

Opis choroby *

Definicja

46,XX disorder of sex development-anorectal anomalies syndrome is a rare developmental defect during embryogenesis syndrome characterized by a normal female karyotype, normal ovaries, male or ambiguous genitalia, urinary tract malformations (ranging from bilateral renal agenesis to mild unilateral hydronephrosis), Müllerian duct anomalies (e.g. complete absence of the uterus and vagina, bicornuate uterus), and imperforate anus. Additional features may include tracheoesophageal fistula, radial aplasia, and malrotation of the gut.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Pseudohermafrodytyzm żeński - wady odbytnicy i odbytu
	46,XX disorder of sex development-anorectal anomalies syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
2973	-	Q56.2

Kod ICD11
LD2F.1Y

*Źródło

orphanet