

Rare Disease Health Care Access Study

2020 RESULTS



ABOUT THE COUNCIL

In 2019 the rare disease community passed bill SF973 calling for the creation of a Rare Disease Advisory Council.

The bill passed unanimously and was endorsed by 42 organizations across patient advocacy groups, hospital systems, industry, and medical associations...

the [Chloe Barnes Advisory Council on Rare Diseases](#) was formed.



THE WHY

The Chloe Barnes Advisory Council on Rare Diseases has been 'diagnosing' the rare disease patients' barriers to care in Minnesota since 2019.

Made up of 24 members that span five health care institutions and multiple patient groups, the vision of the Council is a society where every patient with a rare disease has access to a timely diagnosis, appropriate care, and effective treatment.

Recognizing persistent barriers to health care for the rare disease community, the council needed **health care access research to effectively change the trajectory of rare disease patients' diagnosis odyssey.**

Rare Disease Frontline Provider Survey

KEY TAKEAWAYS



LIMITED
SPECIALTY
KNOWLEDGE

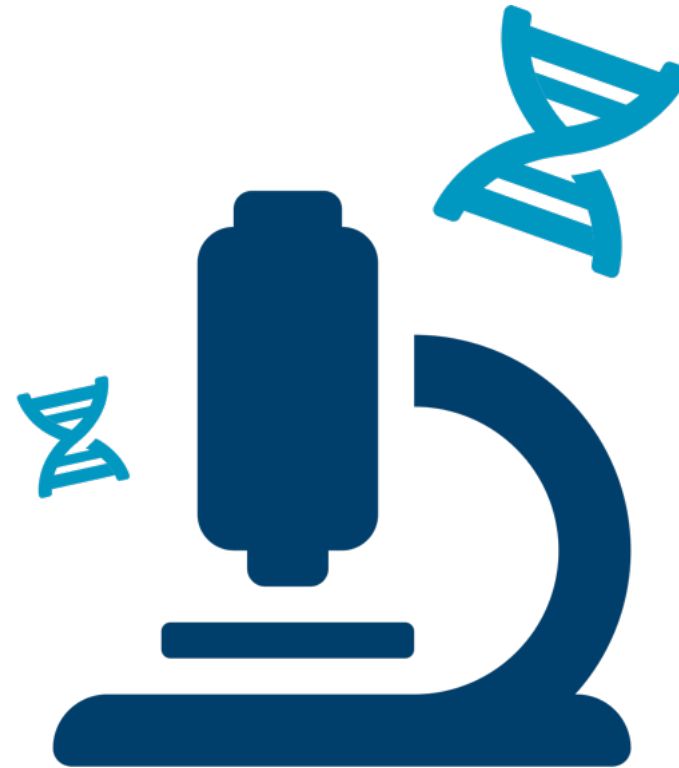
Nearly a quarter of providers do not think
their knowledge of rare diseases meets the needs
of their practice.



