

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

Date of Report 29.06.2026  
Sample Received 22.06.2026  
Date of Sampling 16.06.2026  
LAB-ID 262028128

## Medical Report

Patient name	<b>HUGHES JOSEPHINE WHITNEY</b>	Sample-ID	A0340581
Date of Birth	<b>26.05.2026</b>	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)	3470	g	-
17-hydroxyprogesterone (17OHP)	9.5	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	0.8	μU/mL	< 15.0
Biotinidase	262.7	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	8.3	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	21.8	μmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

**Interpretation:** NEGATIVE RESULT

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## Results:

### Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	21.8	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.29	µmol/L	< 2.20
Tyrosine (Tyr)	75.3	µmol/L	< 200.0
Leucine (Leu)	92.7	µmol/L	< 270.0
Valine (Val)	54.8	µmol/L	< 200.0
Methionine (MET)	12.6	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.58	µmol/L	< 1.60
Citrulline (Cit)	16.0	µmol/L	< 50.0
Ornithine (Orn)	53.2	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	3.33	µmol/L	1.50 - 20.00
Proline (Pro)	82.6	µmol/L	< 350.0
Alanine (Ala)	131.5	µmol/L	< 750.0
Arginine (Arg)	18.1	µmol/L	< 100.0
Aspartic acid (Asp)	31.8	µmol/L	< 100.0
Glutamic acid (Glu)	242.1	µmol/L	< 600.0
Glycamine (Gly)	149.6	µmol/L	< 700.0

### Acylcarnitines

Free carnitine (C0)	12.76	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	5.46	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	0.53	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.11	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.06	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.01	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.29	µmol/L	0.02 - 0.57
glutarylacarnitine (C5DC)	0.04	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.03	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.04	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.02	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.06	µmol/L	< 0.30
dodecanoylcarnitine (C12)	0.05	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.04	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.05	µmol/L	0.10 - 0.43
palmitoylcarnitine (C16)	0.30	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.19	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	0.50	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.

**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