Opis choroby *

Definicja

A rare autosomal anomaly syndrome, with a highly variable phenotype, typically characterized by short length, joint abnormalities (e.g. dysplasia, hyperextensibility, contractures, dislocation), congenital cardiac defects, and craniofacial dysmorphism (incl. microcephaly, a high, prominent, narrow and/or hairy forehead, epicanthus, upward-slanting and/or small palpebral fissures, broad, high or depressed nasal bridge and malformed ears). Delayed motor development and intellectual disability is observed in patients not presenting early demise.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(22)
Del(22)
Delecja 22
Deletion 22

Kod ORPHA
96123

Kod OMIM
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Kod ICD10
Q93.0

Kod ICD11
-

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*Źródło

orphanet