

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

173430
Gael Mathias Ruge Chiquiza
09/03/2026
ANGIE MARISOL CHIQUIZA ORTIZ
1,070,330,944
15/05/2026
01/06/2026

SEXO : MASCULINO
REGISTRO CIVIL: 1,025,335,709
TIPO DE MUESTRA : TALÓN
PESO : 2395



TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	2.21 µl/mL	>= 6 µl/mL talón en prematuros >= 10 µl/mL talón >= 15 µl/mL cordón	Normal
Deficiencia de G6PDH	6.30 U/gHb	< 2.6 U/gHb	Normal

TÉCNICA: Fluoroimmunoensayo (Delfia).

Procesado en Colombia por PREGEN.

Hemoglobinopatías	FA	Ausencia de hemoglobinas anormales	Normal
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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
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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
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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.

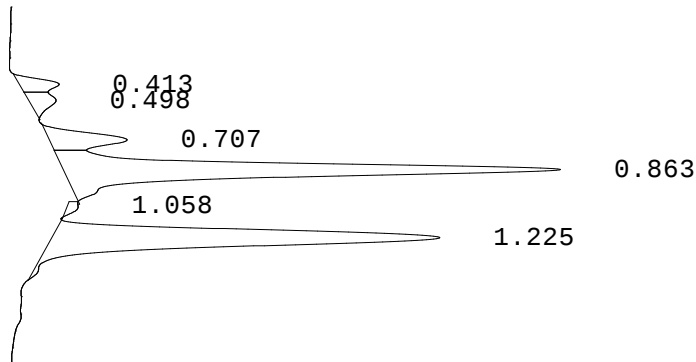
REVISADO :	EDUVILIA JOHANA GOMEZ	PROCESADO :	MARIA JOSE PINZON GARCIA	FECHA :
	Bacterióloga		Bacterióloga	
Reg.	40.936.003	Reg.	1.015.469.392	01/06/2026

LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2149, Rack A, Plate 1, Well E10, 173430
[DD6DE0494B2D999D] May 20, 2026 14:03:53

Pressure = 65 bar (64 to 66)

FA



PEAK	RT	REL	RT	% CONC	AREA	COMMENT
1	0.413	F	0.48	2.9%	13588	
2	0.498	F	0.58	2.8%	13042	
3	0.707	F	0.82	7.1%	33433	Acetylated F peak
4	0.863	F	1.01	45.0%	211450	Consistent with F
5	1.058	A	0.87	0.9%	4290	
6	1.225	A	1.01	41.3%	194058	A peak - REVIEW
Total Area: 469861						

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 29.05.2026
Sample Received 26.05.2026
Date of Sampling 15.05.2026
LAB-ID 262023699

Medical Report

Patient name	RUGE CHIQUIZA GAELE MATHIAS	Sample-ID	A0341547
Date of Birth	09.03.2026	Gender	M

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)	2395	g	-
17-hydroxyprogesterone (17OHP)	6.9	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	1.3	μU/mL	< 15.0
Biotinidase	205.6	U	> 51.0
Galactose-1-P-uridylyltransferase (GALT)	5.9	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	19.1	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	RUGE CHIQUIZA GAEI MATHIAS
Date of Birth	09.03.2026

Sample-ID	A0341547
Gender	M

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	28.2	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.55	µmol/L	< 2.20
Tyrosine (Tyr)	50.9	µmol/L	< 200.0
Leucine (Leu)	112.6	µmol/L	< 270.0
Valine (Val)	38.6	µmol/L	< 200.0
Methionine (MET)	16.6	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.59	µmol/L	< 1.60
Citrulline (Cit)	9.2	µmol/L	< 50.0
Ornithine (Orn)	73.5	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	7.99	µmol/L	1.50 - 20.00
Proline (Pro)	123.3	µmol/L	< 350.0
Alanine (Ala)	162.2	µmol/L	< 750.0
Arginine (Arg)	20.7	µmol/L	< 100.0
Aspartic acid (Asp)	60.1	µmol/L	< 100.0
Glutamic acid (Glu)	202.9	µmol/L	< 600.0
Glycamine (Gly)	207.3	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	39.81	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	19.07	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	1.76	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.11	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)	0.39	µmol/L	0.02 - 0.57
glutaryl carnitine (C5DC)	0.04	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.04	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.03	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.03	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.11	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.06	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.11	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.08	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	0.90	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.03	µmol/L	< 0.10
stearoylcarnitine (C18)	0.42	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	2.51	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.04	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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