Provider Fact Sheet

Borderline Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Immunoreactive Trypsinogen (IRT) ≥ 120 ng/mL with No *CFTR* Variants Identified

Next Steps

This week, you should take the following recommended actions:

- **Contact** family to notify them of the newborn screening result and while unlikely, assess for symptoms (poor weight gain, absent stooling, abdominal pain, voracious appetite); arrange immediate referral if symptomatic.
- **Order** a repeat newborn screen.

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 when to contact you with concerns.

Possible Explanations for Result

Screening result is most likely due to meconium contamination during specimen collection. Additionally, many marked elevations of IRT are found in NICU graduates due to a range of health problems, including birth asphyxia, chromosome abnormalities, congenital anomalies, and intestinal perforation.

The Minnesota Newborn Screening Program only screens for a panel of the 39 most common cystic fibrosis (CF) variants. Rarely, this screening result can be due to the child having cystic fibrosis caused by variants not on this panel.

Clinical Summary

Most borderline results have normal repeat screens. Children with a borderline result who have a normal repeat screen require no additional follow-up or treatment. If repeat screening is abnormal, a Newborn Screening Program genetic counselor will contact you to discuss the next steps.

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

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