Essential Fructosuria

Essential fructosuria is an autosomal recessive metabolic disorder caused by a deficiency of the enzyme fructokinase, which catalyzes the degradation of fructose to fructose-1-phosphate in the liver. Unlike other defects in fructose metabolism, this deficiency causes a clinically benign condition characterized by incomplete metabolism of fructose and excretion of fructose in the urine without excess fructose entering cells. A diagnosis of essential fructosuria is typically made after a positive test for reducing substances in the urine. Because the disease is clinically benign, no treatment is indicated.



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Autosomal Recessive
Recessive-chocolate
This disorder is inherited in an autosomal recessive pattern.

Defect in fructokinase
Fruit-toast Kite-ace Broken by lightning
Essential fructosuria is caused by a deficiency of the enzyme fructokinase which catalyzes the reaction fructose to fructose-1-phosphate.

Fructose to fructose 1-P
Fruit-toast to fruit-toast (1) Wand P
Fructokinase is the first enzyme involved in the degradation of fructose to fructose-1-phosphate.

Benign
Bunny
Unlike other defects in fructose metabolism, essential fructosuria causes a clinically benign condition.

Fructose appears in blood and urine
Fruit-toast on Blood-red and Urine-yellow river
Although fructose accumulates in blood and urine, it cannot enter cells and therefore causes a clinically benign condition. Fructose is excreted through the urine, causing a positive test for reducing substances.

Fructose does not enter cells
Stop sign that prevents fruit-toast from entering cell
Although fructose accumulates in blood and urine, it cannot enter cells and therefore causes a clinically benign condition.

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