Opis choroby *

Definicja

Hawkinsinuria is an inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin ((2-l-cystein-S-yl, 4-dihydroxycyclohex-5-en-1-yl)acetic acid), in the urine.

Dane

Klasyfikacja
Choroba

Synonimy
4-HPPD deficiency
Niedobór 4-HPPD
Niedobór dioksygenazy kwasu 4-hydroksyfenylpirogronowego
Niedobór hydroksylazy 4-alfa-hydroksyfenylpirogronianowej
4-alpha-hydroxyphenylpyruvate hydroxylase deficiency
4-hydroxyphenylpyruvic acid dioxygenase deficiency

Kod ORPHA
2118

Kod OMIM
140350

Kod ICD10
E70.2

Kod ICD11
5C50.1Y

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*Źródło

orphanet