Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by congenital, total, cortical blindness, intellectual disability, postaxial polydactyly of the hands and feet, pre- and postnatal growth delay, psychomotor developmental retardation, and mild facial dysmorphism (incl. prominent forehead, short nose, long philtrum, high-arched palate, and microretrognathia). Recurrent respiratory and intestinal infections, as well as moderate hypertonia and hyperreflexia, are also associated. There have been no further descriptions in the literature since 1985.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1389

Kod OMIM
218010

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet