

## Opis choroby \*

### Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by congenital microcephaly, infantile-onset epileptic encephalopathy, and profound developmental delay. Additional reported features include cortical visual impairment, sensorineural hearing loss, increased muscle tone, limb contractures, scoliosis, and dysmorphic features like midface hypoplasia, narrow forehead, short nose, narrowed nasal bridge, and small chin. Brain imaging may show thin corpus callosum and delayed myelination.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

RNF13-related severe EOEE

RNF13-related severe EOEE

#### Kod ORPHA

544503

#### Kod OMIM

618379

#### Kod ICD10

G40.4

#### Kod ICD11

-

---

#### \*Źródło

orphanet