



CODIGO : 173184
NOMBRE PACIENTE : EMILY LUCIANA DOMINGUEZ JIMENEZ **SEXO :** FEMENINO
FECHA DE NACIMIENTO : 21/02/2026 **REGISTRO CIVIL :** 1,233,528,459
NOMBRE RESPONSABLE : ERIKA PAOLA JIMENEZ RAMIREZ
DOC.IDENTIDAD DE LA MADRE : 1,108,151,273
FECHA TOMA DE MUESTRA : 24/04/2026 **TIPO DE MUESTRA :** TALÓN
FECHA DE IMPRESIÓN : 13/05/2026 **PESO :** 2435

TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	6.98	>= 6 µl/mL talón en prematuros >= 10 µl/mL talón >= 15 µl/mL cordón	Normal
Deficiencia de G6PDH	5.10	> 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 Archimedlife international medical laboratory. 1110 Vienna.

DESORDENES DE AMINOÁCIDOS

Citrulina, Metionina, Leucina, Isoleucina, Valina, Fenilalanina, Tirosina.

Ausencia de metabolitos anormales Normal

PERFIL DE ACILCARNITINAS

C16,C18,C18:1,C16OH,C18:1OH,C8,C10:1,C5,C5DC,C4,C14,C14:1,C50H,C3,C5:1

Ausencia de metabolitos anormales Normal

RESULTADOS NORMALES

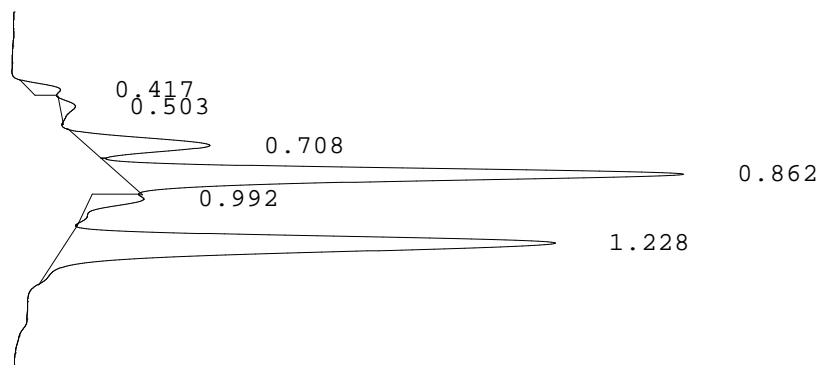
Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta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

REVISADO : EDUVILIA JOHANA GOMEZ **PROCESADO :** MARIA JOSE PINZON GARCIA **FECHA :**
Bacterióloga Bacterióloga
Reg. 40.936.003 Reg. 1.015.469.392 13/05/2026

LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2133, Rack A, Plate 1, Well C05, 173184
[89674BFC1096A640] Apr 29, 2026 12:07:47 Pressure = 66 bar (65 to 67)

FA



PEAK	RT	REL RT	% CONC	AREA	COMMENT
1	0.417	F 0.48	1.7%	9432	
2	0.503	F 0.59	1.1%	6318	
3	0.708	F 0.82	9.5%	52355	Acetylated F peak
4	0.862	F 1.00	39.9%	220529	Consistent with F
5	0.992	F 1.15	3.8%	21217	
6	1.228	A 1.00	44.0%	243462	A peak - REVIEW
Total Area:				553313	

- 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 08.05.2026
Sample Received 04.05.2026
Date of Sampling 24.04.2026
LAB-ID 262019147

Medical Report

Patient name	DOMINGUEZ JIMENEZ EMILY LUCIANA	Sample-ID	A0340889
Date of Birth	21.02.2026	Gender	F

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)	2435	g	-
17-hydroxyprogesterone (17OHP)	5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	1.0	µU/mL	< 15.0
Biotinidase	240.1	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	8.0	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	27.0	µmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

Interpretation: NEGATIVE RESULT

Patient name	DOMINGUEZ JIMENEZ EMILY LUCIANA
Date of Birth	21.02.2026

Sample-ID	A0340889
Gender	F

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	27.0	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.42	µmol/L	< 2.20
Tyrosine (Tyr)	63.8	µmol/L	< 200.0
Leucine (Leu)	109.2	µmol/L	< 270.0
Valine (Val)	48.4	µmol/L	< 200.0
Methionine (MET)	18.6	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.69	µmol/L	< 1.60
Citrulline (Cit)	18.7	µmol/L	< 50.0
Ornithine (Orn)	99.8	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	5.34	µmol/L	1.50 - 20.00
Proline (Pro)	100.5	µmol/L	< 350.0
Alanine (Ala)	160.4	µmol/L	< 750.0
Arginine (Arg)	8.0	µmol/L	< 100.0
Aspartic acid (Asp)	81.6	µmol/L	< 100.0
Glutamic acid (Glu)	393.5	µmol/L	< 600.0
Glycamine (Gly)	135.5	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	24.81	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	11.49	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	1.35	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.18	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.13	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.02	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.36	µmol/L	0.02 - 0.57
glutarylacarnitine (C5DC)	0.04	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.05	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.05	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.05	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.13	µmol/L	< 0.30
decadienoylcarnitine (C10:2)	0.13	µmol/L	< 0.10
dodecanoylcarnitine (C12)	0.07	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.09	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.08	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	0.84	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.45	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	1.49	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.07	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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