DISEASE:
Dopa-responsive dystonia due to sepiapterin reductase deficiency

NAME:	Dopa-responsive dystonia due to sepiapterin reductase deficiency
DESCRIPTION:	Dopa-responsive dystonia (DRD) due to sepiapterin reductase deficiency (SRD) is a very rare neurometabolic disorder characterized by dystonia with diurnal fluctuations, axial hypotonia, oculogyric crises, and delays in motor and cognitive development.
ORPHACODE:	70594
SYNONYMS:	Autosomal recessive sepiapterin reductase-deficient DRD DRD due to SRD SPR deficiency Sepiapterin reductase deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SPR
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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