MINNESOTA
RARE DISEASE ADVISORY COUNCIL

Rare is not rare.

MINNESOTA RARE DISEASE ADVISORY COUNCIL POLICY FOCUS GUIDE

2024

WHO IS THE RARE DISEASE COMMUNITY?

The FDA defines a rare disease as a condition that affects fewer than 200,000 people in the US. There are over 7,000 rare diseases, and an estimated 25-30 million Americans living with a rare disease. While each individual community may be small, collectively, they represent 8-10% of the population. Despite this large collective number, individuals with rare diseases encounter numerous inequities when seeking care in our health system, such as:

- Significant delay in receiving a diagnosis (7-8 years)
- Significant number of visits before diagnosis (8 different clinicians)
- Numerous misdiagnosis (2-3)
- Lack of effective treatment options (only 5% of rare diseases have an FDA-approved treatment)

RARE DISEASE PATIENT SURVEY INFOGRAPHIC. Click *here* to see it.

THE MINNESOTA RARE DISEASE ADVISORY COUNCIL BACKGROUND

In 2019, recognizing that the current structures of the health system are inequitable for individuals with rare diseases, a grassroots group of patient advocates called on the state of Minnesota to pass legislation creating a Rare Disease Advisory Council. The bill passed unanimously, and 42 organizations across patient advocacy groups, hospital systems, and industry endorsed it. Effective July 2022, the Council transitioned to an executive branch state agency.

In the 2023 legislative session, the Council successfully supported legislation to increase network access to clinicians with knowledge of rare diseases and to increase rare disease community representation on various committees within the state.

VISION AND MISSION

The Council envisions a world where every Minnesotan with a rare disease has access to a timely diagnosis, comprehensive care, and effective treatment. It seeks to be a comprehensive policy resource for legislators, other state agencies, and all stakeholders who shape policy in Minnesota. Its mission is to provide advice on research, diagnosis, treatment, and education related to rare diseases.

WHERE WE ARE GOING

As a newly formed state agency, the Minnesota Rare Disease Advisory Council will continue to be a voice for the rare disease community. We will focus on the following pillars to ensure that the state of Minnesota takes a comprehensive approach to improving care for the rare disease community.

DEEPER UNDERSTANDING OF THE COLLECTIVE BARRIERS ACROSS DIAGNOSIS-SPECIFIC RARE DISEASE COMMUNITIES

The adage "if you can't measure it, you can't improve it" is especially true for the rare disease community. For many rare diseases, basic data such as disease prevalence and the natural history of the disease (what happens to an individual with a disease without any intervention) are non-existent. This lack of data makes developing standards of care and effective treatment options difficult. The Council supports increasing programs that increase our knowledge of rare diseases. Surveillance studies such as the Sickle Cell Data Collection (SCDC) program conducted by the Minnesota Department of Health are a key example. See it *here*.

REDUCED TIME TO DIAGNOSIS

The journey of rare disease patients and families to obtain a diagnosis has been coined the "diagnostic odyssey" due to the long and challenging process of getting a diagnosis. There are many contributing factors, and the Council supports policies that fosters early diagnosis. Some examples are:

- Expanded newborn screening
- Increased access to genetic testing (roughly 80% of all rare diseases have a genetic component)
- Better access to specialists with expertise in care for rare diseases

INCREASED COORDINATION OF CARE

The rare disease community represents a fragmented patient population in an already fragmented healthcare system. The Council supports changes to reduce the burden on patients and families to coordinate their own care:

- Improvement in transition of care from pediatrics to adult care settings
- Reduction in geographical barriers to care through expanded telehealth.

ACCELERATION OF RESEARCH / INCREASED ACCESS TO EFFECTIVE TREATMENTS

While the rare disease community continues to have a limited number of FDA-approved treatments, in recent years, rare disease research and drug development have accelerated at an unprecedented pace, leading to breakthroughs such as gene therapies that were unknown a generation ago. The Council supports efforts to increase the number of available treatments and recognizes that availability does not mean access. To address the gap between efforts to increase available treatments and accessible treatments, the Council supports the following policy areas:

- Value-based arrangements for rare disease treatments when appropriate
- Reduction in cost-sharing burdens for patients
- Expanded coverage for medically necessary treatments and procedures
- Reduction in burdensome prior authorization requirements not designed for the rare disease community

WHAT YOU CAN DO

Become a community champion.

Many medical professionals recall being told, *"When you hear hoof beats, look for horses and not zebras,"* a warning to focus on the likelihood that most people will be diagnosed with a common disease and not a rare one. The Council believes that the healthcare system should accurately diagnose and treat all patients, so we proudly use zebra stripes to symbolize our community. As you consider policies, we encourage you to determine their impact on the rare disease community.

Contact the agency.

The Council is a resource for rare disease technical expertise, collaboration, and briefings. We welcome the opportunity to work with policymakers and all stakeholders as they craft healthcare policy and legislation.

> *"Families affected by rare diseases represent a medically disenfranchised population that falls through the cracks of every healthcare system in the world."*
>
> -Abbey Meyers,
> Founder of the National Organization for Rare Disorders (NORD)