Opis choroby *

Definicja

Hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency is a hereditary disorder of purine metabolism associated with uric acid overproduction and a continuum spectrum of neurological manifestations depending on the degree of the enzyme deficiency.

Dane

Klasyfikacja
Grupa fenomenów

Synonimy
HPRT deficiency
Niedobór fosforybozyltransferazy hipoksantynowo-guaninowej 1
Niedobór HPRT deficiency
Niedobór HPRT1
HPRT1 deficiency
Hypoxanthine-guanine phosphoribosyltransferase 1 deficiency

Kod ORPHA
206428

Kod OMIM
-

Kod ICD10
E79.8

Kod ICD11
-

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*Źródło

orphanet