Classic Galactosemia

Classic galactosemia is a rare autosomal recessive metabolic disorder caused by an absence of the enzyme galactose-1-phosphate uridyltransferase. This enzyme normally catalyzes the reaction of galactose-1-phosphate to glucose-1-phosphate in the galactose degradation pathway. An absence of the enzyme causes an accumulation of galactose-1-phosphate in various tissues to toxic levels. Individuals commonly present with infantile cataracts, due to accumulation of galactitol in the lens, failure to thrive, hepatomegaly, and jaundice. Severe elevations of galactose-1-phosphate can also cause brain damage, leading to intellectual disability. Without treatment, mortality in infants with galactosemia is about 75%. The only treatment for classic galactosemia is the complete elimination of lactose and galactose from the diet. It is important not to confuse classic galactosemia with galactokinase deficiency or lactose intolerance.



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Pathophysiology

Autosomal Recessive
Recessive-chocolate
This disease is inherited in an autosomal recessive fashion.

Galactose-1-phosphate uridyltransferase is Absent (GALT)
Galactic-toast 1-P with U-transformer being Absent
This enzyme normally catalyzes galactose-1-phosphate to UDP-galactose in the galactose degradation pathway. This enzyme is absent in classic galactosemia.

Impaired Galactose-1-P to UDP-Galactose
Galactic-toast 1-P is Impaired from becoming Upside-Down-Pineapple Galactic-toast
The enzyme galactose-1-P uridyltransferase catalyzes the reaction galactose-1-phosphate to UDP-galactose in the galactose degradation pathway. This can then be converted to UDP-glucose or lactose.

Galactitol Accumulation in Lens
Galaxy Accumulates in Lens
Accumulation of galactose causes an increase in galactitol, which can accumulate in the lens of the eye and cause infantile cataracts.

Signs and Symptoms

Infantile Cataracts
Baby Cadillac-cataracts
Accumulation of galactose causes an increase in galactitol, which can accumulate in the lens of the eye and cause infantile cataracts.

Failure to Thrive
Very Skinny Baby
Infants with classic galactosemia display failure to thrive early in life due to toxic accumulations of galactose-1-phosphate in tissues.

Hepatomegaly
Liver-balloon
Galactose-1-phosphate uridyl transferase is particularly prevalent in the liver, and deficiency can cause enlargement.

Jaundice
Jaundice-janitor
Jaundice is yellowing of the skin and conjunctival membranes caused by hyperbilirubinemia and often liver disease. Galactose-1-phosphate uridyl transferase is particularly prevalent in the liver, and deficiency can cause liver disease leading to jaundice.

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Intellectual Disability

Tar Covered Book

Severe elevations of galactose-1-phosphate can also cause brain damage, leading to intellectual disability.

Increased Risk E. Coli Sepsis

Up-arrow Risk of E Coal-eye with Sepsis-snake

Due to the inhibition of leukocyte bactericidal activity, patients are more prone to developing E. coli sepsis. This is responsible for a high frequency of neonatal death in classic galactosemia.

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