

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	Synonimy
Zespół wad wrodzonych	Del(22)
	Del(22)
	Delecja 22
	Deletion 22

Kod ORPHA	Kod OMIM	Kod ICD10
96123	-	Q93.0

Kod ICD11

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*Źródło

orphanet