

## DIHYDROXYACETONE PHOSPHATE ACYLTRANSFERASE (DHAP-AT)

### Relevant disorders

Peroxisomal biogenesis disorders.  
Rhizomelic chondrodysplasia punctata (RCDP)

### Related Metabolic Tests

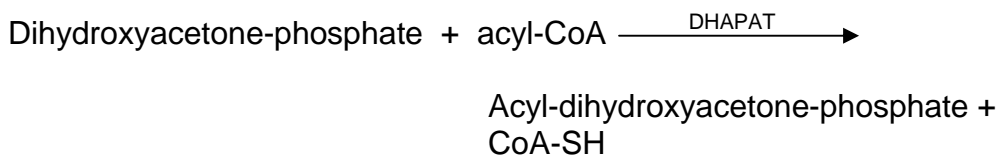
Plasma and fibroblast very long chain fatty acids  
Catalase immunofluorescence  
Plasmaolgens

### 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



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

The [<sup>14</sup>C]DHAP is then incubated with palmitoyl-CoA as the second substrate. The product acyl-[<sup>14</sup>C]dihydroxyacetone-phosphate is separated from the substrate [<sup>14</sup>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