



**CODIGO :** 172965  
**NOMBRE PACIENTE :** SARAH GABRIELA DIAZ MORENO **SEXO :** FEMENINO  
**FECHA DE NACIMIENTO :** 21/02/2026 **REGISTRO CIVIL :** 1,021,516,140  
**NOMBRE RESPONSABLE :** ZOILA ROSA MORENO ALFONSO  
**DOC.IDENTIDAD DE LA MADRE :** 1,051,286,069  
**FECHA TOMA DE MUESTRA :** 07/04/2026 **TIPO DE MUESTRA :** TALÓN  
**FECHA DE IMPRESIÓN :** 25/04/2026 **PESO :** 2780

## TAMIZAJE NEONATAL

### ANÁLISIS MUESTRA DE SANGRE

|   | RESULTADO | VALORES DE REFERENCIA   | INTERPRETACIÓN                           |
|---|-----------|---|--|
| T.S.H Neonatal  | 1.67      | >= 6 µl/mL talón en prematuros<br>>= 10 µl/mL talón<br>>= 15 µl/mL cordón | Normal                                   |
| Deficiencia de G6PDH  | 7.00      | > 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

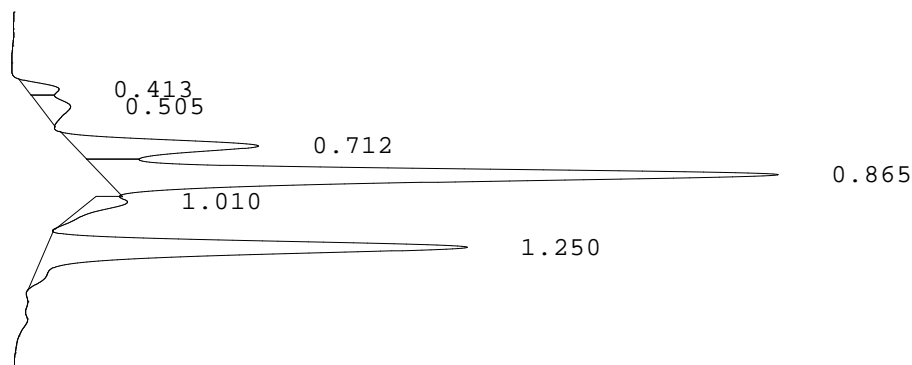
Recuerde que estas son pruebas de tamizaje que solo indican la probabilidad de que el recién nacido tenga una de las enfermedades estudiadas por el programa y pueden requerir pruebas adicionales para la confirmación de algún diagnóstico. La sensibilidad de estas pruebas se reduce a medida que aumenta 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 25/04/2026

LABORATORIO PREGEN  
Carrera 15a No 106-42  
BOGOTA

Batch 2121, Rack A, Plate 1, Well D06, 172965  
[9D220303035E046A] Apr 13, 2026 11:56:20 Pressure = 58 bar (57 to 59)

FA



| PEAK        | RT    | REL RT | % CONC | AREA   | COMMENT           |
|-------------|-------|--------|--------|--------|-------------------|
| 1           | 0.413 | F 0.47 | 1.8%   | 10412  |                   |
| 2           | 0.505 | F 0.58 | 3.0%   | 17634  |                   |
| 3           | 0.712 | F 0.81 | 13.2%  | 76461  | Acetylated F peak |
| 4           | 0.865 | F 0.99 | 47.8%  | 277305 | Consistent with F |
| 5           | 1.010 | F 1.16 | 3.2%   | 18573  |                   |
| 6           | 1.250 | A 1.01 | 31.0%  | 179844 | A peak - REVIEW   |
| Total Area: |       |        |        | 580229 |                   |

- 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 24.04.2026  
Sample Received 17.04.2026  
Date of Sampling 07.04.2026  
LAB-ID 262017194

