

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	Synonimy
Zespół wad wrodzonych	Battaglia-Neri syndrome
	Zespół Battaglia i Neri

Kod ORPHA	Kod OMIM	Kod ICD10
1948	601352	Q87.8

Kod ICD11

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*Źródło

orphanet