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Case Report

True hermaphrodite with teratoma

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Abstract

A 32 years old healthy male, married for 6 years presented with infertility. Clinical examination revealed normal external genitalia with underdeveloped empty scrotum. Radiological investigations reported bilateral intra abdominal testis with left testis showing features of teratoma. Intra-operative findings showed bilateral intra abdominal testis along with uterus and fallopian tube and left testis with features of teratoma. Histopathology confirmed the presence of uterus, cervix, right adnexa with atropic testis with tube, seminal vesical, epididymis, and left adnexa ovotestis with mature teratoma, tube and epididymis. Cytogenetics reported 46, XY karyotype. This case is rare presentation of combination of male phenotype true hermaphrodite with presence of teratoma.

Key words

True hermaphrodite, Teratoma, Ovotestis, Karyotype, Histopathology.

Introduction

True hermaphroditism is a rare form of ambiguous genitalia characterized by simultaneous presence of both normal male and female gonadal tissues [1, 2]. Although western literature recommends for early gender assignment surgeries in such patients keeping in mind the psycho-sexual development and risk of neoplasia, the subject still remains an enigma in Indian society where social taboos prevent such patients from having medical treatment. We have reported here one such case of true hermaphroditism who presented to us in middle age with infertility.

Case report

A middle aged male married for 6 years presented with infertility and bilateral undescended testis. Physical examination was normal. Secondary sexual characters were normal for male. Penis well developed; Scrotum infantile; and rugosity absent and absent testis. **(Photo – 1, Photo - 2)**

Radiological investigations **(Photo – 3A, 3B)** showed the presence of bilateral undescended testis with features suggestive of teratoma on left side with possibility of uterus with was considered. Patient underwent metastatic workup for possibility of tumor and was negative (Serum LDH normal, Alpha fetoprotein

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normal and Serum beta HCG normal). Patient underwent exploratory laparotomy and findings suggested of uterus with bilateral fallopian tubes with left vas deferens with Left side mass (? ovarian/ testicular tumor) and Right (? ovary/ testis). **(Photo – 4A, 4B)**

Photo - 1: Normal secondary sexual characters of male.

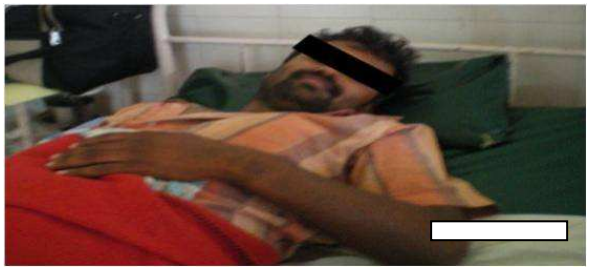
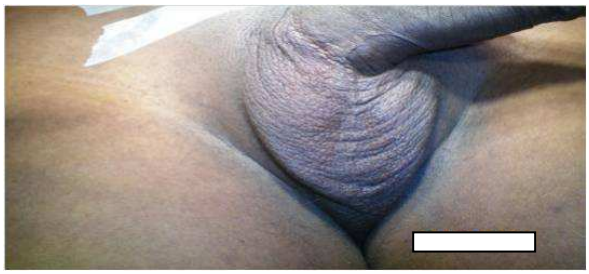


Photo - 2: Infantile scrotum with absent testis.

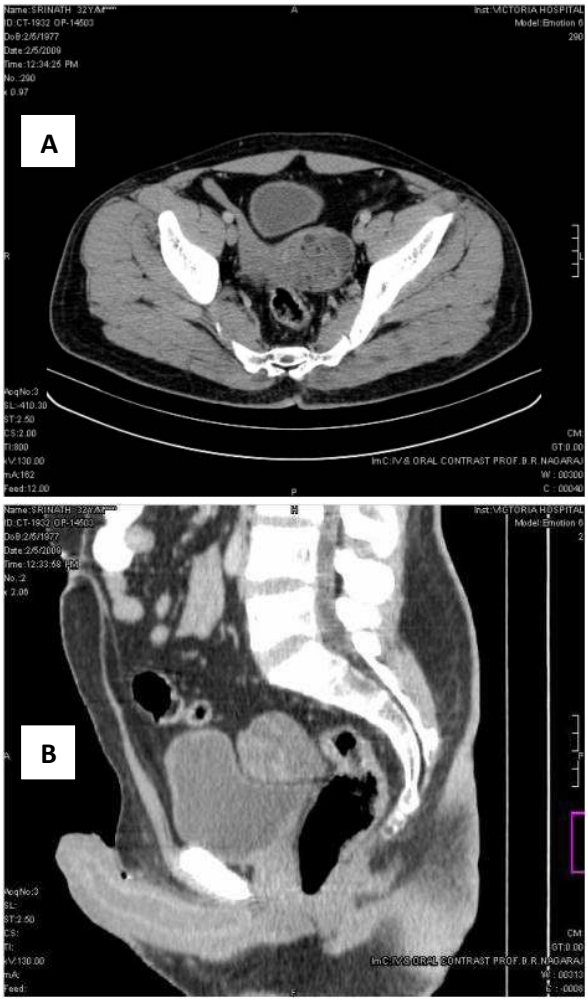


Histopathological examination revealed uterus with normal myometrium, cervix and endometrium. Left adnexa showed presence of ovotestis with mature teratoma, tubes and epididymis. Right adnexa showed presence of atrophic testes with tubes and epididymis.

