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

PUBLIC HEALTH LABORATORY DIVISION
CLIA #45D0660644

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PO BOX 149347
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CONFIDENTIAL LABORATORY REPORT

SUBMITTER NAME - 00000000
123 MEDICAL STREET
AUSTIN, TX 78756

NEWBORN SCREENING REPORT - 1

Patient Name:	GIRL TEXAN	MRN:	334455B	Lab Number:	2025 188 7001
Date Of Birth:	07/04/2025	Birth Order:	2 Sex: FEMALE	Form Serial No:	23-0083021
Mother Name:	MOTHER TEXAN	Birthweight:	3,000 grams	Date Collected:	07/05/2025
Mother Phone:	(512) 999 - 9999	Feed:	Breastmilk Only	Date Received:	07/07/2025
PCP Name:	DOCTOR, MEDICAL	Status:	NORMAL	Date Reported:	07/07/2025
PCP Phone:	(512) 777 - 7777				

ABNORMAL SCREEN

Overall Specimen Result

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

Disorder	Screening 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
SCID	Normal
X-ALD	Normal
SMA	Abnormal: See Note 2
Lysosomal Diseases	Abnormal: See Note 3
	Abnormal: See Note 4

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. One copy 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.
3. Possible Mucopolysaccharidosis type I (Hurler syndrome). IDUA activity Low. GAGs Slightly Elevated. If this is the second screen, follow recommendations received from Clinical Care Coordination. Otherwise, repeat the newborn screen within 7 days.
4. Probable Krabbe disease. GALC activity Low. Psychosine Elevated. A homozygous 30KB Deletion was detected. Recommend immediate consultation with a Krabbe Referral Center. Follow recommendations received from Clinical Care Coordination.

The Screening Result Notes provide additional information on possible disorders, analyte results for abnormal screening results, recommendations for follow-up testing and reasons for unsatisfactory specimens. Notes may continue on a second page.

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

Reference Lab statement included when a specimen is sent to an outside lab for GAGs reflex testing for MPS I and MPS II

Scope of NBS Testing and lab developed testing explanation

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

Reference Lab statement included when a specimen is sent to an outside lab for Psychosine reflex testing for Krabbe

Disorders Screened: Navigate to the webpage or scan the QR code for a complete listing of disorders screened in each category appearing in the result table

-- The newborn screen identifies newborns at increased risk for specified disorders. The reference value for all screened disorders is 'Normal'. Analyte results are only reported 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. List of disorders screened available at www.dshs.state.tx.us/lab/NBSDisordersScreened.

--The SCID / SMA test is performed by multiplex 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, Hemoglobinopathy, and Lysosomal Diseases screening tests and CAH, X-ALD, and Lysosomal Diseases 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 droplet digital 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).

-- With the exception of unsatisfactory results, Glycosaminoglycans (GAGs) reflex testing for Mucopolysaccharidosis (MPS I and MPS II), Lysosomal Diseases, was completed by Revvity Omics, 250 Industry Drive, Pittsburgh, PA 15275, CLIA 39D0673919.

-- With the exception of unsatisfactory results, Psychosine reflex testing for Krabbe, a Lysosomal Disease, was completed by Revvity Omics, 250 Industry Drive, Pittsburgh, PA 15275, CLIA 39D0673919.