Galactokinase Deficiency

Galactokinase deficiency is an autosomal recessive metabolic disorder caused by a deficiency in the enzyme galactokinase. Galactokinase catalyzes the first step of galactose metabolism from galactose to galactose-1-phosphate. Deficiency of this enzyme causes marked accumulation of galactose and galactitol in the blood and urine. Unlike classic galactosemia, which is caused by a deficiency of galactose-1-phosphate uridyltransferase, galactokinase deficiency does not present with severe manifestations in early infancy. Although the disease is mostly clinically benign, patients do develop cataracts early due to accumulation of galactitol in the lens. Sometimes, this presents with failure to develop a social smile. Treatment include elimination of galactose in the diet.



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

Galactose to Galactose 1P
Galaxy-toast to Galaxy-toast (1) P
Galactokinase catalyzes the first step of galactose metabolism from galactose to galactose-1-phosphate.

Galactose Appears in Blood and Urine
Blood-red and Urine-yellow-pipes
A deficiency of galactokinase causes a marked accumulation of galactose and galactitol in the blood and urine.

Benign
Bunny
Unlike classic galactosemia, which is caused by a deficiency of galactose-1-phosphate uridyltransferase, galactokinase deficiency does not present with severe manifestations in early infancy and is relatively benign.

Infantile Cataracts
Baby Cadillac-cataracts
Galactitol, a product of an alternative route of galactose metabolism, can accumulate in the lens of the eye and cause cataracts during the first weeks or months of life.

Failure to Develop a Social Smile
Frowning Baby
Infants with galactokinase deficiency may present with a failure to develop a social smile due to infantile cataracts.

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