

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

*Źródło

orphanet