DEPARTMENT OF HEALTH

Protecting, Maintaining and Improving the Health of All Minnesotans

July 7, 2026

Linda Hasadrsi, MS, MD, PhD
1216 2nd Street SW
Rochester, MN 55902

Dear Dr. Hasadsri and Co-Sponsor (Ms. Rachelen Varghese),

The Advisory Committee on Heritable and Congenital Disorders thanks you for your nomination to include Fabry disease on the Minnesota (MN) newborn screening (NBS) panel. We appreciate your interest and the time you invested in the nomination and review process.

As part of the committee's consideration process, a workgroup was convened to evaluate the current evidence regarding the suitability of population-based screening in MN. This work group consisted of individuals from MN with expertise in Fabry disease. Key findings were presented and discussed at the Advisory Committee meeting held on April 14, 2026. Advisors then completed their own review utilizing the nomination materials submitted, published literature, and the findings from the evidence review. The committee has finished its review and formally voted on June 16, 2026. **The Advisory Committee voted 16:3 to not recommend Fabry disease be added to the MN NBS panel.**

Below are key findings and discussion points:

- Fabry has a broad range of symptoms and a variable age of onset, including late onset.
- The current screening method is feasible, however the below concerns were expressed:
  - NBS will unlikely identify females with Fabry disease (risk of false negatives).
  - Although a biochemical second-tier may become available this summer, the assay may be normal in individuals with Fabry disease (risk of false negatives).
  - While molecular second-tier testing would reduce the number of false negative cases, it poses challenges with variant classification.
- Data on the outcomes of NBS for Fabry disease are limited despite multiple states screening newborns for the disease for a number of years.
- While specialists are available in MN who can diagnose and treat these patients, there are no data-driven clinical guidelines to advise them on the management and treatment of individuals identified through NBS without symptoms.

While Fabry disease is not recommended at this time, re-nomination can occur once some of the above items have been addressed. If you have any questions, please contact the committee coordinators via email at health.nsac@state.mn.us.

Sincerely,

Rae Blaylark

Rae Blaylark, Chairperson
Advisory Committee on Heritable and Congenital Disorders