

**CODIGO :**  
**NOMBRE PACIENTE :**  
**FECHA DE NACIMIENTO :**  
**NOMBRE RESPONSABLE :**  
**DOC.IDENTIDAD DE LA MADRE :**  
**FECHA TOMA DE MUESTRA :**  
**FECHA DE IMPRESIÓN :**

**173891**  
**EMMA SOFIA CASSIANIS MEJIA**  
**27/05/2026**  
**YEISIMAR MEJIA SEVERICHE**  
**1,100,550,001**  
**01/07/2026**  
**22/07/2026**

**SEXO : FEMENINO**  
**REGISTRO CIVIL: 1,025,335,874**  
**TIPO DE MUESTRA : TALÓN**  
**PESO : 3847**



TAMIZAJE NEONATAL

ANÁLISIS MUESTRA DE SANGRE

|                                                            | RESULTADO  | VALORES DE REFERENCIA                                                     | INTERPRETACIÓN |
|------------------------------------------------------------|------------|---------------------------------------------------------------------------|----------------|
| T.S.H Neonatal                                             | 1.56 µU/mL | >= 6 µU/mL talón en prematuros<br>>= 10 µU/mL talón<br>>= 15 µU/mL cordón | Normal         |
| Deficiencia de G6PDH                                       | 6.80 U/gHb | < 2.6 U/gHb                                                               | Normal         |
| TÉCNICA: Fluoroimmunoensayo (Delfia).                      |            | Procesado en Colombia por PREGEN.                                         |                |
| Hemoglobinopatías                                          | FA         | Ausencia de hemoglobinas anormales                                        | Normal         |
| TÉCNICA: Cromatografía Líquida 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, C5OH, 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**

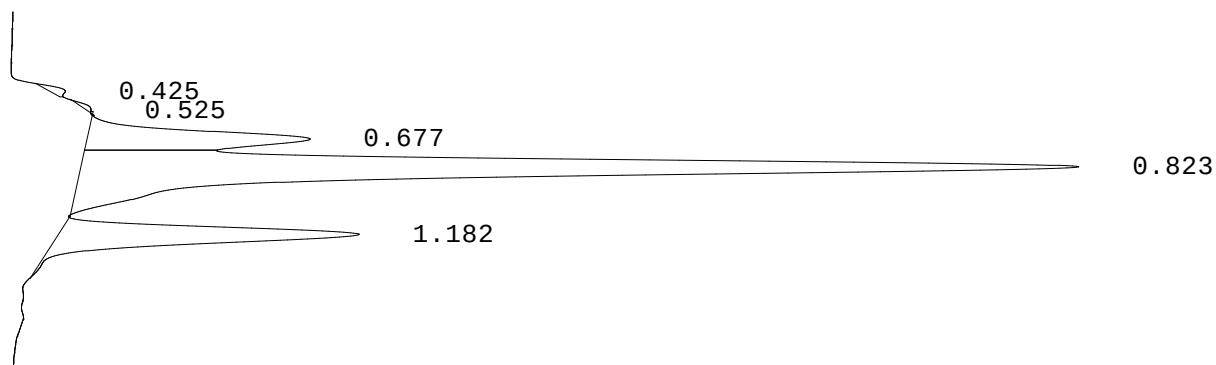
**PROCESADO :**  
**MARIA JOSE PINZON GARCIA**  
**Bacterióloga**  
**Reg. 1.015.469.392**

**FECHA :**  
**22/07/2026**

- - - - -  
`LABORATORIO PREGEN  
Carrera 15a No 106-42  
BOGOTA

Batch 2191, Rack A, Plate 1, Well E09, 173891  
[8D29C57FB12864E2] Jul 06, 2026 13:35:42 Pressure = 79 bar (78 to 81)

FA



| PEAK        | RT    | REL | RT   | % CONC | AREA   | COMMENT           |
|-------------|-------|-----|------|--------|--------|-------------------|
| 1           | 0.425 | F   | 0.51 | 0.5%   | 4335   |                   |
| 2           | 0.525 | F   | 0.63 | 0.3%   | 2550   |                   |
| 3           | 0.677 | F   | 0.82 | 13.9%  | 114260 | Acetylated F peak |
| 4           | 0.823 | F   | 0.99 | 67.0%  | 552556 | Consistent with F |
| 5           | 1.182 | A   | 1.01 | 18.3%  | 150597 | A peak            |
| Total Area: |       |     |      |        | 824298 |                   |

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

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

Date of Report 11.07.2026  
Sample Received 09.07.2026  
Date of Sampling 01.07.2026  
LAB-ID 262031115

