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
Zespół wad wrodzonych

Synonimy
Del(X)(p23)
Del(X)(p23)

Kod ORPHA
1643

Kod OMIM
-

Kod ICD10
Q99.8

Kod ICD11
-

---

*Źródło

orphanet