ORIGINAL RESEARCH PAPER
Volume - 15 | Issue - 09 | September - 2026 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A RARE 46,XY DISORDER OF SEX DEVELOPMENT PRESENTING IN ADOLESCENCE WITH AMBIGUOUS GENITALIA AND BILATERAL INGUINAL TESTES: A CASE REPORT

Paediatric Surgery

Dr. Rohith Yadav G* Assistant Professor, Department of General Surgery, SABVMCRI, Bengaluru, India *Corresponding Author

Dr. Sandhya N Associate Professor, Department of Surgery, KIMS, Hubli, India

Dr. Rajashankar Professor and Head, Department of Paediatric Surgery, KIMS, Hubli, India

ABSTRACT

Background: 46,XY differences/disorders of sex development (DSD) comprise a heterogeneous group of conditions in which chromosomal, gonadal and phenotypic sex are discordant. The presentation may range from severe genital undervirilization to predominantly male external genitalia. Delayed diagnosis may occur when the phenotype is interpreted according to the sex assigned at birth. **Case Presentation:** A 14-year-old individual reared as female presented with primary amenorrhea, an enlarged phallus, male secondary sexual characteristics and libido. Examination demonstrated a rudimentary penis/phallus, perineal hypospadias with a vaginal introitus, and bilateral inguinal swellings measuring approximately 2 × 4 cm on the right and 3 × 4 cm on the left. Ultrasonography and MRI demonstrated a left testis at the superficial inguinal ring and a right testis at the deep inguinal ring, with absence of the uterus and ovaries. Karyotyping demonstrated 46,XY. Urethroscopy revealed a common urogenital tract with a vaginal blind pouch and perineal hypospadias. Laparoscopy confirmed a normally appearing intra-abdominal right testis near the deep ring. In view of the patient's expressed desire to live as male, a masculinizing surgical pathway was undertaken, including stage 1 Fowler–Stephens orchidopexy on the right and left orchidopexy with fixation in a superficial inguinal position. Further reconstruction of the blind vaginal pouch and urethroplasty was planned after six months. **Conclusion:** This case illustrates the importance of a structured multidisciplinary evaluation of adolescents with primary amenorrhea and atypical genitalia. Karyotype, gonadal localization and assessment of Müllerian structures establish the phenotypic framework, while biochemical and molecular studies are important for defining the underlying etiology. Although 5α-reductase type 2 deficiency was considered in the original case presentation, the available records do not contain testosterone/dihydrotestosterone measurements or SRD5A2 molecular confirmation; therefore, the etiologic diagnosis should remain provisional until such testing is performed.

KEYWORDS

46,Xy Dsd; Differences of Sex Development; Ambiguous Genitalia; 5α-Reductase Deficiency; Hypospadias; Orchidopexy; Fowler–Stephens; Adolescent; Primary Amenorrhea

INTRODUCTION

46,XY differences/disorders of sex development (DSD) are a heterogeneous group of congenital conditions characterized by discordance among chromosomal sex, gonadal development and/or genital phenotype. The older term "male pseudohermaphroditism" has historically been used for some 46,XY presentations, but contemporary literature favors the term 46,XY DSD or 46,XY difference of sex development. The 2006 international consensus and subsequent reviews emphasize a multidisciplinary approach, careful characterization of the external genitalia and gonads, biochemical assessment, imaging and genetic investigation. [1–3]

Among the important causes of 46,XY DSD are disorders of androgen synthesis, disorders of androgen action such as androgen insensitivity syndrome (AIS), 5α-reductase type 2 deficiency, disorders of gonadal development and defects in testosterone biosynthesis. The phenotype can overlap substantially, particularly between partial androgen insensitivity syndrome and 5α-reductase type 2 deficiency. Consequently, clinical appearance alone is insufficient for etiologic diagnosis. Contemporary diagnostic pathways recommend formal karyotyping, abdominopelvic imaging, age-appropriate hormonal evaluation and stepwise genetic testing when indicated. [2,4,5]

5α-reductase type 2 deficiency, caused by pathogenic variants in SRD5A2, impairs conversion of testosterone to dihydrotestosterone (DHT), an androgen that has a major role in fetal external genital masculinization. Affected 46,XY individuals may therefore present with varying degrees of undervirilization at birth and may undergo substantial virilization at puberty. However, biochemical thresholds are age-dependent and the testosterone/DHT ratio has important limitations; molecular testing and/or urinary steroid profiling can provide stronger confirmation. [5–7]

We report an adolescent 46,XY DSD case with ambiguous external genitalia, bilateral inguinal testes, absent uterus and ovaries, a common urogenital tract and a male gender identity. The case highlights the diagnostic complexity of late-presenting DSD and the importance of aligning surgical planning with the patient's informed preferences while considering gonadal function, tumor risk, fertility potential and long-term quality of life.

Case Presentation

A 14-year-old individual, documented in the source presentation as a female, presented with primary amenorrhea and male secondary sexual characteristics, including libido. The patient had an enlarged phallus and was noted clinically to have a rudimentary penis with perineal hypospadias and a vaginal introitus. Bilateral inguinal swellings were palpable, measuring approximately 2 × 4 cm on the right and 3 × 4 cm on the left. The clinical findings raised the possibility that the inguinal swellings represented gonads.

The source presentation does not provide a detailed birth history, family history, consanguinity history, previous genital surgery, growth parameters, or a formal external masculinization score. These details should be added from the clinical record before journal submission if available.

Table 1. Clinical and Diagnostic Findings

Domain	Finding
Age / recorded sex	14 years; documented as female in the source presentation
Presenting features	Primary amenorrhea; male secondary sexual characteristics; libido; enlarged phallus
External genitalia	Rudimentary penis/phallus; perineal hypospadias; vaginal introitus
Gonads	Right inguinal swelling ~2 × 4 cm; left inguinal swelling ~3 × 4 cm
Ultrasonography / MRI	Left testis at superficial inguinal ring; right testis at deep inguinal ring; uterus and ovaries absent
Karyotype	46,XY
Urethroscopy	Common urogenital tract; vaginal blind pouch; perineal hypospadias
Laparoscopy	Normally appearing intra-abdominal right testis over deep ring

Investigations

Ultrasonography and MRI localized the left testis to the superficial inguinal ring and the right testis to the deep inguinal ring. The uterus and ovaries were not visualized. Karyotyping demonstrated a 46,XY chromosomal complement. Urethroscopy demonstrated a common urogenital tract with a vaginal blind pouch and perineal hypospadias.

Diagnostic interpretation: The combination of 46,XY karyotype, bilateral testes and absent Müllerian structures establishes a 46,XY DSD phenotype with functioning testicular Sertoli-cell activity

sufficient to prevent Müllerian duct persistence. The available material, however, does not include serum testosterone, DHT, LH, FSH, AMH, inhibin B, 17-hydroxyprogesterone, androstenedione, testosterone/androstenedione ratio, hCG stimulation testing, urinary steroid profile, androgen receptor testing or SRD5A2 sequencing. Thus, the records supplied do not establish whether the underlying diagnosis is 5α-reductase type 2 deficiency, partial androgen insensitivity syndrome, another androgen biosynthetic disorder, or a different form of 46,XY DSD. [4–7]

Management

Because the patient expressed a desire to live as male, the treating team planned a male-oriented reconstructive pathway. Laparoscopy was performed and demonstrated a normally appearing intra-abdominal right testis situated over the deep ring. The risk of gonadal malignancy was explained. A trial of stage 1 Fowler–Stephens orchidopexy was performed for the right testis. The left testis was mobilized and fixed in a superficial inguinal pouch. The patient was counselled regarding subsequent obliteration/management of the vaginal blind pouch and urethroplasty after approximately six months.

The supplied material does not document postoperative hormone therapy, postoperative testicular position, vascularity, urethral outcome, complications, histopathology, fertility assessment, or long-term follow-up. It therefore would be inappropriate to claim definitive surgical success or a completed reconstruction. These outcomes should be added when available.

Figure 1A. Clinical Photograph Demonstrating Atypical External Genitalia with a Prominent Phallus and Perineal Opening.

Figure 1B. Clinical Photograph Demonstrating the External Genital Phenotype and Perineal Hypospadias.

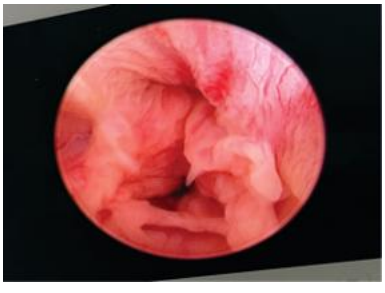

Figure 2A. Endoscopic/Urethroscopic Image from the Source Presentation Demonstrating the Common Urogenital Tract.

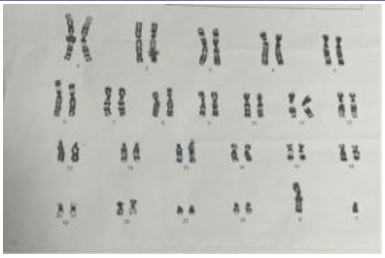

Figure 2B. Karyotype Image from the Source Presentation Demonstrating a 46,XY Complement.

