## Opis choroby *

Definicja

A partial monosomy of the long arm of chromosome 9 characterized by intellectual disability, developmental delay with pronounced speech delay, short stature, and muscular hypotonia. Common craniofacial dysmorphic features consist of microcephaly, prominent forehead, round face, arched eyebrows, upslanting palpebral fissures, strabismus, short nose, and thin upper lip. Other clinical findings include epilepsy, ataxia, unspecific brain MRI findings, early-onset primary dystonia, nail dysplasia, and bone malformations, in particular patellar abnormalities, epistaxis, and cutaneous-mucous telangiectasias.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Del(9)(q33.3q34.11)
Deletion 9q33.3q34.11
Monosomy 9q33.3q34.11
Del(9)(q33.3q34.11)
Deletion 9q33.3q34.11
Monosomy 9q33.3q34.11

**Kod ORPHA**
495818

**Kod OMIM**
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**Kod ICD10**
Q93.5

**Kod ICD11**
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*Źródło

orphanet