TEST

PBG urine test

A PBG urine test measures porphobilinogen to help diagnose an acute porphyria attack.

WHAT IT MEASURES
Porphobilinogen, a heme-making chemical, in urine

SAMPLE
Urine

FASTING NEEDED?
No

HOW LONG IT TAKES
About 5 to 10 minutes to collect

RESULTS IN
Often several days; timing differs by laboratory

USED FOR
Diagnosing an acute porphyria attack

MAIN LIMITATION
Dilute or delayed samples can hide an abnormal result

Overview

A PBG urine test measures porphobilinogen in urine. PBG is a substance made during the body's production of heme, a part of haemoglobin and several important enzymes.

Small amounts of PBG are normally produced. A large increase can occur during attacks of certain acute porphyrias. These are rare inherited conditions involving the pathway that makes heme.

The test is mainly used when symptoms suggest an acute porphyria attack. It is not a routine screening test for people without symptoms. A random, or "spot," urine sample collected while symptoms are happening is usually more useful than a 24-hour collection.

PBG is commonly measured with urine aminolevulinic acid, called ALA, and urine creatinine. Creatinine helps the laboratory judge whether a sample is unusually dilute.

Why the test is ordered

The test may be ordered when symptoms could be caused by an acute hepatic porphyria.

"Hepatic" means related to the liver. These conditions include acute intermittent porphyria, variegate porphyria, and hereditary coproporphyria.

Possible reasons for testing include an otherwise unexplained combination of:

These symptoms have many more common causes. The test helps separate acute porphyria from those other possibilities; symptoms alone cannot confirm it.

PBG may also be checked in someone already known to have acute porphyria when it is unclear whether new symptoms represent an attack. It is generally not useful for monitoring symptoms day to day without a specific clinical reason.

- Severe or repeated abdominal pain
- Nausea, vomiting, or constipation
- Fast heartbeat or high blood pressure
- Muscle weakness, tingling, or numbness
- Confusion, unusual behaviour, or seizures
- Dark or reddish urine, especially during other symptoms

Who should have the test

You may need this test if you are having symptoms that raise concern for an acute porphyria attack, particularly if similar unexplained episodes have happened before.

Testing may also be appropriate if a close biological relative has a confirmed acute porphyria and you develop compatible symptoms. Genetic testing is often used separately to identify relatives who carry a known family variant.

A PBG urine test is not a general test for abdominal pain, fatigue, or dark urine. Other urine, blood, or imaging tests may be more appropriate depending on your symptoms.

How to prepare

You do not usually need to fast. You can normally eat, drink, and take your usual medicines unless you have been given different instructions.

Tell the healthcare team about prescription medicines, non-prescription medicines, supplements, and any recent treatment with hemin. Hemin is a treatment used for acute porphyria attacks and can lower PBG levels.

If possible, collect the sample while significant symptoms are present and before

porphyria-specific treatment begins. Urgent treatment should not be delayed solely to obtain a sample.

Avoid drinking an unusually large amount of water immediately before collection. Excess fluid can make the urine too dilute. Follow the laboratory's container, storage, and transport instructions carefully.

What happens during the test

You will usually provide a fresh urine sample in a labelled container. A midstream clean-catch sample may be requested: begin urinating into the toilet, collect some urine in the container, then finish in the toilet.

A spot sample is generally preferred for investigating a possible acute attack. A 24-hour collection is rarely needed for this purpose and may delay testing or produce misleading results if some urine is missed.

PBG can break down if the sample is left warm for too long. The container is usually protected from light, refrigerated, and sent promptly to the laboratory. The exact handling instructions differ between laboratories.

Understanding your results

Results may be reported as the amount of PBG per litre of urine or relative to urine creatinine. There is no single range that applies everywhere. Ranges and interpretation vary by laboratory, test method, age, sex, pregnancy status, medicines, kidney function, and clinical context.

A clearly raised PBG level during symptoms strongly supports an acute attack caused by acute intermittent porphyria, variegate porphyria, or hereditary coproporphyria. The result does not show which of these conditions is present. Further urine, blood, stool, or genetic testing may be needed.

A normal PBG result from a properly collected sample during active symptoms makes those three acute porphyrias much less likely as the cause of the current episode. However, the result may be misleading if the urine was very dilute, stored incorrectly, collected after symptoms settled, or obtained after hemin treatment.

