

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

Date of Report 06.05.2026
Sample Received 28.04.2026
Date of Sampling 21.04.2026
LAB-ID 262018946

Medical Report

| | | | |
|---------------|----------------------------|-----------|----------|
| Patient name | CRESPO MUÑOZ SALOME | Sample-ID | A0361117 |
| Date of Birth | 29.10.2025 | 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) | 2450 | g | - |
| 17-hydroxyprogesterone (17OHP) | <5.0 | nmol/L | < 90.0 |
| Thyroid-stimulating hormone (TSH) | 0.7 | μU/mL | < 15.0 |
| Biotinidase | 346.6 | U | > 51.0 |
| Galactose-1-P-uridyltransferase (GALT) | 7.0 | U/g Hb | > 2.5 |
| Immunoreactive trypsinogen (IRT) | <15 | ng/mL | < 65.0 |
| Phenylalanine | 47.0 | μmol/L | < 150.0 |
| Amino acid profile | negative | | - |
| Acylcarnitine profile | negative | | - |

Interpretation: NEGATIVE RESULT

| | |
|---------------|----------------------------|
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Results:

| Parameter | Value | Unit | Reference |
|--|-------|--------|---------------|
| Phenylalanine (Phe) | 47.0 | µmol/L | < 150.0 |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.83 | µmol/L | < 2.20 |
| Tyrosine (Tyr) | 56.3 | µmol/L | < 200.0 |
| Leucine (Leu) | 65.4 | µmol/L | < 270.0 |
| Valine (Val) | 65.2 | µmol/L | < 200.0 |
| Methionine (MET) | 20.9 | µmol/L | < 78.0 |
| Methionine / Phenylalanine (Met/Phe) | 0.44 | µmol/L | < 1.60 |
| Citrulline (Cit) | 21.4 | µmol/L | < 50.0 |
| Ornithine (Orn) | 101.4 | µmol/L | < 250.0 |
| Ornithine / Citrulline ratio (Orn/Cit) | 4.74 | µmol/L | 1.50 - 20.00 |
| Proline (Pro) | 79.2 | µmol/L | < 350.0 |
| Alanine (Ala) | 105.9 | µmol/L | < 750.0 |
| Arginine (Arg) | 31.2 | µmol/L | < 100.0 |
| Aspartic acid (Asp) | 50.5 | µmol/L | < 100.0 |
| Glutamic acid (Glu) | 240.1 | µmol/L | < 600.0 |
| Glycamine (Gly) | 155.9 | µmol/L | < 700.0 |
| Free carnitine (C0) | 36.20 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2) | 10.74 | µmol/L | 1.34 - 48.81 |
| propionylcarnitine (C3) | 0.56 | µmol/L | 0.13 - 6.60 |
| butyryl-/isobutyrylcarnitine (C4) | 0.39 | µmol/L | 0.03 - 0.90 |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.08 | µmol/L | 0.02 - 2.00 |
| tiglylcarnitine (C5:1) | 0.02 | µmol/L | < 0.20 |
| hydroxyvalerylcarnitine (C5OH) | 0.33 | µmol/L | 0.02 - 0.57 |
| glutarylacarnitine (C5DC) | 0.05 | µmol/L | < 0.30 |
| hexanoylcarnitine (C6) | 0.07 | µmol/L | 0.01 - 0.13 |
| octanoylcarnitine (C8) | 0.06 | µmol/L | 0.01 - 0.30 |
| decanoylcarnitine (C10) | 0.06 | µmol/L | 0.01 - 0.36 |
| decenoylcarnitine (C10:1) | 0.11 | µmol/L | < 0.30 |
| decadienoylcarnitine (C10:2) | 0.12 | µmol/L | < 0.10 |
| dodecanoylcarnitine (C12) | 0.06 | µmol/L | 0.10 - 0.60 |
| myristoylcarnitine (C14) | 0.07 | µmol/L | 0.01 - 0.57 |
| tetradecenoylcarnitine (C14:1) | 0.08 | µmol/L | 0.10 - 0.38 |
| palmitoylcarnitine (C16) | 0.53 | µmol/L | 0.62 - 7.81 |
| 3-hydroxypalmitoylcarnitine (C16OH) | 0.03 | µmol/L | < 0.10 |
| stearoylcarnitine (C18) | 0.42 | µmol/L | 0.30 - 2.40 |
| oleylcarnitine (C18:1) | 1.56 | µmol/L | 0.06 - 3.86 |
| 3-hydroxystearoylcarnitine (C18OH) | 0.01 | µmol/L | < 0.09 |
| malonylcarnitine (C3DC) | 0.08 | µmol/L | < 0.50 |

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.