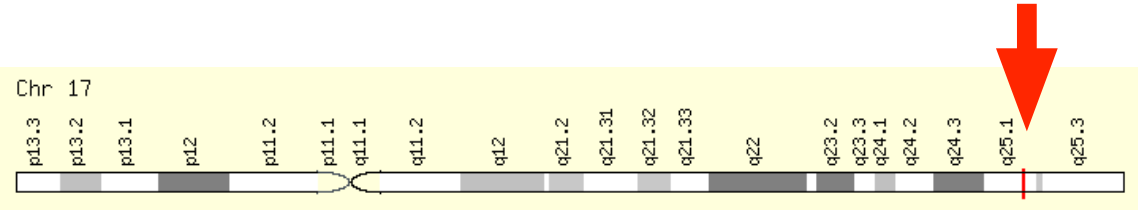
# Galactosemia type II

versione italiana

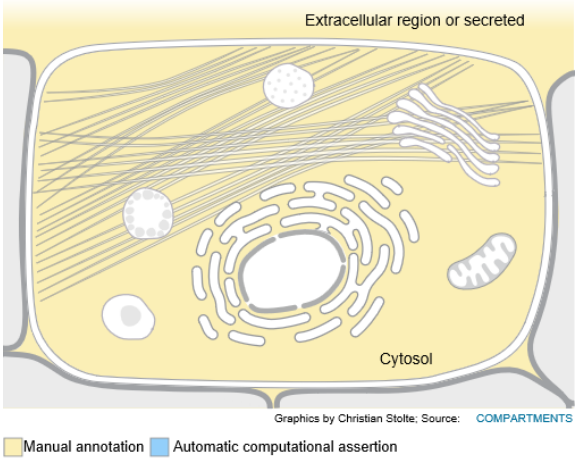
**Gene name:** GALK1 galactokinase 1




**Gene location:** 17q25.1

**Gene function:** the GALK1 gene provides instructions for making an enzyme called galactokinase 1. This enzyme enables the body to process a simple sugar called galactose, which is present in small amounts in many foods. Galactose is primarily part of a larger sugar called lactose, which is found in all dairy products and many baby formulas.

**Protein location:** It's mainly localized in the cytosol and extracellular region. However it's likely to be found in the nucleus and Golgi apparatus at lower levels (https://www.genecards.org/cgi-bin/carddisp.pl?id_type=hgnc&id=4118#localization)




**Protein name:** **Galactokinase**

**Uniprot Accession:** P51570

**Size:** 392 amino acids

**Molecular mass:** 42272 Da

**3D structure**


**1YH7**: Theoretical Model Of Human Galactokinase

## Description and function

Galactokinase 1 is responsible for one step in a chemical process that converts galactose into other molecules that can be used by the body. Specifically, this enzyme modifies galactose to create a similar molecule called galactose-1-phosphate. A series of additional steps converts galactose-1-phosphate to another simple sugar called glucose, which is the main energy source for most cells. Galactose-1-phosphate can also be converted to a form that is used to build galactose-containing glycoproteins and glycolipids. These modified proteins and fats play critical roles in chemical signaling, building cellular structures, transporting molecules, and producing energy.

**Catalytic activity**

ATP + alpha-D-galactose = ADP + alpha-D-galactose 1-phosphate.

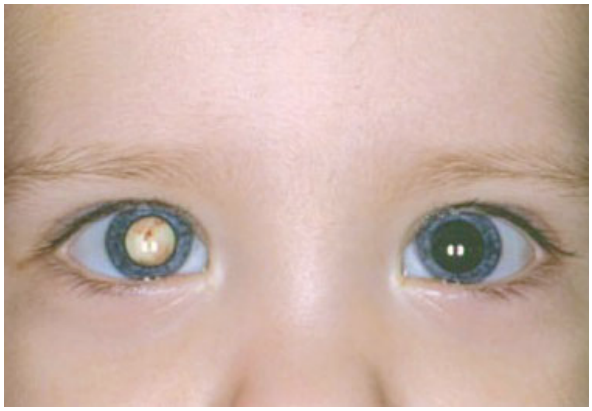
## Mutations and pathologies: Galactosemia type 2 , autosomal recessive disease

## Omim access 230200

More than 30 mutations in the *GALK1* gene have been identified in people with a form of galactosemia called type II or galactokinase deficiency. Affected infants develop clouding of the lens of the eye (cataracts) but otherwise experience few of the long-term complications associated with classic galactosemia (GALT, galactose-1-phosphate uridyltransferase deficiency, the second enzyme in galactose metabolic chain, OMIM access 230400). Classic galactosemia, also known as type I, is the most common and most severe form of the condition.

Most of the mutations change a single aminoacid in galactokinase 1. A few other mutations consist in the deletion of a small amount of genetic material from the *GALK1* gene, in both cases resulting in an unstable or inactive version of this enzyme.

A shortage of functional galactokinase 1 prevents cells from processing galactose obtained from the diet. Galactose is a sugar found in many food, including milk (main food for early childhood). As a result, galactose and a related sugar called galactitol can build up in cells.



Due to osmotic phenomena, the accumulation of galactose and galactitol in cells that make up the lens of the eye damages the lens during the first months of life, causing it to become cloudy and leading to blurred vision.

**Incidence:** Prevalence of galactosemia type II is not known but is estimated to be less than 1/ 100,000 newborns. Classic galactosemia occurs in 1 in 30,000 to 60,000 newborns.

**Therapies:** Development of cataracts appears to be fully preventable if diagnosis is made early and a galactose-restricted diet is implemented and strictly followed.

https://learn.genetics.utah.edu/content/disorders/singlegene/

Patient organisation

**Curiosities:** Congenital cataract
It is a pathology involving about 4 children out of 10,000 and is still one of the main causes of blindness in pediatric age: congenital cataract, defined as crystalline opacity, differs sharply from that of adulthood and, if not properly treated, it can permanently compromise the visual development of the child. The collaboration between pediatricians, ophthalmologists and orthoptists is essential to early detect the disorder and to immediately start a therapeutic process in order to eliminate the problem and to insert the child in a rehabilitation pathway.

Paolo Nucci, professor of visual apparatus pathologies at the University of Milan and director of the University Ophthalmology Clinic of the San Giuseppe Hospital in Milan explains how and when the congenital cataract is manifested and the related surgical treatment.