

CODIGO : 170603
NOMBRE PACIENTE : SOPHIA VICTORIA CEDEÑO RUIZ **SEXO :** FEMENINO
FECHA DE NACIMIENTO : 30/06/2025 **REGISTRO CIVIL :** 1,249,969,707
NOMBRE RESPONSABLE : ZAIR RASHELL CEDEÑO RUIZ
DOC.IDENTIDAD DE LA MADRE : 34,108,630
FECHA TOMA DE MUESTRA : 09/10/2025
FECHA DE IMPRESION : 01/11/2025



TAMIZAJE NEONATAL

ANALISIS MUESTRA DE SANGRE

| | RESULTADO | VALORES DE REFERENCIA | INTERPRETACION |
|---|-----------|---|--|
| Hipotiroidismo congénito | 1.12 | VN: < 10 uUI/ml | Normal |
| Deficiencia de G6PDH | 6.30 | VN : > 2.6 U/gHb | Normal |
| <i>TÉCNICA: Fluoroimmunoensayo (Delfia).</i> | | | <i>Procesado en Colombia por PREGEN.</i> |
| Hemoglobinopatías | ASF | Cromatograma compatible con posible rasgo | Anormal |
| <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

DETECCION DE POSIBLE VARIANTE DE HEMOGLOBINA S

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 **FECHA :** 01/11/2025
 Bacteriologa
 Reg. 40.936.003

Dr. MARIA JOSE PINZON GARCIA
RED COLOMBIANA DE MEDICINA GENETICA SAS - PREGEN
BOGOTA
CARRERA 15 A # 106 - 42
11001 BOGOTA
Colombia

Date of Report 01.11.2025
Sample Received 24.10.2025
Date of Sampling 09.10.2025
LAB-ID 252040812

Medical Report

| | | | |
|---------------|------------------------------------|--------|----------|
| Patient name | CEDEÑO RUIZ SOPHIA VICTORIA | DBS-ID | A0320584 |
| Date of Birth | 30.06.2025 | Gender | F |

Indication: Newborn Screening

Method(s): Immunoassay, Tandem mass spectrometry from Dried Blood Spot

Results:

| Parameter | Value | Unit | Reference |
|--|----------|--------|-----------|
| Birth weight (g) | 2600 | g | - |
| 17-hydroxyprogesterone (17OHP) | <5.0 | nmol/L | < 90.0 |
| Thyroid-stimulating hormone (TSH) | <0.7 | μU/mL | < 15.0 |
| Biotinidase | 293.9 | U | > 51.0 |
| Galactose-1-P-uridyltransferase (GALT) | 8.3 | U/g Hb | > 2.5 |
| Immunoreactive trypsinogen (IRT) | <15 | ng/mL | < 65.0 |
| Phenylalanine | 27.5 | μmol/L | < 150.0 |
| Amino acid profile | negative | | - |
| Acylcarnitine profile | negative | | - |

Interpretation: NEGATIVE RESULT

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.

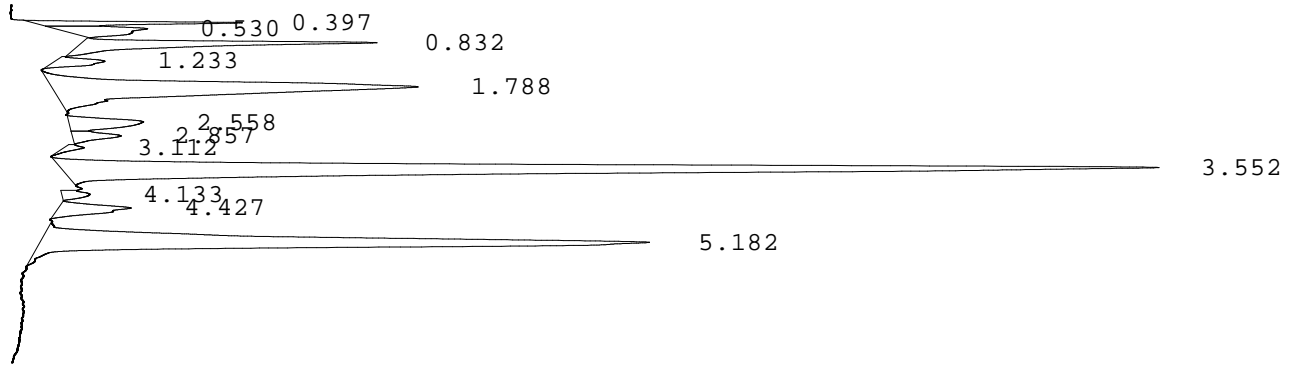
Authorized By: Assoc.-Prof. Dr. Andrea-Romana KASPER, MD, PhD
[Specialist for Pediatrics, Neonatology and Nutrition]

Report was electronically signed and approved.

LABORATORIO PREGEN
 Carrera 15a No 106-42
 BOGOTA

Batch 1951, Rack A, Plate 1, Well A06, 170603
 [8D304EA55ABCF937] Oct 15, 2025 16:10:46 Pressure = 69 bar (67 to 70)

ASF



| PEAK | RT | REL RT | % CONC | AREA | COMMENT |
|---------------------|-------|--------|--------|--------|-------------------|
| 1 | 0.397 | F 0.22 | 2.4% | 47209 | |
| 2 | 0.530 | F 0.29 | 2.5% | 48374 | |
| 3 | 0.832 | F 0.46 | 6.3% | 121416 | |
| 4 | 1.233 | F 0.68 | 1.4% | 26287 | |
| 5 | 1.788 | F 0.99 | 17.7% | 342705 | Consistent with F |
| 6 | 2.558 | F 1.42 | 2.7% | 52086 | |
| 7 | 2.857 | A 0.80 | 1.3% | 24796 | |
| 8 | 3.112 | A 0.87 | 0.4% | 8169 | |
| 9 | 3.552 | A 1.00 | 37.7% | 728075 | A peak - REVIEW |
| 10 | 4.133 | A 1.16 | 0.7% | 13784 | |
| 11 | 4.427 | S 0.87 | 2.4% | 47073 | 3 |
| 12 | 5.182 | S 1.02 | 24.4% | 472040 | Consistent with S |
| Total Area: 1932014 | | | | | A/V=1.55 |

- 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