Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Immunoreactive Trypsinogen (IRT) and 2 *CFTR* Variants (Both CF-Causing)

This screening result is likely a true diagnosis of cystic fibrosis (CF). Medical intervention needs to start as soon as possible.

Next Steps

This week, you should take the following recommended actions:

- **Consult** with a CF specialist. Contact information for accredited CF centers can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Evaluate** infant (poor weight gain, absent stooling, abdominal pain, voracious appetite); arrange immediate referral if symptomatic.
- **Arrange** sweat testing as recommended by the CF specialist.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share the follow-up plan with them. Educate family about signs, symptoms, and need for urgent treatment.

False Positives

Unlikely since two CF-causing variants were found on screening.

Differential Diagnosis

An elevated IRT with two CF-causing variants are primarily associated with:

- Cystic fibrosis — Incidence of 1 in 3,500

Clinical Summary

CF is an autosomal recessive disorder caused by specific cystic fibrosis transmembrane conductance regulator (CFTR) gene variants. *CFTR* variants affect the secretory glands, including those that make mucus and sweat.

Individuals with two CF-causing variants have cystic fibrosis. In infancy, CF is primarily manifested as a disorder of pancreatic insufficiency resulting in poor weight gain. Pulmonary disease manifests in childhood with chronic airway inflammation and infection. Affected children benefit from early dietary intervention and on-going management of pulmonary complications.