## Opis choroby *

Definicja

A rare, genetic, multiple congenital anomalies syndrome characterized by the association of a typical facial phenotype with microcephaly associated with congenital hypothyroidism, skeletal involvement (polydactyly, long thumb(s) and long first toe(s), and patellar hypoplasia/agenesis), and some degree of global developmental delay, hypotonia and intellectual disability. Facial features include an immobile mask-like face, severe blepharophimosis and ptosis, tear duct abnormalities, a broad nasal bridge, bulbous nasal tip, small mouth, thin upper lip, hypoplastic teeth and small, low set ears. Renal and genital anomalies, usually cryptorchidism, are often present in affected males. Congenital heart defects and growth delay are variably present.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Hypothyroidism-dysmorphism-postaxial polydactyly-intellectual disability syndrome
Niedoczynność tarczycy- dysmorfia - polidaktylia pozaosiowa - niepełnosprawność intelektualna
Zespół Say'a, Barbera, Bieseckera, Younga i Simpsona
Zespół zwężenie szpary powiekowej- upośledzenie umysłowe typu zespołu Say'a, Barbera, Bieseckera, Younga i Simpsona
SBBYS variant of Ohdo syndrome
SBBYSS
Say-Barber-Biesecker-Young-Simpson syndrome

**Kod ORPHA**
3047

**Kod OMIM**
603736

**Kod ICD10**
Q87.8

**Kod ICD11**
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*Źródło

orphanet