

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Synonimy

XY sex reversal-adrenal failure

Odwrócenie płci XY - niewydolność nadnerczy

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

Kod ORPHA

168558

Kod OMIM

613743

Kod ICD10

Q56.1

Kod ICD11

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*Źródło

orphanet