Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Immunoreactive Trypsinogen (IRT) ≥ 100 ng/mL with 1 *CFTR* Variant Identified

Next Steps

This week, you should take the following recommended actions:

- **Consult** with a cystic fibrosis (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. Since newborn screening only tests for certain variants, explain the importance of pursuing a sweat chloride test to confirm or rule out a diagnosis. Educate family about signs, symptoms, and when urgent treatment may be needed.

False Positives

Most infants with only one *CFTR* variant found on screening are unaffected carriers.

Differential Diagnosis

An elevated IRT with at least one *CFTR* variant is primarily associated with:

- Cystic fibrosis carrier — about 1 in 25 Caucasians are carriers
- Cystic fibrosis — Incidence of 1 in 3,500
- *CFTR*-related metabolic syndrome (CRMS)

Clinical Summary

CF is an autosomal recessive disorder caused by specific cystic fibrosis transmembrane conductance regulator (CFTR) gene variants.

Individuals with only one variant in the CFTR gene are considered carriers. A CF carrier is healthy and does not have cystic fibrosis. Because the Minnesota Newborn Screening Program only screens for a panel of the 39 most common CF variants, it is possible that a second *CFTR* variant exists that is not identifiable by the variant panel.

Individuals with two *CFTR* variants have cystic fibrosis or *CFTR*-related metabolic syndrome (CRMS). In infancy, CF is primarily manifested as a disorder of pancreatic insufficiency resulting in poor weight gain. An IRT ≥100 ng/mL heightens concern for pancreatic insufficiency. Children with CF need medical intervention right away. Individuals with CRMS have less severe symptoms and many are completely asymptomatic. Children with CRMS should be monitored by a CF Center for symptom development.