



CODIGO : 173248
NOMBRE PACIENTE : MIA ISABELLA BOHORQUEZ TOLEDO **SEXO :** FEMENINO
FECHA DE NACIMIENTO : 01/04/2026 **REGISTRO CIVIL :** 1,031,191,211
NOMBRE RESPONSABLE : KAREN DAYANA BOHORQUEZ TOLEDO
DOC.IDENTIDAD DE LA MADRE : 1,026,592,664
FECHA TOMA DE MUESTRA : 30/04/2026 **TIPO DE MUESTRA :** TALÓN
FECHA DE IMPRESIÓN : 21/05/2026 **PESO :** 3520

TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

| | RESULTADO | VALORES DE REFERENCIA | INTERPRETACIÓN |
|---|-----------|---|--|
| T.S.H Neonatal | 1.05 | >= 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 | 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

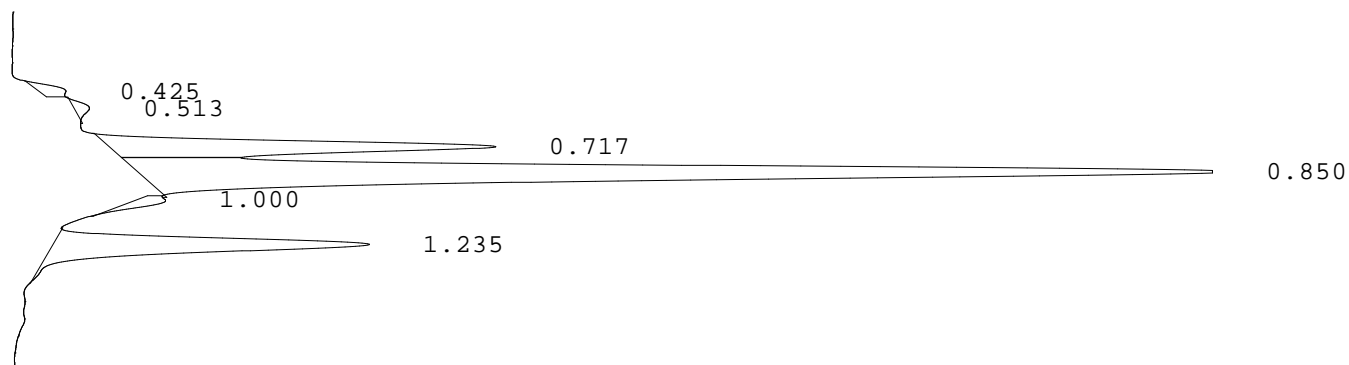
Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta 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 21/05/2026

LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

Batch 2140, Rack A, Plate 1, Well B11, 173248
[9C675A2E643626EA] May 06, 2026 12:12:12 Pressure = 64 bar (64 to 66)

FA



| PEAK | RT | REL RT | % CONC | AREA | COMMENT |
|-------------|-------|--------|--------|--------|-------------------|
| 1 | 0.425 | F 0.50 | 1.1% | 8985 | |
| 2 | 0.513 | F 0.60 | 0.8% | 6419 | |
| 3 | 0.717 | F 0.84 | 17.9% | 142977 | Acetylated F peak |
| 4 | 0.850 | F 1.00 | 61.4% | 490337 | Consistent with F |
| 5 | 1.000 | F 1.17 | 1.6% | 12525 | |
| 6 | 1.235 | A 1.02 | 17.2% | 136905 | A peak |
| Total Area: | | | | 798148 | |

- 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 17.05.2026
Sample Received 11.05.2026
Date of Sampling 30.04.2026
LAB-ID 262020463

Medical Report

| | | | |
|---------------|--------------------------------------|-----------|----------|
| Patient name | BOHORQUEZ TOLEDO MIA ISABELLA | Sample-ID | A0341479 |
| Date of Birth | 01.04.2026 | 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) | 3520 | g | - |
| 17-hydroxyprogesterone (17OHP) | <5.0 | nmol/L | < 90.0 |
| Thyroid-stimulating hormone (TSH) | <0.7 | μU/mL | < 15.0 |
| Biotinidase | 192.2 | U | > 51.0 |
| Galactose-1-P-uridyltransferase (GALT) | 8.7 | U/g Hb | > 2.5 |
| Immunoreactive trypsinogen (IRT) | <15 | ng/mL | < 65.0 |
| Phenylalanine | 30.9 | μmol/L | < 150.0 |
| Amino acid profile | negative | | - |
| Acylcarnitine profile | negative | | - |

