46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency. ORPHA:168558

46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency is a rare, genetic, developmental defect during embryogenesis disorder characterized by severe, early-onset, salt-wasting adrenal insufficiency and ambiguous/female external genitalia (irrespective of chromosomal sex) due to mutations in the CYP11A1 gene. Milder cases may present delayed onset of adrenal gland dysfunction and genitalia phenotype may range from normal male to female in individuals with 46,XY karyotype. Imaging studies reveal hypoplastic/absent adrenal glands and biochemical findings include low serum cortisol, mineralocorticoids, androgens, and sodium, with elevated potassium levels.