46,XY disorder of sex development due to isolated 17,20-lyase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 46,XY disorder of sex development due to isolated 17,20-lyase deficiency. ORPHA:90796

46,XY disorder of sex development due to isolated 17,20-lyase deficiency is a rare disorder of sex development due to reduced 17,20-lyase activity that affects individuals with 46,XY karyotype and is characterized by ambiguous external genitalia, including micropenis, perineal hypospadias, bifid scrotum, cryptorchidism, and a blind vaginal pouch. Blood pressure and electrolytes are normal whilst hormonal investigations show normal basal and stimulated levels of cortisol, and low basal and stimulated androgen levels.