

**DISEASE:**

**46,XY difference of sex development due to 5-alpha-reductase 2 deficiency**

<b>NAME:</b>	46,XY difference of sex development due to 5-alpha-reductase 2 deficiency
<b>DESCRIPTION:</b>	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.
<b>ORPHACODE:</b>	753
<b>SYNONYMS:</b>	46,XY DSD due to 5-alpha-reductase 2 deficiency 46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency Pseudovaginal perineoscrotal hypospadias Steroid 5-alpha-reductase 2 deficiency
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">OMIM</a> <a href="#">MedDRA</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	<a href="#">SRD5A2</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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