LIMITED
GENETIC TESTING
INTERPRETATION
SKILLS

Over **75%**

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



OUT-OF-STATE
REFERRALS



63%

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

LIMITED
LOCAL
CARE

**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.



INSURANCE BARRIERS



28%

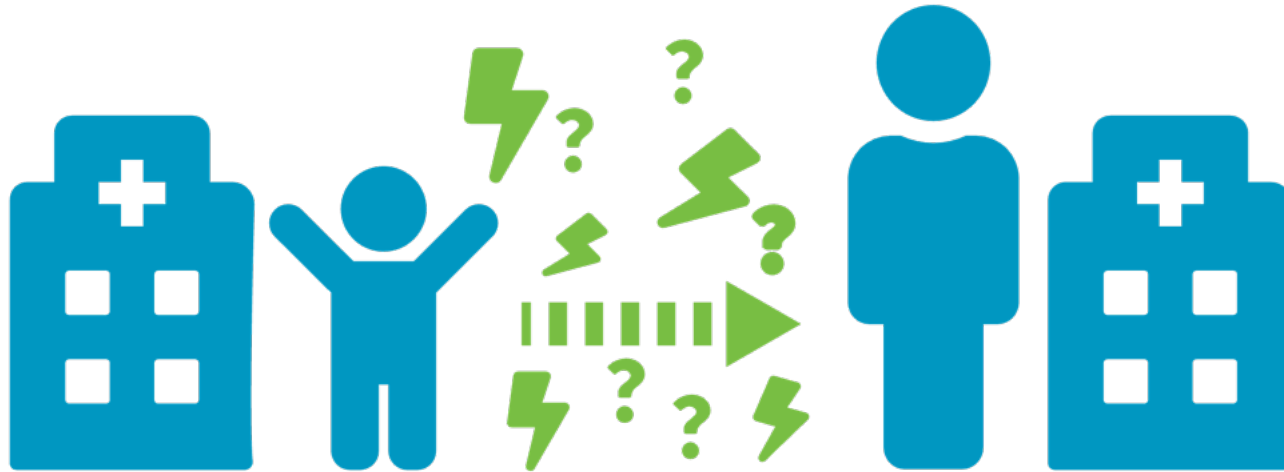


report that **many times**
the **type of insurance**
the patients carries has
affected their ability to
complete the recommended
treatment plan.



