DISEASE:
Biotinidase deficiency

NAME:	Biotinidase deficiency
DESCRIPTION:	A late-onset form of multiple carboxylase deficiency, an inborn error of biotin metabolism that, if untreated, is characterized by seizures, breathing difficulties, hypotonia, skin rash, alopecia, hearing loss and delayed development.
ORPHACODE:	79241
SYNONYMS:	Juvenile-onset multiple carboxylase deficiency Late-onset multiple carboxylase deficiency
XREF(S):	Orphanet ICD-10 OMIM MeSH MedDRA
ANALYTE(S):	BTD
CREATED:	13 May 2019 - 01:02
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