## Medical Report

|               |                                   |           |          |
|---------------|-----------------------------------|-----------|----------|
| Patient name  | <b>DIAZ MORENO SARAH GABRIELA</b> | Sample-ID | A0361572 |
| Date of Birth | <b>21.02.2026</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)                       | 2780     | g      | -         |
| 17-hydroxyprogesterone (17OHP)         | <5.0     | nmol/L | < 90.0    |
| Thyroid-stimulating hormone (TSH)      | <0.7     | µU/mL  | < 15.0    |
| Biotinidase                            | 283.2    | U      | > 51.0    |
| Galactose-1-P-uridyltransferase (GALT) | 9.0      | U/g Hb | > 2.5     |
| Immunoreactive trypsinogen (IRT)       | <15      | ng/mL  | < 65.0    |
| Phenylalanine                          | 25.7     | µmol/L | < 150.0   |
| Amino acid profile                     | negative |        | -         |
| Acylcarnitine profile                  | negative |        | -         |

**Interpretation:** NEGATIVE RESULT

|               |                                   |
|---------------|-----------------------------------|
| Patient name  | <b>DIAZ MORENO SARAH GABRIELA</b> |
| Date of Birth | <b>21.02.2026</b>                 |

|           |          |
|-----------|----------|
| Sample-ID | A0361572 |
| Gender    | F        |

## Results:

### Amino Acids

| Parameter                                | Value | Unit   | Reference    |
|--|-------|--------|--------------|
| Phenylalanine (Phe)                      | 25.7  | µmol/L | < 150.0      |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.63  | µmol/L | < 2.20       |
| Tyrosine (Tyr)                           | 40.6  | µmol/L | < 200.0      |
| Leucine (Leu)                            | 79.1  | µmol/L | < 270.0      |
| Valine (Val)                             | 30.8  | µmol/L | < 200.0      |
| Methionine (MET)                         | 17.8  | µmol/L | < 78.0       |
| Methionine / Phenylalanine (Met/Phe)     | 0.69  | µmol/L | < 1.60       |
| Citrulline (Cit)                         | 14.1  | µmol/L | < 50.0       |
| Ornithine (Orn)                          | 86.4  | µmol/L | < 250.0      |
| Ornithine / Citrulline ratio (Orn/Cit)   | 6.13  | µmol/L | 1.50 - 20.00 |
| Proline (Pro)                            | 90.4  | µmol/L | < 350.0      |
| Alanine (Ala)                            | 126.0 | µmol/L | < 750.0      |
| Arginine (Arg)                           | 8.0   | µmol/L | < 100.0      |
| Aspartic acid (Asp)                      | 51.1  | µmol/L | < 100.0      |
| Glutamic acid (Glu)                      | 201.1 | µmol/L | < 600.0      |
| Glycamine (Gly)                          | 103.4 | µmol/L | < 700.0      |

### Acylcarnitines

|  |       |        |               |
|--|-------|--------|---------------|
| Free carnitine (C0)                      | 15.13 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2)                     | 9.90  | µmol/L | 1.34 - 48.81  |
| propionylcarnitine (C3)                  | 0.75  | µmol/L | 0.13 - 6.60   |
| butyryl-/isobutyrylcarnitine (C4)        | 0.08  | µmol/L | 0.03 - 0.90   |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.12  | µmol/L | 0.02 - 2.00   |
| tiglylcarnitine (C5:1)                   | 0.01  | µmol/L | < 0.20        |
| hydroxyvalerylcarnitine (C5OH)           | 0.08  | 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)                | 0.08  | µmol/L | < 0.30        |
| decadienoylcarnitine (C10:2)             | 0.03  | µmol/L | < 0.10        |
| dodecanoylcarnitine (C12)                | 0.02  | µmol/L | 0.10 - 0.60   |
| myristoylcarnitine (C14)                 | 0.04  | µmol/L | 0.01 - 0.57   |
| tetradecenoylcarnitine (C14:1)           | 0.09  | µmol/L | 0.10 - 0.38   |
| palmitoylcarnitine (C16)                 | 0.28  | µmol/L | 0.62 - 7.81   |
| 3-hydroxypalmitoylcarnitine (C16OH)      | 0.01  | µmol/L | < 0.10        |
| stearoylcarnitine (C18)                  | 0.22  | µmol/L | 0.30 - 2.40   |
| oleylcarnitine (C18:1)                   | 2.80  | µmol/L | 0.06 - 3.86   |
| 3-hydroxystearoylcarnitine (C18OH)       | 0.01  | µmol/L | < 0.09        |
| malonylcarnitine (C3DC)                  | 0.03  | 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.

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