

GALACTOSE-1-PHOSPHATE URIDYL TRANSFERASE

Also known as:

Gal-1-PUT
Galactosaemia screen
Beutler test

Relevant disorders

Classical galactosaemia (Galactose-1-phosphate uridylyltransferase deficiency);

Indication for Test

Galactose is normally metabolised to glucose via galactose-1-phosphate to glucose-1-phosphate and requires the enzyme galactose-1-phosphate uridylyl transferase (GIPUT).

Deficiency of this enzyme, known as classical galactosaemia is the most common disorder of galactose metabolism (incidence approx 1 in 40,000), and is inherited as an autosomal recessive condition. Affected infants appear normal at birth but symptoms appear in the first few days of life and include jaundice, FTT, hepatomegaly and liver failure, vomiting, cataracts and septicaemia. If the disease is undiagnosed and untreated, death may ensue, so early detection and removal of galactose from the diet is essential; soya milk is an alternative.

Screening tests based on the detection of galactose in blood or urine may be useful but will only be reliable for babies on full milk feeds. The 'Beutler' screening test for the enzyme galactose-1-phosphate uridylyl transferase (GIPUT) is far more reliable and allows screening while the baby is on a galactose-free diet, provided that an exchange transfusion has not been given (it will be necessary to wait at least 3-4 months for all donor erythrocytes to be replaced).

Methodology

When normal red cell haemolysates are provided with NADP, galactose -1-phosphate and UDP glucose at a suitable pH, then NADPH will be produced. The NADPH fluoresces when excited with long wave-length U.V. light; NADP does not. If uridylyl transferase is absent no NADPH and hence no fluorescence is produced. The fluorescence is conveniently seen if the substrate/haemolysate mixture is spotted onto a filter paper and the paper examined under U.V. light.

Sample requirements

0.5ml Lithium heparin whole blood – venous or capillary (NOT EDTA, Fluoride or Citrate).

Dried blood spot - Guthrie card, 2 spots (if no whole blood is available or if this is the only sample available prior to a transfusion).

N.B.: This test is not valid if a blood transfusion has been given in the preceding 3 – 4 months.

Turn Around Time

2 days if whole blood is sent

1 week if blood spots are sent

Transport information/Contact details

Send by first class post.

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Reference Ranges

Interpretation will be provided as part of the report.

References

- Beutler and Baluda (J. Lab. Med. 68 (1966) 137)