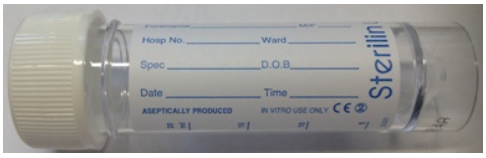
Glycosaminoglycans (GAGS)

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 phenoytpe 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

UKAS MEDICAL
8692

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ISO 15189:2022