Opis choroby *

Definicja

A rare syndrome with 46,XY disorder of sex development characterized by mild developmental delay and streak gonads associated with short stature, cardiac, renal, musculoskeletal, and ectodermal abnormalities (the latter including scalp defects and unusual hair whorls), and dysmorphic facial features (such as preauricular pits, short columella, and small nares). There have been no further descriptions in the literature since 1980.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1770

Kod OMIM
233430

Kod ICD10
Q99.1

Kod ICD11
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*Źródło

orphanet