What is Galactosemia?

Galactosemia is an inherited metabolic disorder caused by changes, or mutations, in both copies of an individual’s GALT gene (genes are the instructions that tell our body how to grow and function). Typically children with Galactosemia do not have parents with Galactosemia; instead, each parent is a silent carrier of the condition, which is expressed in the child. This type of inheritance is also called autosomal recessive.

Lactose is a sugar found in milk products. The body cuts lactose into two types of sugar: glucose and galactose. These two sugars are one of the ways our bodies get energy. People with Galactosemia cannot break down the galactose and turn it into energy for the body; instead, the galactose builds up in the brain, eyes, liver, and kidneys. This can result in serious health problems.

Infants with Galactosemia may be identified through newborn screening programs or by symptoms that present during the first few weeks of life. Untreated infants develop liver and kidney disease, cataracts in their eyes, and serious infections.

5 Questions to Ask Your Health Care Provider

1. How is Galactosemia affecting my health right now?
2. How does Galactosemia affect my future fertility?
3. What therapies may help me regain or prolong ovarian function?
4. What fertility preservation options are available?
5. Can I have children in the future?
How is Galactosemia treated?
A strict galactose-free (dairy-free) diet prevents the development of liver and kidney disease, eye cataracts, and serious infections.

What are the Long-Term Complications of Galactosemia?
Unfortunately, diet cannot prevent all complications of Galactosemia later in life. Individuals with Galactosemia may develop cognitive impairment and problems with bone health. Females with Galactosemia are at increased risk to develop primary ovarian insufficiency (POI) with infertility. Galactosemia is not known to affect the fertility of males.

What is Primary Ovarian Insufficiency (POI)?
Primary ovarian insufficiency (POI) is also called premature menopause, because menstrual periods stop prior to age 40 years, or in some cases menstrual periods do not begin. Women with POI have ovaries that are not producing normal amounts of the hormone estrogen. A lack of estrogen prevents ovaries from releasing eggs regularly, or in some cases, at all. POI varies among individuals. Some women may have subfertility, or decreased fertility, meaning that it is more difficult than expected to get pregnant naturally. Some women may have irregular menstrual cycles with infertility, meaning they are unable to get pregnant naturally. The most severe form of POI is primary amenorrhea, or a complete lack of menstrual cycles and no initiation of puberty.

What is the Risk of Developing Primary Ovarian Insufficiency (POI)?
Almost every female with classic Galactosemia develops POI, regardless of diet.

Female Fertility Preservation Options

Embryo Banking
Embryo banking is an option for females who have gone through puberty. First, a woman’s ovaries are stimulated to mature multiple eggs, which are then removed and fertilized with sperm using in vitro fertilization (IVF) to create embryos. The embryos are frozen for future use. Embryo banking can take up to one month.

Egg Banking
Egg banking is an option for females who have gone through puberty. It is very similar to embryo banking, except the eggs are not fertilized before freezing. Egg banking is a good option for women who do not have a male partner and do not want to use a sperm donor at the time of the procedure. Egg banking can take up to one month.

Ovarian Tissue Banking
Ovarian tissue banking is an experimental option for females of any age. This is the only option for girls who have not started puberty. Part or all of an ovary is surgically removed and frozen for future use. The procedure can be done any time, as no stimulation of eggs is needed.

Information found here or elsewhere on the oncofertility.northwestern.edu website should not be considered medical advice, diagnosis, or treatment. Any information on this document or website should not be used in lieu of consultation with your healthcare provider or physician. Before starting any course of treatment, always consult a qualified health care provider. Do not delay seeking or disregard medical advice because of anything you have read or seen here. For information regarding fertility options contact the FERT line at 866-708-FERT (3378).
Galactosemia

Alternative Options

Women with Galactosemia may choose to use donor eggs, or consider adoption when family planning.

Questions?

Call the 24-hour FERTLINE to ask your fertility preservation questions, get connected with a fertility preservation program near you, and access resources, tools, and support!

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