Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, hypotonia, seizures, microcephaly, delayed bone maturation, and skeletal abnormalities (such as scoliosis or pectus excavatum, among others). Dysmorphic features include coarse face, hirsutism, thick eyebrows, broad nasal septum, short philtrum, large mouth, and prominent ears. There have been no further descriptions in the literature since 1996.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Battaglia-Neri syndrome
Zespół Battaglia i Neri

Kod ORPHA
1948

Kod OMIM
601352

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet