Opis choroby *

Definicja

Oculocerebral hypopigmentation syndrome, Preus type is a rare congenital syndrome characterized by skin and hair hypopigmentation, growth retardation, and intellectual deficit that are associated with a combination of various additional clinical anomalies such as ocular albinism, cataract, delayed neuropsychomotor development, sensorineural hearing loss, dolicocephaly, high arched palate, widely spaced teeth, anemia, and/or nystagmus.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2720

Kod OMIM
257790

Kod ICD10
E70.3

Kod ICD11
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*Źródło

orphanet