Provider Fact Sheet

# Positive Result:

## Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

# Elevated Immunoreactive Trypsinogen (IRT) and 2 *CFTR* Variants (One or Both of Varying Clinical Consequence) Identified

## 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.

## False Positives

Unlikely since two variants were found on screening.

## Differential Diagnosis

An elevated IRT with two *CFTR* variants (at least one of varying clinical consequence) is primarily associated with:

- *CFTR*-related metabolic syndrome (CRMS)
- 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 found by newborn screening can either be CF-causing variants or variants of varying clinical consequence.

Individuals with two *CFTR* variants where one or both are of varying clinical consequence can have *CFTR*-related metabolic syndrome (CRMS) or cystic fibrosis.

Children with cystic fibrosis experience poor weight gain, absent stooling, abdominal pain, and need medical intervention as soon as possible.

However, this result is most often associated with CRMS. Individuals with CRMS have less severe CF symptoms, including mild respiratory problems, sinusitis, pancreatitis, or infertility. Many individuals are completely asymptomatic. Children with CRMS should have regular check-ups at a CF Center to monitor symptom development.