



# ANNUAL REPORT FFY 2022

Summary of activities undertaken by the Council for FFY 2022  
Created December 2022

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*Envisioning a world where every Minnesota citizen living with  
a rare disease has access to a timely diagnosis, comprehensive care,  
and an effective treatment.*

## Table of Contents

<b>BACKGROUND .....</b>	<b>2</b>
<i>The Community Served .....</i>	<i>2</i>
<i>Council Description.....</i>	<i>2</i>
<b>COUNCIL MEMBERSHIP 2022.....</b>	<b>4</b>
<b>COUNCIL ACTIVITIES AND ACCOMPLISHMENTS .....</b>	<b>5</b>
<i>Transition to the State .....</i>	<i>5</i>
<i>Collaborations with Stakeholders .....</i>	<i>5</i>
Transition of Care Roundtable .....	5
Project ECHO, Minnesota Medical Association .....	6
RARE: Stories of DIS-EASE Theater Production.....	6
Students for Rare Club, University of Minnesota Chapter .....	7
University of Minnesota GCC1907 Undergraduate Grand Challenge Course.....	7
<b>EDUCATION AND AWARENESS .....</b>	<b>8</b>
<b>PUBLICATIONS/EDUCATIONAL RESOURCE DEVELOPMENT .....</b>	<b>10</b>
<b>APPENDIX A — Provider Survey Infographic.....</b>	<b>11</b>
<b>APPENDIX B — Patient Survey Infographic .....</b>	<b>12</b>

## **BACKGROUND**

### ***The Community Served***

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 population. Their etiologies are diverse, but each community's barriers 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".

### ***Council Description***

The Minnesota Rare Disease Advisory Council is a newly established state agency that advocates for the one in ten Minnesotans affected by rare diseases. The Council was legislatively created in 2019 as a result of grassroots efforts by Minnesota patients and families whose lives have been affected by rare diseases and initially housed at the University of Minnesota Medical School under the governance of the Board of Regents. In July 2022, the Legislature transitioned the Council to the State of Minnesota. The Council operates on a \$150,000 annual appropriation set to expire at the end of the current fiscal year.

The Council's mission is to provide advice on research, diagnosis, treatment, and education related to rare diseases. Its goals are:

- ***Be a comprehensive policy and information resource in the state of Minnesota for all stakeholders that have engagement with the rare disease community.*** The Council builds coalitions and engages in collaborative activities that deepen our understanding of the barriers to care individuals with rare diseases encounter. The Council then seeks to shape policy through communicating the unique healthcare disparities faced by the rare disease community and solutions to address these disparities to policymakers, state agencies, the medical community, and the public.
- ***Support and empower the rare disease patient community to advocate for improved quality of life.*** The rare disease community is a fragmented patient population in a fragmented healthcare system. The Council seeks to increase collaboration 1.) across disease-specific patient populations to increase resource sharing and reduce duplication of efforts and 2.) between the rare disease community and researchers, clinicians, and industry to accelerate research and promote clinical trial readiness. The Council executes this by promoting Rare Disease Day events and activities across all rare disease communities as well as being a hub of information and resources related to rare disease care.
- ***Support and equip the medical community to better address the unique needs of the rare disease community to reduce health disparities.*** Knowledge and information related to rare disease diagnosis and treatment can be difficult to find. The Council seeks to make education and resources available to clinicians that identify and promote best practices and increase the ease of getting individuals with rare diseases access to appropriate care. In addition, the Council seeks to influence the next generation of medical professionals.

## COUNCIL MEMBERSHIP in 2022

### MEMBERS

**David Tilstra, MD** | Hospital Administrator (Greater Minnesota)  
**Abigail Miller, MD** | Health Plan  
**Amy Gaviglio, MS CGC** | Genetic Counselor  
**Angela Cowen, LCSW** | Social Worker  
**Art Beisang, MD** | Physician (Pediatrics)  
**Barbara Joers** | Hospital Administrator  
**Dawn Stenstrom** | Industry  
**Jackie Foster, MPH, RN, OCN** | Rare Disease Non-profit  
**Karl Nelsen, PA-C** | Rare Disease Patient/Caregiver  
**Kerry Hansen, RN** | Nurse  
**Kris Ann Schultz, MD** | Physician  
**Maureen Alderman** | Rare Disease Patient/Caregiver  
**Paul Orchard, MD** | Researcher  
**Rae Blaylark** | Rare Disease Patient/Caregiver  
**Sheldon Berkowitz, MD** | Physician  
**Soraya Beiraghi, DDS, MSD, MS, MSD** | Dentist  
**Tom Blissenbach** | Pharmacist

