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

Newborn Screening ACT Sheet

Cystic Fibrosis

Elevated Immunoreactive Trypsinogen (IRT) and

Two Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) Variants (Both Cystic Fibrosis-Causing)

This screening result is likely a true diagnosis of cystic fibrosis (CF). Evaluation at a CF Foundation-Accredited Center and additional testing needed to confirm diagnosis.

Provider Next Steps

Within 24 hours, you should take the following recommended actions:

- **Contact** family to notify them of the newborn screening (NBS) result and assess symptoms;
- **Evaluate** infant (poor weight gain, poor feeding, abdominal pain, constipation, and/or frequent foul-smelling greasy stools, cough, wheezing, and/or congestion);
- **Consult** with CF Specialist and arrange for IMMEDIATE REFERRAL. Find contact information for accredited CF centers on the provided resource list; and
- **Arrange** sweat testing as recommended by the CF Center.

If you have questions about the NBS 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 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.

False Positives

Unlikely — the screening showed two CF-causing variants. *CF is found in all races and ethnicities.*

Differential Diagnosis

An elevated IRT with two CF-causing variants is primarily associated with CF (1 in 3,500 incidence).

Review with Family

Discuss this result with the family as the Texas Department of State Health Services (DSHS) Newborn Screening Program has **NOT** notified them. Share the FACT sheet with them. Educate the family about signs, symptoms, and need for follow-up with CF Center.

Family Discussion Points

- DSHS identified two changes in your baby's CF gene. These changes are called variants or mutations;
- Further assessment, evaluation, and a **SWEAT TEST** at a CF Center is needed to confirm the diagnosis;
- CF is a genetic disease that primarily causes thick, sticky mucus and can impact breathing and how food is digested.
- CF is a chronic disease that will require life-long treatment. There are medications and therapies that allow most CF patients to live healthy lives;

- At the initial visit at the CF center, the team will cover the basics of CF and likely initiate medication. Genetic counseling will be available; 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 that may provide inaccurate or outdated information.

Revised: 6/5/2024