

## Opis choroby \*

### Definicja

A rare developmental defect during embryogenesis characterized by microcephaly, characteristic facial features, hypotonia, non-progressive intellectual deficit, myopia and retinal dystrophy, neutropenia and truncal obesity.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

193

#### Kod OMIM

216550

#### Kod ICD10

Q87.8

#### Kod ICD11

LD90.Y

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### \*Źródło

orphanet