GALACTITOL

Relevant disorders

Classical galactosaemia (Galactose-1-phosphate uridyltransferase deficiency);
Galactokinase deficiency; UDP-galactose epimerase deficiency

Related Metabolic Tests

Galactose-1-phosphate uridyl transferase (GAL-1-PUT) (aka Beutler test)

Indication for Test

In patients in whom the differential diagnosis includes galactosaemia but who have had a blood transfusion in the last 3 months.
The usual screening test for galactosaemia (i.e. the Beutler test) may be invalidated if recently transfused with erythrocytes, while urinary galactitol measurements are unaffected by a recent transfusion.

Methodology

Urinary galactitol is quantitated by GCMS using a stable isotope of galactitol (D2-galactitol) by single ion monitoring (SIM) on the Agilent 5973 GC-MS.

Sample requirements

5 ml random ('spot') urine in a plain universal tube – samples collected into boric acid are unsuitable.

Turn Around Time

5 – 14 days

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

Reference Ranges

Literature values:
Untreated classical galactosaemia: 8,000-69,000 µmol/mmol creatinine
Treated classical galactosaemia: 45-900 µmol/mmol creatinine
Untreated compound heterozygotes: 96-170 µmol/mmol creatinine.

N.B: If the patient hasn't been exposed to galactose (i.e. no milk feeds and galactose is not a constituent of TPN) the result cannot be used to exclude classical galactosaemia.

References

- Jakobs *et al.* (1995) Eur. J. Pediatr. **154 (7 Suppl 2)** S50-2