A rare condition called ALA dehydratase deficiency porphyria may raise ALA without substantially raising PBG. Lead poisoning can also raise ALA. This is one reason ALA is often measured with PBG.

A small or borderline PBG increase is less specific than a large increase. It should be interpreted with the symptoms, urine concentration, testing method, and other laboratory findings.

What affects the results

Several factors can alter the measured level without there being a laboratory mistake:

Give the laboratory and healthcare team an accurate medicine list. Some medicines can also trigger attacks in a person with acute porphyria, although this is different from directly interfering with the test.

- Timing: PBG is most informative during symptoms. Levels can fall after an attack, sometimes quickly in variegate or hereditary coproporphyria.
- Dilute urine: Heavy fluid intake, intravenous fluids, or naturally dilute urine can hide an increase. Reporting PBG relative to creatinine helps but does not remove this problem completely.
- Storage and transport: Warm temperatures, light exposure, and long delays can allow PBG to break down.
- Treatment: Hemin treatment can reduce PBG. A sample collected after treatment may not reflect the earlier level.
- Kidney function: Reduced kidney function can change how PBG and creatinine appear in urine. Blood testing may sometimes be considered.
- Creatinine differences: Age, sex, pregnancy, muscle mass, and severe illness can affect urine creatinine and therefore a creatinine-adjusted result.
- Collection problems: An incomplete 24-hour collection or a sample taken from the wrong container can produce an unreliable measurement.
- Test method: Older screening methods can react with substances other than PBG. Modern quantitative methods are more specific.

Accuracy and limitations

A properly handled quantitative PBG test collected during active symptoms is good at detecting the common acute hepatic porphyrias when they are causing an attack. Published performance cannot be reduced to one reliable percentage because results are strongly affected by timing, urine concentration, handling, and the method used.

The test can miss an attack if the sample is collected too late, after hemin, or after large amounts of fluid. It can also miss ALA dehydratase deficiency porphyria because that condition may not raise PBG.

Older rapid or qualitative screening tests are more likely to report PBG when another substance caused the reaction. Mild increases may also occur in other illnesses and need confirmation with a quantitative method.

PBG cannot identify the exact porphyria type or show whether a person carries a genetic variant when they have no symptoms. Plasma fluorescence testing, stool porphyrins, enzyme studies, or genetic testing may be better for those questions.

Risks and complications

Providing a urine sample has no significant physical risk. The main practical problems are difficulty collecting the sample, contamination, or leakage during transport.

If urine must be collected through a catheter for another medical reason, catheter-related risks are separate from the PBG test itself.

What happens next

A clearly abnormal result is usually followed by confirmatory biochemical testing and testing to identify the porphyria type. Genetic testing may then confirm the responsible variant and help with family testing.

A normal result may lead to investigation of other causes of the symptoms. If the sample was dilute, delayed, or collected after symptoms had improved, repeat testing during symptoms may be considered.

Routine follow-up is appropriate for questions about a result, repeat testing, or family assessment.

Seek urgent medical care for severe or worsening abdominal pain with weakness, persistent vomiting, confusion, or a very fast heartbeat. Call emergency services for trouble breathing, a seizure, fainting, severe confusion, or rapidly worsening muscle weakness. Do not wait for a PBG result if emergency symptoms are present.

Common questions

Can I drink coffee before a PBG urine test?

Usually, yes. Fasting is not normally required. Avoid drinking an unusually large volume of coffee or other fluids just before collection because this can dilute the urine.

Does the time of day matter?

There is usually no required time of day. Collecting the sample while symptoms are active is more important than collecting it in the morning.

Can I exercise beforehand?

Normal activity is not known to directly distort PBG in the way it can affect some blood tests. If exercise worsens your symptoms or leaves you dehydrated, follow the instructions given by the healthcare team.

Why does the sample need protection from light?

PBG and related porphyria markers can become less reliable with poor storage. Laboratories commonly request protection from light, cool storage, and prompt transport.

Do I need to stop my medicines?

Do not stop medicines on your own. Provide a complete medicine and supplement list, and mention any recent hemin treatment.

Is a 24-hour urine collection better?

Usually not for a suspected acute attack. A fresh spot sample collected during symptoms, with PBG reported relative to creatinine, is generally faster and less vulnerable to collection errors.

Sources

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