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Galactose-1-phosphate uridyltransferase blood test

Galactose-1-phosphate uridyltransferase is a blood test that measures the level of a substance called GALT, which helps break down milk sugars in your body. A low level of this substance causes a condition called galactosemia.

How the Test is Performed

A blood sample is needed.

How the Test will Feel

When the needle is inserted to draw blood, some infants feel moderate pain. Others feel only a prick or stinging. Afterward, there may be slight bruising. This soon goes away.

Why the Test is Performed

This is a screening test for galactosemia.

In normal diets, most galactose comes from the breakdown (metabolism) of lactose, which is found in milk and dairy products. One out of 65,000 newborns lack a substance (enzyme) called GALT. Without this substance, the body cannot break down galactose, and the substance builds up in the blood. Continued use of milk products can lead to:

- Clouding of the lens of the eye (cataracts)
- Scarring of the liver (cirrhosis)
- Failure to thrive
- Yellow color of the skin or eyes (jaundice)
- Liver enlargement
- Intellectual disability

This can be a serious condition if not treated.

Every state in the United States provides newborn screening tests to check for this disorder.

Normal Results

The normal range is less than 24.5 nmol/hour/mg of hemoglobin.

Normal value ranges may vary slightly among different labs. Some labs use different measurements or may test different samples. Talk to your health care provider about the meaning of your specific test results.

What Abnormal Results Mean

An abnormal result suggests galactosemia. Further tests must be done to confirm the diagnosis.

If your child has galactosemia, a genetics specialist should be consulted promptly. The child should be put on a no-milk diet right away. This means no breast milk and no animal milk. Soy milk and infant soy formulas are generally used as substitutes.

This test is very sensitive, so it does not miss many infants with galactosemia. But, false-positives can occur. If your child has an abnormal screening result, follow-up tests must be done to confirm the result.

Risks

There is little risk in taking blood from an infant. Veins and arteries vary in size from one infant to another and from one side of the body to the other. Obtaining a blood sample from some infants may be more difficult than from others.

Other risks associated with having blood drawn are slight but may include:

- Excessive bleeding
- Multiple punctures to locate veins
- Fainting or feeling lightheaded
- Hematoma (blood accumulating under the skin, causing bruising)
- Infection (a slight risk any time the skin is broken)

Alternative Names

Galactosemia screen; GALT; Gal-1-PUT

References

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