**CODIGO :** 173586

**NOMBRE PACIENTE :** EILEEN SAMANTHA GUTIERREZ UCHUVO **SEXO :** FEMENINO

**FECHA DE NACIMIENTO :** 24/03/2026 **REGISTRO CIVIL:** 1,023,054,977

**NOMBRE RESPONSABLE :** PAULA ANDREA GUTIERREZ UCHUVO

**DOC.IDENTIDAD DE LA MADRE :** 1,001,270,473

**FECHA TOMA DE MUESTRA :** 28/05/2026 **TIPO DE MUESTRA :** TALÓN

**FECHA DE IMPRESIÓN :** 18/06/2026 **PESO :** 2635

# TAMIZAJE NEONATAL

## ANÁLISIS MUESTRA DE SANGRE

| | RESULTADO | VALORES DE REFERENCIA | INTERPRETACIÓN |
|---|---|---|---|
| T.S.H Neonatal | 1.38 µl/mL | >= 6 µl/mL talón en prematuros<br>>= 10 µl/mL talón<br>>= 15 µl/mL cordón | Normal |
| Deficiencia de G6PDH | 7.30 U/gHb | < 2.6 U/gHb | Normal |

*TÉCNICA: Fluoroinmunoensayo (Delfia).* *Procesado en Colombia por PREGEN.*

| | | | |
|---|---|---|---|
| Hemoglobinopatías | AF | Ausencia de hemoglobinas anormales | Normal |

*TÉCNICA: Cromatografía Liquida de Alto Rendimiento (HPLC).* *Procesado en Colombia por PREGEN.*

# 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
Bacterióloga
Reg. 40.936.003

**PROCESADO :** MARIA JOSE PINZON GARCIA
Bacterióloga
Reg. 1.015.469.392

**FECHA :** 18/06/2026

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`LABORATORIO PREGEN
Carrera 15a No 106-42
BOGOTA

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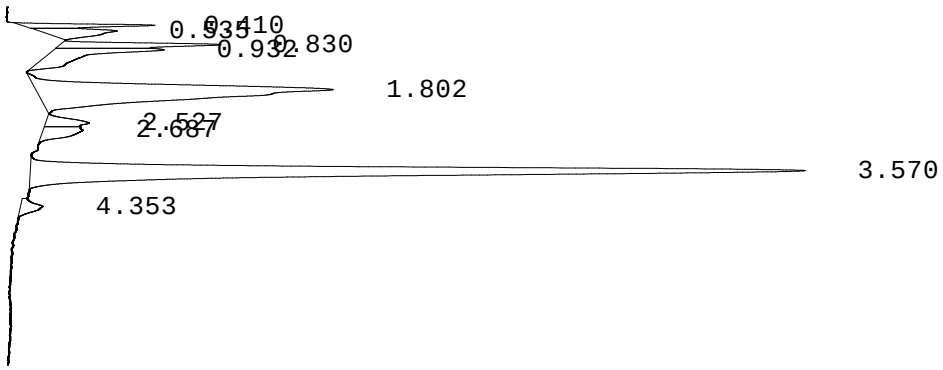
Batch 2167, Rack A, Plate 1, Well C03, 173586 AF
[892CA25A4978E468] Jun 02, 2026 12:42:13 Pressure = 66 bar (62 to 66)




| PEAK | RT | REL RT | % CONC | AREA | COMMENT |
|---|---|---|---|---|---|
| 1 | 0.410 | F 0.22 | 2.8% | 31141 | |
| 2 | 0.535 | F 0.29 | 3.5% | 38174 | |
| 3 | 0.830 | F 0.45 | 4.8% | 53273 | |
| 4 | 0.932 | F 0.50 | 6.3% | 69595 | Acetylated F peak |
| 5 | 1.802 | F 0.98 | 28.3% | 313513 | Consistent with F |
| 6 | 2.527 | F 1.37 | 2.2% | 23874 | |
| 7 | 2.687 | F 1.46 | 2.7% | 29571 | |
| 8 | 3.570 | A 1.00 | 48.0% | 531534 | A peak - REVIEW |
| 9 | 4.353 | S 0.85 | 1.4% | 15799 | 3 |
| | | | Total Area: | 1106474 | |

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Codes: 1) Wide A peak
2) Area of A peak < 80%
3) Peak area greater than expected
4) Peak after A2
5) A1c > 10%
6) HbF or variant present
7) Total sample area too small/big
8) A2 is not within normal range

baby health check©

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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 | 262025006 |

# Medical Report

| | | | |
|---|---|---|---|
| Patient name | **RODRIGUEZ GUTIERREZ EILEEN SAMANTHA** | Sample-ID | A0358651 |
| Date of Birth | **24.03.2026** | 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) | 2635 | g | - |
| 17-hydroxyprogesterone (17OHP) | <5.0 | nmol/L | < 90.0 |
| Thyroid-stimulating hormone (TSH) | 1.0 | µU/mL | < 15.0 |
| Biotinidase | 291.3 | U | > 51.0 |
| Galactose-1-P-uridyltransferase (GALT) | 8.1 | U/g Hb | > 2.5 |
| Immunoreactive trypsinogen (IRT) | <15 | ng/mL | < 65.0 |
| Phenylalanine | 32.6 | µmol/L | < 150.0 |
| Amino acid profile | negative | | - |
| Acylcarnitine profile | negative | | - |

**Interpretation:** NEGATIVE RESULT

**Contact Details**
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| Patient name | **RODRIGUEZ GUTIERREZ EILEEN SAMANTHA** |
|---|---|
| Date of Birth | **24.03.2026** |

| Sample-ID | A0358651 |
|---|---|
| Gender | F |

## Results:

### Amino Acids

| Parameter | Value | Unit | Reference |
|---|---|---|---|
| Phenylalanine (Phe) | 32.6 | µmol/L | < 150.0 |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.69 | µmol/L | < 2.20 |
| Tyrosine (Tyr) | 47.5 | µmol/L | < 200.0 |
| Leucine (Leu) | 92.4 | µmol/L | < 270.0 |
| Valine (Val) | 47.8 | µmol/L | < 200.0 |
| Methionine (MET) | 16.6 | µmol/L | < 78.0 |
| Methionine / Phenylalanine (Met/Phe) | 0.51 | µmol/L | < 1.60 |
| Citrulline (Cit) | 18.9 | µmol/L | < 50.0 |
| Ornithine (Orn) | 85.4 | µmol/L | < 250.0 |
| Ornithine / Citrulline ratio (Orn/Cit) | 4.52 | µmol/L | 1.50 - 20.00 |
| Proline (Pro) | 83.1 | µmol/L | < 350.0 |
| Alanine (Ala) | 108.8 | µmol/L | < 750.0 |
| Arginine (Arg) | 21.4 | µmol/L | < 100.0 |
| Aspartic acid (Asp) | 55.7 | µmol/L | < 100.0 |
| Glutamic acid (Glu) | 233.6 | µmol/L | < 600.0 |
| Glycamine (Gly) | 155.9 | µmol/L | < 700.0 |

### Acylcarnitines

| | | | |
|---|---|---|---|
| Free carnitine (C0) | 37.34 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2) | 15.33 | µmol/L | 1.34 - 48.81 |
| propionylcarnitine (C3) | 1.96 | µmol/L | 0.13 - 6.60 |
| butyryl-/isobutyrylcarnitine (C4) | 0.17 | µmol/L | 0.03 - 0.90 |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.19 | 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 |
| glutarylcarnitine (C5DC) | 0.05 | µmol/L | < 0.30 |
| hexanoylcarnitine (C6) | 0.05 | µmol/L | 0.01 - 0.13 |
| octanoylcarnitine (C8) | 0.04 | µmol/L | 0.01 - 0.30 |
| decanoylcarnitine (C10) | 0.05 | µmol/L | 0.01 - 0.36 |
| decenoylcarnitine (C10:1) | 0.10 | µmol/L | < 0.30 |
| dodecanoylcarnitine (C12) | 0.06 | µmol/L | 0.10 - 0.60 |
| myristoylcarnitine (C14) | 0.12 | µmol/L | 0.01 - 0.57 |
| tetradecenoylcarnitine (C14:1) | 0.10 | µmol/L | 0.10 - 0.43 |
| palmitoylcarnitine (C16) | 1.22 | µmol/L | 0.62 - 7.81 |
| 3-hydroxypalmitoylcarnitine (C16OH) | 0.03 | µmol/L | < 0.10 |
| stearoylcarnitine (C18) | 0.70 | µmol/L | 0.30 - 2.40 |
| oleylcarnitine (C18:1) | 3.22 | µmol/L | 0.06 - 3.86 |
| 3-hydroxystearoylcarnitine (C18OH) | 0.01 | µmol/L | < 0.09 |
| malonylcarnitine (C3DC) | 0.05 | 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, methylglutaconiciduria, 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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