Opis choroby *

Definicja

14q22q23 microdeletion syndrome is a rare partial deletion of the long arm of chromosome 14 characterized by ocular anomalies (anopthalmia/microphthalmia, ptosis, hypertelorism, exophthalmos), pituitary anomalies (pituitary hypoplasia/aplasia with growth hormone deficiency and growth retardation) and hand/foot anomalies (polydactyly, short digits, pes cavus). Other clinical features may include muscular hypotonia, psychomotor development delay/intellectual disability, dysmorphic signs (facial asymmetry, microretrognathia, high-arched palate, ear anomalies), congenital genitourinary malformations, hearing impairment. Smaller 14q22 deletions may have variable expression.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
14q22-q23 microdeletion syndrome
Del(14)(q22q23)
Monosomia 14q22q23
Monosomia 14q22-q23
Zespół mikrodelecji 14q22-q23
Del(14)(q22q23)
Monosomy 14q22-q23
Monosomy 14q22q23

Kod ORPHA
264200

Kod OMIM
609640

Kod ICD10
Q93.5

Kod ICD11
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*Źródło

orphanet