

# Zespół Basel-Vanagaite i Smirin-Yosef

Kod Orpha: 464738 Kod OMIM: 616449

## Opis choroby \*

### Definicja

A rare, genetic intellectual disability syndrome characterized by severe global developmental delay with intellectual disability, microcephaly, growth retardation, ocular defects such as congenital cataract, and nevus flammeus simplex on the forehead. Cardiac, urogenital, and skeletal abnormalities, as well as seizures are present in most patients. Dysmorphic craniofacial features include sparse hair, downslanting palpebral fissures, hypertelorism, broad and overhanging nasal tip and short philtrum, among others.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA  
464738

Kod OMIM  
616449

Kod ICD10  
Q87.8

Kod ICD11  
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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.