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A COMPREHENSIVE REVIEW ON GALACTOSEMIA

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Abstract

Galactosemia is a rare inherited metabolic disorder in which the body is unable to properly metabolize galactose. The disease is divided into three categories depending on the enzyme involved., classic galactosemia (galactose-1-phosphate uridylyltransferase deficiency, GALT), galactokinase deficiency (GALK), and uridine diphosphate galactose-4-epimerase deficiency (GALE). The symptoms of galactosemia start shortly after regular feeding with milk these include feeding intolerance (vomit and diarrhea), liver failure (rare) leading to jaundice, bleeding symptoms, hypoalbuminemia and hypoglycemia, kidney failure congenital bilateral cataracts, and alternatively, fatal gram-negative sepsis, principally Escherichia coli. Galactosemia is a genetic disorder that can be diagnosed at birth even if the symptoms are not present with screening of newborns by assessing the GALT enzyme activity and level of galactose. Children with galactosemia may experience speech delay, learning difficulties and poor motor coordination. The primary treatment for galactosemia is lifelong restriction of lactose and galactose from the diet. Dairy products, breast milk, and lactose containing. formulas must be avoided immediately after diagnosis. Continuous medical monitoring, nutritional support, and rehabilitative therapies are therefore essential for improving quality of life.

INTRODUCTION:

Galactosemia is a rare inherited metabolic disorder in which the body is unable to properly metabolize galactose, a sugar mainly obtained from lactose in milk and dairy products. This disorder is caused by the lack of enzymes in the Leloir pathway of galactose metabolism (Welling et al., 2017). The accumulation of toxic metabolites, such as galactose-1-phosphate and galactitol, can lead to severe organ damage, including in the liver, brain, kidneys and eyes

Galactosemia is an autosomal recessive disease, which means that the defective genes are passed on by both parents(Fridovich-Keil et al., 2025). The disease is basically divided into three categories depending on the enzyme involved: classic galactosemia (galactose-1-phosphate uridylyltransferase deficiency, GALT); galactokinase deficiency (GALK); and uridine diphosphate galactose-4-epimerase deficiency (GALE)(Coelho et al., 2017). Of these, classic galactosemia is believed to be the most severe and clinically significant. If not diagnosed and treated early, the disease may progress to liver failure, cataracts, sepsis, intellectual disability, and even death(Coss et al., 2013) .The prevalence of classic galactosemia was estimated between 1 in 16,000 and 1 in 60,000 live births in the world from 2018–2026(Coelho et al., 2017). Neonatal

screening programmes have made great strides in early detection and clinical treatment of the disorder(Welling et al., 2017) .Use of newborn screening techniques like dried blood spot analysis and enzymatic assays is common for diagnosis(Fridovich-Keil & Berry, 2022). The most important step in managing galactosemia is the dietary management of lactose and galactose (Welling et al., 2017)

Starting a lactose-free diet early will avoid acute neonatal complications and will greatly increase neonatal survival rates (Fridovich-Keil & Walter, 2021). As for long-term complications, many patients have speech disorders, cognitive problems, developmental delays, decreased bone density, and motor abnormalities despite dietary control(Rubio-Gozalbo et al., 2019). In spite of early treatment, many women develop delayed puberty, infertility, or have a decreased reproductive function(Succoio et al., 2022a) .Additional issues commonly reported among affected people are neurological problems such as tremors, anxiety, learning disabilities and impaired speech(Carlock et al., 2019)

Current research into gene therapy, enzyme replacement therapy, substrate reduction therapy and pharmacological chaperones as treatment options is ongoing (Succoio et al., 2022a)New metabolomics and transcriptomics research are also assisting in the identification of biomarkers of disease severity and progression. For reducing disease burden and improving patient outcomes, public health strategies like newborn screening, genetic counselling and awareness programmes are essential(Welling et al., 2017) .The inability to be diagnosed are still a major problem in many developing countries owing to the lack of awareness and poor healthcare facilities(Berry, 1993).Galactosemia is a complex metabolic disorder and has a number of serious biochemical, genetic, and clinical implications(Fridovich-Keil et al., 2025) .Long-term neurological and reproductive side effects are still affecting the quality of life despite the significant improvement in survival with early dietary treatment(Rubio-Gozalbo et al., 2019)

Objectives:

- To review the pathophysiology, clinical manifestation and diagnosis of galactosemia.
- To evaluate current nutritional management strategies and dietary intervention of patient with galactosemia.

