46,XX disorder of sex development-anorectal anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 46,XX disorder of sex development-anorectal anomalies syndrome. ORPHA:2973

46,XX disorder of sex development-anorectal anomalies syndrome is a rare developmental defect during embryogenesis syndrome characterized by a normal female karyotype, normal ovaries, male or ambiguous genitalia, urinary tract malformations (ranging from bilateral renal agenesis to mild unilateral hydronephrosis), Müllerian duct anomalies (e.g. complete absence of the uterus and vagina, bicornuate uterus), and imperforate anus. Additional features may include tracheoesophageal fistula, radial aplasia, and malrotation of the gut.