



**CODIGO :** 172483  
**NOMBRE PACIENTE :** EITHAN DANIEL LOPEZ RODRIGUEZ **SEXO :** MASCULINO  
**FECHA DE NACIMIENTO :** 10/12/2025 **REGISTRO CIVIL :** 1,089,652,597  
**NOMBRE RESPONSABLE :** ALISON DAYANA RODRIGUEZ ARREDONDO  
**DOC.IDENTIDAD DE LA MADRE :** 12,693,187  
**FECHA TOMA DE MUESTRA :** 24/02/2026 **TIPO DE MUESTRA :** TALÓN  
**FECHA DE IMPRESIÓN :** 09/03/2026 **PESO :** 4420

## TAMIZAJE NEONATAL

### ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	1.89	>= 6 µl/mL talón en prematuros >= 10 µl/mL talón >= 15 µl/mL cordón	Normal
Deficiencia de G6PDH	4.30	> 2.6 U/gHb	Normal
<i>TÉCNICA: Fluoroimmunoensayo (Delfia).</i>			<i>Procesado en Colombia por PREGEN.</i>
Hemoglobinopatías	FA	Ausencia de hemoglobinas anormales	Normal
<i>TÉCNICA: Cromatografía Líquida de Alto Rendimiento (HPLC).</i>			<i>Procesado en Colombia por PREGEN.</i>

## 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

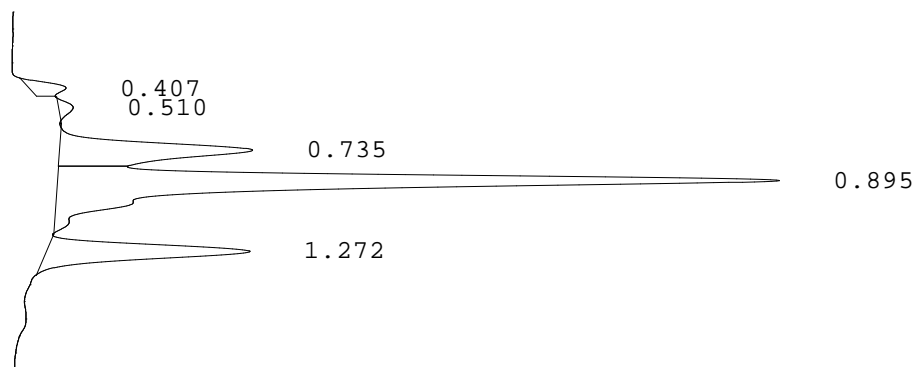
Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta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

**REVISADO :** EDUVILIA JOHANA GOMEZ **PROCESADO :** MARIA JOSE PINZON GARCIA **FECHA :**  
Bacterióloga Bacterióloga  
Reg. 40.936.003 Reg. 1.015.469.392 09/03/2026

LABORATORIO PREGEN  
Carrera 15a No 106-42  
BOGOTA

Batch 2083, Rack A, Plate 1, Well D06, 172483  
[DD3378E7AF99D397] Feb 27, 2026 11:43:22 Pressure = 54 bar (54 to 56)

FA



PEAK	RT	REL RT	% CONC	AREA	COMMENT
1	0.407	F 0.46	2.4%	12676	
2	0.510	F 0.58	1.1%	6060	
3	0.735	F 0.83	18.8%	100267	
4	0.895	F 1.01	60.9%	325318	Consistent with F
5	1.272	A 1.01	16.8%	89571	A peak
Total Area:				533892	

- Codes:
- 1) Wide A peak
  - 2) Area of A peak < 80%
  - 3) Peak area greater than expected
  - 4) Peak after A2
  - 5) Alc > 10%
  - 6) HbF or variant present
  - 7) Total sample area too small/big
  - 8) A2 is not within normal range

Dr. MARIA JOSE PINZON GARCIA  
RED COLOMBIANA DE MEDICINA GENETICA SAS - PREGEN  
BOGOTA  
CARRERA 15 A # 106 - 42  
11001 BOGOTA  
Colombia

