CODIGO : **166638**

NOMBRE PACIENTE : LIAM SANTIAGO BERRIO MELO **SEXO :** MASCULINO

FECHA DE NACIMIENTO : 06/12/2024 **REGISTRO CIVIL :** 1,206,230,989

NOMBRE RESPONSABLE : KAREN LIZETH MELO ALZATE

DOC.IDENTIDAD DE LA MADRE : 1,073,238,467

FECHA TOMA DE MUESTRA : 16/01/2025

FECHA DE IMPRESION : 10/02/2025

TAMIZAJE NEONATAL

ANALISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACION
Hipotiroidismo congénito	3.01	VN: < 15 uUl/ml	Normal
Deficiencia de G6PDH	5.80	VN : > 2.6 U/gHb	Normal
Fenilcetonuria (PKU)	0.80	VN : < 2.1 mg/dL	Normal

TÉCNICA: Fluoroinmunoensayo (Delfia). *Procesado en Colombia por PREGEN.*

TÉCNICA: Cromatografía Liquida de Alto Rendimiento (HPLC). *Procesado en Colombia por PREGEN.*

TAMIZAJE AMPLIADO

ESPECTROMETRIA DE MASAS EN TANDEM

Procesado en Tennessee Department of Healt.

DESORDENES DE AMINOÁCIDOS
Citrulina, Metionina, Leucina, Isoleucina, Valina, Fenilalanina, Tirosina.

Ausencia de metabolitos anormales — Normal

DESORDENES DE LA OXIDACIÓN DE ÁCIDOS GRASOS
C16,C18,C18:1,C16OH,C18:1OH,C8,C10:1,C5,C5DC,C4,C14,C14:1

Ausencia de metabolitos anormales — Normal

ACIDEMIAS ORGÁNICAS
C5OH, C5DC, C5, C3, C5:1

Ausencia de metabolitos anormales — Normal

RESULTADOS NORMALES

Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta la edad del paciente, por esto es conveniente realizarlas dentro del primer mes de nacido.

REVISADO : EDUVILIA JOHANA GOMEZ **FECHA :** 10/02/2025
Bacteriologa
Reg. 40.936.003

SC Department of Public Health
Public Health Laboratory

CLIA ID: 42D0658606

8231 Parklane Road
Columbia, SC 29223
Phone: 803-896-0800
Fax: 803-896-0983

Patient Name :	BERRIO MELO , LIAM	**Baby's Sex :**	Male	**Mother Name :**	MELO , KAREN
				Mother	C11 16 K1126-20
DOB :	12/6/2024 23:02	**Race :**	Afr. Amer.	**Address :**	MOSOLURA ,
Medical Record :		**Birth Weight :**	2500 grams	**Mother Phone :**	(322)447-8683
Specimen				**Gestational**	
Number :	20250241524	**Present Weight :**	3440 grams	**Age :**	36 weeks
		Age at			
Kit Number :	TN0000090129	**Collection :**	970 hours		
Date Collected :	1/16/2025 09:24	**Birth Order :**		**Submitter :**	TN Department of Health-NBS
		Transfusion			
Date Received :	1/24/2025	**Date :**		**Physician :**	TN Department of Health-NBS
Date Reported :	1/28/2025	**Feed Type :**		**Sample Type:**	Dried Blood Spot

INITIAL NEWBORN SCREEN - FINAL

Action Required: Please see Notes

Screening Test	Screening Results	Value	Expected Ranges	Units	Note
Congenital Hypothyroidism					
TSH	Within Acceptable Limits		<20.00, <= 7 days old <10.50, > 7 days old	µIU/mL	
Congenital Adrenal Hyperplasia					
17-OHP	Within Acceptable Limits		<30, Present Wgt >= 2500g <76, Present Wgt <2500g	ng/mL	
Hemoglobinopathies	Within Acceptable Limits	FA	FA		
Biotinidase	Within Acceptable Limits		>70.00	U/dL	
Galactosemia					
GAO	Within Acceptable Limits		<12.00	mg/dl	
GALT	Within Acceptable Limits		>3.75	U/dL	
Cystic Fibrosis					
CF Molecular	Within Acceptable Limits		No Mutations Detected		1
SCID					
TREC	Within Acceptable Limits		>250	copies/10^5 cells	
SMA					
SMN1	Within Acceptable Limits		SMN1 Detected		
X-linked Adrenoleukodystrophy (X-ALD)	Within Acceptable Limits		Within Acceptable Limits		
Amino Acid (AA) Panel (Includes SA)	Within Acceptable Limits		Panel Within Acceptable Limits		
Acylcarnitine (AC) Panel	Within Acceptable Limits		Panel Within Acceptable Limits		
Pompe					
GAA (acid-a-glucosidase)	Within Acceptable Limits		> 20% of Daily Median	%	
Mucopolysaccharidosis Type I (MPSI)					
IDUA (a-L-iduronidase)	Within Acceptable Limits		> 10% of Daily Median	%	
Krabbe					
GALC (ß-galactocerebrosidase)	Within Acceptable Limits		> 15% of Daily Median	%	

Notes

1: The Cystic Fibrosis (CF) 2nd tier molecular testing panel includes mutations and variants in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene recommended by the American College of Medical Genetics and American College of Obstetricians and Gynecologists (ACMG/ACOG), plus some of mutations. The test was performed by polymerase chain reaction (PCR) and allele specific oligonucleotide hybridization, using a Luminex® FDA-cleared assay. The testing panel consists of 60 mutations and 4 variants. A false negative result may occur if other CF causing mutations are present but not detectable by this test and/or rare errors due to mutations in the primer binding region. Test performance specifications were verified by this laboratory according to CLIA. This test is intended for use as a screening tool and should not be considered diagnostic. The results should be interpreted in the context of clinical findings, family history, and other laboratory data. Please contact the laboratory if additional information regarding the test is needed. For a list of mutations and variants detectable by the Luminex xTAG Cystic Fibrosis 60 kit, v2, please refer to the Luminex Corporation website: https://www.luminexcorp.com/?wpdmdl=8568.

If the patient has received a blood transfusion within the last 120 days, the CF molecular assay may yield a false positive or false negative result. If a satisfactory 120-day post-transfusion specimen is submitted, the CF molecular assay will be performed.

Report Sent To:

1. TN Department of Health- NBS, 630 Hart Lane Nashville, TN 37216
2. TN Department of Health- NBS, 630 Hart Lane Nashville, TN 37216