

Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by intellectual disability, growth retardation, unilateral preaxial polydactyly, and colobomatous anomalies (including coloboma of the iris, optic nerve head, choroid, and retina). There have been no further descriptions in the literature since 1987.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych Pfeiffer-Mayer syndrome	Pfeiffer-Mayer syndrome
	Zespół Pfeiffera i Mayera

Kod ORPHA	Kod OMIM	Kod ICD10
2921	-	Q87.2

Kod ICD11

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*Źródło

orphanet