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
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*Źródło

orphanet