

CODIGO :

173823

NOMBRE PACIENTE :

JULIETTA SOPHIA CADENA PAREDES

FECHA DE NACIMIENTO :

04/05/2026

NOMBRE RESPONSABLE :

KERLENIA PAREDES CAPANO

DOC.IDENTIDAD DE LA MADRE :

4,858,446

FECHA TOMA DE MUESTRA :

23/06/2026

FECHA DE IMPRESIÓN :

15/07/2026

SEXO : FEMENINO

REGISTRO CIVIL: 1,011,271,979

TIPO DE MUESTRA : TALÓN

PESO : 3295



TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	0.65 µUL/mL	>= 6 µUL/mL talón en prematuros >= 10 µUL/mL talón >= 15 µUL/mL cordón	Normal
Deficiencia de G6PDH	6.50 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

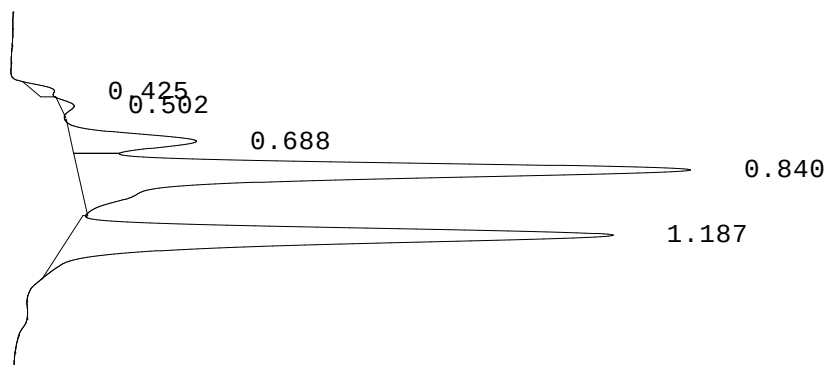
Cra 15A N.106-42 Celulares: 318 735 08 83 - 316 617 25 34

Código:TME-A-FR-050B Fecha versión:20251130

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`LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2185, Rack A, Plate 1, Well F03, 173823
[993DEE0321CD91BF] Jun 25, 2026 13:55:56 Pressure = 72 bar (72 to 74)

FA



PEAK	RT	REL	RT	% CONC	AREA	COMMENT
1	0.425	F	0.50	1.0%	6584	
2	0.502	F	0.59	0.7%	4558	
3	0.688	F	0.81	8.9%	59187	Acetylated F peak
4	0.840	F	0.99	48.3%	322517	Consistent with F
5	1.187	A	1.01	41.2%	275204	A peak - REVIEW
Total Area: 668050						

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 - PREGEN
BOGOTA
CARRERA 15 A # 106 - 42
11001 BOGOTA
Colombia

Date of Report 10.07.2026
Sample Received 30.06.2026
Date of Sampling 23.06.2026
LAB-ID 262029616

Medical Report

Patient name	CADENA PAREDES JULIETTA SOPHIA	Sample-ID	A0358478
Date of Birth	04.05.2026	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)	3295	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	<0.7	μU/mL	< 15.0
Biotinidase	228.9	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	7.7	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	33.4	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	CADENA PAREDES JULIETTA SOPHIA
Date of Birth	04.05.2026

Sample-ID	A0358478
Gender	F

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	33.4	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.52	µmol/L	< 2.20
Tyrosine (Tyr)	63.7	µmol/L	< 200.0
Leucine (Leu)	94.3	µmol/L	< 270.0
Valine (Val)	37.1	µmol/L	< 200.0
Methionine (MET)	28.7	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.86	µmol/L	< 1.60
Citrulline (Cit)	11.9	µmol/L	< 50.0
Ornithine (Orn)	32.9	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	2.76	µmol/L	1.50 - 20.00
Proline (Pro)	88.5	µmol/L	< 350.0
Alanine (Ala)	78.9	µmol/L	< 750.0
Arginine (Arg)	19.0	µmol/L	< 100.0
Aspartic acid (Asp)	54.2	µmol/L	< 100.0
Glutamic acid (Glu)	272.6	µmol/L	< 600.0
Glycamine (Gly)	82.8	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	14.04	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	7.90	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	1.22	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.12	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.08	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.01	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.25	µmol/L	0.02 - 0.57
glutaryl carnitine (C5DC)	0.06	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.03	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.03	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.06	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.19	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.05	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.08	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.07	µmol/L	0.10 - 0.43
palmitoylcarnitine (C16)	0.59	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.22	µmol/L	0.30 - 2.40
oleyl carnitine (C18:1)	1.80	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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