A case report of vanishing testicle: radiological diagnosis and short review

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Abstract
Background: Vanishing testicular syndrome is also known as testicular regression syndrome (TRS) is due to atrophy and disappearance of testis in the fetal life after the formation of the normal testis. It is a spectrum of disorders; clinical features are depending upon the stage of fetal or early neonatal life at which function of testicles ceases.

Case presentation: Young 40-year-old male patient came for a routine master health checkup. On clinical examination he had a nonpalpable left testis, Rest of the clinical examinations are unremarkable. Referred to ultrasound (USG), for testis localization reveals the absence of left testis in the left scrotal sac and inguinal canal. Further MRI of the scrotum reveals the feature of TRS.

Conclusion: TRS in the 4th-decade adult is rarely reported in the literature. The asymptomatic presentation shows the least possibility of neoplasm in TRS patients.

Keywords: Vanishing testis, USG, Cryptorchidism, Testicular regression syndrome

Background
Vanishing testicular syndrome is also known as testicular regression syndrome (TRS) is due to atrophy and disappearance of unilateral testis in the fetal life after the formation of the normal testis. It is a spectrum of disorders; clinical features are depending upon the stage of fetal or early neonatal life at which function of testicles ceases.

Case presentation
Young 40-year-old male patient came to the hospital for a routine master health checkup. No significant past surgical or medical history. He is married 10 years before and had two children. His vital parameters were stable. Examinations of the respiratory, cardiovascular system, central nervous system are within normal limits. The abdomen was soft with no organomegaly. Local examination of the scrotum shows nonpalpable left testis. The right testis appears normal. The patient was referred to USG, USG shows normal right testis with normal echoes and vascularity (Fig. 1). The left scrotal sac is empty. The left testis is not visualized in the left scrotal sac and the left inguinal canal. The patient was further subjected to MRI to find the left testis. MRI shows a normal appearance of the right testis. Left spermatic cord structures are noted within the left inguinal canal. A small soft tissue nodule was noted in the tip of the left spermatic cord (Figs. 2, 3, 4, 5).

Left testis is not separately visualized in the left scrotum, inguinal canal, or pelvis. Features are suggestive of left TRS. The patient is not willing for surgical exploration and was advised to have a follow-up.

Discussion
Vanishing testicular syndrome is also known as testicular regression syndrome (TRS) is due to atrophy and disappearance of unilateral testis in the fetal life.
after the formation of the normal testis [1]. The presence of a blind-ending spermatic cord is a piece of evidence that testis is formed during intrauterine life [2]. Young phenotypically normal male absence of unilateral testis is due to the consequence of perinatal or intrauterine testicular torsion [1]. It is a spectrum of disorders; clinical features are depending upon the stage of fetal or early neonatal life at which function of testicles ceases [3]. TRS is more common than testicular agenesis in the case of the non-palpable testis [3]. The usual protocol to followed in case of nonpalpable testis is given in the below flowchart.
TRS is seen in 5% of the cases of cryptorchidism [4]. Among 10–20% nonpalpable testis in cryptorchidism, about 35–60% cases are TRS [1]. There may complete absence of testis, vas deferens, and epididymis (22.5%), blindly-ending vas deferens (15%), or blindly-ending vas deferens with blood vessels in the inguinal canal (67.5%) [1].

The main postulated pathophysiology is due to late antenatal or perinatal torsion, vascular occlusion due to thrombosis, or endocrine disorder [5]. Incompletely undescended testis is more prone to torsion during the perinatal and fetal period than they normally descended testis [1]. The presence of hemosiderin-laden macrophages in the testis had supported the vascular insult as the cause of TRS [6, 7]. The left side of the testis is more prone because of early descent of the left compared to the right [3, 6]. Cryptorchidism is more common on the right, but Honore and Smith et al. found that the left testis was more susceptible to regression, and then with 68% of cases being left-sided [8]. It may be an anatomical arrangement of the left spermatic vein, draining into the left renal vein, which may predispose to kinking, due to an unusually mobile left kidney. As there is no venous anastomosis across the midline until after 16 weeks, leading to more common left testicular TRS.

It has also been reported with Y chromosome microdeletion [9]. Although most of the cases are sporadic in occurrence few cases also had been reported in the members of the same family [10, 11]. Although TRS is usually unilateral, it can also be bilateral rarely associated with normal external genitalia (R1). The clinical presentation is classified based upon the age of presentation from early embryonic, late embryonic, early fetal, mid fetal, late fetal, and early neonatal [6]. Late presentation is the adult in the 4th decade is rarely described in the literature. Embryonic TRS is part of the partial (46XY) gonadal dysgenesis. Patients will exhibit the features of intersex or severe micropenis or even phenotypical females. The degree of masculinization of external and internal genitalia depends upon the duration of testicular function before regression. Missense mutation of the SF1 gene (V355M) is also reported recently in a boy with micropenis and TRS.

Intrauterine testicular descent occurs in two-phase, first during the 8–15 weeks and second during the 25–35 weeks. Various hormones are involved in the testicular descent are Human chorionic gonadotrophin, testosterone, and the protein hormone Insulin-Like 3 (INSL3).

TRS clinical diagnosis is based upon the clinically nonpalpable testis with blind-ending spermatic cord within the internal inguinal ring or in the retroperitoneum. The small fibrotic nodule is seen at the spermatic cord end which shows areas of fibrosis, dystrophic calcification hemosiderin deposition. Nodule shows viable germ cell or seminiferous tubule in 0–16% of the excised remnant [12].
The presence of germ cells can lead to the development of germ cell neoplasia in certain cases, but the absence of testicular tissue in many cases counterfeit the statement of germ cell neoplasia in the testicular remnant. Gross descriptive pathological features of TRS are several cm of spermatic cord with a small mass of firm, fibrotic tissue at one end. Elements of the vas deferens, spermatic artery, and venous plexuses are usually present [7].

Two essential criteria for TRS are (1) Visualizing the blind-ending spermatic vessels within the retro-peritoneum or visualizing spermatic vessels and vas deferens exiting a closed internal inguinal ring. (2) Testis is not palpated during examination under anesthesia.

Two different type of management exit, some urologist consider surgical exploration either via inguinal or scrotal approach with excision of testicular remnant, while other groups of surgeons consider conservative treatment [6].

The limitation of this case report is that, we had no surgical or pathological confirmation. Surgical exploration in case of TRS is a controversial issue.

Conclusion
What does this case report add to the existing knowledge?

TRS can even occur in adult upto 4th Decade.

Unilateral TRS in adult will not affect the sexual characteristics of the patient.

Germ Cell neoplasm is rare in TRS even in adults, although more common in cryptorchidiism.

Surgical exploration is not needed in all cases, especially in patients having normal physical and sexual characteristics.

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