BIOCHEMISTRY AND GENETICS BRANCH
LABORATORY SERVICES
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CHANGES TO NEWBORN SCREENING RESULT REPORTING STATEMENTS

NEWBORN SCREENING LABORATORY – DEPARTMENT OF STATE HEALTH SERVICES

In early June 2013, the following changes (in red) will be applied to Newborn Screening result reporting statements:

Changes to Screening Result Notes:

Disorder	CURRENT Screening Result Note	NEW Screening Result Note
Amino Acid Disorders, Organic Acid Disorders, and Fatty Acid Disorders	Borderline Result. Possible Metabolic Disorder. Please repeat the newborn screen.	Borderline Result. Possible Metabolic Disorder. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.
Hypothyroidism	Possible Hypothyroidism. Please repeat the newborn screen.	Possible Hypothyroidism. If this is the second screen, please follow recommendations received from Clinical Care Coordination. Otherwise, please repeat the newborn screen.

Changes to Specimen Result, Screening Result and associated Screening Result Note:

Disorder	CURRENT Specimen Result	NEW Specimen Result	CURRENT Screening Result	NEW Screening Result	CURRENT Screening Result Note	NEW Screening Result Note
SCID	Abnormal	Unsatisfactory	Indeterminate	Unsatisfactory	Inadequate sample for analysis of T-cell receptor excision circles (TREC). Please repeat the newborn screen.	Unsatisfactory – Please resubmit: Specimen inadequate for accurate detection of TREC (T-cell receptor excision circles)

Changes to a Screening Result Note for Cystic Fibrosis (CF) [used for specimens that had an initial indeterminate CF result and for which a second screen has not been received]:

Disorder	Screening Result	CURRENT Screening Result Note	NEW Screening Result Note
Cystic Fibrosis	Result may be: Inconclusive or Abnormal	*(Example:) No further evaluation necessary unless clinically indicated. Although there is a minimal risk for Cystic Fibrosis (CF) in the absence of detected mutations, an elevated immunoreactive trypsinogen (IRT) result may be indicative of CF due to a mutation not included in the 40-mutation panel. Recommend sweat testing and possible genetic evaluation only if clinically indicated.* [The original (previous) result for Cystic Fibrosis Testing was indeterminate.]	Revised Screening Result for Cystic Fibrosis. Additional testing using a CFTR 40 Mutation Panel has been performed. *(Example:) No further evaluation necessary unless clinically indicated. Although there is a minimal risk for Cystic Fibrosis (CF) in the absence of detected mutations, an elevated immunoreactive trypsinogen (IRT) result may be indicative of CF due to a mutation not included in the 40-mutation panel. Recommend sweat testing and possible genetic evaluation only if clinically indicated.* [The specimen was originally reported as Indeterminate for Cystic Fibrosis showing Immunoreactive Trypsinogen as Elevated. The original screening result note read "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. Please repeat the newborn screen."]
Note: *Actual result note will vary depending on results for CFDNA; text in blue is currently hard-coded for reflexed IRT specimens; text in red is the revised hard-coded text. For more detailed information on this change – please see List of all possible NBS Results, Analytes & Notes.*			

Resources:

- Healthcare Provider Resources: http://www.dshs.state.tx.us/lab/nbsHCRes.shtm
- List of all possible NBS Results, Analytes & Notes (NEW format)
- List of all possible NBS Results, Analytes & Notes (OLD format)
- Example Abnormal Result Report
- Example Normal Result Report

Reminders:

- Read Screening Result Notes fully before taking action.
- Screening Result Notes may continue on Page 2 of the Result Report.

Please contact the laboratory with any questions:
Telephone: 1-888-963-7111 X7585 or x2638 / Email: NewbornScreeningLab@dshs.state.tx.us