

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

Date of Report 04.11.2025
Sample Received 29.10.2025
Date of Sampling 22.10.2025
LAB-ID 252042255

Medical Report

| | | | |
|---------------|----------------------------|--------|----------|
| Patient name | ASTAKHOVA ALEXANDRA | DBS-ID | A0320696 |
| Date of Birth | 09.10.2025 | Gender | F |

Indication: Newborn Screening

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

Results:

| Parameter | Value | Unit | Reference |
|--|----------|--------|-----------|
| Birth weight (g) | 3280 | g | - |
| 17-hydroxyprogesterone (17OHP) | 5.1 | nmol/L | < 90.0 |
| Thyroid-stimulating hormone (TSH) | <0.7 | µU/mL | < 15.0 |
| Biotinidase | 252.4 | U | > 51.0 |
| Galactose-1-P-uridyltransferase (GALT) | - | U/g Hb | > 2.5 |
| Immunoreactive trypsinogen (IRT) | 30.6 | ng/mL | < 65.0 |
| Phenylalanine | 29.7 | µmol/L | < 150.0 |
| Amino acid profile | negative | | - |
| Acylcarnitine profile | negative | | - |

Interpretation: **INCONCLUSIVE RESULT for GALT.**

Please send a 2nd DBS card for cost free re-testing. Please write "2n DBS" on the card. All other tests were negative.

Please note: Inconspicuous biochemical results cannot exclude any metabolic disease with certainty. We recommend genetic testing if clinical symptoms suggest any inborn error of metabolism. For further information please contact us: info@archimedlife.com.

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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