

Opis choroby *

Definicja

A rare syndrome with 46,XY disorder of sex development characterized by variable degrees of intellectual disability, short stature, severe genital anomalies resulting in sexual ambiguity (such as pseudovaginal perineoscrotal hypospadias and persistence of Müllerian structures), and ocular anomalies (microphthalmia, coloboma). Craniofacial peculiarities (coarse features, deep set eyes), spina bifida, imperforate anus, and sensorineural hearing loss were also described. No new cases have been reported since 1994.

Dane

Klasyfikacja

Choroba

Synonimy

Verloes-Gillerot-Fryns syndrome

Pseudohermafrodytyzm - upośledzenie umysłowe

Zespół Verloesa, Gillerota i Frynsa

Disorder of sex development-intellectual disability syndrome

Kod ORPHA

2983

Kod OMIM

600122

Kod ICD10

Q56.3

Kod ICD11

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*Źródło

orphanet