Date of Report 06.03.2026  
Sample Received 03.03.2026  
Date of Sampling 25.02.2026  
LAB-ID 262009450

## Medical Report

Patient name	<b>LOPEZ RODRIGUEZ EITHAN DANIEL</b>	Sample-ID	A0361372
Date of Birth	<b>10.12.2025</b>	Gender	M

**Indication:** Newborn Screening

**Method(s):** Immunoassay, Tandem mass spectrometry from Dried Blood Spot. qPCR from Dried Blood Spot.

**Results:**

Parameter	Value	Unit	Reference
Birth weight (g)	4420	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	1.6	µU/mL	< 15.0
Biotinidase	270.5	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	7.3	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	41.9	ng/mL	< 65.0
Phenylalanine	41.3	µmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

**Interpretation:** NEGATIVE RESULT

Patient name	<b>LOPEZ RODRIGUEZ EITHAN DANIEL</b>
Date of Birth	<b>10.12.2025</b>

Sample-ID	A0361372
Gender	M

## Results:

### Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	41.3	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.66	µmol/L	< 2.20
Tyrosine (Tyr)	62.2	µmol/L	< 200.0
Leucine (Leu)	120.2	µmol/L	< 270.0
Valine (Val)	60.5	µmol/L	< 200.0
Methionine (MET)	33.6	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.81	µmol/L	< 1.60
Citrulline (Cit)	16.0	µmol/L	< 50.0
Ornithine (Orn)	154.8	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	9.68	µmol/L	1.50 - 20.00
Proline (Pro)	102.5	µmol/L	< 350.0
Alanine (Ala)	176.4	µmol/L	< 750.0
Arginine (Arg)	6.3	µmol/L	< 100.0
Aspartic acid (Asp)	89.1	µmol/L	< 100.0
Glutamic acid (Glu)	249.2	µmol/L	< 600.0
Glycamine (Gly)	231.4	µmol/L	< 700.0

### Acylcarnitines

Free carnitine (C0)	29.34	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	24.00	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	2.34	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.10	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.11	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.02	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.19	µmol/L	0.02 - 0.57
glutarylacetyl carnitine (C5DC)	0.03	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.04	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.02	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.03	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.10	µmol/L	< 0.30
decadienoylcarnitine (C10:2)	0.04	µmol/L	< 0.10
dodecanoylcarnitine (C12)	0.07	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.14	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.09	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	1.07	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.04	µmol/L	< 0.10
stearoylcarnitine (C18)	0.43	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	4.27	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.04	µmol/L	< 0.50

Amino acid levels are indicators of phenylketonuria, tyrosinemia, MSUD, hydroxyprolinuria, hypermethioninemia (homocystinuria), citrullinemia, argininosuccinate aziduria, hyperargininemia, and hyperprolinemia. Acylcarnitine levels are indicators of carnitine uptake disorders, CPT-I deficiency, CPT-II deficiency, CAT deficiency, propionaciduria, methylmalonic aciduria, malonic aciduria, SCAD deficiency/ethylmalonic aciduria, isovaleric aciduria, HMG-CoA lyase deficiency, 3-methylcrotonyl-CoA carboxylase deficiency, methylglutaconiduria, MCAD deficiency, VLCAD deficiency, LCHAD deficiency, glutaraziduria I, multiple acyl-CoA dehydrogenase deficiency (MAD deficiency/glutaraziduria II), and Beta-ketothiolase deficiency.

**Please note:** Inconspicuous negative biochemical results cannot exclude any inborn error of metabolism or endocrine disorder with certainty in newborns. We recommend any follow-up or genetic testing if any clinical symptoms are present.

**Authorized By:** Assoc.-Prof. Dr. Andrea-Romana KASPER, MD, PhD  
[Specialist for Pediatrics, Neonatology and Nutrition]

Report was electronically signed and approved.

#### Contact Details

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