DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

PRIMARY CARNITINE DEFICIENCY

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

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

- Treem WR, Stanley CA, Finegold DN, Hale DE, and Coates PM. Primary Carnitine Deficiency due to a Failure of Carnitine Transport in Kidney, Muscle and Fibroblasts. NEJM. 1988: 319, 20; 1331-1335.

- Lamhonwah A-M, Olpin SE, Pollitt RJ, Vianey-Saban C, Divry P, Guffon N, Besley GTN, Onizuka R, De Meirleir LJ, Cvitanovic-Sojat L, Baric I, Dionisi-Vici C, Fumic K, Maradin M, Tein I. Lack of Genotype-Phenotype Correlation in Eleven Individuals with Novel OCTN2 Mutations: Early Carnitine Therapy Prevents Cardiomyopathy. American Journal of Medical Genetics 2002.

- Lamhonwah A-M, Onizuka R, Olpin SE, Muntoni F, Tein I. OCTN2 mutation (R254X) found in Saudi Arabian Kindred: Recurrent mutation or founder mutation? J Inher Metab Dis 2004; 27: 473-476.