

VERY LONG CHAIN FATTY ACIDS

Relevant disorders

- Peroxisomal biogenesis disorders
(Zellweger syndrome, Neonatal Adrenoleukodystrophy, Infantile Refsum disease)
- X-Linked Adrenoleukodystrophy (X-ALD) and Adrenomyeloneuropathy (AMN)
- Refsum disease (phytanic alpha oxidase deficiency)
- α Methyl-acyl-CoA racemase deficiency
- Rhizomelic Chondrodysplasia Punctata (RCDP)
- Trihydroxycholestanoyl CoA oxidase deficiency
- 2-Methylacyl-CoA racemase deficiency

Related Metabolic Tests

Plasmalogens

Bile acids

Pipecolic acid

DHAP-AT assay

Indication for Test

Very long chain fatty acids (VLCFAs) are essential components of cell membranes especially in neurones. Present in many foods they are also synthesized by chain elongation of short chain fatty acids. Catabolism of VLCFAs occurs by conversion to CoA esters followed by beta-oxidation in peroxisomes.

Inherited disorders of peroxisomal biogenesis / function are multi-system diseases, a significant proportion of which feature abnormal VLCFA oxidation and thence abnormal concentrations of VLCFAs in blood and tissues.

This assay measures the following VLCFAs:

- | | |
|-----------------------|---------------------------|
| - docosahexanoic acid | (C22 or behenic acid); |
| - tetracosanoic acid | (C24 or lignoceric acid); |
| - hexacosanoic acid | (C26 or cerotic acid) |

The following branched-chain fatty acids are also measured:

- Phytanic acid [C20] (3,7,11,15-tetramethylhexadecanoic acid) and
- Pristanic acid [C19] (2,6,10,14-tetramethylpentadecanoic acid) are metabolites of phytol and oxidised in peroxisomes by alpha- and beta-oxidation respectively. They are both potential markers for peroxisome biogenesis disorders as well as specific isolated defects affecting their oxidation.

Methodology

Stable isotope dilution GCMS.

Sample requirements

1ml venous blood in a Lithium heparin or EDTA tube. Serum and fluoride oxalate samples are also accepted. Separate and send the plasma.

Fibroblasts, amniocyte and chorionic villus cell assay also available. Please contact laboratory.

Transport information/Contact details

Send all samples by first class post to:

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

Joanne Croft (Clinical Scientist)
0114 2717307

Turn Around Time

4 – 6 weeks (for plasma assay)
2 months (for fibroblast assay)

Reference Ranges

Interpretation of the results will be provided with the report.

References

- Gould SJ, Raymond GV, Valle D (2001) The Peroxisome Biogenesis Disorders. In: Scriver C, Beaudet A, Valle D, Sly W et al (eds) The Metabolic and Molecular Basis of Disease, 8th edition. McGraw Hill Medical Publishing, pp 3181 – 3217
- Wanders RJA, Barth PG, Heymans HSA (2001) Single Peroxisomal Enzyme Deficiencies. In: Scriver C, Beaudet A, Valle D, Sly W et al (eds) The Metabolic and Molecular Basis of Disease, 8th edition. McGraw Hill Medical Publishing, pp 3219 – 3256