

## 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