

Opis choroby *

Definicja

Isolated glycerol kinase deficiency (GKD) is a very rare X-linked disorder of glycerol metabolism characterized biochemically by elevated plasma and urine glycerol levels, and clinically by variable neurometabolic manifestations, depending on the age of onset, and varying from a life-threatening childhood metabolic crisis to an asymptomatic adult form (infantile GKD, juvenile GKD, and adult GKD (see these terms)).

Dane

Klasyfikacja

Choroba

Synonimy

Hyperglycerolemia

Hiperglicerolemia

Kod ORPHA

408

Kod OMIM

307030

Kod ICD10

E74.8

Kod ICD11

5C51.1

*Źródło

orphanet