### LEGISLATIVE APPOINTEES (VOTING)

**Sen Julia Coleman** | Legislator  
**Sen Matt Klein, MD** | Legislator  
**Rep Kelly Morrison, MD** | Legislator  
**Rep Tony Albright** | Legislator

### EX OFFICIO (NON-VOTING)

**Nicole Brown, RN** | Commissioner of Health designee  
**Tim Schacker, MD** | University of Minnesota Medical School  
**Lisa Schimmenti, MD** | Mayo Clinic

## COUNCIL ACTIVITIES AND ACCOMPLISHMENTS

### *Transition to the State*

In July 2022, the Minnesota Rare Disease Advisory Council became a small state agency. Prior to the establishment of the Council as a state agency, it was housed at the University of Minnesota Medical School under the direction of the Board of Regents.

To fulfill the original intent of legislation that the Council would engage on policy and advise the state legislature, the Council transitioned out of the University. Since July, and with the support of the Minnesota Council on Disability, the Council has carried out numerous activities to establish operations and implement processes that ensure compliance with all state requirements that state agencies are subject to.

### *Collaborations with Stakeholders*

The Council believes that collaboration and multi-stakeholder engagement is crucial to addressing the complex issues that the rare disease community faces. The rare disease community is fragmented across health systems and solutions require the input of many perspectives. Therefore, whenever possible, the Council seeks to engage other organizations. In 2022, the Council engaged in the following five formal collaborative projects.

### **Transition of Care Roundtable**

Transitioning children with complex medical needs from pediatric to adult care is often an informal, complicated process that places stress and burden on the patient, families, and clinicians. For this reason, the Council, in partnership with Gillette Children's Specialty Healthcare, the Minnesota Chapter of the American Academy of Pediatrics, and the Minnesota Department of Health, convened a roundtable of cross-sector leaders to generate solutions designed to break down these barriers. The results of the roundtable and subsequent listening sessions will be summarized in a position paper in 2023.

#### **Collaborative partners:**

Gillette Children's Specialty Healthcare

Minnesota Chapter of the American Academy of Pediatrics

Minnesota Department of Health

#### **Further information:**

[Pediatric to Adult Healthcare Transition Roundtable -June 14, 2022 - YouTube](#)

## **Project ECHO, Minnesota Medical Association**

After completing the 2021 frontline provider survey, the Council identified several clinician-reported barriers in providing care for individuals with rare diseases. A significant reported barrier is the knowledge gap between specialists and primary care clinicians on the complex needs of the rare disease community. Throughout 2022 and continuing into 2023, the Council is collaborating with the Minnesota Medical Association to provide telementoring to the medical community on issues related to transition of care. To date, more than 80 clinicians have received training and education through Project ECHO.

### **Collaborative partners:**

Minnesota Medical Association

PACER Center

Family Voices of Minnesota

Gillette Children's Specialty Healthcare

### **Further information:**

[Minnesota Medical Association - Project ECHO \(mnmed.org\)](https://mnmed.org)

## **RARE: Stories of DIS-EASE Theater Production**

In collaboration with The University of Minnesota's Center for Orphan Drug Research and the Department of Theater Arts and Dance, the Council participated in the creation of a theater production. The goal of the production was to bring greater awareness and a deeper understanding of the medical, economic, psychological, and social challenges facing people with rare diseases. In addition, the Council organized and oversaw an interactive panel discussion following each performance. The play had eight performances in Minnesota, Wisconsin, and North Dakota communities, with a total of over 625 attendees.

### **Collaborative partners:**

Sod House Theater

University of Minnesota's Center for Orphan Drug Research

University of Minnesota's Department of Theater Arts and Dance

### **Further information:**

[RARE: Stories of Dis-Ease Full Length Documentary - YouTube](https://www.youtube.com/watch?v=...)

## **Students for Rare Club, University of Minnesota Chapter**

The Council serves as an advisor to the student club whose goal is to spread awareness and recognition of rare diseases in the academic setting, establish professional relationships and partnerships and encourage students to pursue future opportunities within the rare disease space.

### **Collaborative Partners:**

National Organization for Rare Disorders

Reena Kartha, PhD (co-advisor)

### **Further information:**

[Students For Rare at UMN \(@nordsfr.umn\) • Instagram photos and videos](#)

## **University of Minnesota GCC1907 Undergraduate Grand Challenge Course**

The Council worked with Dr. Reena Kartha at the University of Minnesota to develop a curriculum for a Freshman Seminar "Rare Diseases: What it Takes to Be a Medical Orphan." Grand Challenge Freshman Seminars engage first-year students in complex issues facing the world and introduce the importance of interdisciplinary approaches. In addition, the Council worked with a student group to create a project related to rare disease awareness.

