

PLASMALOGENS

Relevant disorders

Peroxisomal biogenesis defects
Rhizomelic chondrodysplasia punctata

Related Metabolic Tests

Very Long Chain Fatty Acids; Bile Salts

Indication for Test

As part of the investigations for Peroxisomal Biogenesis defects (Zellweger spectrum) or if Rhizomelic chondrodysplasia punctata is suspected.

Methodology

Gas Chromatography Mass Spectrometry

Sample requirements

Red blood cells are required for this assay.
2 ml EDTA blood should be centrifuged at 3000 rpm, plasma and buffy coat removed and the red blood cells washed three times with isotonic saline and stored at -20deg.C. Samples sent by post should be prepared this way before dispatch and can be frozen as red blood cell pellets. Samples can be sent first class post, at room temperature.

Transport information/Contact details

Send by first class post to:

Department of Clinical Chemistry
Sheffield Children's NHS Foundation Trust
Western Bank, Sheffield
S10 2TH, UK

Jane Dalley (Clinical Scientist)
0114 2717307

Turn Around Time

4 – 6 weeks

Reference Ranges

Interpretation will be provided as part of the report.

References

- Dacremont and Vincent 1995 J.Inher.Metab.Dis.18.Suppl.1 p85