DISEASE:
Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome

NAME:	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome
DESCRIPTION:	Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome is a rare, genetic, neuro-ophthalmological syndrome characterized by post-natal, progressive microcephaly and early-onset seizures, associated with delayed global development, bilateral cortical visual impairment and moderate to severe intellectual disability. Additional manifestations include short stature, generalized hypotonia and pulmonary complications, such as recurrent respiratory infections and bronchiectasis. Auditory and metabolic screenings are normal.
ORPHACODE:	477814
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	DIAPH1
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