

## Opis choroby \*

### Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by overgrowth and macrocephaly with megalencephaly apparent at birth, global developmental delay, intellectual disability, and dysmorphic facial features (including frontal bossing, long face, sparse eyebrows, hypertelorism, downslanting palpebral fissures, and prognathism). Patients may exhibit tall stature with dolichostenomelia, arachnodactyly, kyphoscoliosis, and joint laxity, as well as neurologic manifestations, such as hypotonia, gait ataxia, or seizures. Brain imaging may show increased white matter volume, thick corpus callosum, or small cerebellum.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

457359

#### Kod OMIM

617011

#### Kod ICD10

Q87.3

#### Kod ICD11

LD2C

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### \*Źródło

orphanet