

[Open Peer Review on Qeios](#)

# Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.*

*Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome.*

*ORPHA:477814*

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.