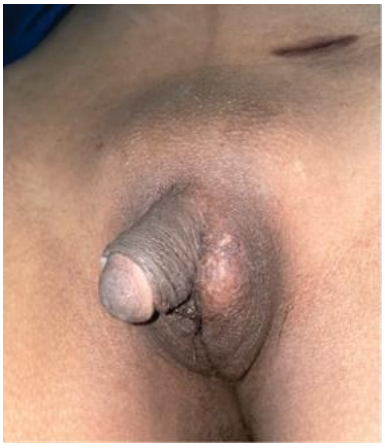

Figure 3. Additional Clinical Photograph Demonstrating the External Genital Phenotype and Palpable Gonadal Region.

DISCUSSION

The principal clinical lesson is that an adolescent presenting with primary amenorrhea and virilization should not be assumed to have a conventional female reproductive anatomy. The finding of palpable bilateral inguinal gonads, followed by imaging demonstrating testes and absence of a uterus, should prompt a 46,XY DSD evaluation. Current consensus recommendations emphasize coordinated assessment by experienced endocrinology, genetics, radiology, urology/paediatric surgery and psychological or psychosocial services. [2,3]

From a developmental perspective, the presence of testes in a 46,XY individual provides a plausible source of anti-Müllerian hormone (AMH), explaining the absence of a uterus and ovaries. External genital masculinization, however, depends on adequate androgen production, conversion and tissue responsiveness. Testosterone is converted to DHT by 5α-reductase type 2, encoded by SRD5A2. Deficiency can result in a spectrum from predominantly female external genitalia to more substantially masculinized genitalia, with variable pubertal virilization. [6,7]

An important issue in this case is the distinction between 5α-reductase type 2 deficiency and partial androgen insensitivity syndrome (PAIS). These disorders can produce overlapping genital phenotypes. Published Indian data also demonstrate that clinical/hormonal provisional diagnoses may correlate imperfectly with molecular findings; in one South Indian cohort of 37 children with 46,XY DSD, only 22.2% of provisional diagnoses based on clinical and hormonal assessment were concordant with a pathogenic molecular diagnosis. [8]

Where 5α-reductase deficiency is suspected, an age-appropriate endocrine evaluation may include testosterone and DHT measurement, with careful interpretation of the testosterone/DHT ratio, and may be supplemented by urinary steroid profiling and SRD5A2 molecular analysis. Recent evidence emphasizes that stimulated androgen ratios can be informative but are not infallible; genetic confirmation is particularly valuable when the biochemical phenotype is equivocal. [6,7,8]

The surgical component of this case also illustrates the need for individualized planning. Orchidopexy can improve the position of an undescended testis, may facilitate surveillance and may preserve endocrine function and potential fertility when the gonads are

functional. The decision to retain or remove testes in a 46,XY DSD patient should, however, be based on the specific molecular diagnosis, gonadal histology/function, tumor risk, patient's gender identity and informed preferences. Reviews emphasize that gonadal tumor risk varies widely among DSD etiologies rather than being uniform across all patients with Y-chromosome material. [9,10]

In the present case, the right testis was intra-abdominal near the deep ring and the patient was counselled regarding malignancy risk. This was appropriate to document because tumor risk is an important component of long-term DSD care. Continued specialist surveillance and individualized discussion of future gonadal management are appropriate.

Another important issue is the timing and sequencing of genital reconstruction. The patient had a common urogenital tract, a vaginal blind pouch and perineal hypospadias. A staged approach was selected, with further management planned after six months. Contemporary DSD care increasingly emphasizes functional goals, avoidance of unnecessary irreversible procedures, preservation of fertility and sexual function where possible, and participation of the patient in decisions as developmentally appropriate. [3,9]

Finally, the adolescent presentation emphasizes the psychosocial dimension of DSD. Gender identity, previous sex of rearing, expectations about sexual function and fertility, family dynamics, disclosure, stigma and body image can all affect outcomes. A multidisciplinary team should provide age-appropriate counselling and shared decision-making rather than treating the condition solely as an anatomical surgical problem. [3,9]

Clinical Timeline

Event	Timing	Details
Presentation	14 years	Primary amenorrhea, virilization, enlarged phallus and bilateral inguinal swellings.
Imaging	At presentation	USG/MRI localized bilateral testes; uterus and ovaries absent.
Cytogenetics	During evaluation	Karyotype: 46,XY.
Endoscopy	During evaluation	Common urogenital tract, vaginal blind pouch and perineal hypospadias.
Operative assessment	During surgical management	Laparoscopy confirmed right intra-abdominal testis near deep ring.
Surgery	Index operation	Stage 1 Fowler–Stephens orchidopexy on right; left orchidopexy to superficial inguinal pouch.
Planned second stage	~6 months	Further management of vaginal blind pouch and urethroplasty planned.

