DISEASE:
Adenosine monophosphate deaminase deficiency

NAME:	Adenosine monophosphate deaminase deficiency
DESCRIPTION:	A rare metabolic disorder for which two forms have been described. Lack of activity of the erythrocyte isoform of adenosine monophosphate (AMP) deaminase has been described in subjects with low plasma uric acid levels without obvious clinical relevance and will not be described further. Myoadenylate deaminase deficiency is an inherited disorder of muscular energy metabolism with a lack of AMP deaminase activity in skeletal muscle. It is characterised by exercise-induced muscle pain, cramps and/or early fatigue.
ORPHACODE:	45
SYNONYMS:	AMP deaminase deficiency Myoadenylate deaminase deficiency
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10
ANALYTE(S):	AMPD1 AMPD3
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