

PROPIONYL CoA CARBOXYLASE

Relevant disorders

Propionic acidaemia (propionic aciduria)

Related Metabolic Tests

Organic acids
Acylcarnitines

Indication for Test

To aid in the confirmation of propionic acidaemia after detection of increased excretion of typical organic acids (3 hydroxypropionate, methylcitrate, propionylglycine, tiglylglycine) and/or increased propionylcarnitine on an acylcarnitine profile.

Methodology



* $\text{NaH}^{14}\text{CO}_3$

In the presence of excess propionyl-CoA and Mg^{++} ions (and K^+) fibroblast propionyl-CoA carboxylase 'fixes' the labelled $^{14}\text{CO}_2$ from labelled sodium bicarbonate into methylmalonyl-CoA. The reaction is stopped by 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 will be provided with the report.

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
- Gibson KM, Lee CF, Kamali V, and Oddmund S. *Clinica Chimica Acta.* 205: 127 - 135, 1992
- Sweetman FR, Gibson KM et al. (1986) *Prenatal Diagnosis* 6:187-194