

Opis choroby *

Definicja

Pentosuria is an inborn error of metabolism which is characterized by the excretion of 1 to 4 g of the pentose L-xylulose in the urine per day.

Dane

Klasyfikacja

Choroba

Synonimy

Essential pentosuria

Niedobór dehydrogenazy ksylitolu

Pentozuria

Xylitol dehydrogenase deficiency

Kod ORPHA

2843

Kod OMIM

260800

Kod ICD10

E74.8

Kod ICD11

5C51.0

*Źródło

orphanet