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
Zespół wad wrodzonych

Synonimy
Pfeiffer-Mayer syndrome
Zespół Pfeiffera i Mayera

Kod ORPHA
2921

Kod OMIM
-

Kod ICD10
Q87.2

Kod ICD11
-

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*Źródło

orphanet