

Opis choroby *

Definicja

46,XY ovotesticular disorder of sex development is a rare, genetic disorder of sex development characterized by either the coexistence of both male and female reproductive gonads or, more frequently, by the presence of one or both gonads containing a mixture of both testicular and ovarian tissue (ovotestes) in an individual with a normal male 46, XY karyotype. External genitalia are usually ambiguous, but can range from normal male to normal female and if a uterus and/or fallopian tubes are present, they are generally hypoplastic. Cryptorchidism, hypospadias, infertility and increased risk of gonadal tumours are frequently associated.

Dane

Klasyfikacja

Choroba

Synonimy

46,XY ovotesticular DSD

Jajnikowo-jądrowe ZRP z karyotypem 46,XY

Kod ORPHA

325345

Kod OMIM

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Kod ICD10

Q56.0

Kod ICD11

LD2A.0

*Źródło

orphanet