Gandhi D *et al* JPCHR (2025) 01:e25041501
doi: 10.63486/jpchr.25041501

Journal of Public and Clinical Health Research

Case Report

A Rare Case Report of Ovo testicular Disorder of Sex Development in an Ambiguous Genitalia Case

Dulari Gandhi[1], Poojan Joshi[1], Yuvraj Vala[1], Rishilkumar Patel[1], Aftabhusen Memon[1]

[1]Shree C.A. Patel Hospital & Community health centre, Mota Fofadiya, Vadodara, India

Correspondence: Dr Poojan Joshi, Email: poojanjoshi111@gmail.com

Abstract

Usually, the distinction between male and female is considered absolute, allowing for instantaneous sex assignment at birth. This process is based primarily on external genitalia, which are typically classified as either male or female. However, in rare instances, ambiguity in external genitalia prevents immediate sex assignment. This necessitates further clinical evaluation commonly known as Disorders of Sex Development (DSD).

Among various types of DSDs, Ovo testicular DSD is a particularly rare condition, with approximately only 500 cases reported worldwide. Among various types of DSDs, a case of Ovo testicular DSD with a 46, XX/46, XY karyotype has been presented in this article. A comprehensive analysis, including detailed investigation and diagnostic evaluation, is provided. Additionally, we discuss various management options, emphasizing the clinical challenges and considerations associated with this rare disorder.

Key Words: Disorders of Sex Development, 46, XX/46, XY chimerism, Ovo-testicular Disorder

Introduction

Disorders of sex development (DSD) can have a wide range of presenting phenotypes depending on the underlying condition as well as its severity. It is estimated that abnormalities requiring formal investigation occur in approximately 1 in 4,000 births.[1] Defining the exact basis of DSDs can have important implications for gender assignment, predicting response to treatment (e.g., androgen supplementation), assessing associated features (e.g., adrenal dysfunction) or the risk of tumorigenesis, and determining likely fertility options as well as long-term counselling for the individuals and their family.

Most patients with Ovo testicular DSD have a 46, XX karyotype, particularly in South and Western Africa.[2–4] whereas 46, XX/46, XY chimerism has been in individuals from North America and Europe. The molecular basis of this disorder is known in developed countries, but familial cases have been reported, and both autosomal recessive and sex-limited autosomal dominant transmission have been proposed. SRY translocations are rare.[5] Ovo testicular DSD associated with a 46, XY karyotype is rare, and may represent cryptic gonadal mosaicism for a Y chromosome deletion or early sex-determining gene mutation. Thus, Ovo testicular DSD likely represents several different aetiologies.

Case Report

One and a half year old child presented with a history of ambiguous genitalia. He/she was born out of a non-consanguineous marriage at full term, with a birth weight of 2.8 kg without any perinatal complications/ uneventful. On birth noticed to have genital ambiguity.

Clinical examination of the child revealed a nutritionally normal child without any dysmorphic features excepting sexual ambiguity. Vitals and systemic examination were normal. External genitalia revealed ill formed bifid scrotum with absent scrotal rugosity, tiny hypoplastic gonads palpable on both sides with gonadal volume of less than 1 ml, microphallus <1 cm size, urethral orifice was seen at the tip of phallus, Vaginal orifice visible and there no localized area of hyperpigmentation (Figure 1). With the above clinical findings, a provisional diagnosis of 46, XY, DSD, under virilized male was made. Clinical diagnosis of under virilized DSD either a 46, XY or a mosaic.

Investigations

Table 1:
List of Investigations and Results

Investigation	Results
Karyotype	46XX /46XY (72% and 28%)
Hormonal Assays	Testosterone: 0.55 (1.68-7.58 ng/ml) Serum FSH: 16.38 (1.27-19.26 mIU/ml) Serum LH: 1.80 (1.24-8.62 mIU/ml) Serum Estradiol: 175 (20-75 pg/ml)
USG Abdomen	Rudimentary uterus with absence of ovaries and prostate

Article Received: 20-02-2025 | Accepted: 14-04-2025 | Published: 15-04-2025

Investigation	Results
Genitogram	Patency of urogenital tract showed enhancement of contrast posterior to the urethra
MRI pelvis	Presence of rudimentary uterus and ovaries and presence of right testis in the inguinal ring

