

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

Date of Report 08.01.2026  
Sample Received 05.01.2026  
Date of Sampling 19.12.2025  
LAB-ID 252052972

## Medical Report

|               |                           |           |          |
|---------------|---------------------------|-----------|----------|
| Patient name  | <b>GRUBER LEON FRANCO</b> | Sample-ID | A0321375 |
| Date of Birth | <b>31.10.2025</b>         | Gender    | M        |

**Indication:** Newborn Screening

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

**Results:**

| Parameter                              | Value    | Unit   | Reference |
|--|----------|--------|-----------|
| Birth weight (g)                       | 1260     | g      | -         |
| 17-hydroxyprogesterone (17OHP)         | 11.9     | nmol/L | < 90.0    |
| Thyroid-stimulating hormone (TSH)      | <0.7     | μU/mL  | < 15.0    |
| Biotinidase                            | 207.4    | U      | > 51.0    |
| Galactose-1-P-uridyltransferase (GALT) | 8.1      | U/g Hb | > 2.5     |
| Immunoreactive trypsinogen (IRT)       | <15      | ng/mL  | < 65.0    |
| Phenylalanine                          | 38.8     | μmol/L | < 150.0   |
| Amino acid profile                     | negative |        | -         |
| Acylcarnitine profile                  | negative |        | -         |

**Interpretation:** NEGATIVE RESULT

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.