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

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CONFIDENTIAL LABORATORY REPORT

SUBMITTER NAME - 00000004
123 MEDICAL STREET
AUSTIN, TX 78758

NEWBORN SCREENING REPORT -

Patient's Name:	GIRL TEXAN	Laboratory Number:	2021 152 7001
Mother's Name:	MOTHER TEXAN	Form Serial No:	20-0123458
Date Of Birth:	05/24/2021	Date Collected:	05/31/2021
Medical Record:	334455B	Date Received:	06/01/2021
Birth Weight:	2,750 grams	Date Reported:	06/04/2021
Race/Ethnicity:			
Sex:	Birth Order:	Test:	
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	Normal		
Galactosemia	Normal		
Biotinidase Deficiency	Normal		
Hypothyroidism	Normal		
CAH	Normal		
Hemoglobinopathies	Normal		
Cystic Fibrosis	Abnormal: See Note 1	Immunoreactive Trypsinogen	Elevated
		CFTR Mutation	2 Mutations Detected
SCID	Normal		
X-ALD	Normal		
SMA	Abnormal: See Note 2		

Screening Result Notes:

1. Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G551D (c.1652G>A), in the CFTR gene were identified.
2. Probable Spinal Muscular Atrophy. Deletion of SMN1 exon 7 detected. Zero copies of SMN2 detected. Recommend rapid molecular confirmation including SMN1 and SMN2 copy number and telephone consultation and referral to a neurologist or neurogeneticist within 24 hours.

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 may include an "Analyte" and "Analyte Result" column.

The Analyte column may list which analyte results were used to determine the screening result.

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

Cystic Fibrosis DNA testing methodology statement (for abnormal CF results tested for DNA)

SMN2 (SMA) DNA testing methodology statement (for abnormal SMA results)

-- 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 / SMA test is performed by multiplex quantitative real-time PCR to detect the presence of T-cell receptor excision circles (TRECs) and SMN1 gene homozygous exon 7 deletion. The detection rate is estimated to be 95% of SMA cases. SCID, SMA, 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 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.

--The SMN2 copy number assay was performed by qualitative real-time polymerase chain reaction analysis to detect the copy number of SMN2 gene. It was developed by DSHS and its performance characteristics are determined by DSHS. This test has not been approved by the U.S. Food and Drug Administration (FDA).

***** 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. **CYSTIC FIBROSIS. SCID and T-Cell related Lymphopenias. X-ALD. SMA.** List of disorders screened available at www.dshs.state.tx.us/lab/NBSdisorderList.pdf