

FUMARATE HYDRATASE

Relevant disorders

Fumarate hydratase deficiency

Related Metabolic Tests

Urine organic acids

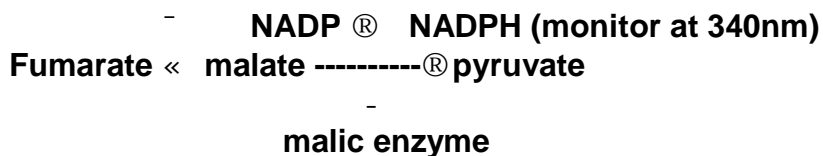
Indication for Test

Fumarate hydratase deficiency is a rare autosomal recessive disorder of Krebs's citric acid cycle. Most patients have severe neurologic impairment including hypotonia, seizures, and cerebral atrophy. The deficiency is usually confirmed in fibroblasts or lymphocytes.

Methodology

This kinetic assay involves monitoring the production of NADPH at 340nm using a coupled enzyme system.

Fumarate Hydratase (amniocytes, fibs, lymphocytes)



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

Joanne Croft (Clinical Scientist)
0114 2717307

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

- Hatch MD. 1978 A simple spectrophotometric assay for fumarate hydratase in crude tissue extracts. *Anal. Biochem* 1978, 85(1); 271-275.