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Vanishing Testes Syndrome: Report of Two Cases

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Abstract
Vanishing testes syndrome is a clinical condition found in phenotypic male with karyotype 46 XY in which testes is not palpable and it is believed that testes is lost after once it has formed. The manifestation of this disorder depends on the stage of fetal development at which testes is lost and also if this is unilateral or bilateral.

Introduction

Vanishing testes syndrome, also known as Testicular Regression Syndrome (TRS), is a condition which is considered to be due to subsequent atrophy and disappearance in fetal life of an initially normal testes(1). This absence of testes in otherwise 46XY male is assumed to be a consequence of intrauterine or perinatal torsion or infarction. We present here two cases of vanishing testes syndrome.

Case 1

23 year old phenotypic male came with underdeveloped sexual characteristics. His scrotum was not well developed, testes not palpable (since birth) and he had micropenis with single urogenital opening at tip of penis(SMR P2 ,SPL 3 cm) and B/L Gynaecomastia with disc size 4 cm . He was found to have hypergonadotropic hypogonadism with karyotype 46 XY. On MRI, a small hypointense lesion was found in left inguinal region. The tissue was surgically removed. Histopathological examination of the tissue was marked by presence of vascularised fibrous nodule (VFN) in proximity of spermatic cord. Vascularised fibrous nodule is hallmark of Testicular regression syndrome (TRS). There was no feature of malignanacy in the tissue sample. Patient is on testosterone replacement therapy and his present SMR is P5, with SPL 10 cm.

