



**CODIGO :** 173059  
**NOMBRE PACIENTE :** ANTONELLA RODRIGUEZ ABRIL **SEXO :** FEMENINO  
**FECHA DE NACIMIENTO :** 16/01/2026 **REGISTRO CIVIL :** 1,030,719,772  
**NOMBRE RESPONSABLE :** LEIDY JOHANA RODRIGUEZ ABRIL  
**DOC.IDENTIDAD DE LA MADRE :** 1,014,250,618  
**FECHA TOMA DE MUESTRA :** 14/04/2026 **TIPO DE MUESTRA :** TALÓN  
**FECHA DE IMPRESIÓN :** 09/05/2026 **PESO :** 3000

## TAMIZAJE NEONATAL

### ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	0.32	>= 6 µl/mL talón en prematuros >= 10 µl/mL talón >= 15 µl/mL cordón	Normal
Deficiencia de G6PDH	5.70	> 2.6 U/gHb	Normal
<i>TÉCNICA: Fluoroimmunoensayo (Delfia).</i>			<i>Procesado en Colombia por PREGEN.</i>
Hemoglobinopatías	AF	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 Tennessee Department of Health.

#### DESORDENES DE AMINOÁCIDOS

Citrulina, Metionina, Leucina, Isoleucina, Valina, Fenilalanina, Tirosina.

Ausencia de metabolitos anormales Normal

#### DESORDENES DE LA OXIDACIÓN DE ÁCIDOS GRASOS

C16,C18,C18:1,C16OH,C18:1OH,C8,C10:1,C5,C5DC,C4,C14,C14:1

Ausencia de metabolitos anormales Normal

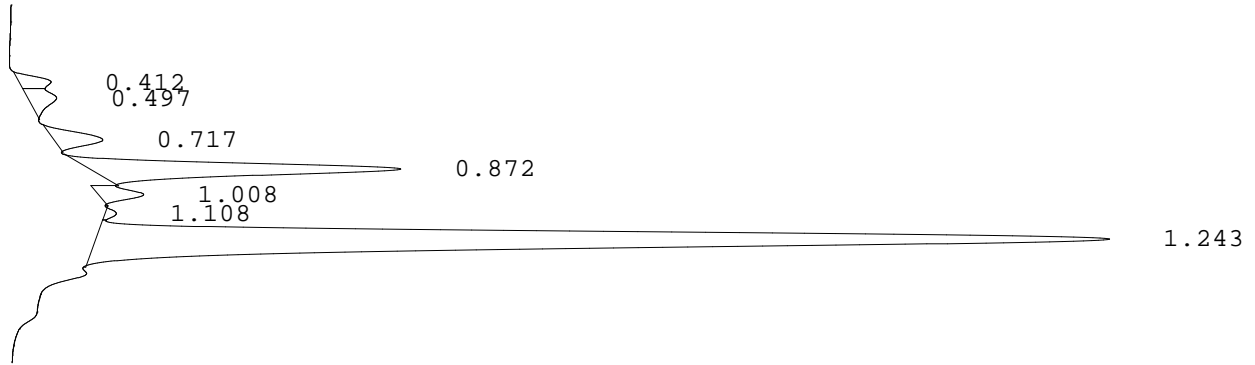
Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta la edad del paciente, por esto es conveniente realizarlas dentro del primer mes de nacido.

**REVISADO :** EDUVILIA JOHANA GOMEZ **PROCESADO :** MARIA JOSE PINZON GARCIA **FECHA :**  
Bacterióloga Bacterióloga  
Reg. 40.936.003 Reg. 1.015.469.392 09/05/2026

LABORATORIO PREGEN  
Carrera 15a No 106-42  
BOGOTA

Batch 2124, Rack A, Plate 1, Well H11, 173059  
[DC735DA3079CACC8] Apr 16, 2026 13:26:33 Pressure = 62 bar (62 to 63)

AF



PEAK	RT	REL RT	% CONC	AREA	COMMENT
1	0.412	F 0.47	1.5%	10254	
2	0.497	F 0.57	2.2%	14779	
3	0.717	F 0.83	3.0%	20424	
4	0.872	F 1.01	16.8%	113727	Consistent with F
5	1.008	F 1.16	2.4%	16436	
6	1.108	A 0.89	0.5%	3593	
7	1.243	A 1.00	73.6%	498341	A peak - REVIEW
Total Area:				677554	

- 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 06.05.2026  
Sample Received 23.04.2026  
Date of Sampling 14.04.2026  
LAB-ID 262017948

## Medical Report

Patient name	<b>RODRIGUEZ ABRIL ANTONELLA</b>	Sample-ID	A0341227
Date of Birth	<b>16.01.2026</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)	3000	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	<0.7	μU/mL	< 15.0
Biotinidase	254.3	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	7.4	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	34.6	μmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

**Interpretation:** NEGATIVE RESULT

Patient name	<b>RODRIGUEZ ABRIL ANTONELLA</b>
Date of Birth	<b>16.01.2026</b>

Sample-ID	A0341227
Gender	F

## Results:

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	34.6	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.64	µmol/L	< 2.20
Tyrosine (Tyr)	54.4	µmol/L	< 200.0
Leucine (Leu)	90.2	µmol/L	< 270.0
Valine (Val)	49.2	µmol/L	< 200.0
Methionine (MET)	12.6	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.36	µmol/L	< 1.60
Citrulline (Cit)	15.8	µmol/L	< 50.0
Ornithine (Orn)	75.4	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	4.77	µmol/L	1.50 - 20.00
Proline (Pro)	74.9	µmol/L	< 350.0
Alanine (Ala)	104.8	µmol/L	< 750.0
Arginine (Arg)	23.8	µmol/L	< 100.0
Aspartic acid (Asp)	87.5	µmol/L	< 100.0
Glutamic acid (Glu)	186.2	µmol/L	< 600.0
Glycamine (Gly)	134.9	µmol/L	< 700.0
Free carnitine (C0)	26.04	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	14.23	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	1.64	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.16	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.17	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.02	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.34	µmol/L	0.02 - 0.57
glutaryl carnitine (C5DC)	0.05	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.05	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.05	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.05	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.11	µmol/L	< 0.30
decadienoylcarnitine (C10:2)	0.12	µmol/L	< 0.10
dodecanoylcarnitine (C12)	0.06	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.15	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.08	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	1.06	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.03	µmol/L	< 0.10
stearoylcarnitine (C18)	0.44	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	2.54	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.06	µmol/L	< 0.50

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.