Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by severe global developmental delay, osteogenesis imperfecta, presence of wormian bones, seizures, ocular abnormalities (blue sclerae, optic atrophy, retinal detachment), and dysmorphic facial features (including frontal bossing, low anterior hairline, medial flare of the eyebrows, long eyelashes, hypertelorism, depressed nasal bridge, and low-set, large ears). There have been no further descriptions in the literature since 1994.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Al Gazali-Nair syndrome
Zespół Al Gazali i Naira

Kod ORPHA
2773

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

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*Źródło

orphanet