

CODIGO :

173826

NOMBRE PACIENTE :

VALERY THALIANA HERNANDEZ GONZALEZ

FECHA DE NACIMIENTO :

30/04/2026

NOMBRE RESPONSABLE :

LUZ MARY GONZALEZ QUINTERO

DOC.IDENTIDAD DE LA MADRE :

1,007,417,983

FECHA TOMA DE MUESTRA :

23/06/2026

FECHA DE IMPRESIÓN :

15/07/2026

SEXO : FEMENINO

REGISTRO CIVIL: 1,077,979,044

TIPO DE MUESTRA : TALÓN

PESO : 3800



TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	1.68 µU/mL	>= 6 µU/mL talón en prematuros >= 10 µU/mL talón >= 15 µU/mL cordón	Normal
Deficiencia de G6PDH	5.60 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

Bacterióloga

Reg. 40.936.003

PROCESADO :

MARIA JOSE PINZON GARCIA

Bacterióloga

Reg. 1.015.469.392

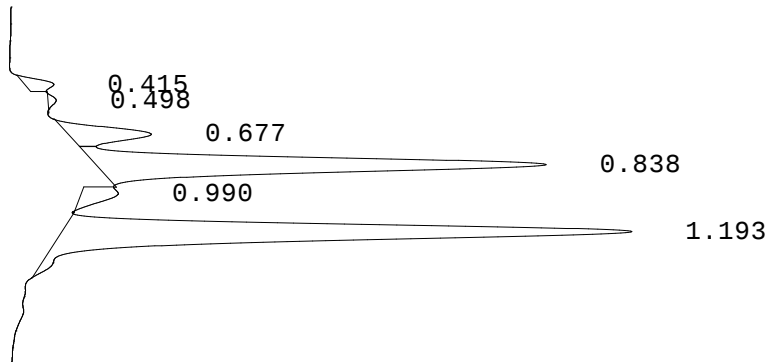
FECHA :

15/07/2026

LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2185, Rack A, Plate 1, Well F06, 173826
[C83CAE0321C5139F] Jun 25, 2026 14:02:00 Pressure = 72 bar (72 to 74)

AF



PEAK	RT	REL	RT	% CONC	AREA	COMMENT
1	0.415	F	0.49	1.6%	8975	
2	0.498	F	0.59	0.5%	2651	
3	0.677	F	0.80	6.5%	35651	Acetylated F peak
4	0.838	F	0.99	37.0%	201827	Consistent with F
5	0.990	F	1.16	3.2%	17494	
6	1.193	A	1.01	51.2%	279455	A peak - REVIEW
Total Area: 546053						

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 262029619

Medical Report

Patient name	HERNANDEZ GONZALEZ VALERY THALIANA
Date of Birth	30.04.2026

Sample-ID	A0358477
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)	3800	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	1.4	μU/mL	< 15.0
Biotinidase	330.3	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	8.2	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	15.2	ng/mL	< 65.0
Phenylalanine	26.7	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	HERNANDEZ GONZALEZ VALERY THALIANA
Date of Birth	30.04.2026

Sample-ID	A0358477
Gender	F

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	26.7	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.41	µmol/L	< 2.20
Tyrosine (Tyr)	65.9	µmol/L	< 200.0
Leucine (Leu)	83.2	µmol/L	< 270.0
Valine (Val)	29.5	µmol/L	< 200.0
Methionine (MET)	17.4	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.65	µmol/L	< 1.60
Citrulline (Cit)	11.3	µmol/L	< 50.0
Ornithine (Orn)	50.8	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	4.50	µmol/L	1.50 - 20.00
Proline (Pro)	79.9	µmol/L	< 350.0
Alanine (Ala)	146.9	µmol/L	< 750.0
Arginine (Arg)	26.4	µmol/L	< 100.0
Aspartic acid (Asp)	38.0	µmol/L	< 100.0
Glutamic acid (Glu)	172.9	µmol/L	< 600.0
Glycamine (Gly)	136.0	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	18.27	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	6.06	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	1.61	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.09	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.12	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.01	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.26	µmol/L	0.02 - 0.57
glutarylacarnitine (C5DC)	0.04	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.03	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.01	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.01	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.07	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.03	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.05	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.04	µmol/L	0.10 - 0.43
palmitoylcarnitine (C16)	0.65	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.40	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	1.39	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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