## Medical Report

|               |                                   |           |          |
|---------------|-----------------------------------|-----------|----------|
| Patient name  | <b>CASSIANIS MEJIA EMMA SOFIA</b> | Sample-ID | A0358455 |
| Date of Birth | <b>27.05.2026</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)                       | 3847     | g      | -         |
| 17-hydroxyprogesterone (17OHP)         | 6.9      | nmol/L | < 90.0    |
| Thyroid-stimulating hormone (TSH)      | 1.0      | μU/mL  | < 15.0    |
| Biotinidase                            | 262.7    | U      | > 51.0    |
| Galactose-1-P-uridyltransferase (GALT) | 6.8      | U/g Hb | > 2.5     |
| Immunoreactive trypsinogen (IRT)       | 22.7     | ng/mL  | < 65.0    |
| Phenylalanine                          | 30.7     | μmol/L | < 150.0   |
| Amino acid profile                     | negative |        | -         |
| Acylcarnitine profile                  | negative |        | -         |

**Interpretation:** NEGATIVE RESULT

### Contact Details

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E-Mail: [info@archimedlife.com](mailto:info@archimedlife.com)

|               |                                   |
|---------------|-----------------------------------|
| Patient name  | <b>CASSIANIS MEJIA EMMA SOFIA</b> |
| Date of Birth | <b>27.05.2026</b>                 |

|           |          |
|-----------|----------|
| Sample-ID | A0358455 |
| Gender    | F        |

## Results:

### Amino Acids

| Parameter                                | Value | Unit   | Reference    |
|------------------------------------------|-------|--------|--------------|
| Phenylalanine (Phe)                      | 30.7  | µmol/L | < 150.0      |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.23  | µmol/L | < 2.20       |
| Tyrosine (Tyr)                           | 132.7 | µmol/L | < 200.0      |
| Leucine (Leu)                            | 145.0 | µmol/L | < 270.0      |
| Valine (Val)                             | 73.5  | µmol/L | < 200.0      |
| Methionine (MET)                         | 31.3  | µmol/L | < 78.0       |
| Methionine / Phenylalanine (Met/Phe)     | 1.02  | µmol/L | < 1.60       |
| Citrulline (Cit)                         | 15.6  | µmol/L | < 50.0       |
| Ornithine (Orn)                          | 86.1  | µmol/L | < 250.0      |
| Ornithine / Citrulline ratio (Orn/Cit)   | 5.52  | µmol/L | 1.50 - 20.00 |
| Proline (Pro)                            | 122.3 | µmol/L | < 350.0      |
| Alanine (Ala)                            | 201.6 | µmol/L | < 750.0      |
| Arginine (Arg)                           | 40.2  | µmol/L | < 100.0      |
| Aspartic acid (Asp)                      | 73.6  | µmol/L | < 100.0      |
| Glutamic acid (Glu)                      | 212.8 | µmol/L | < 600.0      |
| Glycamine (Gly)                          | 176.4 | µmol/L | < 700.0      |

### Acylcarnitines

|                                          |       |        |               |
|------------------------------------------|-------|--------|---------------|
| Free carnitine (C0)                      | 20.30 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2)                     | 8.88  | µmol/L | 1.34 - 48.81  |
| propionylcarnitine (C3)                  | 1.25  | µmol/L | 0.13 - 6.60   |
| butyryl-/isobutyrylcarnitine (C4)        | 0.11  | µmol/L | 0.03 - 0.90   |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.09  | µmol/L | 0.02 - 2.00   |
| tiglylcarnitine (C5:1)                   | 0.01  | µmol/L | < 0.20        |
| hydroxyvalerylcarnitine (C5OH)           | 0.35  | µmol/L | 0.02 - 0.57   |
| glutaryl carnitine (C5DC)                | 0.04  | µmol/L | < 0.30        |
| hexanoylcarnitine (C6)                   | 0.04  | µmol/L | 0.01 - 0.13   |
| octanoylcarnitine (C8)                   | 0.03  | µmol/L | 0.01 - 0.30   |
| decanoylcarnitine (C10)                  | 0.04  | µmol/L | 0.01 - 0.36   |
| decenoylcarnitine (C10:1)                | 0.09  | µmol/L | < 0.30        |
| dodecanoylcarnitine (C12)                | 0.04  | µmol/L | 0.10 - 0.60   |
| myristoylcarnitine (C14)                 | 0.05  | µmol/L | 0.01 - 0.57   |
| tetradecenoylcarnitine (C14:1)           | 0.03  | µmol/L | 0.10 - 0.43   |
| palmitoylcarnitine (C16)                 | 0.49  | µmol/L | 0.62 - 7.81   |
| 3-hydroxypalmitoylcarnitine (C16OH)      | 0.02  | µmol/L | < 0.10        |
| stearoylcarnitine (C18)                  | 0.25  | µmol/L | 0.30 - 2.40   |
| oleylcarnitine (C18:1)                   | 1.28  | 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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