

## 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

1948

#### Kod OMIM

601352

#### Kod ICD10

Q87.8

#### Kod ICD11

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#### \*Źródło

orphanet