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Date of Report 08.05.2026
Sample Received 04.05.2026
Date of Sampling 22.04.2026
LAB-ID 262019153

Medical Report

| | | | |
|---------------|--------------------------------------|-----------|----------|
| Patient name | RODRIGUEZ RUIZ GAEL ALEXANDER | Sample-ID | A0341280 |
| Date of Birth | 30.03.2026 | 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) | 2980 | g | - |
| 17-hydroxyprogesterone (17OHP) | 5.5 | nmol/L | < 90.0 |
| Thyroid-stimulating hormone (TSH) | <0.7 | µU/mL | < 15.0 |
| Biotinidase | 223.5 | U | > 51.0 |
| Galactose-1-P-uridylyltransferase (GALT) | 6.0 | U/g Hb | > 2.5 |
| Immunoreactive trypsinogen (IRT) | <15 | ng/mL | < 65.0 |
| Phenylalanine | 29.1 | µmol/L | < 150.0 |
| Amino acid profile | negative | | - |
| Acylcarnitine profile | negative | | - |

Interpretation: NEGATIVE RESULT

| | |
|---------------|--------------------------------------|
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| Date of Birth | 30.03.2026 |

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| Sample-ID | A0341280 |
| Gender | M |

Results:

Amino Acids

| Parameter | Value | Unit | Reference |
|--|-------|--------|--------------|
| Phenylalanine (Phe) | 29.1 | µmol/L | < 150.0 |
| Phenylalanine / Tyrosine ratio (Phe/Tyr) | 0.62 | µmol/L | < 2.20 |
| Tyrosine (Tyr) | 47.3 | µmol/L | < 200.0 |
| Leucine (Leu) | 109.0 | µmol/L | < 270.0 |
| Valine (Val) | 53.0 | µmol/L | < 200.0 |
| Methionine (MET) | 18.9 | µmol/L | < 78.0 |
| Methionine / Phenylalanine (Met/Phe) | 0.65 | µmol/L | < 1.60 |
| Citrulline (Cit) | 12.3 | µmol/L | < 50.0 |
| Ornithine (Orn) | 84.4 | µmol/L | < 250.0 |
| Ornithine / Citrulline ratio (Orn/Cit) | 6.86 | µmol/L | 1.50 - 20.00 |
| Proline (Pro) | 87.6 | µmol/L | < 350.0 |
| Alanine (Ala) | 139.4 | µmol/L | < 750.0 |
| Arginine (Arg) | 3.7 | µmol/L | < 100.0 |
| Aspartic acid (Asp) | 31.8 | µmol/L | < 100.0 |
| Glutamic acid (Glu) | 262.3 | µmol/L | < 600.0 |
| Glycamine (Gly) | 169.0 | µmol/L | < 700.0 |

Acylcarnitines

| | | | |
|--|-------|--------|---------------|
| Free carnitine (C0) | 19.19 | µmol/L | 6.00 - 100.00 |
| acetylcarnitine (C2) | 7.81 | µmol/L | 1.34 - 48.81 |
| propionylcarnitine (C3) | 0.64 | µmol/L | 0.13 - 6.60 |
| butyryl-/isobutyrylcarnitine (C4) | 0.11 | µmol/L | 0.03 - 0.90 |
| isovaleryl-/2-methylbutyrylcarnitine(C5) | 0.12 | µmol/L | 0.02 - 2.00 |
| tiglylcarnitine (C5:1) | 0.02 | µmol/L | < 0.20 |
| hydroxyvalerylcarnitine (C5OH) | 0.32 | µmol/L | 0.02 - 0.57 |
| glutarylacetyl carnitine (C5DC) | 0.04 | µmol/L | < 0.30 |
| hexanoylcarnitine (C6) | 0.04 | µmol/L | 0.01 - 0.13 |
| octanoylcarnitine (C8) | 0.04 | 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 |
| decadienoylcarnitine (C10:2) | 0.12 | µmol/L | < 0.10 |
| dodecanoylcarnitine (C12) | 0.06 | µmol/L | 0.10 - 0.60 |
| myristoylcarnitine (C14) | 0.08 | µmol/L | 0.01 - 0.57 |
| tetradecenoylcarnitine (C14:1) | 0.06 | µmol/L | 0.10 - 0.38 |
| palmitoylcarnitine (C16) | 0.44 | µmol/L | 0.62 - 7.81 |
| 3-hydroxypalmitoylcarnitine (C16OH) | 0.02 | µmol/L | < 0.10 |
| stearoylcarnitine (C18) | 0.30 | µmol/L | 0.30 - 2.40 |
| oleylcarnitine (C18:1) | 0.86 | 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, 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.

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