## 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