OTHERS:

GALACTOSEMIA

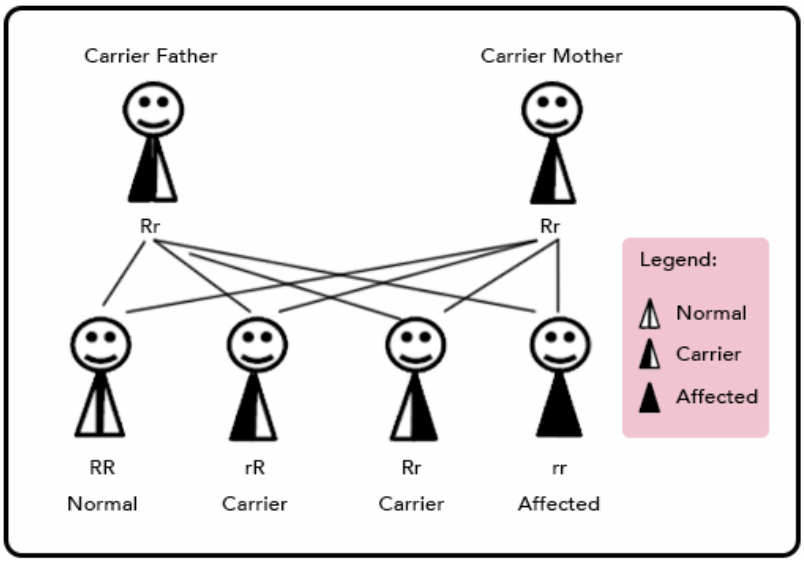
What is Galactosemia?

Galactosemia is one of the conditions that can be detected through newborn screening. Untreated children may develop cataracts, liver failure and developmental delay. Early detection can prevent the complications of this condition.

What causes Galactosemia?

To efficiently use the food we eat, our body has enzymes or chemical scissors which break these down to smaller units. Carbohydrates are broken down into sugars. A milk sugar called lactose is primarily affected in galactosemia. Due to a lack of chemical scissors, lactose cannot be broken down. This causes a sugar known as galactose to accumulate in the body.

The chemical process to break down galactose is complicated and involves several enzymes. Whether your child has classical or non-classical galactosemia will depend on which enzyme is deficient or missing. You may consult your doctor to know which type of galactosemia your child has.



Galactosemia is an inherited condition. The gene for the enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Galactosemia have one working and one non-working gene coding for the enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.

If the child inherits the non-working gene from both parents, he or she will have Galactosemia. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

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WHAT ARE THE SIGNS AND SYMPTOMS OF GALACTOSEMIA?

Upon starting feeding with milk, the baby with galactosemia may have vomiting, irritability, yellow tinge of the skin (jaundice), severe infection (sepsis), enlarged liver and signs of liver failure such as bleeding. After a few weeks, patients may have cataracts. Without treatment, patients may die of liver failure or have cataracts and developmental delays.

WHAT IS THE TREATMENT OF GALACTOSEMIA?

The main treatment of Galactosemia is through control of the diet. Because children with galactosemia cannot efficiently breakdown lactose, treatment is through the elimination of lactose from the diet. Babies are given soy-based milk formula. As their diet progresses, food which contain dairy and lactose are avoided altogether. It is important that the diet be followed to avoid any complications. Please consult with your attending physician prior to starting treatment.

The control of galactosemic patients are evaluated with the help of blood spots, similar to the samples taken during their newborn screening. The levels of galactose metabolites are measured. As part of their monitoring, children with galactosemia should have their levels taken regularly and visit their physicians to assess their growth and development.

OTHER IMPORTANT CONSIDERATIONS

Have the child vaccinated according to the regular childhood schedule. Children with CF need all the usual childhood vaccinations. It is especially important to have a measles vaccine. In addition, the primary physician may suggest that the child have vaccinations against influenza and pneumonia on a yearly basis. Children with CF should also be protected against RSV, a respiratory illness that can be severe, and sometimes life-threatening, in children with chronic lung disease.

Keep the child away from all forms of smoke, especially cigarette smoke. It can add to lung damage. Teach good hand washing habits to prevent infection.

If the child has a respiratory infection and is too sick to eat or follow regular health habits, call the attending physician right away. During some illnesses, the child may need to be seen in the hospital for treatment.

Encourage the child to get plenty of exercise. This will help maintain the child's lung function and improve overall health.