



**CODIGO :** 172284  
**NOMBRE PACIENTE :** MARIANA RAMIREZ AMORTEGUI **SEXO :** FEMENINO  
**FECHA DE NACIMIENTO :** 04/01/2026 **REGISTRO CIVIL :** 1,016,757,204  
**NOMBRE RESPONSABLE :** MARLI YAIRA AMORTEGUI MONROY  
**DOC.IDENTIDAD DE LA MADRE :** 1,071,170,487  
**FECHA TOMA DE MUESTRA :** 10/02/2026 **TIPO DE MUESTRA :** TALÓN  
**FECHA DE IMPRESIÓN :** 27/02/2026 **PESO :** 2800

## TAMIZAJE NEONATAL

### ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	3.23	>= 6 µl/mL talón en prematuros >= 10 µl/mL talón >= 15 µl/mL cordón	Normal
Deficiencia de G6PDH	7.30	> 2.6 U/gHb	Normal
<i>TÉCNICA: Fluoroimmunoensayo (Delfia).</i>			<i>Procesado en Colombia por PREGEN.</i>
Hemoglobinopatías	FA	Ausencia de hemoglobinas anormales	Normal
<i>TÉCNICA: Cromatografía Líquida de Alto Rendimiento (HPLC).</i>			<i>Procesado en Colombia por PREGEN.</i>

## 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,C50H,C3,C5:1

Ausencia de metabolitos anormales Normal

#### RESULTADOS NORMALES

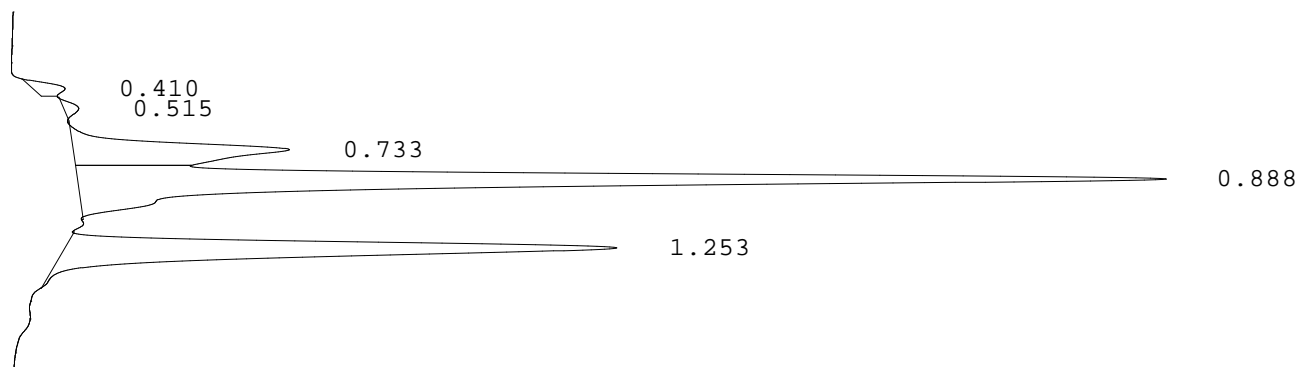
Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta 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 27/02/2026

LABORATORIO PREGEN  
Carrera 15a No 106-42  
BOGOTA

Batch 2073, Rack A, Plate 1, Well E05, 172284  
[CD32650E618C0CE2] Feb 13, 2026 14:18:54 Pressure = 51 bar (51 to 52)

FA



PEAK	RT	REL RT	% CONC	AREA	COMMENT
1	0.410	F 0.46	1.3%	10753	
2	0.515	F 0.58	0.7%	5804	
3	0.733	F 0.83	14.2%	118226	Acetylated F peak
4	0.888	F 1.00	54.7%	456644	Consistent with F
5	1.253	A 1.00	29.2%	243674	A peak - REVIEW
Total Area:				835101	

- Codes:
- 1) Wide A peak
  - 2) Area of A peak < 80%
  - 3) Peak area greater than expected
  - 4) Peak after A2
  - 5) Alc > 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 27.02.2026  
Sample Received 19.02.2026  
Date of Sampling 10.02.2026  
LAB-ID 262006998

## Medical Report

Patient name	<b>RAMIREZ AMORTEGUI MARIANA</b>	Sample-ID	A0361371
Date of Birth	<b>04.01.2026</b>	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)	2800	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	1.6	μU/mL	< 15.0
Biotinidase	299.2	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	10.8	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	17.0	ng/mL	< 65.0
Phenylalanine	32.2	μmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

**Interpretation:** NEGATIVE RESULT

Patient name	<b>RAMIREZ AMORTEGUI MARIANA</b>
Date of Birth	<b>04.01.2026</b>

Sample-ID	A0361371
Gender	F

## Results:

### Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	32.2	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.42	µmol/L	< 2.20
Tyrosine (Tyr)	77.4	µmol/L	< 200.0
Leucine (Leu)	126.0	µmol/L	< 270.0
Valine (Val)	61.7	µmol/L	< 200.0
Methionine (MET)	31.8	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.99	µmol/L	< 1.60
Citrulline (Cit)	17.0	µmol/L	< 50.0
Ornithine (Orn)	162.3	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	9.55	µmol/L	1.50 - 20.00
Proline (Pro)	122.4	µmol/L	< 350.0
Alanine (Ala)	171.5	µmol/L	< 750.0
Arginine (Arg)	34.6	µmol/L	< 100.0
Aspartic acid (Asp)	107.2	µmol/L	< 100.0
Glutamic acid (Glu)	301.8	µmol/L	< 600.0
Glycamine (Gly)	154.1	µmol/L	< 700.0

### Acylcarnitines

Free carnitine (C0)	15.74	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	9.78	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)	0.13	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.01	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.16	µmol/L	0.02 - 0.57
glutarylacetyl carnitine (C5DC)	0.03	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)	0.10	µmol/L	< 0.30
decadienoylcarnitine (C10:2)	0.03	µmol/L	< 0.10
dodecanoylcarnitine (C12)	0.04	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.05	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.09	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	0.53	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.23	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	1.79	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.02	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.

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