Relevant disorders
Primary carnitine deficiency

Related Metabolic Tests
Acylcarnitines

Indication for Test

L-carnitine (β-hydroxy-γ-trimethylaminobutyric acid) is an essential requirement for transporting long-chain fatty acids across the inner mitochondrial membrane prior to β-oxidation. L-carnitine is transported by a high affinity (Km 3.2µm in fibroblasts) cationic transporter OCTN2 which is present on the plasmalemma membrane of most cell types including heart, gut, renal tubule, muscle and fibroblasts. Defects in this cationic transporter give rise to Primary Carnitine Deficiency or OCTN2 transporter deficiency (CTD). Patients generally present in the first few years of life with cardiomyopathy and/or Reye-like illness with hypoketotic hypoglycaemia often in association with intercurrent illness or other metabolic stress. If diagnosed in time the disorder is adequately treated by oral carnitine (50mg/kg/day) in bolus doses. More recently some asymptomatic adults with OCTN2 transporter deficiency have been described.

Methodology

Measurement of the rate of uptake of labelled L-carnitine into cultured fibroblasts in culture medium containing L-carnitine at a concentration of 5µm detects patients with CTD.

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
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

