TEST

Galactose-1-phosphate uridyltransferase blood test

A blood test that measures an enzyme needed to process the milk sugar galactose

WHAT IT MEASURES
Activity of the galactose-processing enzyme GALT

SAMPLE
Blood

FASTING NEEDED?
No

HOW LONG IT TAKES
About five minutes for a blood draw

RESULTS IN
Several days; timing varies by laboratory

USED FOR
Newborn screening and diagnosis of galactosemia

MAIN LIMITATION
Recent transfusion can hide low enzyme activity

Overview

The galactose-1-phosphate uridyltransferase blood test measures the activity of an enzyme called GALT. GALT helps process galactose, a sugar found in milk, breast milk, standard infant formula, and some other foods.

When GALT activity is very low, galactose and related substances can build up in the body. The test is mainly used to screen for or help diagnose classic galactosemia, a rare inherited condition that is present from birth. Galactosemia is different from lactose intolerance.

Newborn screening often uses blood collected by heel prick and dried on a special card. Depending on the screening program, the laboratory may measure GALT activity, total galactose, or both. An abnormal screening result is not a diagnosis and requires further testing.

Diagnostic testing usually measures GALT activity in red blood cells. It may be combined with galactose-1-phosphate measurement and genetic testing of the GALT gene.

Why the test is ordered

The test may be ordered after newborn screening suggests low GALT activity or raised galactose levels. Confirmatory testing should be arranged promptly because early feeding decisions may be important.

It may also be ordered when a baby has symptoms that could have several causes, including:

These findings do not prove that a baby has galactosemia. Testing may also be considered when galactosemia runs in a family or parents carry changes in the GALT gene. Genetic testing is often more useful than an enzyme test for identifying carriers.

- Poor feeding or repeated vomiting
- Yellow skin or eyes, called jaundice
- Poor weight gain
- Unusual sleepiness or low energy
- Liver problems or a serious infection

Who should have the test

Many babies have GALT-related testing through routine newborn screening. Methods and follow-up processes vary by location.

A diagnostic blood test may be appropriate for:

A normal newborn screen makes classic galactosemia less likely, but screening does not identify every affected baby. Diagnostic testing may still be needed if the medical or family history is concerning.

- A baby with an abnormal newborn screening result
- A baby with symptoms that could fit galactosemia
- A sibling of someone with confirmed classic galactosemia
- Someone with a family history who needs clarification of their result

How to prepare

Fasting is not needed. You or your baby can usually eat and drink normally before the sample is taken.

Do not change an infant's milk or formula unless the clinical team responsible for the baby advises it. If there is strong concern about classic galactosemia, temporary feeding changes may be recommended while confirmatory results are pending.

Tell the testing team about any recent blood transfusion. Donor red blood cells may contain normal GALT activity, making the result look more reassuring than it should.

Also mention previous newborn screening results, family history, medicines, and the current feeding plan. Other blood tests collected at the same time may have separate preparation instructions.

What happens during the test

For newborn screening, a clinician warms and cleans the baby's heel, makes a small prick, and places drops of blood onto a screening card. The card is dried and sent to the laboratory.

For diagnostic testing, blood is usually taken from a vein in the arm or hand. The skin is cleaned, and a small needle collects blood into a tube. The draw usually takes only a few minutes. The laboratory then measures GALT activity in the red blood cells.

Understanding your results

Results may be reported as normal, reduced, or very low GALT activity. Some laboratories provide a number with their own reference range.

Very low or absent activity can support a diagnosis of classic galactosemia, but it must be interpreted alongside symptoms, feeding history, galactose-1-phosphate levels, and GALT gene testing.

Partial activity may occur with certain inherited variants, including the Duarte variant, or in carriers with one working and one altered gene copy. These findings do not all have the same health implications, so genetic testing may be needed.

A normal result usually makes classic GALT deficiency unlikely. However, it may be unreliable after a recent transfusion and does not rule out every disorder affecting galactose processing. Always use the reference range and interpretation on the laboratory report.

What affects the results

A recent red blood cell transfusion is the main cause of a misleading result. Donor cells with

normal GALT activity can hide the person's low activity until those cells have cleared. Genetic testing may be more appropriate during this period.

Poor handling may falsely lower enzyme activity. Unsuitable temperatures, delayed transport, or improper storage can damage the sample. The laboratory may request another sample if testing would not be reliable.

Sample type also matters. A dried blood spot used for screening is interpreted differently from a red-cell enzyme test used for diagnosis.

Eating galactose does not correct an inherited lack of GALT. Dietary changes can reduce galactose-1-phosphate and total galactose levels, but GALT activity remains low. Age, pregnancy, ordinary exercise, and time of day are not usually major influences. Report all medicines and supplements.

Accuracy and limitations

A clearly abnormal red-cell GALT result can strongly support classic galactosemia, but it cannot always identify the specific genetic changes or predict the effects of partial activity.

Newborn screening identifies babies who need prompt follow-up; it does not provide a final diagnosis. Screening methods and cutoffs vary. Some unaffected babies screen positive, and an affected baby may occasionally be missed.

False-positive results may be caused by sample handling, screening cutoffs, partial GALT activity, or another cause of raised galactose. Total galactose testing may also detect liver disease or another galactose-processing disorder.

A false-normal enzyme result is a particular concern after transfusion. Genetic testing is generally more reliable in this situation because transfused red cells do not change the baby's inherited DNA.

Genetic testing can help distinguish classic galactosemia, partial deficiency, and carrier status, but it may miss uncommon changes not covered by the chosen method. Enzyme and genetic results are often considered together.

Risks and complications

A heel prick or blood draw may cause brief pain, minor bleeding, or a small bruise. Some people feel faint during a venous blood draw, but serious complications are uncommon.

What happens next

An abnormal newborn screening result usually leads to prompt confirmatory blood tests and specialist assessment. The clinical team may recommend temporary feeding changes while results are pending. Do not make these changes without specific advice.

If GALT activity is low, further testing may include red-cell galactose-1-phosphate, GALT gene analysis, liver tests, and assessments based on the baby's condition. Parents and siblings may be offered testing or genetic counselling.

A normal or mildly reduced result may need only routine follow-up if the baby is well and other tests are reassuring. Repeat testing may be necessary after a transfusion, an unsuitable sample, or an uncertain genetic result.

Seek urgent medical advice if a young baby is feeding poorly, repeatedly vomiting, becoming increasingly sleepy, or developing worsening jaundice. Call emergency services if the baby is difficult to wake, has trouble breathing, has a seizure, becomes limp or unresponsive, or appears severely unwell.

Common questions

Can I drink coffee before the test?

Yes. Fasting is not required. Follow different instructions if other tests are being performed at the same time.

Can my baby feed before the blood test?

Usually, yes. Feeding does not restore missing GALT activity. Do not change breast milk or formula unless the clinical team gives specific instructions.

Does the time of day matter?

No. GALT activity does not change enough during the day to require morning testing.

Can I exercise beforehand?

Ordinary exercise is not expected to affect the result. Ask about restrictions if other blood tests are also being collected.

Why does the laboratory need to know about a transfusion?

The test measures GALT in red blood cells. Donor cells may have normal activity and hide the person's low activity.

Is this the same as a genetic test?

No. The enzyme test measures how well GALT works in red blood cells. A genetic test looks for changes in the GALT gene and may help confirm or explain the result.

Sources

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