Galactosemia Metabolism and Pathophysiology:

Galactosemia Metabolism and Leloir pathway:

Carbohydrates are the key source of energy and the biosynthesis of glycoconjugates required for all living organisms. A monosaccharide which is one of these is called galactose. Humans metabolize galactose via the Leloir pathway of galactose metabolism, a pathway that includes three enzymes acting on galactose which serve to modify it into a molecule species that can participate in glycolysis producing cellular energy (Leslie et al., 2005)

Galactosemia is a metabolic inherited disorder of the enzymes involved in the pathway of galactose metabolism called the Leloir pathway. The four types are galactose-1-phosphate uridylyltransferase (GALT), galactokinase (GALK1), uridine diphosphate (UDP)-galactose 4'-epimerase (GALE), and galactose mutarotase (GALM). Symptoms can range from almost none to liver, kidney and brain damage that can be life-threatening. Severely affected patients may develop cognitive disability(McCorvie & Timson, 2020).

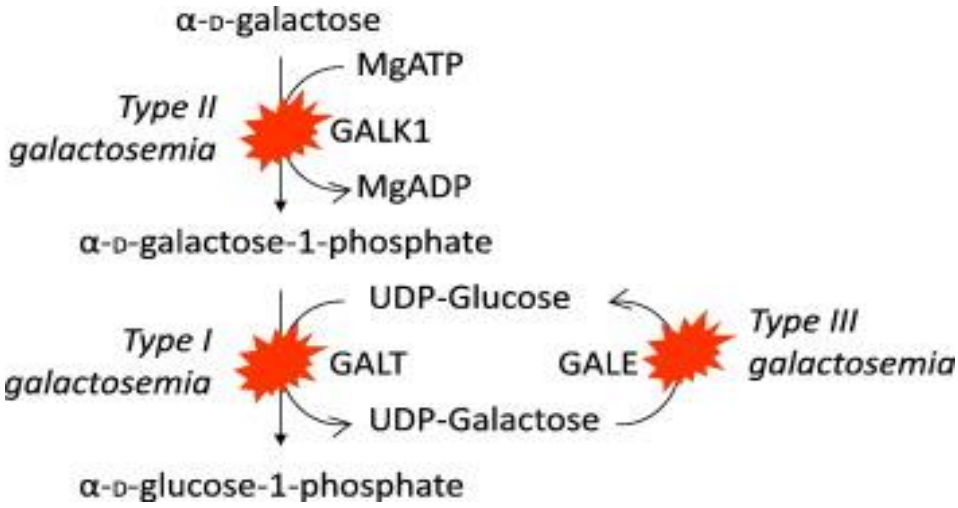

Figure 1. Leloir pathway Representation with several forms of galactosemia Type-1, Type-2, and Type-3

GALM, an aldose epimerase, catalyzes the reversible interconversion of β- and α-D-galactose, the first enzyme in the galactose metabolic pathway. GALK then converts the latter to galactose-1-phosphate (Gal-1-P). GALK belongs to a superfamily of small molecule kinase enzymes called GHMP (Galactokinase, Homoserine kinase, Mevalonate kinase, Phosphomevalonate kinase) that also use ATP as a phosphate donor (Tang et al., 2010)

GALT is involved in the subsequent metabolic step. GALT is a homodimer and belongs to the class of homodimers that share the functional His-Pro-His motif at the dimer interface, the so-called Histidine Triad (HIT) superfamily(Viggiano et al., 2018)

Galactose-1-phosphate uridylyltransferase (GALT): the enzyme plays an important role in galactose metabolism, especially during the neonatal period because of the ingestion of galactose-containing milk. GALT deficiency results in the genetic disorder classic galactosemia(Coelho et al., 2014)

GALT converts Gal-1-P into glucose-1-phosphate (Glc-1-P) with the formation of UDP-galactose (UDP-Gal) from UDP-glucose (UDP-Glc) by a "ping-pong" mechanism. The fourth enzyme in the Leloir pathway is UDP-galactose 4-epimerase (GALE), which is a member of the short-chain dehydrogenase/reductase (SDR) superfamily, and catalyzes the conversion of UDP-Gal to UDP-Glc, with the involvement of the cofactor NAD+. The enzymatic activity is dependent on UDP-Glc. GALE also catalyses the interconversion of UDP-N-acetylglucosamine (UDP-GlcNAc) and UDP-N-acetylgalactosamine (UDP-GalNAc), which are involved in the galactosylation of complex molecules and the synthesis of various glycoproteins and glycolipids(Banford et al., 2021)