Figure 1:
Genital Appearance of the Baby

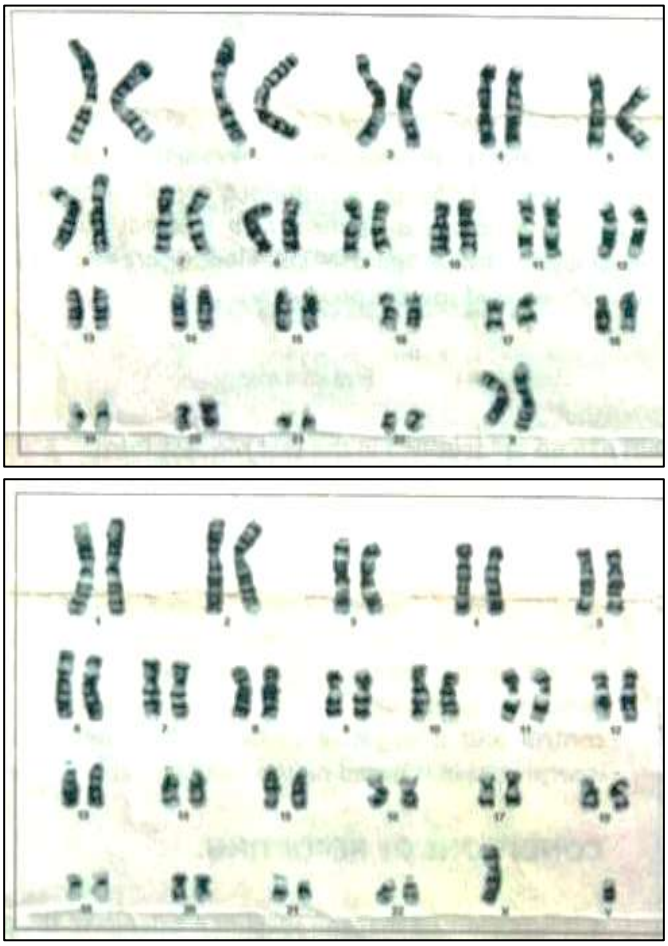
Discussion

The diagnosis of Ovo testicular DSD should be considered in all patients with ambiguous genitalia, especially when there is more pronounced asymmetry of genitalia or detection of a gonad on the right. A 46, XX/46, XY karyotype strongly supports the diagnosis, but the detection of a 46, XX or 46, XY karyotype does not exclude the diagnosis.

Pelvic imaging, with ultrasound or magnetic resonance imaging (MRI) is useful for visualizing internal genitalia. The presence of testicular tissue may also be suggested by the measurement of basal testosterone, AMH and inhibin B in the first months of life, as well as testosterone increase following hCG stimulation.

Ovo testicular DSD has a spectrum of presentation.[6] Most patients who present early have ambiguous genitalia or significant hypospadias.[7] Cryptorchidism is common, but at least one gonad is palpable usually in the labioscrotal fold or inguinal region, more often on the right, and often associated with an inguinal hernia.[8] The differentiation of the genital ducts usually follows that of the gonad, and a hemiuterus or rudimentary uterus is often present on the side of the ovary/ovotestis.[9]

Figure 2:
Chromosomal Analysis of Blood smear



The management of Ovo-testicular DSD varies depending upon the age at diagnosis, genital development, internal structures, and reproductive capacity. Those individuals with a 46, XX karyotype and a uterus are likely to have functional ovarian tissue, and female assignment is likely to be appropriate. Potentially functional testicular tissue should be removed and monitored postoperatively by serum AMH levels and by showing a lack of testosterone response to hCG stimulation. The risk of malignant transformation in the ovarian tissue of 46, XX patients is not known.

A male gender assignment may be more appropriate if there is reasonable phallic development and müllerian structures are absent or very poorly formed. Ovarian tissue is usually removed to prevent estrogenization at

puberty and remnant müllerian structures can be removed by an experienced surgeon. The prevalence of gonad blastoma and/or germinoma arising in the testicular tissue of patients with 46,XX Ovo testicular DSD has been estimated at 3% to 4 % and the Ovo testicular tissue is usually dysgenetic, so removal of this testicular tissue has been advocated.[10] However, the management of a histologically normal scrotal positioned testis is more difficult, and careful monitoring and biopsy for carcinoma in situ in adolescence may be an appropriate strategy.

Gender identity is an important consideration in patients with Ovo testicular DSD who first present in late childhood or adolescence, due to androgenization in girls or estrogenization in boys. In most cases, gender identity is consistent with sex of rearing. The discordant gonad and dysgenetic tissue should be removed to prevent further androgenization in girls and estrogenization in boys, following appropriate counselling. Sex hormone supplementation may be required for complete pubertal development.

References

1. Sax L. How common is intersex? a response to Anne Fausto-Sterling. J Sex Res 2002;39(3):174–8. doi:10.1080/00224490209552139.
2. Wiersma R. Management of the African child with true hermaphroditism. J Pediatr Surg 2001;36(2):397–9. doi:10.1053/JPSU.2001.20726.
3. Spurdle AB, Shankman S, Ramsay M. XX true hermaphroditism in southern African blacks: exclusion of SRY sequences and uniparental disomy of the X chromosome. Am J Med Genet 1995;55(1):53–6. doi:10.1002/AJMG.1320550115.
4. Krob G, Braun A, Kuhnle U. True hermaphroditism: geographical distribution, clinical findings, chromosomes and gonadal histology. Eur J Pediatr 1994;153(1):2–10. doi:10.1007/BF02000779.
5. McElreavey K, Rappaport R, Vilain E, Abbas N, Richaud F, Lortat-Jacob S, et al. A minority of 46,XX true hermaphrodites are positive for the Y-DNA sequence including SRY. Hum Genet 1992;90(1–2):121–5. doi:10.1007/BF00210754.
6. Damiani D, Fellous M, McElreavey K, Barbaux S, Barreto ESA, Dichtchekenian V, et al. True hermaphroditism: clinical aspects and molecular studies in 16 cases. Eur J Endocrinol 1997;136(2):201–4. doi:10.1530/EJE.0.1360201.
7. De MR, Escano J, Mejia Sang ME, Reyes-Mugica M, Colaco M, Fox J. Ovotesticular Disorder of Sex Development: Approach and Management of an Index Case in the Dominican Republic. Cureus 2021;13(10):e18512. doi:10.7759/CUREUS.18512.
8. Kilberg MJ, McLoughlin M, Pyle LC, Vogiatzi MG. Endocrine Management of Ovotesticular DSD, an Index Case and Review of the Literature. Pediatr Endocrinol Rev 2019;17(2):110. doi:10.17458/per.vol17.2019.kmv.endocrineovotesticulardsd.
9. Syryn H, Van De Vijver K, Cools M. Ovotesticular Difference of Sex Development: Genetic Background, Histological Features, and Clinical Management. Horm Res Paediatr 2023;96(2):180–9. doi:10.1159/000519323.
10. Cools M, Drop SLS, Wolffenbuttel KP, Oosterhuis JW, Looijenga LHJ. Germ cell tumors in the intersex gonad: old paths, new directions, moving frontiers. Endocr Rev 2006;27(5):468–84. doi:10.1210/ER.2006-0005.