DEPARTMENT OF CLINICAL CHEMISTRY
SHEFFIELD CHILDREN'S NHS FOUNDATION TRUST

GLYCOSAMINOGLYCANS - Qualitative

Also Known as:

GAGs, Glycosaminoglycan electrophoresis, Mucopolysaccharides

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

Anionic GAGs in urine complex with and precipitate the cationic dye Alcian Blue. This process is enhanced by the addition of magnesium chloride. The precipitated complex is dissolved in sodium chloride and methanol and then disassociated with sodium carbonate. The Alcian Blue precipitates and is centrifuged down leaving the GAGs in solution. These are then isolated by precipitation with ethanol. The aqueous solution of the GAGs are separated by low voltage electrophoresis in two dimensions on cellulose acetate sheets and stained with Alcian Blue.

Sample requirements

10ml random Urine sample or aliquot of a 24 hour 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

Interpretation is provided with the report.

References

- Laboratory Guide to the Methods in Biochemical Genetics. Eds. Nenad Blau, Marinu Duran, K.Michael Gibson. Published by Springer. 2008. Chapter 4.1 Mucopolysaccharides. Written by Zoltan Lukacs