

In 1998, The Australian Society for Inborn Errors of Metabolism published a handbook for people with Galactosaemia

How is Galactosaemia diagnosed?

In South Australia, Galactosaemia is usually diagnosed by the newborn screening programme. All babies have 4 spots of blood collected a few days after they are born. This blood is tested for signs of several different diseases, including Galactosaemia. The chemical which is tested when looking for Galactosaemia is called galactose-1-phosphate. Sometimes, if a baby with Galactosaemia is very sick before the blood spot is collected, the doctors looking after the baby may suspect the diagnosis and measure the **galactose - 1 phosphate**. Once a baby has been found to have a high level of this chemical, further tests will be done. These include measuring the enzyme that is missing in Galactosaemia and gene testing. Not all states screen newborn babies for Galactosaemia.

How is Galactosaemia inherited?

All the cells in a person's body contain many, many sets of instructions on how to make the chemicals which make up our bodies. These instructions are called **genes** and are made of **DNA**. It is estimated that we all have between 50,000 and 100,000 different genes. We each carry two copies of all our genes; one copy of each gene is inherited from our father, and one from our mother. Mistakes can happen in the gene for the enzyme which is missing in Galactosaemia. A child with mistakes in both copies of the gene will have Galactosaemia. If a person has a mistake (also known as a **mutation**) in one copy of the gene, but the other copy of the gene is normal, that person is a 'carrier' of Galactosaemia. Being a carrier has no effect on a person's health. In order for a person to have Galactosaemia, both of their parents must be carriers.

What will happen to future children?

If a couple, both of whom are carriers of mistakes in the Galactosaemia gene, have a child, there are three things that can happen. There is a one in four chance, *every time that they have a child*, that the child will have Galactosaemia. There is a one in two chance that the child will be a healthy carrier, like its parents. Lastly, there is a one in four chance that the child will not even be a carrier. These chances apply for each pregnancy, regardless of what has happened before. While it is impossible to say in advance what will happen each time, it is usually possible to do a test early in pregnancy which will show whether or not the baby is affected by Galactosaemia. Further information is available from your local genetics service.

How do I know if I am a carrier of Galactosaemia?

In Australia, about one person in 100 is a carrier of a mistake in the Galactosaemia gene. However if someone in your family has Galactosaemia, your chance of being a carrier may be higher. For example, the parents of a person with galactosaemia must be carriers. A parent who is a carrier must have inherited the Galactosaemia gene with the mistake in it from one of his or her parents, and so on.

Within a family, even if the particular mutation cannot be identified, testing is usually possible to identify carriers. Not all people want or need to know whether or not they are carriers of Galactosaemia. It is usually helpful to discuss the issues of testing with a geneticist or genetic counsellor, before the test is done.

What is the Duarte Variant?

Sometimes, a baby is found to have a high level of galactose-1-phosphate when their newborn screening tests are done, but when the level of enzyme is measured, instead of no activity (as in galactosaemia) they have a low level of activity - usually about a quarter of normal. **These children do not have Galactosaemia**. They have what is called the "Duarte variant". They do not have any of the problems that people with Galactosaemia have, and do not need to go on a special diet. The reason for the high level of galactose-1-phosphate on their initial test is that although a quarter of the usual amount of enzyme is plenty (most people make far more than they need), when babies are born, they make less enzyme than they will later on - so initially they may have a high galactose-1-phosphate. Generally speaking, people who have the Duarte variant are carriers of Galactosaemia, and on their other copy of the gene there is a mild mistake, which means that a reduced amount of enzyme is made.

How can I find out more about Galactosaemia?

If you have further questions, either speak to your local doctor or contact:

The Department of Genetic Medicine

4th Floor Rogerson Building
Women's and Children's Hospital
72 King William Road
NORTH ADELAIDE SA 5006

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