Positive Result: Blood Spot Screen Result Notification

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.