Open Peer Review on Qeios

Hyperandrogenism due to cortisone reductase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hyperandrogenism due to cortisone reductase deficiency. ORPHA:168588

A rare, genetic, endocrine disease characterized by defect in conversion of cortisone to active cortisol, resulting in ACT H-mediated excessive androgen release from adrenal glands. Premature adrenarche is typical with precocious pseudopuberty, proportionate tall stature and accelerated bone maturation in males, and hirsutism, oligoamenorrhea, central obesity and infertility in females. Imaging studies may indicate adrenal hyperplasia.