

## 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

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#### \*Źródło

orphanet