Differential Diagnosis and Recommended Completion of Etiologic Work-up

Most Relevant Differentials: 5α-reductase type 2 deficiency; partial androgen insensitivity syndrome; defects of testosterone biosynthesis including 17β-hydroxysteroid dehydrogenase type 3 deficiency; and selected disorders of gonadal development.

CONCLUSION

46,XY DSD can remain unrecognized until adolescence when atypical genital anatomy is not investigated earlier. In an adolescent with primary amenorrhea, virilization and inguinal gonads, the combination of careful genital examination, karyotyping, gonadal localization and assessment of Müllerian structures can rapidly establish the phenotypic diagnosis. Definitive etiologic diagnosis requires age-appropriate endocrine and, when indicated, molecular testing. In patients who express a male gender identity, orchidopexy and staged genital reconstruction may be considered within a multidisciplinary, shared-decision framework. In this case, 5α-reductase type 2 deficiency remains a differential diagnosis rather than a confirmed molecular diagnosis.

Declarations

Patient Consent: written informed consent for publication was taken.

Conflict of Interest: Authors declared no conflict of interests.

REFERENCES

1. Hughes IA, Houk C, Ahmed SF, Lee PA; LWPES/ESPE Consensus Group. Consensus statement on management of intersex disorders. J Pediatr Urol. 2006;2(3):148-162. doi:10.1016/j.jpurol.2006.03.004.
2. Baidya A, Basu AK, Bhattacharjee R, et al. Diagnostic approach in 46, XY DSD: an Endocrine Society of Bengal consensus statement. J Pediatr Endocrinol Metab. 2023;36(1):4-18. doi:10.1515/jpem-2022-0515.
3. Mendonca BB, Domenice S, Arnhold IJP, Costa EMF. 46,XY disorders of sex development: hormonal, molecular and anatomical aspects. Best Pract Res Clin Endocrinol Metab. 2019;33(3):101270. (Use the journal's preferred citation format after final verification.)
4. Audi L, Ahmed SF, Krone N, et al. GENETICS IN ENDOCRINOLOGY: Approaches to molecular genetic diagnosis in the management of disorders of sex development. Eur J Endocrinol. 2018;179(4):R197-R209.
5. Hughes LA, McKay-Bounford K, Webb EA, et al. Next generation sequencing (NGS) to improve the diagnosis and management of patients with disorders of sex development (DSD). Endocr Connect. 2019;8(4):100-110.
6. Balagamage C, Igbokwe R, Cole T, et al. The diagnostic value of stimulated androgen ratios in 5-alpha reductase type 2 (SRD5A2) deficiency: a case series and review of the literature. J Pediatr Endocrinol Metab. 2025;38(9):922-930. doi:10.1515/jpem-2025-0195.
7. Wong P, et al. Diagnosis of 5α-reductase 2 deficiency: is measurement of dihydrotestosterone essential? Clin Chem. 2013. doi:10.1373/clinchem.2012.196501.
8. Palui R, Ravichandran L, Kamalanathan S, et al. Clinical, Hormonal, and Genetic Spectrum of 46 XY Disorders of Sexual Development (DSD) Patients. Indian J Pediatr. 2025;92(8):836-843. doi:10.1007/s12098-024-05144-8.
9. Moser C, et al. Management of 46,XY differences/disorders of sex development (DSD) throughout life. Endocr Rev. 2019;40(6):1547-1572. doi:10.1210/er.2019-00083.
10. Cools M, et al. A practical guide for evaluating gonadal germ cell tumor predisposition in differences of sex development. Am J Med Genet C Semin Med Genet. 2017. doi:10.1002/ajmg.c.31562.
11. Gagnier JJ, Kienle G, Altman DG, et al. The CARE guidelines: consensus-based clinical case report guideline development. J Clin Epidemiol. 2014;67(1):46-51. doi:10.1016/j.jclinepi.2013.08.003.
12. Agha RA, Franchi T, Sohrabi C, Mathew G, Kerwan A; SCARE Group. The SCARE 2023 guideline: updating consensus Surgical CAse REport (SCARE) guidelines. Int J Surg. 2023;109(5):1136-1140. (Verify final bibliographic details against the target journal.)