BIOCHEMISTRY AND GENETICS BRANCH
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CHANGES TO NEWBORN SCREENING CYSTIC FIBROSIS TESTING AND RESULT REPORTING STATEMENTS

NEWBORN SCREENING LABORATORY – DEPARTMENT OF STATE HEALTH SERVICES

The Texas DSHS Newborn Screening (NBS) Laboratory is pleased to announce full in-house Cystic Fibrosis 2nd-tier DNA testing has resumed. Several Newborn Screening Cystic Fibrosis result reporting statements will be updated to reflect the change in methodology. These updates are scheduled to go into effect Tuesday, May 24, 2016. All changes are indicated in red**.** A full list of all possible results including all Cystic Fibrosis results including mutation names can be found here: **http://www.dshs.texas.gov/lab/docs/SubmitterMailer-May2016.pdf**.

Overall Specimen Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	Cystic Fibrosis	Normal	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	Normal 0 Mutations Detected	No further evaluation necessary unless clinically indicated. The immunoreactive trypsinogen (IRT) result was normal. Tests for a ~~40~~-mutation panel in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were negative, however, the presence of other mutations cannot be ruled out.
Abnormal	Cystic Fibrosis	Result may be Abnormal or Inconclusive	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	IRT Elevated CFTR Mutation Panel may be 0, 1, or 2 Mutations Detected	Revised Screening Result for Cystic Fibrosis. Additional testing using a CFTR ~~40~~ Mutation Panel has been performed. *(Note: Result notes vary depending on the results applied for CFTR Mutation Panel)* [The specimen was originally reported as Indeterminate for Cystic Fibrosis showing Immunoreactive Trypsinogen as Elevated. The original screening result note read "Please repeat the newborn screen within 7 days. Many unaffected infants have an elevated immunoreactive trypsinogen (IRT) level on the first specimen. The second screening specimen (collected after 7 days of age) is required to determine if result is significant."]
Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	Elevated 0 Mutations Detected	No further evaluation necessary unless clinically indicated. None of the ~~40~~ CFTR mutations in the DSHS panel were detected, but there is a minimal risk for Cystic Fibrosis due to a mutation not included in the panel. Clinical evaluation not necessary unless symptomatic.

Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	Very Elevated 0 Mutations Detected	Although there is a minimal risk for Cystic Fibrosis (CF) in the absence of detected mutations, a very elevated immunoreactive trypsinogen (IRT) result may be indicative of CF due to mutations not included in the ~~40~~ mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	Elevated or Normal 1 Mutation Detected	One mutation, (Note: Mutation Name will display here.), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene was identified. Cystic Fibrosis cannot be ruled out due to a possibility of a second mutation which is not included in the ~~40~~ mutation panel. Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	Elevated or Normal 2 Mutations Detected	A homozygous Cystic Fibrosis-causing mutation, (Note: Mutation Name will display here.), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR ~~40~~ Mutation ~~Panel~~	Elevated or Normal 2 Mutations Detected	Two potential Cystic Fibrosis-causing mutations, (Note: Mutation Name will display here.) and (Note: Mutation Name will display here.), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days.

Reminders:

- Read Screening Result Notes fully before taking action.
- Contact the laboratory with any questions:
    - Telephone: 1-888-963-7111 X7585 or x2638
    - Email: NewbornScreeningLab@dshs.state.tx.us