



**CODIGO :** 173129  
**NOMBRE PACIENTE :** GEILANY CAROLINA DIAZ SANCHEZ **SEXO :** FEMENINO  
**FECHA DE NACIMIENTO :** 03/03/2026 **REGISTRO CIVIL :** 1,026,311,874  
**NOMBRE RESPONSABLE :** GENESIS DEL VALLE DIAZ SANCHEZ  
**DOC.IDENTIDAD DE LA MADRE :** 22,136,568  
**FECHA TOMA DE MUESTRA :** 20/04/2026 **TIPO DE MUESTRA :** TALÓN  
**FECHA DE IMPRESIÓN :** 07/05/2026 **PESO :** 2750

## TAMIZAJE NEONATAL

### ANÁLISIS MUESTRA DE SANGRE

|   | RESULTADO | VALORES DE REFERENCIA   | INTERPRETACIÓN                           |
|---|-----------|---|--|
| T.S.H Neonatal  | 1.96      | >= 6 µl/mL talón en prematuros<br>>= 10 µl/mL talón<br>>= 15 µl/mL cordón | Normal                                   |
| Deficiencia de G6PDH  | 6.20      | > 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.

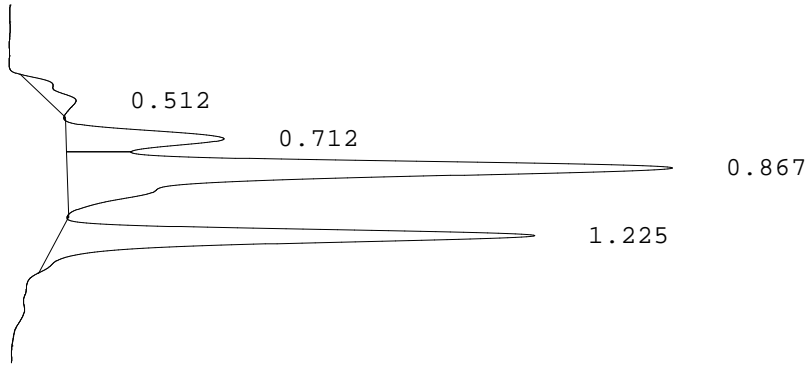
**REVISADO :** EDUVILIA JOHANA GOMEZ **PROCESADO :** MARIA JOSE PINZON GARCIA **FECHA :**  
Bacterióloga Bacterióloga  
Reg. 40.936.003 Reg. 1.015.469.392 07/05/2026

LABORATORIO PREGEN  
Carrera 15a No 106-42  
BOGOTA

Batch 2130, Rack A, Plate 1, Well A11, 173129  
[C8671C3F9996A648] Apr 24, 2026 11:31:29

Pressure = 61 bar (61 to 63)

FA



| PEAK        | RT    | REL RT | % CONC | AREA   | COMMENT           |
|-------------|-------|--------|--------|--------|-------------------|
| 1           | 0.512 | F 0.59 | 3.3%   | 21592  |                   |
| 2           | 0.712 | F 0.83 | 12.3%  | 79926  | Acetylated F peak |
| 3           | 0.867 | F 1.01 | 52.6%  | 343107 | Consistent with F |
| 4           | 1.225 | A 1.00 | 31.8%  | 207155 | A peak - REVIEW   |
| Total Area: |       |        |        | 651780 |                   |

- 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 28.04.2026  
Date of Sampling 20.04.2026  
LAB-ID 262018938

## Medical Report

|               |                                      |           |          |
|---------------|--------------------------------------|-----------|----------|
| Patient name  | <b>DIAZ SANCHEZ GEILANY CAROLINA</b> | Sample-ID | A0340900 |
| Date of Birth | <b>03.03.2026</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)                       | 2750     | g      | -         |
| 17-hydroxyprogesterone (17OHP)         | <5.0     | nmol/L | < 90.0    |
| Thyroid-stimulating hormone (TSH)      | 1.6      | µU/mL  | < 15.0    |
| Biotinidase                            | 291.3    | U      | > 51.0    |
| Galactose-1-P-uridyltransferase (GALT) | 9.6      | U/g Hb | > 2.5     |
| Immunoreactive trypsinogen (IRT)       | <15      | ng/mL  | < 65.0    |
| Phenylalanine                          | 27.0     | µmol/L | < 150.0   |
| Amino acid profile                     | negative |        | -         |
| Acylcarnitine profile                  | negative |        | -         |

**Interpretation:** NEGATIVE RESULT

|               |                                      |
|---------------|--------------------------------------|
| Patient name  | <b>DIAZ SANCHEZ GEILANY CAROLINA</b> |
| Date of Birth | <b>03.03.2026</b>                    |

|           |          |
|-----------|----------|
| Sample-ID | A0340900 |
| Gender    | F        |

## Results:

### Amino Acids

| Parameter                                | Value | Unit   | Reference    |
|--|-------|--------|--------------|
| Phenylalanine (Phe)                      | 27.0  | µmol/L | < 150.0      |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.48  | µmol/L | < 2.20       |
| Tyrosine (Tyr)                           | 56.4  | µmol/L | < 200.0      |
| Leucine (Leu)                            | 78.6  | µmol/L | < 270.0      |
| Valine (Val)                             | 50.5  | µmol/L | < 200.0      |
| Methionine (MET)                         | 15.2  | µmol/L | < 78.0       |
| Methionine / Phenylalanine (Met/Phe)     | 0.56  | µmol/L | < 1.60       |
| Citrulline (Cit)                         | 10.9  | µmol/L | < 50.0       |
| Ornithine (Orn)                          | 72.1  | µmol/L | < 250.0      |
| Ornithine / Citrulline ratio (Orn/Cit)   | 6.61  | µmol/L | 1.50 - 20.00 |
| Proline (Pro)                            | 86.3  | µmol/L | < 350.0      |
| Alanine (Ala)                            | 133.0 | µmol/L | < 750.0      |
| Arginine (Arg)                           | 8.7   | µmol/L | < 100.0      |
| Aspartic acid (Asp)                      | 42.3  | µmol/L | < 100.0      |
| Glutamic acid (Glu)                      | 218.6 | µmol/L | < 600.0      |
| Glycamine (Gly)                          | 124.0 | µmol/L | < 700.0      |

### Acylcarnitines

|  |       |        |               |
|--|-------|--------|---------------|
| Free carnitine (C0)                      | 20.55 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2)                     | 9.32  | µmol/L | 1.34 - 48.81  |
| propionylcarnitine (C3)                  | 1.07  | µmol/L | 0.13 - 6.60   |
| butyryl-/isobutyrylcarnitine (C4)        | 0.14  | µmol/L | 0.03 - 0.90   |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.15  | µmol/L | 0.02 - 2.00   |
| tiglylcarnitine (C5:1)                   | 0.01  | µmol/L | < 0.20        |
| hydroxyvalerylcarnitine (C5OH)           | 0.34  | µmol/L | 0.02 - 0.57   |
| glutarylacetyl carnitine (C5DC)          | 0.03  | µmol/L | < 0.30        |
| hexanoylcarnitine (C6)                   | 0.03  | µmol/L | 0.01 - 0.13   |
| octanoylcarnitine (C8)                   | 0.02  | µmol/L | 0.01 - 0.30   |
| decanoylcarnitine (C10)                  | 0.02  | µmol/L | 0.01 - 0.36   |
| decenoylcarnitine (C10:1)                | 0.07  | µmol/L | < 0.30        |
| decadienoylcarnitine (C10:2)             | 0.14  | µmol/L | < 0.10        |
| dodecanoylcarnitine (C12)                | 0.03  | µmol/L | 0.10 - 0.60   |
| myristoylcarnitine (C14)                 | 0.07  | µmol/L | 0.01 - 0.57   |
| tetradecenoylcarnitine (C14:1)           | 0.04  | µmol/L | 0.10 - 0.38   |
| palmitoylcarnitine (C16)                 | 0.57  | µmol/L | 0.62 - 7.81   |
| 3-hydroxypalmitoylcarnitine (C16OH)      | 0.02  | µmol/L | < 0.10        |
| stearoylcarnitine (C18)                  | 0.29  | µmol/L | 0.30 - 2.40   |
| oleylcarnitine (C18:1)                   | 0.98  | 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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