

Opis choroby *

Definicja

Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis (FHHNC) is a form of familial primary hypomagnesemia (FPH, see this term), characterized by renal magnesium (Mg) and calcium (Ca) wasting, nephrocalcinosis, kidney failure and, in some cases, severe ocular impairment. Two subtypes of FHHNC are described: FHHNC with severe ocular involvement (FHHNCOI) and without severe ocular involvement (FHHN) (see these terms).

Dane

Klasyfikacja

Choroba

Synonimy

FHHNC

FHHNC

Zespół Michellisa i Castrillo

Pierwotna hipomagnezemia z hiperkalciurią i nefrokalcynozą

Michellis-Castrillo syndrome

Kod ORPHA

306516

Kod OMIM

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Kod ICD10

E83.4

Kod ICD11

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*Źródło

orphanet