training and Ad Hoc workgroups. She is now an employee of.

Lee A. Jones - Biotech Industry Appointee



Position:

- 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 - Payer Appointee



Position:

- 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 - Researcher Appointee



Position:

- Professor of Pediatrics - Division of Blood and Marrow Transplantation
- Medical Director - Inherited Metabolic and Storage Disease Bone Marrow Transplantation Program
- BMT Specialist, Pediatric Blood and Marrow Transplant (BMT) Center
- Director, Mucopolysaccharidosis (MPS) Center
- Team Member, Leukodystrophy Center

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 regard 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 in regard to 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 - Non-Profit Organization Appointee



Position:

- Manager, Jason Carter Clinical Trials Program and Education
- Patient Advocacy and Navigation, Be the Match

Education:

- BA, Nursing, St. Olaf College
- MPH, Community Health Education - University of MN, Twin Cities

Further Information:

Ms. Foster has numerous publications addressing strategies and tools to increase patient engagement and increase health literacy in-bone marrow transplant patients

Item 4: Patient Survey Methodology

Minnesota Rare Disease Health Care Access Study

This study is a collaboration between Amanda Hemmesch, Ph.D., at St. Cloud State University, Kathleen Bogart, Ph.D., at Oregon State University, and the Chloe Barnes Rare Disease Advisory Council hosted by the University of Minnesota Medical School. We designed an online survey to collect information from adults with rare conditions and parents of children with rare conditions. The questions in the survey were primarily selected from previously published research investigating rare disease. This study was approved by the St. Cloud State University and the University of Minnesota institutional review boards and is currently under review at Oregon State University.

Participants

We began recruiting for this study on October 2, 2020. Participants are being recruited through diverse channels to try to reach as many participants or parents with rare diagnoses as possible, including support groups and online communities for rare conditions, doctors and other health care providers, hospital systems' media departments, and through word of mouth. The team has identified wide range of patient advocacy organizations across the highly heterogeneous rare disease patient populations to ensure that diverse perspectives are being captured. Our goal is to recruit approximately 500 participants from Minnesota, as well as a comparison sample from across the United States.

Measures

Patient-Reported Outcomes Measurement Information System (PROMIS) Profile-29 (Adult and Parent Proxy). PROMIS-29 assesses seven health domains (physical function, fatigue, pain interference, depression, anxiety, ability to participate in social roles and activities, and sleep disturbance) using four items per domain, and pain intensity using a single item (Cella et al., 2019). PROMIS is a National Institutes of Health (NIH) initiative with excellent validity and reliability (Cella et al., 2019). Scales are scored using published T scores calibrated such that a mean of 50 and standard deviation of 10 is representative of the U.S. general population.

Patient Satisfaction Questionnaire Short Form. This 18 item questionnaire assesses access to and satisfaction with healthcare generally (Marshall & Hays, 1994). It yields the following seven subscales: general satisfaction, technical quality, interpersonal manner, communication, financial aspects, time spent with doctor, and accessibility. It shows good reliability and validity (Marshall & Hays, 1994).

Chronic Illness Anticipated Stigma Scale Healthcare subscale. This 4-item scale examines the extent to which participants expect that they will experience stigma or discrimination in a healthcare situation. It has been validated in a chronic illness population and shows good reliability (Earnshaw et al., 2013).

Stigma scale for Chronic Illness (SSCI). The eight-item SSCI is a unidimensional measure of enacted and internalized stigma validated for use in a variety of chronic illnesses (Molina et al., 2013). This scale is part of the Neuro-QOL NIH toolkit. Following Neuro-QOL scoring guidelines, domains are scored using the published T scores calibrated such that a mean of 50 and standard deviation of 10 is representative of a clinical neurologic sample norm.

Questions concerning the nature of the rare disease; length of time to diagnosis; access to insurance, care coordination, and specialist centers; barriers and facilitators to access; and demographics were developed by the research team or adapted from Molster et al., (2016).

Procedure

Participants received a paper or electronic flyer inviting them to participate in this study and follow a link to the online survey. The survey is hosted on the Qualtrics platform, which is password-protected and encrypted. The first page of the online survey is a consent form explaining the study; participants are asked to click ‘I agree to participate in the study’ to indicate consent. After that, participants are asked to answer questions about their rare diagnosis and general physical and mental health status, access to health care, and questions about stigma. Interested participants who do not want to complete the survey online can request a paper copy (including a large print version) to be mailed to them. Participation in this study is estimated to take 20-30 minutes.

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