46,XY ovotesticular disorder of sex development

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 46,XY ovotesticular disorder of sex development. ORPHA:325345

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.