DISEASE:
Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement

NAME:	Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
DESCRIPTION:	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement (FHHNCOI) is a form of familial primary hypomagnesemia (FPH, see this term), characterized by excessive magnesium and calcium renal wasting, bilateral nephrocalcinosis, progressive renal failure and severe ocular abnormalities.
ORPHACODE:	2196
SYNONYMS:	FHHNC with severe ocular involvement Hypercalciuria-bilateral macular coloboma syndrome Meier-Blumberg-Imahorn syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CLDN19
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