DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

CARNITINE PALMITOYL TRANSFERASE TYPE I

Relevant disorders

Carnitine Palmitoyl Transferase Type 1 deficiency

Related Metabolic Tests

Plasma acylcarnitines

Indication for Test

CPTI is a key enzyme of the carnitine shuttle and is the major control point for long chain fatty acid oxidation in the mitochondria. A deficiency of this enzyme constitutes a rare disorder of fatty acid oxidation presenting as liver disease with hypoglycaemia and Reye-like illness. The disease is usually readily amenable to dietary therapy.

Methodology

In the presence of TWEEN-20 CPTI is not active so only CPTII is being measured.

Palmitoyl-CoA + *carnitine $\rightarrow$ palmitoyl*carnitine + CoA

* = [methyl^{14}C]

The assay is performed with and without TWEEN-20. CPTI activity is calculated by subtracting the activity with TWEEN (CPTII activity) from the activity without TWEEN (CPTII + CPTI.

Sample requirements

Skin biopsy for fibroblast culture or cultured 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

Simon Olpin (Consultant Clinical Scientist)
0114 2717267

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

- Method from Professor RJA. Wanders Amsterdam (not published)
- Olpin SE, Allen JC, Bonham JR, Clark S, Clayton PT, Calvin J, Downing M, Ives K, Jones S, Manning NJ, Pollitt RJ, Standing SJ, Tanner MS. Features of CPTI deficiency. J Inher Metab Dis 24:35-42 2001.
- The paradox of the carnitine palmitoyltransferase type 1a P479L variant in the Canadian Aboriginal population. Greenberg CR, Dilling LA, Thompson GR, Seargeant LE, Haworth JC, Phillips S, Chan A, Vallance HD, Waters PP, Sinclair G, Lillquist Y, Wanders RJ, Olpin SE. Mol Genet Metab. 2009 Apr;96(4):201-7. Epub 2009 Feb 13