

CODIGO :

173876

NOMBRE PACIENTE :

ISABELLA JIMENEZ MARTINEZ

SEXO : FEMENINO

FECHA DE NACIMIENTO :

03/04/2026

REGISTRO CIVIL:

1,019,768,642

NOMBRE RESPONSABLE :

LINA MARIA MARTINEZ PEREZ

DOC.IDENTIDAD DE LA MADRE :

1,032,431,028

FECHA TOMA DE MUESTRA :

03/04/2026

TIPO DE MUESTRA :

TALÓN

FECHA DE IMPRESIÓN :

22/07/2026

PESO :

3550



TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	0.54 µUL/mL	>= 6 µUL/mL talón en prematuros >= 10 µUL/mL talón >= 15 µUL/mL cordón	Normal
Deficiencia de G6PDH	6.00 U/gHb	< 2.6 U/gHb	Normal
TÉCNICA: Fluoroimmunoensayo (Delfia).		Procesado en Colombia por PREGEN.	
Hemoglobinopatías	AF	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

PROCESADO :

MARIA JOSE PINZON GARCIA

FECHA :

Bacterióloga

Bacterióloga

Reg. 40.936.003

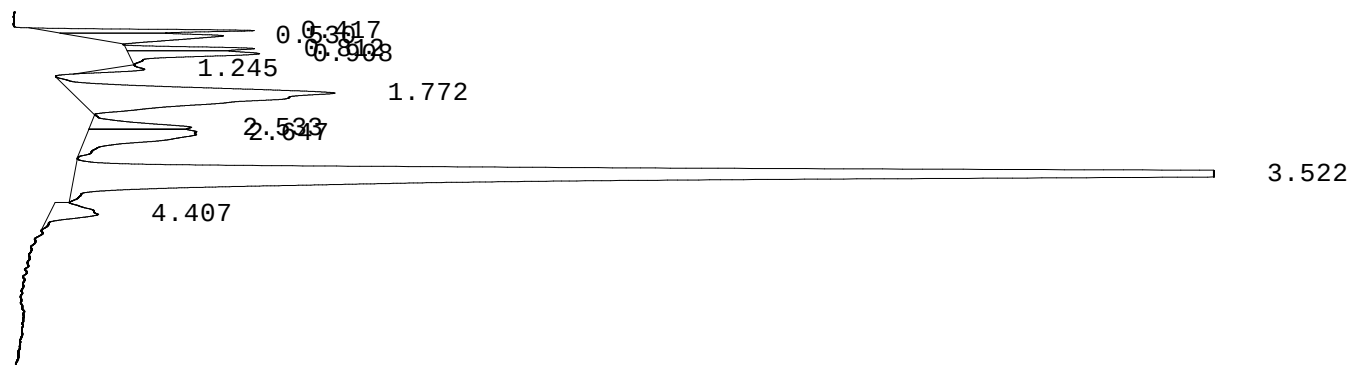
Reg. 1.015.469.392

22/07/2026

LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2192, Rack A, Plate 1, Well A04, 173876
[986CC1455EF7BB3F] Jul 06, 2026 15:23:32 Pressure = 80 bar (78 to 81)

AF



PEAK	RT	REL	RT	% CONC	AREA	COMMENT
1	0.417	F	0.23	2.5%	50313	
2	0.530	F	0.29	3.4%	68328	
3	0.812	F	0.45	1.7%	34919	
4	0.908	F	0.50	2.7%	55444	
5	1.245	F	0.69	0.8%	15283	
6	1.772	F	0.98	15.0%	302849	Consistent with F
7	2.533	F	1.39	2.4%	48070	
8	2.647	F	1.46	5.5%	110992	3
9	3.522	A	1.00	63.9%	1293211	A peak - REVIEW
10	4.407	S	0.87	2.2%	44865	3
Total Area: 2024274						

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 11.07.2026
Sample Received 07.07.2026
Date of Sampling 27.06.2026
LAB-ID 262030692

Medical Report

Patient name	JIMENEZ MARTINEZ ISABELLA	Sample-ID	A0358503
Date of Birth	03.04.2026	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)	3550	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	<0.7	μU/mL	< 15.0
Biotinidase	287.8	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	8.0	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	36.1	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	JIMENEZ MARTINEZ ISABELLA
Date of Birth	03.04.2026

Sample-ID	A0358503
Gender	F

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	36.1	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.51	µmol/L	< 2.20
Tyrosine (Tyr)	70.1	µmol/L	< 200.0
Leucine (Leu)	107.4	µmol/L	< 270.0
Valine (Val)	53.3	µmol/L	< 200.0
Methionine (MET)	18.7	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.52	µmol/L	< 1.60
Citrulline (Cit)	27.3	µmol/L	< 50.0
Ornithine (Orn)	110.5	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	4.05	µmol/L	1.50 - 20.00
Proline (Pro)	116.6	µmol/L	< 350.0
Alanine (Ala)	169.2	µmol/L	< 750.0
Arginine (Arg)	32.3	µmol/L	< 100.0
Aspartic acid (Asp)	51.5	µmol/L	< 100.0
Glutamic acid (Glu)	209.6	µmol/L	< 600.0
Glycamine (Gly)	173.6	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	46.92	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	19.02	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	2.79	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.24	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.22	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.03	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.48	µmol/L	0.02 - 0.57
glutarylacetylcholine (C5DC)	0.06	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.06	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.05	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.06	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.12	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.10	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.17	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.14	µmol/L	0.10 - 0.43
palmitoylcarnitine (C16)	1.65	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	1.12	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	3.14	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.08	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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