### **Collaborative partner:**

*Reena Kartha, PhD*

### **Further information:**

Video 1:

<https://www.tiktok.com/t/ZTRVuMfkG/>

Video 2:

<https://www.tiktok.com/t/ZTRVu2GVk/>

Video 3:

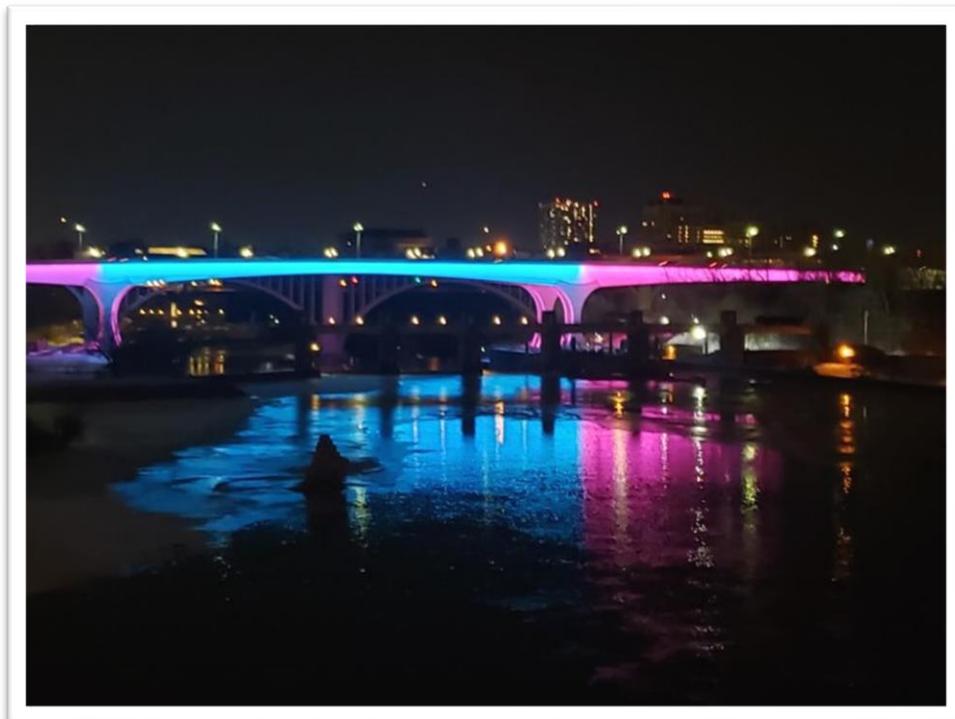
<https://www.tiktok.com/t/ZTRVu2vkN/>

## EDUCATION AND CONSULTATION

The Council provided education to a number of stakeholders by participating in or facilitating a total of 17 educational and awareness opportunities, both within the state of Minnesota as well as nationally in 2022. In addition, the Council engaged with various state agencies to provide advice and consultation for programming related to rare diseases.



MNRDAC created awareness with the "Light Up for Rare" campaign that NORD launches nationally. Using the amazing student group, the Council had 4 different Minneapolis landmarks lit up in rare





RARE DISEASE ADVISORY COUNCIL

disease colors of blue, green, pink and purple. The campaign received coverage on traditional news outlets and social media.

## **PUBLICATIONS/EDUCATIONAL RESOURCE DEVELOPMENT**

A stated duty of the Rare Disease Advisory Council is to provide advice on education related to rare diseases. With this in mind, the Council undertook the following activities:

***Publication reporting the results of a 2021 survey of the Minnesota medical community.*** The goal of the survey was to identify the level of knowledge and awareness the Minnesota medical community currently has as well as the method of education about rare diseases that will be most preferred. A publication analyzing the implications of dental care is pending.

### ***Infographics***

The Council created a series of publicly available infographics (see appendix) with the goal of providing resources for the medical community and general public.

### **For more information, please contact:**

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For accessible formats of this information or assistance with additional equal access to human services, write to [DHS.info@state.mn.us](mailto:DHS.info@state.mn.us), call 651-431-2504, or use your preferred relay service. ADA1 (2-18)

APPENDIX A — Provider Survey Infographic



## Rare Disease Frontline Providers Address Patients' Barriers to Care



SPECIALISTS PROVIDERS DENTISTS

There are over 7,000 rare diseases in existence with over 25 million patients affected. Yet, healthcare access and quality of life are elusive to many rare disease patients, and only 5% of rare disease patients have an approved treatment option. Even when compared to adults with common chronic diseases, adults in Minnesota with rare diseases had significantly worse stigma, physical function, fatigue, and depression, and marginally worse anxiety.

