CARNITINE-ACYLCARNITINE TRANSLOCASE

Relevant disorders

Carnitine translocase deficiency

Related Metabolic Tests

Plasma acylcarnitine

Indication for Test

Carnitine-acylcarnitine translocase is a key enzyme of the carnitine shuttle. It acts between the outer mitochondrial membrane enzyme CPTI and the inner mitochondrial membrane enzyme CPTII and functions in transporting acylcarnitines of C10-C20 between these two sites in exchange for free carnitine. Deficiency of this translocase usually results in severe neonatal disease. Cardiac dysfunction with hypoketotic hypoglycaemia and hyperammonaemia are prominent features of the disorder, often leading to sudden cardiac arrest. Milder forms of the disease have been described where infantile hypoketotic hypoglycaemia without cardiac involvement forms the typical presentation.

Methodology

$$[2\text{-}^{14}C]\text{pyruvate is oxidised to } [^{14}C]\text{acetyl-CoA} \xrightarrow{\text{CATR}} [^{14}C]\text{acetylcarnitine}$$

[^{14}C]acetylcarnitine is produced by carnitine-acylcarnitine translocase and this compound is separated from untransformed label by an ion exchange column. Activity is calculated in relation to the fibroblast protein used in the assay.

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

- Pande et al. 1993 J. Clin. Invest. 91 1247-1252.
- Olpin SE, Bonham RJ, Downing M, Manning NJ, Pollitt RJ, Sharrard M, Tanner MS. Carnitine-acylcarnitine translocase deficiency- a mild phenotype. J. Inher. Metab. Dis 20, 714 - 715; 1997