

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

Date of Report 20.03.2026  
Sample Received 12.03.2026  
Date of Sampling 03.03.2026  
LAB-ID 262010632

## Medical Report

|               |                                |           |          |
|---------------|--------------------------------|-----------|----------|
| Patient name  | <b>RUBIONA VARRETE SALOMON</b> | Sample-ID | A0321872 |
| Date of Birth | <b>31.01.2026</b>              | Gender    | M        |

**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)                       | 3580     | g      | -         |
| 17-hydroxyprogesterone (17OHP)         | <5.0     | nmol/L | < 90.0    |
| Thyroid-stimulating hormone (TSH)      | 0.8      | μU/mL  | < 15.0    |
| Biotinidase                            | 223.8    | U      | > 51.0    |
| Galactose-1-P-uridyltransferase (GALT) | 6.7      | U/g Hb | > 2.5     |
| Immunoreactive trypsinogen (IRT)       | <15      | ng/mL  | < 65.0    |
| Phenylalanine                          | 31.8     | μmol/L | < 150.0   |
| Amino acid profile                     | negative |        | -         |
| Acylcarnitine profile                  | negative |        | -         |

**Interpretation:** NEGATIVE RESULT

|               |                                |
|---------------|--------------------------------|
| Patient name  | <b>RUBIONA VARRETE SALOMON</b> |
| Date of Birth | <b>31.01.2026</b>              |

|           |          |
|-----------|----------|
| Sample-ID | A0321872 |
| Gender    | M        |

## Results:

### Amino Acids

| Parameter                                | Value | Unit   | Reference    |
|--|-------|--------|--------------|
| Phenylalanine (Phe)                      | 31.8  | µmol/L | < 150.0      |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.40  | µmol/L | < 2.20       |
| Tyrosine (Tyr)                           | 79.6  | µmol/L | < 200.0      |
| Leucine (Leu)                            | 125.0 | µmol/L | < 270.0      |
| Valine (Val)                             | 59.1  | µmol/L | < 200.0      |
| Methionine (MET)                         | 24.4  | µmol/L | < 78.0       |
| Methionine / Phenylalanine (Met/Phe)     | 0.77  | µmol/L | < 1.60       |
| Citrulline (Cit)                         | 13.4  | µmol/L | < 50.0       |
| Ornithine (Orn)                          | 95.9  | µmol/L | < 250.0      |
| Ornithine / Citrulline ratio (Orn/Cit)   | 7.16  | µmol/L | 1.50 - 20.00 |
| Proline (Pro)                            | 113.3 | µmol/L | < 350.0      |
| Alanine (Ala)                            | 130.5 | µmol/L | < 750.0      |
| Arginine (Arg)                           | 11.9  | µmol/L | < 100.0      |
| Aspartic acid (Asp)                      | 58.9  | µmol/L | < 100.0      |
| Glutamic acid (Glu)                      | 226.7 | µmol/L | < 600.0      |
| Glycamine (Gly)                          | 134.5 | µmol/L | < 700.0      |

### Acylcarnitines

|  |       |        |               |
|--|-------|--------|---------------|
| Free carnitine (C0)                      | 13.45 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2)                     | 6.30  | µmol/L | 1.34 - 48.81  |
| propionylcarnitine (C3)                  | 0.48  | µmol/L | 0.13 - 6.60   |
| butyryl-/isobutyrylcarnitine (C4)        | 0.09  | µmol/L | 0.03 - 0.90   |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.10  | µmol/L | 0.02 - 2.00   |
| tiglylcarnitine (C5:1)                   | 0.01  | µmol/L | < 0.20        |
| hydroxyvalerylcarnitine (C5OH)           | 0.16  | µmol/L | 0.02 - 0.57   |
| glutarylacetyl carnitine (C5DC)          | 0.03  | µmol/L | < 0.30        |
| hexanoylcarnitine (C6)                   | 0.04  | µmol/L | 0.01 - 0.13   |
| octanoylcarnitine (C8)                   | 0.02  | µmol/L | 0.01 - 0.30   |
| decanoylcarnitine (C10)                  | 0.03  | µmol/L | 0.01 - 0.36   |
| decenoylcarnitine (C10:1)                | 0.09  | µmol/L | < 0.30        |
| decadienoylcarnitine (C10:2)             | 0.02  | µmol/L | < 0.10        |
| dodecanoylcarnitine (C12)                | 0.05  | µmol/L | 0.10 - 0.60   |
| myristoylcarnitine (C14)                 | 0.06  | µmol/L | 0.01 - 0.57   |
| tetradecenoylcarnitine (C14:1)           | 0.08  | µmol/L | 0.10 - 0.38   |
| palmitoylcarnitine (C16)                 | 0.52  | µmol/L | 0.62 - 7.81   |
| 3-hydroxypalmitoylcarnitine (C16OH)      | 0.02  | µmol/L | < 0.10        |
| stearoylcarnitine (C18)                  | 0.40  | µmol/L | 0.30 - 2.40   |
| oleylcarnitine (C18:1)                   | 1.64  | µmol/L | 0.06 - 3.86   |
| 3-hydroxystearoylcarnitine (C18OH)       | 0.01  | µmol/L | < 0.09        |
| malonylcarnitine (C3DC)                  | 0.02  | 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

Assoc.-Prof. Dr. Andrea-Romana KASPER, MD, PhD  
E-Mail: info@archimedlife.com

**ARCHIMEDlife GmbH**  
International Medical Laboratory+  
Leberstrasse 20/2 | 1110 Vienna, Austria  
www.archimedlife.com