GLYCOSAMINOGLYCANS - Quantitative

Also Known as:

GAGs, DMB test

Relevant disorders

Mucopolysaccharidoses

Indication for Test

Mucopolysaccharidoses (MPS) are a group of inherited diseases which are characterised by a progressive accumulation of partially degraded glycosaminoglycans (GAGs) in the lysosomes. The abnormal accumulation is caused by a deficiency in one of the enzymes responsible for GAG degradation and results in cell, tissue and organ dysfunction. The defects are inherited recessively except MPS II (Hunter) which is X-linked. There is a wide spectrum in the clinical phenotypes within a single enzyme deficiency ranging from mild to severe. Affected children usually appear normal at birth but (in most MPS) subsequently develop progressive connective tissue changes, skeletal deformities as well as hepatomegaly. Depending on the MPS type there may be progressive psychomotor retardation and cardiac disease.

Methodology

The dimethyl methylene blue assay provides a direct dye-binding method for quantification of urinary glycosaminoglycans; this uses the metachromatic properties of the histochemical dye 1,9 dimethylmethylene blue.

Sample requirements

10ml random urine or aliquot of 24 hour urine collection.

Turn Around Time

4 – 6 weeks

Transport/Contact information

Send by first class post to:

Department of Clinical Chemistry
Sheffield Children's NHS Foundation Trust
Western Bank, Sheffield
S10 2TH, UK

Jennifer Watkinson (BMS 3)
0114 2717445

Reference Ranges

Urines from children with mucopolysaccharide disorders typically have elevated results of at least twice the upper limit of normal, however the increase may be less marked in older children with MPS Types III and IV, and also those with milder disorders. All significantly elevated results should be followed up with electrophoresis of the glycosaminoglycans.
Some of the other lysosomal storage disorders may show slightly elevated results but this is NOT a reliable screening test for these disorders, enzyme analysis is required.
Children on heparin therapy will have elevated results.

Reference ranges valid only with results produced by Clinical Chemistry, Sheffield Children's Hospital.

Age		**Glycosaminoglycans mg/mmol creatinine**
0-1	mth	22.1-40.8
1-3	mths	9.2-38.8
3-6	mth	11.9-34.5
6-12	mth	4.2-30.5
1-2	yrs	6.8-21.7
2-3	yrs	9.7-19.5
3-5	yrs	6.2-15.4
5-7	yrs	6.2-12.1
7-9	yrs	4.1-10.8
9-11	yrs	4.5-10.8
11-13	yrs	2.8-10.4
13-15	yrs	2.0-7.6
over 15	yrs	1.7-4.4

References

- Whitley et al. CLIN. CHEM. 35/3,374-9 (1989)