TEST

Acid mucopolysaccharides

This urine test measures glycosaminoglycans to help screen for certain inherited storage disorders.

WHAT IT MEASURES
Glycosaminoglycans released into urine

SAMPLE
Urine

FASTING NEEDED?
No

HOW LONG IT TAKES
About 5 to 10 minutes

RESULTS IN
Often several days, varying by laboratory and method

USED FOR
Screening and supporting diagnosis of mucopolysaccharidoses

MAIN LIMITATION
Normal levels can miss some mucopolysaccharidoses

Overview

Acid mucopolysaccharides is an older name for glycosaminoglycans , often shortened to GAGs. These are long sugar molecules found in cartilage, skin, connective tissue and the fluid around joints.

The test usually measures GAGs in urine. Some versions measure the total amount. More detailed methods identify particular GAGs, such as dermatan sulfate, heparan sulfate and keratan sulfate.

Your body normally breaks down old GAGs inside structures called lysosomes. Lysosomes act as recycling centres within cells. In a group of inherited conditions called mucopolysaccharidoses , or MPS disorders, a missing or poorly working enzyme interrupts this process. GAGs can then build up in tissues and pass into urine.

This is mainly a screening test. An abnormal result may support concern about an MPS disorder, but it does not usually confirm the condition or identify the exact type by itself.

Why the test is ordered

A clinician may order urine acid mucopolysaccharides when a person has a combination of findings that could occur with an MPS disorder. These may include unusual bone or joint changes, reduced joint movement, frequent ear or airway problems, hearing loss, enlarged liver or spleen, clouding at the front of the eye, or changes in development.

Many more common conditions can cause individual findings on this list. The test is used as one part of a broader assessment rather than as a diagnosis based on symptoms alone.

The test may also be ordered:

Not every laboratory uses the same screening method. Some centres now use tests that measure specific GAG fragments rather than only measuring total GAGs.

- After an abnormal newborn screening result
- When an MPS disorder is present in the family
- Alongside enzyme or genetic testing
- To follow GAG levels during treatment

Who should have the test

This test is most useful when a paediatrician, metabolic specialist or genetics team thinks an MPS disorder is possible. It is not generally used as a routine health screening test.

Relatives of someone with a confirmed MPS disorder may need testing. The appropriate test often depends on the exact family diagnosis. Testing the relevant enzyme or family genetic change may be more direct than a urine screen.

A normal urine result may not be enough to rule out an MPS disorder when clinical concern remains. This is particularly important with milder disease and with some MPS types that may release only small amounts of GAGs into urine.

How to prepare

Fasting is not normally needed. You can usually eat and drink as usual unless your collection instructions say otherwise.

Avoid deliberately drinking a large amount of water immediately before providing the sample. Very dilute urine can make substances harder to detect. Normal hydration is appropriate.

Tell the requesting team about medicines and supplements, pregnancy, kidney problems, and any treatment for an MPS disorder. Do not stop prescribed medicine unless you have been specifically told to do so.

The laboratory may request a random sample, a first-morning sample or, less commonly, urine collected over a set period. Follow the instructions supplied with your container because collection requirements differ between laboratories.

What happens during the test

You will usually provide urine in a clean specimen container. Wash your hands and avoid touching the inside of the container or lid.

For a clean-catch sample, you may be asked to begin urinating into the toilet and then collect urine partway through the flow. Young children may need a collection bag or another age-appropriate collection method.

The container should be closed firmly and labelled as instructed. Some samples must be refrigerated or delivered promptly because storage conditions can affect the GAGs being measured.

No blood draw or imaging is needed for a urine-only test.

Understanding your results

Results may report a total GAG concentration, a GAG-to-creatinine ratio, individual GAGs, or a pattern produced by electrophoresis or mass spectrometry. Creatinine is a waste product used to help account for how concentrated the urine is.

An increased result means more GAG material was detected than expected for the laboratory's comparison group. The pattern may suggest a particular MPS group, but other tests are needed to confirm the cause.

A result within the laboratory's expected range makes some MPS disorders less likely. It does not exclude every type, especially a milder form or a disorder that produces less urinary GAG.

There is no single universal reference range. Ranges and interpretation vary by laboratory, test method, age, sex, pregnancy status, medicines and clinical context. Age is especially important because young children normally release more GAGs in urine than older children and adults.

Your result should be interpreted with your medical history, examination findings and any enzyme or genetic test results.

