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 lifethreatening childhood metabolic crisis to an asymptomatic adult form (infantile GKD, juvenile GKD, and adult GKD (see these terms)).

Dane

Klasyfikacja

Synonimy

Choroba

Hyperglycerolemia Hiperglicerolemia

Kod ORPHA

408

Kod OMIM

Kod ICD10 E74.8

307030

Kod ICD11 5C51.1

*Źródło

orphanet