Opis choroby *

Definicja

A rare, hereditary disorder of pentose phosphate metabolism characterized by increased urine levels of sedoheptulose and erythritol, and low-to-normal excretion of sedoheptulose-7P. Clinical presentation of this disorder is currently unclear.

Dane

Klasyfikacja
Choroba

Synonimy
Isolated SHPK deficiency
Izolowany Niedobór SHPK

Kod ORPHA
440713

Kod OMIM
617213

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet