

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

Kod ORPHA
206428

Kod OMIM
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Kod ICD10
E79.8

Kod ICD11
-

*Źródło

orphanet