DISEASE:
Classic phenylketonuria

NAME:	Classic phenylketonuria
DESCRIPTION:	A severe form of phenylketonuria (PKU) due to phenylalanine hydroxylase deficiency, an inborn error of amino acid metabolism, characterized in untreated patients by severe intellectual deficit and neuropsychiatric complications.
ORPHACODE:	79254
SYNONYMS:	Classic PKU
XREF(S):	Orphanet MedDRA ICD-10
ANALYTE(S):	PAH
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