Galactosemia Pathophysiology:

If the Leloir pathway is defective then the cells accumulate galactose and some other galactose metabolism pathways become activated. Thereafter, these pathways lead to the production of harmful metabolites (galactitol and D-galactonate) which accumulates in various tissues and induces tissue damage(Lai et al., 2009)

In the Leloir metabolic pathway, there are three galactose metabolizing enzymes, including galactosugar kinase (GALK), galactose-1-phosphate uridylyltransferase (GALT), and uridine diphosphate-GALACTOSE 4-epimerase (GALE). When any of these enzymes is deficient, galactose accumulates and galactosemia is the consequence(Cerone & Rios, 2019a)

Infant carriers of galactosemia are born with a problem that prevents them from breaking down galactose, a sugar present in milk, into glucose,

the sugar the body uses primarily for energy. GALT defect is the most common cause of galactosemia, and galactosemia can be diagnosed at birth, even if the baby doesn't have symptoms, through newborn screening that tests the level of galactose in the blood and the activity of the GALT enzyme(Succoio et al., 2022a)

The symptoms of galactosemia start shortly after regular feeding with milk; these include feeding intolerance (vomit and diarrhea as their gut cannot ferment galactose); liver failure (rare) leading to jaundice; bleeding symptoms; low blood protein levels (hypoalbuminemia) and low blood sugar (hypoglycemia); kidney failure (rare) with loss of phosphate, glucose, and amino acids; congenital bilateral cataracts; and alternatively, fatal gram-negative sepsis, principally Escherichia coli. The hepatopanchromatosis and the renal defects result from an overaccumulation of GALT's substrate, galactose-1-phosphate, in the cells of both organs. The cause of the cataract is an alternate metabolic pathway in the lens due to accumulation of the metabolite galactose called galactitol. The treatment is comparatively easy. After possible or confirmed diagnosis, lactose and lactose-containing products are excluded from the diet to decrease the metabolic load, and most of the neonatal toxicities are mitigated(Bennett, 2010)

Screening and Diagnosis of Galactosemia:

Galactosemia is a genetic disorder that can be diagnosed at birth even if the symptoms are not present with screening of newborns by assessing the GALT enzyme activity and level of galactose as the most common cause of galactosemia is the deficiency of GALT(Viggiano et al., 2018). Newborn screening is the preventive intervention carried out on newborns for diagnosis of inborn metabolic disorders. Newborn screening (NBS)is performed by non-invasive methods on dry blood spot samples within first 24 hours after birth. Newborn screening for galactosemia is performed by measuring total blood galactose or by measuring GALT level both on a dried blood spot using a fluorometer assay (Rybová et al., 2018).

The neonatal screening for galactosemia can appeal early identification and intervention that leads to prevention from complications. All galactosemia types may be detected during the screening of newborns for this disorder. The major focus is galactosemia caused by deficiency of galactose-1-phosphate uridyltransferase (GALT), which is identified by using a combination of total galactose and analysis of GALT enzyme, while in certain cases, mutation screening is used for identification(Badiu Tişa et al., 2022). The identification of classical galactosemia, including a subjective deficiency, is then validated by a laboratory test for galactosemia to identify high level of erythrocyte galactose-1-phosphatase, galactitol concentration in urine and by the recognition of bi-allelic pathogenic variants in the GALT gene. A new method uses non-radioactive ultraviolet light and high-performance chromatography to detect in an accurate manner the GALT levels in erythrocytes(Kikuchi et al., 2021)(Kotb et al., 2018).

