Opis choroby *

Definicja

A very rare, chromosomal anomaly characterized by an intrauterine and postnatal growth retardation, short stature, developmental delay, learning difficulties, hearing loss, hypermetropia,and a recognisable facial dysmorphism including prominent forehead, long, myopathic facies, fine eyebrows, small mouth and micrognathia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(1)(p35.2)
Del(1)(p35.2)
Delecja 1p35.2
Monosomia 1p35.2
Deletion 1p35.2
Monosomy 1p35.2

Kod ORPHA
456298

Kod OMIM
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Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet