## Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, seizures, abnormal gait, and craniofacial dysmorphism (including coarse features, depressed nasal bridge, anteverted nares, broad nasal tip, prominent maxilla and upper lip, wide mouth, abnormal gingiva, and widely spaced teeth). Additional reported manifestations are ocular anomalies, cardiac defects, gastrointestinal problems, and autistic features. Brain imaging may show thin corpus callosum, white matter abnormalities, or dilated ventricles.

Dane

**Klasyfikacja**
Choroba

Synonimy
Skraban-Deardorff syndrome
zespół Skraban i Deardorffa

**Kod ORPHA**
513456

**Kod OMIM**
617616

**Kod ICD10**
Q87.0

**Kod ICD11**
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*Źródło

orphanet