

**CODIGO :** 173579  
**NOMBRE PACIENTE :** GAEL ANDRES RUIZ BOYACA **SEXO :** MASCULINO  
**FECHA DE NACIMIENTO :** 06/03/2026 **REGISTRO CIVIL:** 1,070,998,464  
**NOMBRE RESPONSABLE :** ANDREA STEFANNYA BOYACA MENDEZ  
**DOC.IDENTIDAD DE LA MADRE :** 1,070,961,585  
**FECHA TOMA DE MUESTRA :** 28/05/2026 **TIPO DE MUESTRA :** TALÓN  
**FECHA DE IMPRESIÓN :** 24/06/2026 **PESO :** 2340



## TAMIZAJE NEONATAL

### ANÁLISIS MUESTRA DE SANGRE

	RESULTADO	VALORES DE REFERENCIA	INTERPRETACIÓN
T.S.H Neonatal	2.97 µl/mL	>= 6 µl/mL talón en prematuros >= 10 µl/mL talón >= 15 µl/mL cordón	Normal
Deficiencia de G6PDH	6.50 U/gHb	< 2.6 U/gHb	Normal

*TÉCNICA: Fluoroimmunoensayo (Delfia).*

*Procesado en Colombia por PREGEN.*

Hemoglobinopatías FAS Cromatograma compatible con posible rasgo Anormal \*

*TÉCNICA: Cromatografía Líquida de Alto Rendimiento (HPLC).*

*Procesado en Colombia por PREGEN.*

\* DETECCIÓN DE POSIBLE VARIANTE DE HEMOGLOBINA S

## 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

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.

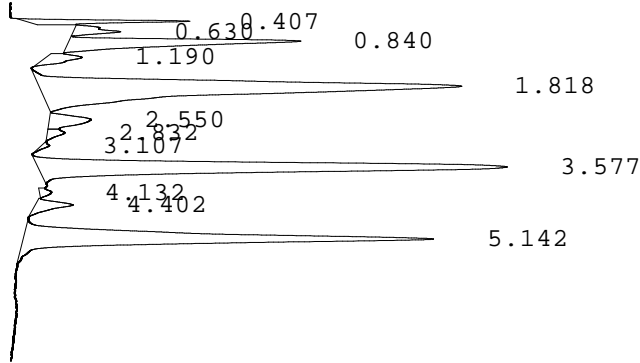
**REVISADO :** EDUVILIA JOHANA GOMEZ **PROCESADO :** MARIA JOSE PINZON GARCIA **FECHA :**  
 Bacterióloga Bacterióloga  
 Reg. 40.936.003 Reg. 1.015.469.392 24/06/2026

LABORATORIO PREGEN  
 Carrera 15a No 106-42  
 BOGOTA

Batch 2167, Rack A, Plate 1, Well B08, 173579  
 [DC6DA70F1C0F391D] Jun 02, 2026 12:18:09

Pressure = 65 bar (63 to 66)

FAS



PEAK	RT	REL RT	% CONC	AREA	COMMENT
1	0.407	F 0.22	2.9%	38416	
2	0.630	F 0.34	1.5%	19714	
3	0.840	F 0.46	8.8%	118261	
4	1.190	F 0.64	1.6%	20927	
5	1.818	F 0.99	31.9%	426733	Consistent with F
6	2.550	F 1.38	2.3%	30319	
7	2.832	A 0.79	0.6%	8667	
8	3.107	A 0.87	0.2%	3068	
9	3.577	A 1.00	26.1%	349293	A peak - REVIEW
10	4.132	A 1.16	0.4%	5503	
11	4.402	S 0.86	1.6%	21634	3
12	5.142	S 1.00	22.1%	295906	Consistent with S
Total Area: 1338441					<b>A/V=1.18</b>

- 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 17.06.2026  
Sample Received 09.06.2026  
Date of Sampling 28.05.2026  
LAB-ID 262025012

## Medical Report

Patient name	<b>RUIZ BOYACA GAEL ANDRES</b>	Sample-ID	A0358658
Date of Birth	<b>06.03.2026</b>	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)	2340	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	2.3	µU/mL	< 15.0
Biotinidase	371.5	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	9.1	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	54.5	ng/mL	< 65.0
Phenylalanine	30.4	µmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

**Interpretation:** NEGATIVE RESULT

Patient name	<b>RUIZ BOYACA GAEL ANDRES</b>
Date of Birth	<b>06.03.2026</b>

Sample-ID	A0358658
Gender	M

## Results:

### Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	30.4	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.65	µmol/L	< 2.20
Tyrosine (Tyr)	46.5	µmol/L	< 200.0
Leucine (Leu)	121.9	µmol/L	< 270.0
Valine (Val)	43.9	µmol/L	< 200.0
Methionine (MET)	16.5	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.54	µmol/L	< 1.60
Citrulline (Cit)	10.4	µmol/L	< 50.0
Ornithine (Orn)	75.6	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	7.27	µmol/L	1.50 - 20.00
Proline (Pro)	98.2	µmol/L	< 350.0
Alanine (Ala)	149.1	µmol/L	< 750.0
Arginine (Arg)	38.2	µmol/L	< 100.0
Aspartic acid (Asp)	58.4	µmol/L	< 100.0
Glutamic acid (Glu)	241.3	µmol/L	< 600.0
Glycamine (Gly)	179.2	µmol/L	< 700.0

### Acylcarnitines

Free carnitine (C0)	42.34	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	16.55	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	1.88	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.15	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)	0.33	µmol/L	0.02 - 0.57
glutaryl carnitine (C5DC)	0.05	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.06	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)	0.13	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.09	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.24	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.10	µmol/L	0.10 - 0.43
palmitoylcarnitine (C16)	1.13	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.03	µmol/L	< 0.10
stearoylcarnitine (C18)	0.72	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	4.37	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, 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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