DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

PYRUVATE CARBOXYLASE

Relevant disorders

Pyruvate carboxylase deficiency

Related Metabolic Tests

Plasma lactate, Pyruvate, Ammonia

Indication for Test

The possibility of pyruvate carboxylase deficiency should be considered in any child presenting with lactic acidosis and neurological abnormalities, especially if associated with hypoglycaemia, hyperammonaemia or ketosis.

Methodology

In the presence of excess pyruvate and acetyl CoA, fibroblast pyruvate carboxylase 'fixes' the $^{14}CO_2$ from labelled sodium bicarbonate into oxaloacetate. The reaction is stopped by adding acid and the protein precipitated. Unfixed label is driven off by drying down and the fixed label counted. Activity is related to the amount of 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 is provided with the report. PC activity in fibroblasts is severely decreased to less than 5% of normal in all patients with the French phenotype, varies from 5 to 23% of controls in patients with the North American phenotype and is less than 10% of controls in patients with the benign variant.

References

- Bartlett K., Dale G., Green A., Leonard LV. Studies on cultured fibroblasts from patients with defects of biotin-dependent carboxylation. J. Inher. Metab. Dis. **4** (1981) 183-189

- Fernandes, Saudubray, van den Berghe, Walter. Inborn Metabolic Diseases, 4th Edition. 2006. pages 162 – 165.

- Dr Peter Berman, Red Cross Children's Hospital, South Africa. Personal communication