

Galactosemia



Does this test have other names?

Galactosemia newborn screening test

What is this test?

This test is part of screening done on all newborns. It looks for high levels of galactose and low galactose-1 phosphate uridytransferase (GALT) in your child's blood. This may mean your child has a condition called galactosemia.

Galactosemia is a rare inherited disorder. It keeps the body from breaking down galactose. Galactose is a sugar found in many foods and in all dairy products. An enzyme called GALT normally breaks down galactose. Low levels of the enzyme cause the high galactose level in the blood. Galactosemia can cause serious problems to the liver, kidneys, and brain.

Why does my child need this test?

This test is part of screening done on all newborns. If the newborn screening test is positive, it doesn't mean your child has galactosemia. More tests are needed to make the diagnosis. If your child has galactosemia, early treatment can help to prevent serious problems.

What other tests might my child have along with this test?

If your child has a positive galactosemia test, they may need other tests, such as:

- Galactosemia reflex test
- Other GALT enzyme tests
- GALT gene blood panel
- Other gene blood tests

What do my child's test results mean?

Test results may vary depending on your child's age, gender, health history, and other things. Your child's test results may be different depending on the lab used. They may not mean your child has a problem. Ask your healthcare provider what your child's test results mean for them.

Positive galactosemia test results usually mean that your child needs more blood tests. Negative results usually mean your child doesn't need more tests to check for this condition.

How is this test done?

This test is done with a blood sample. This is usually taken by a heel prick in the first week of life. For a heel prick, a small needle is used to poke the bottom of your baby's foot. A small drop of blood is taken and tested.

Does this test pose any risks?

Taking a blood sample with a needle carries risks that include bleeding, infection, bruising, or feeling dizzy. When the needle pricks the skin, your child may feel a slight stinging sensation or pain. Afterward, the site may be slightly sore.

What might affect my child's test results?

Factors that may affect your child's newborn screening results include age, other health problems, and treatment, such as blood transfusions or total parenteral nutrition.

How do I get my child ready for this test?

You don't need to do anything to get your child ready for this test.

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