

Opis choroby *

Definicja

46,XY disorder of sex development due to isolated 17,20-lyase deficiency is a rare disorder of sex development due to reduced 17,20-lyase activity that affects individuals with 46,XY karyotype and is characterized by ambiguous external genitalia, including micropenis, perineal hypospadias, bifid scrotum, cryptorchidism, and a blind vaginal pouch. Blood pressure and electrolytes are normal whilst hormonal investigations show normal basal and stimulated levels of cortisol, and low basal and stimulated androgen levels.

Dane

Klasyfikacja

Choroba

Synonimy

46,XY disorder of sex development due to isolated 17,20-lyase deficiency

Kod ORPHA

90796

Kod OMIM

202110

Kod ICD10

E29.1

Kod ICD11

LD2A.3

*Źródło

orphanet