Interpretation: NEGATIVE RESULT

| | |
|---------------|--------------------------------------|
| Patient name | BOHORQUEZ TOLEDO MIA ISABELLA |
| Date of Birth | 01.04.2026 |

| | |
|-----------|----------|
| Sample-ID | A0341479 |
| Gender | F |

Results:

Amino Acids

| Parameter | Value | Unit | Reference |
|--|-------|--------|--------------|
| Phenylalanine (Phe) | 30.9 | µmol/L | < 150.0 |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.63 | µmol/L | < 2.20 |
| Tyrosine (Tyr) | 49.4 | µmol/L | < 200.0 |
| Leucine (Leu) | 111.1 | µmol/L | < 270.0 |
| Valine (Val) | 45.4 | µmol/L | < 200.0 |
| Methionine (MET) | 21.2 | µmol/L | < 78.0 |
| Methionine / Phenylalanine (Met/Phe) | 0.69 | µmol/L | < 1.60 |
| Citrulline (Cit) | 20.8 | µmol/L | < 50.0 |
| Ornithine (Orn) | 92.7 | µmol/L | < 250.0 |
| Ornithine / Citrulline ratio (Orn/Cit) | 4.46 | µmol/L | 1.50 - 20.00 |
| Proline (Pro) | 115.4 | µmol/L | < 350.0 |
| Alanine (Ala) | 142.8 | µmol/L | < 750.0 |
| Arginine (Arg) | 12.2 | µmol/L | < 100.0 |
| Aspartic acid (Asp) | 103.4 | µmol/L | < 100.0 |
| Glutamic acid (Glu) | 391.0 | µmol/L | < 600.0 |
| Glycamine (Gly) | 194.0 | µmol/L | < 700.0 |

Acylcarnitines

| | | | |
|--|-------|--------|---------------|
| Free carnitine (C0) | 13.73 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2) | 9.43 | µmol/L | 1.34 - 48.81 |
| propionylcarnitine (C3) | 0.29 | µmol/L | 0.13 - 6.60 |
| butyryl-/isobutyrylcarnitine (C4) | 0.08 | µmol/L | 0.03 - 0.90 |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.16 | µmol/L | 0.02 - 2.00 |
| tiglylcarnitine (C5:1) | 0.01 | µmol/L | < 0.20 |
| hydroxyvalerylcarnitine (C5OH) | 0.32 | µmol/L | 0.02 - 0.57 |
| glutarylacetyl carnitine (C5DC) | 0.16 | µmol/L | < 0.30 |
| hexanoylcarnitine (C6) | 0.04 | µmol/L | 0.01 - 0.13 |
| octanoylcarnitine (C8) | 0.02 | µmol/L | 0.01 - 0.30 |
| decanoylcarnitine (C10) | 0.13 | µmol/L | 0.01 - 0.36 |
| decenoylcarnitine (C10:1) | 0.02 | µmol/L | < 0.30 |
| decadienoylcarnitine (C10:2) | 0.08 | µmol/L | < 0.10 |
| dodecanoylcarnitine (C12) | 0.12 | µmol/L | 0.10 - 0.60 |
| myristoylcarnitine (C14) | 0.02 | µmol/L | 0.01 - 0.57 |
| tetradecenoylcarnitine (C14:1) | 0.04 | µmol/L | 0.10 - 0.38 |
| palmitoylcarnitine (C16) | 0.49 | µmol/L | 0.62 - 7.81 |
| 3-hydroxypalmitoylcarnitine (C16OH) | 0.02 | µmol/L | < 0.10 |
| stearoylcarnitine (C18) | 0.02 | µmol/L | 0.30 - 2.40 |
| oleylcarnitine (C18:1) | 0.30 | µmol/L | 0.06 - 3.86 |
| 3-hydroxystearoylcarnitine (C18OH) | 0.02 | µmol/L | < 0.09 |
| malonylcarnitine (C3DC) | 0.01 | 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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