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PATIENT REPORT

Patient Age/Sex: 13 days Unknown

Specimen Collected: 04-Nov-25 11:12

Plasmalogens (Red Blood Cells) | **Received: 05-Nov-25 09:24** **Report/Verified: 06-Nov-25 09:31**

Procedure	Result	Units	Reference Interval
Plasmalogen PE (RBC) Interpretation	See Note f1 i1		
16:0(plasm)-PE Total	9.70 L f2	nmol/mL	[89.09-165.13]
18:0(plasm)-PE Total	7.00 L f2	nmol/mL	[124.85-251.80]
18:1(plasm)-PE Total	7.17 L f2	nmol/mL	[45.38-111.35]
Total Plasmalogen PE (RBC)	23.87 L f2	nmol/mL	[275.36-484.68]

Result Footnote

f1: Plasmalogen PE (RBC) Interpretation
The total concentrations of the 16:0, 18:0 and 18:1 plasmalogen species, and total plasmalogens were markedly reduced. These results are most consistent with a severe defect of peroxisomal plasmalogen biosynthesis, such as Zellweger spectrum disorder or rhizomelic chondrodysplasia punctata. Would evaluate very long-chain fatty acids in plasma/serum. Consideration should be given to molecular genetic testing to confirm this finding and identify the specific defect. Genetic and metabolic evaluations are recommended.

Results reviewed and interpreted by Irene De Biase, MD, PhD, FACMG

f2: 16:0(plasm)-PE Total, 18:0(plasm)-PE Total, 18:1(plasm)-PE Total, Total Plasmalogen PE (RBC)
One or more analytes are below the Analytical Measurement Range (AMR). Total was calculated using the value corresponding to the lower AMR limit for analytes below the AMR.

Test Information

i1: Plasmalogen PE (RBC) Interpretation
INTERPRETIVE INFORMATION: Plasmalogens (Red Blood Cells)

This test measures eighteen individual phosphoethanolamine plasmalogen species by LC-MS/MS. Total values calculated by adding each of the six 16:0, 18:0 or 18:1 species and total plasmalogens, obtained by adding all 18 species, are reported.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab