DISEASE:
Pterin-4 alpha-carbinolamine dehydratase deficiency

NAME:	Pterin-4 alpha-carbinolamine dehydratase deficiency
DESCRIPTION:	A rare genetic, transient and benign form of hyperphenylalaninemia due to tetrahydrobiopterin deficiency and characterized by muscular hypotonia, irritability (detected by EEG), slow acquisition of psychomotor skills, age-dependent movement disorders, including dystonia and an accompanying excretion of 7-substituted pterins. Neurological developement is normal with dietary control of blood phenyalanine.
ORPHACODE:	1578
SYNONYMS:	Hyperphenylalaninemia due to dehydratase deficiency Hyperphenylalaninemia due to pterin-4-alpha-carbinolamine dehydratase deficiency Hyperphenylalaninemia with primapterinuria
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	PCBD1
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RELATED CONTENT

Related Genetic Tests

- Tubulopathy (gene panel)

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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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