## 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

---

*Źródło

orphanet