

Opis choroby *

Definicja

Xp22.3 microdeletion syndrome is a microdeletion syndrome resulting from a partial deletion of the chromosome X. Phenotype is highly variable (depending on length of deletion), but is mainly characterized by X linked ichthyosis, mild-moderate intellectual deficit, Kallmann syndrome, short stature, chondrodysplasia punctata and ocular albinism. Epilepsy, attention deficit-hyperactivity disorder, autism and difficulties with social communication can be associated.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Del(X)(p23) Del(X)(p23)

Kod ORPHA	Kod OMIM	Kod ICD10
1643	-	Q99.8

Kod ICD11
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*Źródło

orphanet