

DISEASE:**Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome**

NAME:	Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome
DESCRIPTION:	A rare, syndromic intellectual disability characterized by hypotonia, global developmental delay, limited or absent speech, intellectual disability, macrocephaly, mild dysmorphic features, seizures and autism spectrum disorder. Associated ophthalmologic, heart, skeletal and central nervous system anomalies have been reported.
ORPHACODE:	457279
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>PPP2R5D</u>
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