Zaburzenie różnicowania płci z kariotypem 46,XY spowodowane izolowanym niedoborem 17, 20-liazy Kod Orpha: 90796 Kod OMIM: 202110

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.