TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

Cystic Fibrosis

Elevated Immunoreactive Trypsinogen (IRT) and
One Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) Variant Identified

Provider Next Steps

This week, you should take the following recommended actions:

- Contact family to notify them of the newborn screening (NBS) result and assess symptoms;
- Evaluate infant for poor weight gain, poor feeding, abdominal pain, constipation, and/or frequent foul-smelling greasy stools, cough, wheezing, and/ or congestion. Arrange immediate consultation with a Cystic Fibrosis (CF) center if symptomatic; and
- Arrange for sweat test. Find contact information for accredited CF Centers on the provided resource list.

If you have questions about the NBS result or your next steps, please call **(512) 298-9696**.

Clinical Summary

CF is an autosomal recessive disorder caused by specific CFTR gene variants. CFTR variants affect the secretory glands, including those that make mucus and sweat.

Individuals with only one variant in the CFTR gene are considered carriers. A CF carrier is healthy and does not have CF. Because the Texas NBS Program panel includes the 60 most common CF mutations and four additional variants, it is possible that a second CFTR variant exists that was not identified by the variant panel.

Individuals with two CFTR variants have cystic fibrosis or CFTR-related metabolic syndrome (CRMS). Children with CF experience poor weight gain, have abnormal stooling (constipation or frequent foul-smelling greasy stool), 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. Most individuals with CRMS are asymptomatic, and some can develop signs and symptoms later in life. Children with CRMS should have regular check-ups at a CF center to monitor for CF conversion.

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:

- CF carrier – about 1 in 25 Caucasians are carriers;
- CF — 1 in 3,500 incidence; and
- CRMS.

CF is found in all races and ethnicities.

Review with Family

Discuss this result with the family. The Texas Department of State Health Services NBS Program has not notified the family of a CF-related screening result that should be discussed with their provider. Share the FACT sheet with the

family. Educate family about signs, symptoms, and need for follow-up with CF Center.

Family Discussion Points

- Since Texas NBS only tests for the **60 CF mutation panel with four additional variants**, explain the **importance of a SWEAT TEST** to confirm or rule out a diagnosis;
- Babies with CF do better if diagnosed and treated as early as possible;
- CF is a genetic disease that primarily causes thick, sticky mucus and can impact breathing and how food is digested; and
- For current information on CF, including testing, diagnosis, caring for a child with CF, and living with CF, please visit the CF Foundation website at cff.org. Avoid web searches on other sites that may provide inaccurate or outdated information.