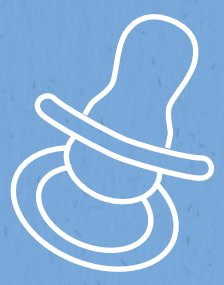
GALACTOSEMIA SYMPTOMS

AT DIFFERENT STAGES OF LIFE

Classic Galactosemia is a rare condition in which the body has trouble processing a sugar called galactose. Everyone with Galactosemia will experience it differently, but below are some common health issues that can occur at different stages of life. Some people with Galactosemia may not experience any of these symptoms. Some symptoms may get better. Others may be permanent.

NEWBORNS



Newborns are screened for Galactosemia, but sometimes symptoms can develop before the results are available. Complications may be serious, requiring intensive care or even causing death.

Acute Symptoms

- Liver failure
- Jaundice
- Kidney problems
- Sepsis
- Swelling of brain (edema)
- Pressure around brain that causes neurological problems (pseudotumor cerebri)

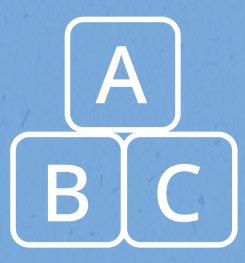
Chronic Symptoms

- Feeding difficulties
- Growth problems
- Cataracts

Monitoring

- Newborns are monitored at least every 3 months for the first year

INFANTS AND TODDLERS



At this stage, there may be early signs of developmental delays and speech problems.

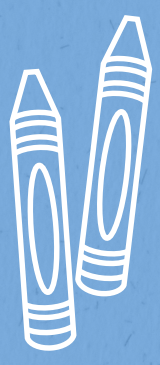
Symptoms

- Speech/language delays
- Coordination problems (fine and gross motor skills)
- Developmental delays
- Attention issues
- Growth problems

Monitoring

- **7 to 12 months:** Begin testing for cognitive and developmental delays
- **2 to 3 years:** Begin assessments of early speech/language and motor development

PRIMARY SCHOOL CHILDREN



Developmental delays may become more noticeable during this stage as children go to school.

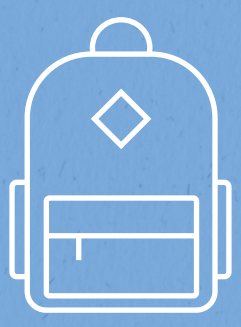
Symptoms

- Learning delays
- Issues with fine and gross motor skills (eg, handwriting)
- Growth problems
- Speech/language problems
- Behavioral and emotional issues

Monitoring

- **4 to 5 years:** Begin neuropsychological tests, evaluations of school readiness, and early educational support
- **8 to 10 years:** Begin assessment of cognitive development
  - Regular neurological and psychological evaluations, bone density screening, eye tests, and dietary monitoring throughout this stage

TEENS



Teens with Galactosemia can struggle with social problems as a result of behavioral, cognitive, or developmental issues they experience.

Symptoms

- Puberty and fertility problems (females)
- Growth delays
- Anxiety
- Social problems
- Learning difficulties

Monitoring

- **12 to 14 years:** Begin neuropsychological assessment for executive function
- Bone density screening and additional psychological, cognitive, and endocrine/hormone testing (for girls) throughout this stage
- It's recommended to see an ophthalmologist periodically throughout childhood and adulthood to monitor for cataracts

ADULTS

Some symptoms that begin earlier in life may continue into adulthood. Because of long-term health issues, it may be difficult for adults with Galactosemia to become independent.

Symptoms

- Anxiety
- Depression
- Attention deficit hyperactivity disorder (ADHD)
- Tremor
- Seizures
- Cataracts

Sources: Berry GT. Classic galactosemia and clinical variant galactosemia. In: Adam MP, et al, eds. *GeneReviews®*. University of Washington: Seattle, WA: 993-2020; Berry GT, et al. *Lancet*. 1995;346(8982):1073-1074; Coelho A, et al. *J Inherit Metab Dis*. 2017;40(3):325-342; Rubio-Gozalbo ME, et al. *Orphanet J Rare Dis*. 2019;14(1):86; Welling L, et al. *J Inherit Metab Dis*. 2017;40(2):171-176.

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