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46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency. ORPHA:753

46, XY disorder of sex development (DSD; see this term) due to 5-alpha-reductase 2 (SRD5A2) deficiency is a disorder of sex development due to a defect in testosterone (T) metabolism resulting in incomplete intrauterine masculinization. Patients present an ambiguous external genitalia which varies from a female with a blind vaginal pouch to a fully male phenotype with pseudovaginal posterior hypospadias (see this term) or only micropenis.