

CODIGO :
NOMBRE PACIENTE :
FECHA DE NACIMIENTO :
NOMBRE RESPONSABLE :
DOC.IDENTIDAD DE LA MADRE :
FECHA TOMA DE MUESTRA :
FECHA DE IMPRESIÓN :

173838
MIA ISABELLA BOHORQUEZ MARTINEZ
26/05/2026
JUNNY CAMILA MARTINEZ GUERRERO
1,014,251,440
24/06/2026
15/07/2026

SEXO : FEMENINO
REGISTRO CIVIL: 1,032,801,270
TIPO DE MUESTRA : TALÓN
PESO : 3380



TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	3.11 µU/mL	>= 6 µU/mL talón en prematuros >= 10 µU/mL talón >= 15 µU/mL cordón	Normal
Deficiencia de G6PDH	7.10 U/gHb	< 2.6 U/gHb	Normal
TÉCNICA: Fluoroimmunoensayo (Delfia).		Procesado en Colombia por PREGEN.	
Hemoglobinopatías	FA	Ausencia de hemoglobinas anormales	Normal
TÉCNICA: Cromatografía Líquida de Alto Rendimiento (HPLC).		Procesado en Colombia por PREGEN.	

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, C5OH, 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
Bacterióloga
Reg. 40.936.003

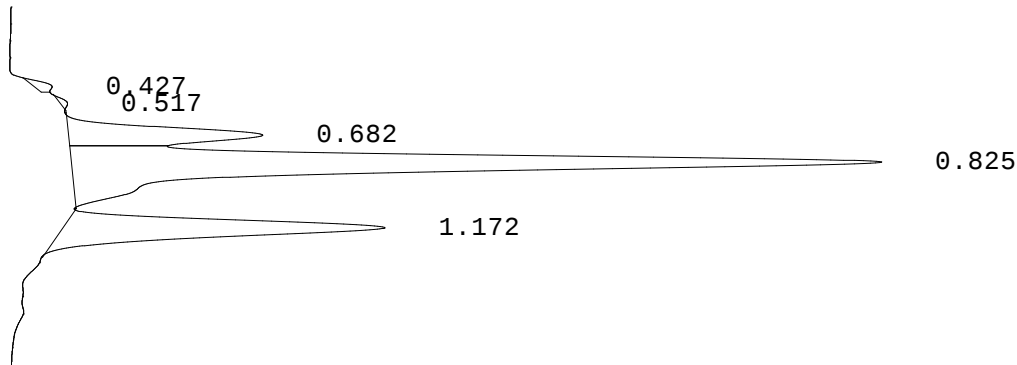
PROCESADO :
MARIA JOSE PINZON GARCIA
Bacterióloga
Reg. 1.015.469.392

FECHA :
15/07/2026

LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2188, Rack A, Plate 1, Well B10, 173838
[8D7C90C32A5D911D] Jul 01, 2026 11:34:57 Pressure = 79 bar (79 to 82)

FA



PEAK	RT	REL	RT	% CONC	AREA	COMMENT
1	0.427	F	0.51	0.7%	5181	
2	0.517	F	0.62	0.3%	2383	
3	0.682	F	0.81	13.1%	91084	Acetylated F peak
4	0.825	F	0.98	62.1%	431486	Consistent with F
5	1.172	A	1.01	23.7%	165012	A peak - REVIEW
Total Area:					695146	

Codes: 1) Wide A peak
2) Area of A peak < 80%
3) Peak area greater than expected
4) Peak after A2
5) A1c > 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 - PREGN
BOGOTA
CARRERA 15 A # 106 - 42
11001 BOGOTA
Colombia

Date of Report 10.07.2026
Sample Received 30.06.2026
Date of Sampling 24.06.2026
LAB-ID 262029629

Medical Report

Patient name	BOHORQUEZ MARTINEZ MIA ISABELLA	Sample-ID	A0358521
Date of Birth	26.05.2026	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)	3380	g	-
17-hydroxyprogesterone (17OHP)	6.2	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	1.3	μU/mL	< 15.0
Biotinidase	242.5	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	8.0	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	28.2	μmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

Interpretation: NEGATIVE RESULT

Contact Details

Assoc.-Prof. Dr. Andrea-Romana KASPER, MD, PhD
E-Mail: info@archimedlife.com

Patient name	BOHORQUEZ MARTINEZ MIA ISABELLA
Date of Birth	26.05.2026

Sample-ID	A0358521
Gender	F

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	28.2	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.50	µmol/L	< 2.20
Tyrosine (Tyr)	56.3	µmol/L	< 200.0
Leucine (Leu)	80.3	µmol/L	< 270.0
Valine (Val)	51.5	µmol/L	< 200.0
Methionine (MET)	11.9	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.42	µmol/L	< 1.60
Citrulline (Cit)	16.7	µmol/L	< 50.0
Ornithine (Orn)	63.1	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	3.78	µmol/L	1.50 - 20.00
Proline (Pro)	115.4	µmol/L	< 350.0
Alanine (Ala)	209.4	µmol/L	< 750.0
Arginine (Arg)	27.6	µmol/L	< 100.0
Aspartic acid (Asp)	46.1	µmol/L	< 100.0
Glutamic acid (Glu)	270.1	µmol/L	< 600.0
Glycamine (Gly)	190.9	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	15.70	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	6.55	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	0.45	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.09	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.07	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.01	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.24	µmol/L	0.02 - 0.57
glutaryl carnitine (C5DC)	0.04	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.03	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.04	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
dodecanoylcarnitine (C12)	0.03	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.09	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.06	µmol/L	0.10 - 0.43
palmitoylcarnitine (C16)	0.63	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.03	µmol/L	< 0.10
stearoylcarnitine (C18)	0.29	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	1.09	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.05	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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