

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

Kod ORPHA	Kod OMIM	Kod ICD10
90796	202110	E29.1

Kod ICD11  
LD2A.3

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\*Źródło

orphanet

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie [www.orphanet.pl](http://www.orphanet.pl)