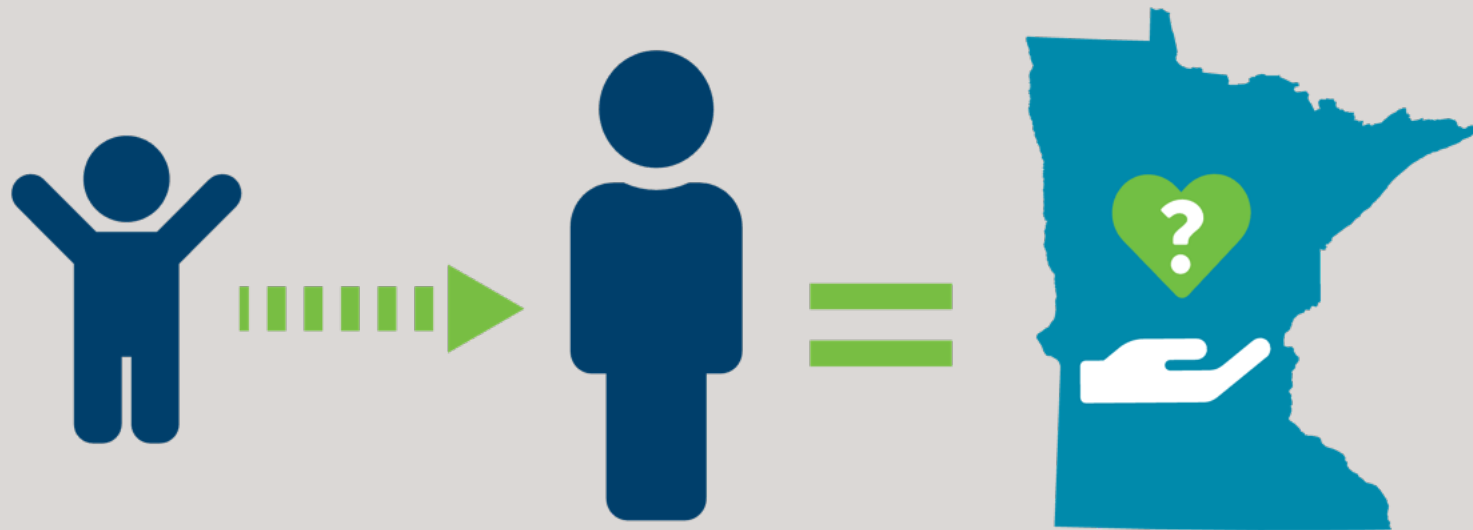
TRANSITION OF CARE

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



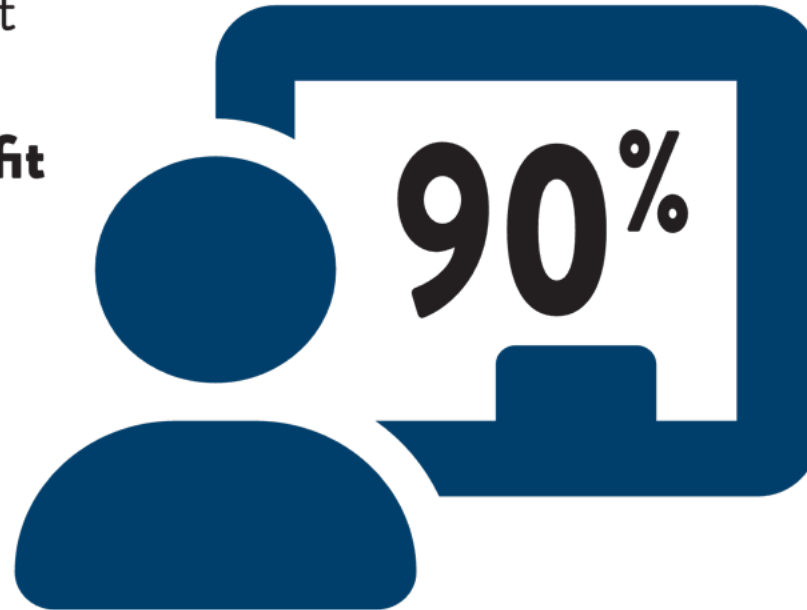
TRANSITION OF CARE REFUSALS

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.



ADDITIONAL TRAINING NEEDS

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



INABILITY
TO PROVIDE
CARE

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.

ADDITIONAL STUDIES TO BE LEVERAGED

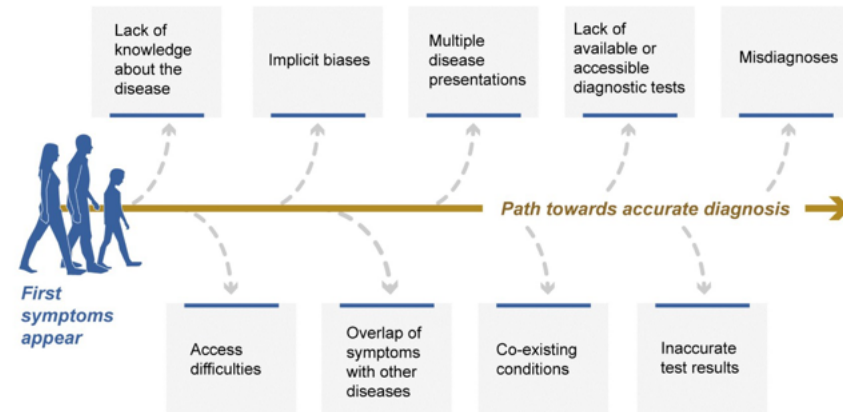
GAO, October 2021

Rare Diseases: Although Limited, Available Evidence Suggests Medical and Other Costs Can Be Substantial

What GAO Found

According to the literature GAO reviewed, diagnosis of any disease can be complicated, and diagnosis of rare diseases can be particularly difficult because of a variety of factors. (See figure.) Although some rare diseases may be detected quickly, in other cases years may pass between the first appearance of symptoms and a correct diagnosis of a rare disease, and misdiagnoses—and treatments based on them—occur with documented frequency. According to the literature GAO reviewed and GAO's interviews, those with undiagnosed, misdiagnosed, or untreated rare diseases may face various negative outcomes. For example, a person's health can suffer when appropriate, timely interventions are not provided or when interventions based on misdiagnoses cause harm. In addition, multiple diagnostic tests, medical appointments, and ultimately unwarranted interventions can add to the costs of the disease.

Examples of Factors That May Interfere with Accurate Diagnosis



Source: GAO analysis of published literature on rare diseases. | GAO-22-104235

The National Economic Burden of Rare Disease Study



FEBRUARY 25, 2021

PREPARED FOR:



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