Opis choroby *

Definicja

Branchioskeletogenital syndrome is a rare multiple congenital anomalies/dysmorphic syndrome characterized by moderate intellectual disability, distinctive craniofacial features (including brachycephaly, facial asymmetry, marked hypertelorism, blepharochalasis, proptosis, a broad nose with concave nasal ridge and bulbous nasal tip, midface hypoplasia, bifid uvula or partial cleft palate, and prognathism), progressive dental anomalies (dentigerous cysts, radicular dentin dysplasia and early tooth loss), vertebral fusions (particularly of C2-C3), and hypospadias. Hearing loss is an additional observed feature.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
BSG syndrome
Zespół BSG
Zespół Elsahy i Watersa
Elsahy-Waters syndrome

Kod ORPHA
1299

Kod OMIM
211380

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet