

## 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.



PLAY PICMONIC

### 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.