Opis choroby *

Definicja

The 19q13.11 microdeletion is characterized by several major features including pre and postnatal growth retardation, slender habitus, severe postnatal feeding difficulties, microcephaly, intellectual deficit with speech disturbance, hypospadias and ectodermal dysplasia presented by scalp aplasia, thin and sparse hair, eyebrows and eyelashes, thin and dry skin and dysplasic nails.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(19)(q13.11)
Del(19)(q13.11)
Monosomia 19q13.11
Monosomy 19q13.11

Kod ORPHA
217346

Kod OMIM
613026

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet