DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

CARNITINE PALMITOYL TRANSFERASE TYPE II

Relevant disorders

Carnitine Palmitoyl Transferase Type II (CPTII)

Related Metabolic Tests

Organic acids
Acylcarnitines

Indication for Test

Carnitine palmitoyl transferase type II is a rare inherited disorder of the carnitine shuttle resulting in defective long chain fatty acid oxidation. The severe neonatal form with very low residual enzyme activity (<10%) leads to severe neonatal disease, sometimes with congenital abnormalities, and hepatic, cardiac and muscle involvement which is invariably fatal. There is however a spectrum of disease with a less severe infantile/juvenile form and these patients may survive if detected before significant clinical damage has occurred. The 'adult' form of the disease has higher residual enzyme activity with only skeletal muscle involvement which manifests as exercise intolerance, muscle pains on prolonged exercise which can result in rhabdomyolysis and renal failure. The 'adult' form of the disease should be considered as part of the differential diagnosis in any older child or young adult presenting with exercise induced muscle pains and rhabdomyolysis. Muscle symptomatology can also be induced by viral infections in some instances. Some heterozygotes can be symptomatic.

Methodology

$[^{14}C]$-Palmitoyl-carnitine + CoA ----$\rightarrow$ $[^{14}C]$-Palmitoyl-CoA + carnitine

^{14}C radiolabeled palmitoyl-carnitine is incubated with fibroblast cell sonicate. The radiolabelled palmitoyl-CoA produced is extracted from the non-used palmitoyl carnitine by means of DEAE filter paper that binds CoA esters.

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

- Personal communication from Professor RJA Wanders ARC Amsterdam (1997)

- Olpin et al (2003) Mutation and biochemical analysis in CPT II deficiency. J Inher Metab Dis **26**: 543-557.