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# PYCR2-related microcephaly-progressive leukoencephalopathy

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. PYCR2-related microcephaly-progressive leukoencephalopathy. ORPHA:481152*

PYCR2-related microcephaly-progressive leukoencephalopathy is a rare, genetic, syndromic intellectual disability disorder characterized by progressive postnatal microcephaly, cerebral hypomyelination and severe psychomotor developmental delayed with absent speech, as well as axial hypotonia, appendicular hypertonia with hyperextensibility of the wrists and ankles, hyperreflexia, severe muscle wasting and failure to thrive. Associated craniofacial dysmorphism includes triangular facies with bitemporal narrowing, down- or upslanting palpebral fissures, malar hypoplasia, large malformed ears with overfolded helices, upturned bulbous nose, long smooth philtrum and thin vermilion borders.