The level of GALT in blood is detected by high performance chromatography or by using non radioactive ultraviolet rays. The newborns who is suffering from galactosemia mostly develop the infection caused by Escherichia coli as this infection preceding the detection of galactosemia(Pasquali et al., 2018). The complete elimination of lactose from the diet and the use of soy milk formulas precedes to the recovery of mild symptoms. If the conditions left untreated, than due to kidney failure, liver failure or infections due to Escherichia coli leads toward death within a few weeks(Pasquali et al., 2018)

Galactosemia testing may also be started by a positive family history, or by general features of the disorder in an older patient. The diagnostic examination is relatively uncomplicated, significant changeability can result from preanalytic and analytic effects if not properly measured(Richards et al., 2015). The measurement of total amount of galactose alone can precedes to false-negative assessment results in infants suffering from galactosemia whose diet is lactose-free formula and in those whose

method of feeding is total parenteral nutrition. False-positive assessment results can be observe in newborns with deficiency of citrin, Fanconi Bickel disease, or diseases of liver along with other prescriptions(Pasquali et al., 2018). Following an irregular screening of newborn, the identification of classic galactosemia is validated by the illustration of intense deficiency of the GALT enzyme in red blood cells and recognition of infective variants within the GALT gene by molecular sequencing. The total galactose in their assessment procedure can validate or eliminate GALK or GALE deficiency by linking results from the molecular and enzymatic assessment. Duarte galactosemia (DG) is diagnosed by isoelectric focusing or by electrophoresis(Frederick et al., 2017).

The cause of type IV galactosemia is mutation in the GALM gene which leads to decline the GALM enzyme activity(Badiu Tişa et al., 2023). The type II and type IV galactosemia shows similar symptoms which leads to risk of misdiagnosis of type II as type IV galactosemia. The sequencing of GALM and GALK is important to avoid misdiagnosis. The positive family history or non specific features of disease in older people patients can also be used for diagnosis of galactosemia. The result of false positive screening observed in newborns with deficiency of citrine and liver diseases(Amjed Torki Al-Rudaini, 2025).

Nutritional Management

Diet is of paramount importance in Galactosemia. Galactosemia patients need to be put on lifelong restriction of lactose and galactose(Succoio et al., 2022b). When patients follow low galactose diets during the 1st week of life, most desirable results are achieved.(Rubio-Gozalbo et al., 2019) Breastfeeding and whey based infant formulas are ceased.(Succoio et al., 2022b) Before the weaning stage, soy based or elemental formulas are suggested. These measures help prevent acute neonatal fatal symptoms of metabolic acidosis, abnormal clotting, vomiting, jaundice and the like.(Cerone & Rios, 2019b) Casein protein hydrolysate formulas can be considered as an option for Classic Galactosemia as it has been deemed safe even though traces of lactose are present in it.(*No Title*, n.d.)(Bosch, 2010) Previously patients had a highly restrictive diet but over time, it was found that fruits, vegetables and legumes can safely be incorporated in the diet as galactose found in these is undigestable, unabsorbable as a result of absence of the required enzymes. Dairy products and animal milk still need to be restricted. Due to the low galactose content of mature cheeses and soy products, they are safe to be consumed. These are recommended with the aim to overcome the Calcium and Vitamin D deficiency, supplementation can be considered if required. Reports of cohorts suggested that those on restrictive diets exhibited higher frequency of neurological impairments.(Caro N. et al., 2022) The best way forward is case by case analysis using for example serum N-glycan profiling to allow for personalized management of galactosemia as glycosylation capability varies from person to person.(Colhoun et al., 2018)

Monitoring and Follow-Up

Long-term monitoring is necessary even in treated patients because complications may still occur despite dietary restriction. Regular follow-up includes:

- Liver function assessment
- Growth and developmental monitoring
- Speech and cognitive evaluation
- Ophthalmologic examination for cataracts
- Bone health assessment

Management of Developmental and Neurological Problems

Children with galactosemia may experience:

- Speech delay
- Learning difficulties
- Poor motor coordination
- Tremors and neurological impairment

Therapeutic interventions include:

- Speech therapy
- Occupational therapy
- Educational support programs

- Neurological rehabilitation

Hormonal and Reproductive Management

Females with classic galactosemia commonly develop premature ovarian insufficiency. Hormonal monitoring, fertility counseling, and hormone replacement therapy may be required.

Emerging and Experimental Therapies

Recent research focuses on advanced therapeutic strategies including:

- Gene therapy
- Enzyme replacement therapy
- Pharmacological chaperone therapy
- Aldose reductase inhibitors

These approaches aim to reduce long-term complications that persist despite dietary treatment. Although it is a serious inherited metabolic disorder requiring early diagnosis and lifelong dietary management. Although lactose and galactose restriction significantly improve survival and reduce acute complications, long-term neurological and reproductive complications may still occur. Continuous medical monitoring, nutritional support, and rehabilitative therapies are therefore essential for improving quality of life. Ongoing research into gene and pharmacological therapies may offer improved future treatment options.

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