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Date of Report 27.02.2026
Sample Received 19.02.2026
Date of Sampling 11.02.2026
LAB-ID 262006982

Medical Report

Patient name	COSTA FRANCISCO XAVIER	Sample-ID	A0321855
Date of Birth	31.01.2026	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)	3270	g	-
17-hydroxyprogesterone (17OHP)	<5.0	nmol/L	< 90.0
Thyroid-stimulating hormone (TSH)	<0.7	μU/mL	< 15.0
Biotinidase	160.2	U	> 51.0
Galactose-1-P-uridyltransferase (GALT)	11.7	U/g Hb	> 2.5
Immunoreactive trypsinogen (IRT)	<15	ng/mL	< 65.0
Phenylalanine	27.9	μmol/L	< 150.0
Amino acid profile	negative		-
Acylcarnitine profile	negative		-

Interpretation: NEGATIVE RESULT

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Date of Birth	31.01.2026

Sample-ID	A0321855
Gender	M

Results:

Amino Acids

Parameter	Value	Unit	Reference
Phenylalanine (Phe)	27.9	µmol/L	< 150.0
Phenylalanine / Tyrosine ratio (Phe/Tyr)	0.30	µmol/L	< 2.20
Tyrosine (Tyr)	94.3	µmol/L	< 200.0
Leucine (Leu)	129.7	µmol/L	< 270.0
Valine (Val)	67.4	µmol/L	< 200.0
Methionine (MET)	25.2	µmol/L	< 78.0
Methionine / Phenylalanine (Met/Phe)	0.90	µmol/L	< 1.60
Citrulline (Cit)	9.8	µmol/L	< 50.0
Ornithine (Orn)	74.0	µmol/L	< 250.0
Ornithine / Citrulline ratio (Orn/Cit)	7.55	µmol/L	1.50 - 20.00
Proline (Pro)	110.6	µmol/L	< 350.0
Alanine (Ala)	143.3	µmol/L	< 750.0
Arginine (Arg)	10.8	µmol/L	< 100.0
Aspartic acid (Asp)	120.3	µmol/L	< 100.0
Glutamic acid (Glu)	419.4	µmol/L	< 600.0
Glycamine (Gly)	174.8	µmol/L	< 700.0

Acylcarnitines

Free carnitine (C0)	6.34	µmol/L	6.00 - 100.00
acetylcarnitine (C2)	7.73	µmol/L	1.34 - 48.81
propionylcarnitine (C3)	0.42	µmol/L	0.13 - 6.60
butyryl-/isobutyrylcarnitine (C4)	0.11	µmol/L	0.03 - 0.90
isovaleryl-/2-methylbutyrylcarnitine(C5)	0.07	µmol/L	0.02 - 2.00
tiglylcarnitine (C5:1)	0.01	µmol/L	< 0.20
hydroxyvalerylcarnitine (C5OH)	0.20	µmol/L	0.02 - 0.57
glutarylacetylacetylcarnitine (C5DC)	0.02	µmol/L	< 0.30
hexanoylcarnitine (C6)	0.01	µmol/L	0.01 - 0.13
octanoylcarnitine (C8)	0.01	µmol/L	0.01 - 0.30
decanoylcarnitine (C10)	0.02	µmol/L	0.01 - 0.36
decenoylcarnitine (C10:1)	0.07	µmol/L	< 0.30
decadienoylcarnitine (C10:2)	0.02	µmol/L	< 0.10
dodecanoylcarnitine (C12)	0.05	µmol/L	0.10 - 0.60
myristoylcarnitine (C14)	0.11	µmol/L	0.01 - 0.57
tetradecenoylcarnitine (C14:1)	0.07	µmol/L	0.10 - 0.38
palmitoylcarnitine (C16)	1.50	µmol/L	0.62 - 7.81
3-hydroxypalmitoylcarnitine (C16OH)	0.02	µmol/L	< 0.10
stearoylcarnitine (C18)	0.79	µmol/L	0.30 - 2.40
oleylcarnitine (C18:1)	3.86	µmol/L	0.06 - 3.86
3-hydroxystearoylcarnitine (C18OH)	0.01	µmol/L	< 0.09
malonylcarnitine (C3DC)	0.02	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.

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[Specialist for Pediatrics, Neonatology and Nutrition]

Report was electronically signed and approved.

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