

# Glycosaminoglycans (GAGS)

## Synonyms

GAGS, mucopolysaccharides

## Clinical Indication

For the investigation of possible mucopolysaccharidosis. These are rare disorders due to defects in lysosomal enzymes responsible for the hydrolysis of long chain mucopolysaccharides (MPS)/glycosaminoglycans (GAGS). These polymers accumulate in cells, blood and connective tissues resulting in permanent and progressive damage. The clinical phenotype is heterogenic. Patients, typically children present with symptoms of:

- Hepatomegaly & cardiomyopathy
- Skeletal deformities
- Abnormal faces – macrocephaly, coarse features, short nose, flat face
- Behavioural problems – developmental delay, SAL problems, hyperactivity
- Inguinal and umbilical hernias
- Loss of developmental skills

## Part of Profile / See Also

## Request Form

Combined Pathology manual blood request form or ICE request

## Availability / Frequency of Analysis

Referred test: Sample sent to Great Ormond Street Hospital [8692](#) for analysis

## Turnaround Time

GAG quantification - 7 days  
 GAG typing - 21 days

## Patient Preparation

## Sample Requirements

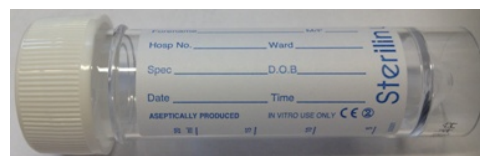
### Specimen Type

Urine

### Volume

5.0 ml

### Container



White-top universal

**Samples should be transported to laboratory immediately.**

## Reference Range & Units

Please see the report.

## Interferences

## Interpretation & Clinical

## Decision Value (if applicable)

Initially, samples are screened for total glycosaminoglycan concentration. False positive results are common, particularly in young infants. Positive results will be referred for typing by electrophoresis if clinical details suggestive of a mucopolysaccharidosis are given. False negative quantitative results may also be encountered, therefore if there is a strong clinical suspicion of a mucopolysaccharidosis, please specifically request GAG typing. If urine glycosaminoglycan typing and white cell enzymes are normal and a storage disorder is still suspected clinically, urinary oligosaccharide and sialic acid analysis should be considered.

## References

Test code

Lab Handling



8692

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ISO 15189:2012