

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB 1/11/2026
Sex: Unknown
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Plasmalogens (Red Blood Cells)

ARUP test code 3019936

Plasmalogen PE (RBC) Interpretation**See Note**

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

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.

16:0(plasm)-PE Total**9.70 nmol/mL L (Ref Interval: 89.09-165.13)**

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.

18:0(plasm)-PE Total**7.00 nmol/mL L (Ref Interval: 124.85-251.80)**

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.

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

18:1(plasm)-PE Total

7.17 nmol/mL L (Ref Interval: 45.38-111.35)

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.

Total Plasmalogen PE (RBC)

23.87 nmol/mL L (Ref Interval: 275.36-484.68)

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Plasmalogen PE (RBC) Interpretation	26-027-119564	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
16:0(plasm)-PE Total	26-027-119564	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
18:0(plasm)-PE Total	26-027-119564	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
18:1(plasm)-PE Total	26-027-119564	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Total Plasmalogen PE (RBC)	26-027-119564	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at: