Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by severe white matter hypoplasia, corpus callosum agenesis or extreme hypoplasia, severe intellectual disability, failure to thrive and minor midline facial dysmorphism (including hypertelorism, broad nasal root, micrognathia). There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Curatolo-Cilio-Pessagno syndrome
Zespół Curatolo, Cilio i Pessagno

Kod ORPHA
3207

Kod OMIM
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Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet