

## Opis choroby \*

### Definicja

A rare disorder of sex development (DSD) due to a defect in metabolizing testosterone to dihydrotestosterone and characterized by incomplete intrauterine masculinization which ranges from a female genitalia with a blind vaginal pouch to a fully male phenotype with pseudovaginal posterior hypospadias and micropenis.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

46,XY DSD due to 5-alpha-reductase 2 deficiency  
Niedobór 5-alfa-reduktazy steroidowej  
Pseudohermafrodytyzm męski spowodowany niedoborem 5-alfa-reduktazy 2  
ZRP 46,XY spowodowane niedoborem 5-alfa-reduktazy 2  
Pseudovaginal perineoscrotal hypospadias  
Steroid 5-alpha-reductase 2 deficiency  
46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency

#### Kod ORPHA

753

#### Kod OMIM

264600

#### Kod ICD10

E29.1

#### Kod ICD11

LD2A.3

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\*Źródło

orphanet