EXAMPLE REPORT INDICATING ABNORMAL SCREENING

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

PHYSICAL ADDRESS
1100 W. 49th St
Austin, TX 78756

LABORATORY SERVICES SECTION
CLIA #45D0660644

CONFIDENTIAL LABORATORY REPORT

MAILING ADDRESS
PO BOX 149347
AUSTIN, TEXAS 78714-9347
1-888-963-7111

SUBMITTER NAME - 00000004
123 MEDICAL STREET
AUSTIN, TX 78758

NEWBORN SCREENING REPORT -

Patient's Name:	GIRL TEXAN	Laboratory Number:	2018 343 0003
Mother's Name:	MOTHER TEXAN	Form Serial No:	18-0123458
Date Of Birth:	02/27/2019	Date Collected:	02/28/2019
Medical Record:	334455B	Date Received:	03/01/2019
Birth Weight:		Date Reported:	03/04/2019
Race/Ethnicity:			
Sex: FEMALE	Birth Order:	Test:	1ST TEST (UNDER 7 DAYS)
Feed:			
Status:		Mother's Address:	
		Mother's Telephone :	
		Physician's Name:	
		Physician's Telephone	

Overall Specimen Result

The Screening Result column indicates if the disorder category tested is Normal, Abnormal, non-specific, Possible TPN, Indeterminate, Inconclusive, or Unsatisfactory.

ABNORMAL SCREEN

Disorder *	Screening Result	Analyte	Analyte Result
Amino Acid Disorders	Normal		
Fatty Acid Disorders	Normal		
Organic Acid Disorders	Abnormal: See Note 1	C5:1	Borderline
		C5-OH	Normal
		C6DC	Normal
Galactosemia	Normal		
Biotinidase Deficiency	Normal		
Hypothyroidism	Normal		
CAH	Normal		
Hemoglobinopathies	Normal		
Cystic Fibrosis	Inconclusive: See Note 2	Immunoreactive Trypsinogen	Elevated
		CFTR Mutation	0 Mutations Detected
SCID	Normal		
X-ALD	Normal		

Screening Result Notes:

1. Possible Organic Acid Disorder. C5:1 Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
2. No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected. However, there is a minimal risk for Cystic Fibrosis due to variants not included in the panel.

The Screening Result Notes provide additional information on possible disorders, recommendations for follow-up testing and reasons for unsatisfactory specimens. Notes may continue on Page 2.

The Result Table includes an "Analyte" and "Analyte Result" column for Abnormal Screens.

The Analyte column lists which analyte's results were used to determine that a screening result was not normal

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Scope of NBS Testing, lab developed testing explanation, and List of Disorders

Cystic Fibrosis DNA testing explanation (for abnormal CF results tested for CFDNA)

-- The newborn screen identifies newborns at increased risk for specified disorders. The reference value for all screened disorders is 'Normal'. Analyte results are only listed for abnormal disorder screening results. The recommended collection time period and the testing methodologies have been designed to minimize the number of false negative and false positive results in newborns and young infants. When the newborn screen specimen is collected before 24 hours of age or on older children, the test may not identify some of these conditions. If there is a clinical concern, diagnostic testing should be initiated. Specimens that are unacceptable are reported as Unsatisfactory.

--The SCID / TREC (T-cell receptor excision circles) test is performed by quantitative real-time polymerase chain reaction analysis to detect the number of TRECs. SCID, Biotinidase deficiency, and Hemoglobinopathy screening tests and CAH and X-ALD reflex panels were developed / modified and performance characteristics determined by DSHS. These tests have not been cleared or approved by the US Food and Drug Administration (FDA). The FDA has determined that such approval is not necessary if performance characteristics are verified at the testing laboratory.

--The Cystic Fibrosis molecular testing panel consists of 60 mutations and 4 variants in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene and is performed using the Luminex xTAG Cystic Fibrosis (CFTR) 60 kit v2 assay. Depending on the patient's ethnicity, the mutation detection rate is estimated to be 54.5-95.9% and the residual risk of carrying a CFTR mutation not included on the panel is approximately 0.2-0.5%. Test results should not be used to diagnose but should be interpreted in the context of clinical findings, family history, and other laboratory data.

* Disorders Screened: **AMINO ACID DISORDERS:** ARG, ASA, CIT, CIT II, BIOPT(BS), BIOPT(REG), HCY, H-PHE, MET, MSUD, PKU, TYRI, TYRII, and TYRIII. **FATTY ACID DISORDERS:** CACT, CPT IA, CPT II, CUD, DE RED, GA2, LCHAD, MCAD, MCAT, M/SCHAD, SCAD, TFP, VLCAD. **ORGANIC ACID DISORDERS:** 2M3HBA, 2MBG, 3MCC, 3MGA, BKT, GA1, HMG, IBG, IVA, MAL, MMA (MUT, Cbl A, B, C, D), MCD, PROP. **GALACTOSEMIA. BIOTINIDASE DEFICIENCY. HYPOTHYROIDISM. CAH. HEMOGLOBINOPATHIES:** Hb S/S, Hb S/C, Hb S-Beta Th, Var Hb. Hb S-Beta Th, Var Hb. **CYSTIC FIBROSIS. SCID and T-Cell related Lymphopenias. X-LINKED ADRENOLEUKODYSTROPHY**. List of disorders screened available at www.dshs.state.tx.us/lab/NBSdisorderList.pdf