

Opis choroby *

Definicja

Essential fructosuria is a rare autosomal recessive disorder of fructose metabolism (see this term) caused by a deficiency of fructokinaseenzyme activity. It is characterized by elevated fructosemia and presence of fructosuria following ingestion of fructose and related sugars (sucrose, sorbitol). Essential fructosuria is clinically asymptomatic and harmless. Dietary restriction is not indicated.

Dane

Klasyfikacja

Choroba

Synonimy

Fructokinase deficiency

Niedobór fruktokinazy

Niedobór ketoheksokinazy

Ketohexokinase deficiency

Kod ORPHA

2056

Kod OMIM

229800

Kod ICD10

E74.1

Kod ICD11

5C51.5Y

*Źródło

orphanet