Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
477814

Kod OMIM
616632

Kod ICD10
Q02

Kod ICD11
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*Źródło

orphanet