

## 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

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#### \*Źródło

orphanet