

Opis choroby *

Definicja

Hereditary fructose intolerance (HFI) is an autosomal recessive disorder of fructose metabolism (see this term), resulting from a deficiency of hepatic fructose-1-phosphate aldolase activity and leading to gastrointestinal disorders and postprandial hypoglycemia following fructose ingestion. HFI is a benign condition when treated, but it is life-threatening and potentially fatal if left untreated.

Dane

Klasyfikacja

Choroba

Synonimy

Hereditary fructose-1-phosphate aldolase deficiency
Dziedziczna fruktozemia
Dziedziczny Niedobór aldolazy fruktozo-1-fosforanu
Hereditary fructosemia

Kod ORPHA

469

Kod OMIM

229600

Kod ICD10

E74.1

Kod ICD11

5C51.50

*Źródło

orphanet