The *Frontline Provider Survey* was conducted to gain a healthcare professional's perspective on challenges to addressing the needs of the rare disease community. For more information on this study and the council, please scan the QR code or visit [www.cbacraredisease.org](http://www.cbacraredisease.org)



Nearly a quarter of providers do not think their knowledge of rare diseases meets the needs of their practice.



Over **75%** of clinicians are somewhat or not at all comfortable interpreting genetic testing results.



**63%** Receiving care in state say that they never or very rarely have to refer difficult to diagnose rare disease patients out of state.



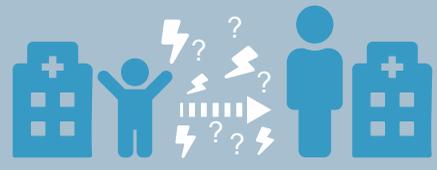
Rare disease specialists are mainly in the Twin Cities metro area. There are a limited number of specialists to treat rare disease outside the metro area.



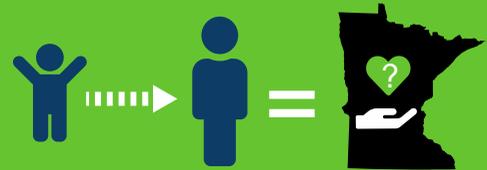
**28%** report that many times the type of insurance the patient carries has affected their ability to complete the recommended treatment plan.



**60%** of providers and dentists identify transitioning care from pediatrics to adult as difficult.



**56% and 60%** of medical and dental providers who reported having difficulty transitioning their patients reported it was due to a local practitioner being unwilling to take the patient.



Over 90% of providers felt (depending on the type of training) they would benefit from additional training.

**90%**



**30%** of medical and dental providers did not feel that they had the ability to care for rare disease patients who have contacted them.

77% because the disease was too complex and lesser numbers reported time constraints, out of the scope, and/or ability of their practice.



APPENDIX B — Patient Survey Infographic



## Healthcare access in rare disease: A PUBLIC HEALTH PRIORITY!

There are over 7,000 rare diseases in existence with over 25 million patients affected. Yet, healthcare access and quality of life are elusive to many rare disease patients, and only 5% of rare disease patients have an approved treatment option. Even when compared to adults with common chronic diseases, adults in Minnesota with rare diseases had significantly worse stigma, physical function, fatigue, and depression, and marginally worse anxiety.

The 2020 Rare Diseases Health Care Access Study set out to learn more about rare disease patients' barriers to care. For more information on this study and the council, please scan the QR code or visit [www.cbacraredisease.org](http://www.cbacraredisease.org)



### LOCAL CARE? ONLY FOR SOME.

Participants living in outstate Minnesota were more likely to report traveling over 60 miles for rare disease care.

47% of participants living in outstate Minnesota reported traveling over 60 miles for rare disease care.

Rare disease care.....60 miles  
Chronic disease care.....3 miles

### DELAYED TIME TO DIAGNOSIS

56% of rare disease patients waited over one year and up to ten years to receive a diagnosis.

### 37% of patients saw 4-10 PROVIDERS prior to receiving a diagnosis.

### 44% of participants reported problems with transition from pediatric to adult care.

### INVESTIGATIONAL DRUG ACCESS

72% MN adult respondents  
79% MN pediatric respondents reported delays or denials for use of an investigational treatment.

### CARE COORDINATORS: EQUALLY RARE

12% of rare disease patients surveyed have a care coordinator.

### TELEHEALTH'S USEFULNESS

58% of all participants reported using telehealth visits for specialist care appointments.

### PERCEPTIONS OF PROVIDER KNOWLEDGE

40% rated the initial provider as "poor" on their knowledge of rare diseases.

### INSUFFICIENT PATIENT EDUCATION FROM PROVIDERS

21% GENERAL PRACTITIONERS  
15% SPECIALISTS did not provide sufficient information.

### OUT OF NETWORK ACCESS

27% pediatric  
19% adult  
Respondents reported being initially denied seeing a specialist due to that specialist being out of network.

[www.cbacraredisease.org](http://www.cbacraredisease.org) © 2022 Minnesota Rare Disease Advisory Council