CODIGO : 173510

NOMBRE PACIENTE : EMILIA ROCHA TORRES **SEXO :** FEMENINO

FECHA DE NACIMIENTO : 19/04/2026 **REGISTRO CIVIL:** 1,233,937,357

NOMBRE RESPONSABLE : JESICA PAOLA ROCHA TORRES

DOC.IDENTIDAD DE LA MADRE : 1,019,088,541

FECHA TOMA DE MUESTRA : 22/05/2026 **TIPO DE MUESTRA :** TALÓN

FECHA DE IMPRESIÓN : 16/07/2026 **PESO :** 2690

TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	0.39 µUl/mL	>= 6 µUl/mL talón en prematuros >= 10 µUl/mL talón >= 15 µUl/mL cordón	Normal
Deficiencia de G6PDH	6.40 U/gHb	< 2.6 U/gHb	Normal

TÉCNICA: Fluoroinmunoensayo (Delfia). *Procesado en Colombia por PREGEN.*

Hemoglobinopatías	FAC	Cromatograma compatible con posible rasgo	Anormal	*

TÉCNICA: Cromatografía Liquida de Alto Rendimiento (HPLC). *Procesado en Colombia por PREGEN.*

** DETECCION DE POSIBLE VARIANTE DE HEMOGLOBINA C*

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

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 : 16/07/2026

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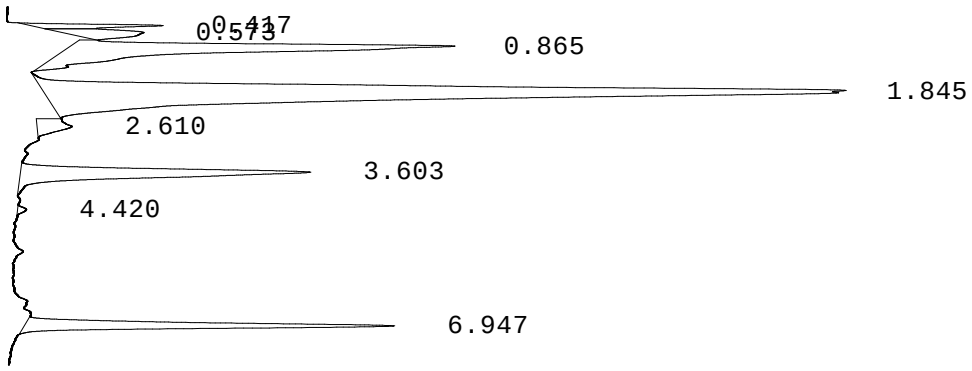
`LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2157, Rack A, Plate 1, Well E02, 173510 FAC
[8C28A3D81FF84C42] May 26, 2026 16:13:54 Pressure = 66 bar (63 to 67)



PEAK	RT	REL RT	% CONC	AREA	COMMENT
1	0.417	F 0.23	2.1%	32843	
2	0.573	F 0.31	2.8%	44497	
3	0.865	F 0.47	17.7%	277736	
4	1.845	F 1.00	52.9%	829351	Consistent with F
5	2.610	F 1.42	1.8%	28645	
6	3.603	A 0.99	12.0%	188775	A peak
7	4.420	S 0.85	0.2%	3369	
8	6.947	C 1.00	10.3%	161912	Consistent with C
			Total Area:	1567128	**A/V=1.17**

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

baby health check©

+ARCHIMEDlife Laboratory has been designated as a medical laboratory with the identification number 0407 from Accreditation Austria/Federal Ministry for Digitization and Business Location for the areas listed and published at: www.bmaw.gv.at/akkreditierung.

Dr. MARIA JOSE PINZON GARCIA
RED COLOMBIANA DE MEDICINA GENETICA SAS - PREGEN
BOGOTA
CARRERA 15 A # 106 - 42
11001 BOGOTA
Colombia

Date of Report	08.06.2026
Sample Received	29.05.2026
Date of Sampling	22.05.2026
LAB-ID	262023981

Medical Report

Patient name	**ROCHA TORRES EMILIA**	Sample-ID	A0341537
Date of Birth	**19.04.2026**	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)	2690	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	<0.7	µU/mL	< 15.0
Biotinidase	333.4	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	7.5	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	37.4	µmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

Interpretation: NEGATIVE RESULT

Contact Details
Assoc.-Prof. Dr. Andrea-Romana KASPER, MD, PhD
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International Medical Laboratory+
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Patient name	**ROCHA TORRES EMILIA**	Sample-ID	A0341537
Date of Birth	**19.04.2026**	Gender	F

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	37.4	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.41	µmol/L	< 2.20
Tyrosine (Tyr)	92.2	µmol/L	< 200.0
Leucine (Leu)	164.6	µmol/L	< 270.0
Valine (Val)	65.6	µmol/L	< 200.0
Methionine (MET)	21.4	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.57	µmol/L	< 1.60
Citrulline (Cit)	18.5	µmol/L	< 50.0
Ornithine (Orn)	73.6	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	3.98	µmol/L	1.50 - 20.00
Proline (Pro)	113.5	µmol/L	< 350.0
Alanine (Ala)	129.1	µmol/L	< 750.0
Arginine (Arg)	26.2	µmol/L	< 100.0
Aspartic acid (Asp)	72.7	µmol/L	< 100.0
Glutamic acid (Glu)	348.2	µmol/L	< 600.0
Glycamine (Gly)	112.8	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	11.51	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	6.48	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	0.75	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.12	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.15	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.02	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.37	µmol/L	0.02 - 0.57
glutarylcarnitine (C5DC)	0.07	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)	0.09	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.05	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.05	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.06	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	0.34	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.18	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	0.73	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.06	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, methylglutaconiciduria, 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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