What affects the results

Several factors can change the result without indicating a laboratory error:

Pregnancy can change kidney handling and urine concentration, so the laboratory should know if you are pregnant. There are few common day-to-day activities known to have a large direct effect, but following the collection instructions remains important.

A recent blood transfusion does not usually change urinary GAGs directly. However, it can affect some other metabolic or genetic investigations performed at the same time.

- Age: GAG excretion is normally highest in young children and decreases with age.
- Urine concentration: A very dilute sample may produce a lower measured concentration. Very concentrated urine can produce a higher one.
- Creatinine: Kidney function and very low muscle mass can affect urine creatinine, which may alter a GAG-to-creatinine ratio.
- Collection and storage: Contamination, an incomplete timed collection, delayed delivery or incorrect storage may make the sample unsuitable.
- Test method: Older dye-based tests measure total GAGs and may be less able to detect small or unusual increases. Electrophoresis and mass spectrometry provide different information.
- MPS type and severity: Some types, particularly milder forms and some cases of MPS III or MPS IV, may not produce a clearly abnormal total urine result.
- Treatment: Enzyme replacement therapy or a stem cell transplant can lower urinary GAG levels. Results must be compared with treatment timing and previous measurements.

Accuracy and limitations

Accuracy differs according to the method, the person's age and the MPS type being investigated. There is no single useful percentage describing how often every version of this test is right.

A total urinary GAG screen can miss disease when GAG levels are only mildly increased, when the urine is very dilute or when the disorder releases GAGs that the method detects poorly. This means a negative result can occasionally occur even when an MPS disorder is present.

An increased result does not prove that a person has an MPS disorder. Total GAG measurements may be affected by age, urine concentration and other biological factors. A pattern suggesting one type may also overlap with another type.

Tests that directly measure selected GAG fragments can detect some disorders more reliably than older total-GAG methods. Even so, a screening result normally needs confirmation.

The better confirmatory test is usually an assay of the relevant enzyme in blood cells, plasma or cultured skin cells. Genetic testing may then identify changes in the associated gene. If clinical concern remains despite a normal urine screen, these more specific tests may still be appropriate.

Risks and complications

Providing a urine sample is non-invasive and has no significant physical risk. Collection bags or cleaning products may occasionally irritate sensitive skin.

The main practical risk is an inaccurate result from contamination, dilution or incorrect storage, which may mean the sample needs to be collected again.

What happens next

A result within the expected range may be reviewed at a routine follow-up appointment. If concern is low, no further testing may be needed.

If the result is abnormal or unclear, the next steps may include repeat urine testing, enzyme activity testing, genetic testing or referral to a metabolic or genetics service. These tests help determine whether an MPS disorder is present and, if so, which type.

Do not wait for a routine test result if the person being assessed develops severe breathing difficulty, blue or grey lips, becomes difficult to wake, or has sudden major weakness. These symptoms need emergency assessment regardless of the reason the urine test was ordered. New but non-emergency concerns, such as worsening walking, strength or breathing during sleep, should be reported promptly to the clinical team.

Common questions

Can I drink coffee before the test?

Usually, yes. Fasting is not normally required. Avoid excessive coffee or water immediately beforehand because producing very dilute urine may affect the measurement.

Does the time of day matter?

Many laboratories accept a random urine sample. Others prefer first-morning urine because it

is often more concentrated. Use the collection time stated on your request or container.

Can I exercise before providing the sample?

Ordinary activity is not known to have a major direct effect on urinary GAGs. If the laboratory gives activity restrictions, follow those instructions.

Should I stop my medicines?

No, not unless the requesting clinician specifically tells you to. Provide an up-to-date list of medicines, supplements and any treatment for an MPS disorder.

Will I need to collect urine for 24 hours?

Usually not. Many current tests use one random urine sample, but some laboratories may request a timed collection.

Does a normal result rule out an MPS disorder?

No. Some milder forms and certain MPS types can produce a normal or borderline urine screen. More specific enzyme or genetic testing may be needed when clinical concern remains.

Sources

- ARUP Consult: Mucopolysaccharidoses - Choose the Right Test
- Mayo Clinic Laboratories: Mucopolysaccharidosis, Quantitative, Random, Urine
- GeneReviews: Mucopolysaccharidosis Type I
- GeneReviews: Mucopolysaccharidosis Type II
- GeneReviews: Mucopolysaccharidosis Type III
- Stapleton M, Arunkumar N, Kubaski F, et al. "Clinical presentation and diagnosis of mucopolysaccharidoses." Molecular Genetics and Metabolism . 2018;125(1-2):4-17.