Karyotyping revealed 46 XY, genotype and peripheral blood smear study showed that 10% of the peripheral blood neutrophils had barr bodies.

Post-operative period was uneventful. Patient was referred to endocrinologist for testosterone replacement. Patient is on regular follow up.

Photo – 3A, 3B: Malignant change in left undescended testis with (?) uterus.



Discussion

True hermaphrodites are individuals who have both testicular and ovarian tissue (with germ cells) which may take the form of one ovary and one testis or, more commonly, one or two ovotestes. The most common variant found in a true hermaphrodite is an ovotestis, with 50% being found in ovarian position on the right side. Ovaries are present in 33% of cases while testicles are found in 22% [3]. The most common combination is ovotestis-ovary, followed by bilateral ovotestis [2].

Both the external genitalia and internal duct structures of true hermaphrodites display

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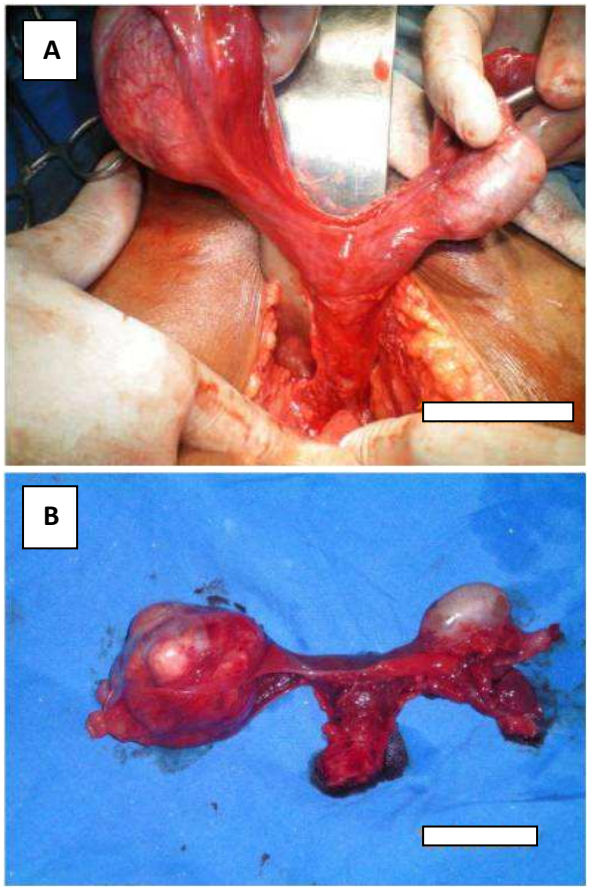
gradations between male and female and is related to the function of the ipsilateral gonad. In most patients, the external genitalia are ambiguous but masculinized to variable degrees and 75% are raised as male. Virtually all patients have a urogenital sinus, and in most cases a uterus is present [4].

Photo – 4A, 4B: Uterus with bilateral fallopian tubes with left vas deferens with Left side mass (? ovarian/ testicular tumor) and Right (? ovary/ testis).



The descent and position of the gonad depend on the amount of testicular tissue present [5]. 50% of the ovotestes are found in an abdominal position, while 25% are positioned in the inguinal region and the other 25% are labioscrotal in position. 85% of ovaries are found in the abdomen and 50% of the testes are labioscrotal [5].

Approximately 60% of true hermaphrodites have a 46, XX karyotype; 33% are mosaics with a second cell line containing a Y chromosome (46, XX/ 46, XY; 46, XX/ 46, XXY), and 7% are 46, XY [6].

The ovarian portion of the ovotestis is frequently normal; whereas the testicular portion is typically dysgenetic. Fertility potential does exist in true hermaphrodites. Phenotypic male true hermaphrodites, however, seldom produce adequate quality sperm to be considered fertile. While ovulation is not uncommon, spermatogenesis has been reported in only 12% of cases of true hermaphroditism [7].

The risk of malignancy ranges from 2.6% to 4.6%, although in true hermaphrodites it is lower than in other types of DSD [8]. Since the chance of malignancy is low, prophylactic removal of the gonad is not indicated [9]. The most common neoplasm is a Germ cell tumour, with dysgerminoma being the most common histological type [8].

There is an increased incidence of seminomas, gonadoblastomas, and teratomas in male hermaphrodites. There is 1.9 to 2.6 percent risk of malignant degeneration in true hermaphroditism, usually occurs in phenotype males and exclusively in mosaics and the risk may reflect the ectopic location of the gonads in these individuals [10, 11].

Conclusion

Our case illustrates the need for early identification and treatment. Ideally, diagnosis and management of patients should begin at birth to avoid gender identity disturbances. Late management should consider both the risk of malignancy and the potential for fertility in affected individuals. Complete investigations

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including chromosomal studies should be performed in all patients with true hermaphroditism. Psychological counseling, cosmetic, and extirpative surgery all play important roles in patients initially diagnosed in adult life.

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