DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

DIHYDROXYACETONE PHOSPHATE ACYLTRANSFERASE (DHAP-AT)

Relevant disorders

Peroxisomal biogenesis disorders.
Rhizomelic chondrodysplasia punctate (RCDP)

Related Metabolic Tests

Plasma and fibroblast very long chain fatty acids
Catalase immunofluorescence
Plasmaolgens

Indication for Test

Dihydroxyacetone phosphate acyltransferase (DHAP-AT) is a peroxisomal enzyme catalysing the first step in ether-phospholipid biosynthesis. DHAP-AT deficiency is indicative of a peroxisomal disorder, either as a single enzyme defect resulting in rhizomelic chondrodysplasia punctata or a generalised peroxisomal defect e.g. Zellwegers syndrome. DHAP-AT can be measured in cultured skin fibroblasts or cultured chorionic villus fibroblasts.

Methodology

Principle of the reactions of DHAP-AT

Dihydroxyacetone-phosphate + acyl-CoA $\xrightarrow{\text{DHAPAT}}$ Acyl-dihydroxyacetone-phosphate + CoA-SH

Radiolabelled DHAP-AT is not available commercially. [^{14}C]DHAP is prepared from [^{14}C]glycerol-3-phosphate in the presence of pyruvate and lactate dehydrogenase during the pre-incubation stage of the assay.

The [^{14}C]DHAP is then incubated with palmitoyl-CoA as the second substrate. The product acyl-[^{14}C]dihydroxyacetone-phosphate is separated from the substrate [^{14}C]DHAP and quantified.

Sample requirements

Skin biopsy for fibroblast culture or cultured fibroblasts.
Cultured chorionic villus fibroblasts

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

Camilla Scott (Consultant Clinical Scientist)
0114 2717307

Turn Around Time

6 – 8 weeks. This may be longer if the cells do not grow adequately.

Reference Ranges

Interpretation will be provided with the report.

References

- Measurement of dihydroxyacetone-phosphate acyltransferase (DHAPAT) in chorionic villous samples, blood cells and cultured cells. RJA Wanders et al. J. Inher. Metab. Dis. 18 Suppl. 1(1995) 90 - 100