Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated Immunoreactive Trypsinogen (IRT) and 1 *CFTR* Variant Identified

Next Steps

This week, you should take the following recommended actions:

- **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 consultation if symptomatic.
- **Arrange** sweat testing with cystic fibrosis (CF) center when infant is about one month of age, earlier if symptoms are present. Contact information for accredited CF centers can be found on the resource list provided.

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). Children with cystic fibrosis experience poor weight gain, absent stooling, abdominal pain, and need medical intervention as soon as possible. Individuals with CRMS have less severe CF symptoms, including mild respiratory problems, sinusitis, pancreatitis, or infertility. Many individuals with CRMS are completely asymptomatic. Children with CRMS should have regular check-ups at a CF Center to monitor symptom development.

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155
Phone (800) 664-7772, Fax (651) 215-6285

REV 05/2019