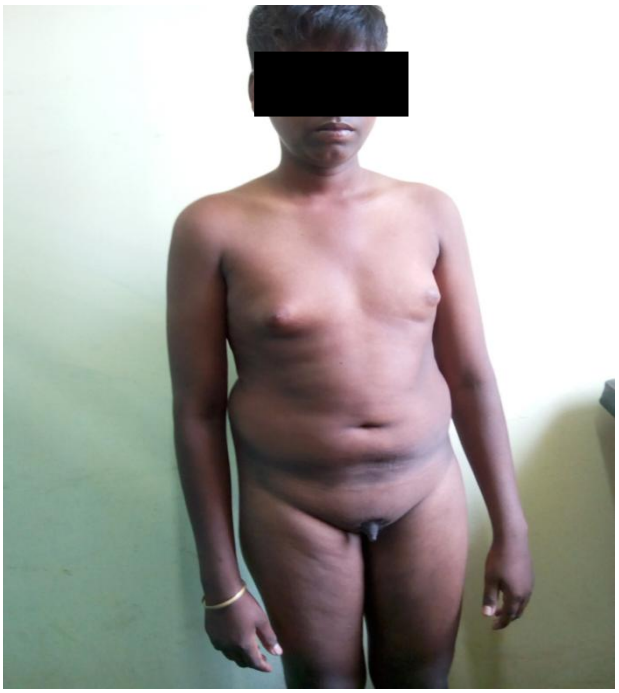

Fig.1 b/l gynaecomastia, micropenis

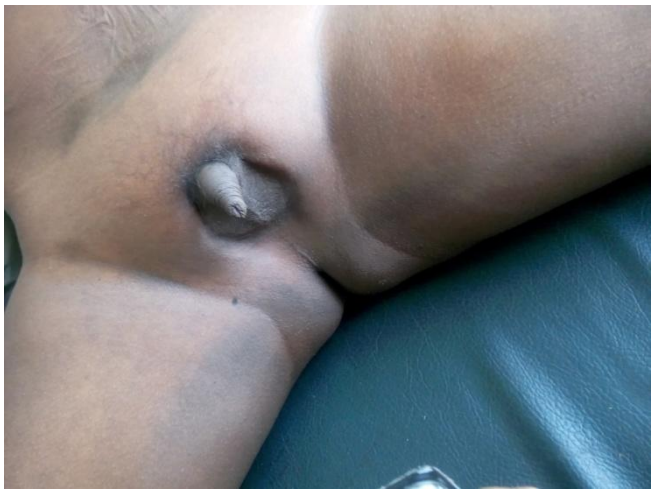

Fig 2 Absent testes in scrotum in phenotypic male

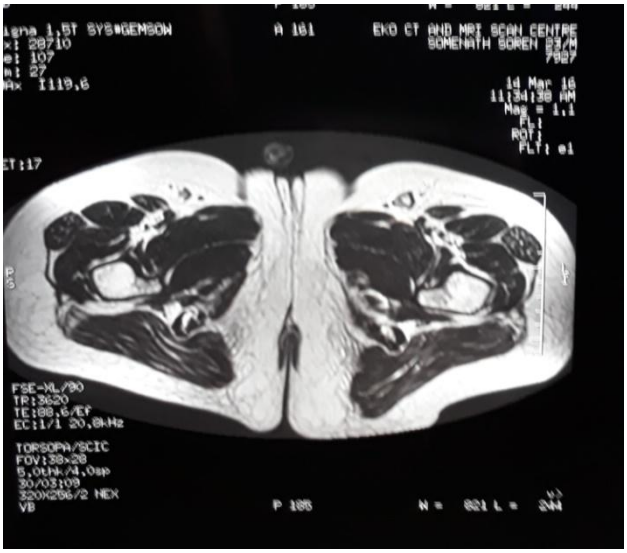

Fig 3.hypointense lesion in left inguinal region

Case 2

25 year old male comes with complain of unilateral testes since birth. His SMR was P5, SPL11 cm, scrotum well developed, right testes not palpable, left testes volume 25 ml ,no gynaecomastia. His testosterone levels were normal. MRI revealed testicular like tissue on right inguinal region which on biopsy revealed presence of vasularised fibrous nodule and hence diagnosis of unilateral TRS was considered. There was no feature of malignancy. As his sexual development were adequate and serum testosterone levels were normal, he did not require testosterone replacement therapy and was reassured of the condition.

Fig 4.single testes in scrotum in well developed male

Discussion

These two cases highlight various manifestation of vanishing testes syndrome. This syndrome is actually a spectrum of disorder with maximum and minimum variations which highly depend on the fetal age when the testicular regression occurs. If the loss of testicular function occurs before the 8th week of fetal development there is female differentiation of the internal and external sex organs. However this is a very rare condition with vanishing testes syndrome. More commonly the testicular regression occurs after fetal male phenotype is already developed. So sex

assignment in these cases is not difficult and is straightforward. Unilateral and bilateral loss of testes are seen in this condition .So it is not uncommon to see many undiagnosed men with singular testes.

As for cause of this condition, it is widely accepted that this happens due to vascular accident and antenatal torsion. This is evidenced by presence of hemosiderin laden macrophages in the tissue removed surgically, which is suggestive of venous congestion and hemorrhagic infarction secondary to torsion of a structure. Other causes suggested are trauma , endocrinopathy and genetic factors[(2)]. However most cases are sporadic and there is no similar family history. Causes of nonpalpable testes can be cryptorchidism, retractile testes, testicular agenesis or testicular regression syndrome (TRS). In case of TRS, spermatic cord ends blindly and a small fibrotic nodule may be seen. In histology, calcification and hemosiderin is identified. Viable germ cells have been reported in 0 to 16% of reported series[(3,4,5,6,7)]. However malignant potential of testicular remnants in this syndrome is not well established.

In our case 1, patient had male phenotype with micropenis and absence of palpable testes, meaning to suggest that sexual development in utero till week 14 was normal male type . As this requires paracrine action of testosterone secretions from testes, it can be assumed that testes were normally present and functioning till atleast week 14. However patient by history had small penis since birth. Beginning of penile growth in utero starts at age of 24 weeks[(8)] and so it can be assumed that patients pathology occurred bilaterally during that period of in utero life. Patient presented to us at age of 25 years and was found to have hypergonadotropic hypogonadism with karyotype 46 XY. Patient was treated with testosterone replacement therapy which resulted in increase in length and circumference of penis along with muscular development and improved masculine psyche.

In case 2, scrotum had singular testes. As no mullerian duct were found inside, it can be assumed that the patient pathology occurred only after 14 weeks. With a single normal sized testes and normal serum testosterone levels , his sexual maturity was normal to his age and hence did not require testosterone replacement.

Conclusion

Testicular regression syndrome (TRS) or vanishing testes syndrome is not an uncommon disorder. It should form important differential diagnosis of cryptorchidism, specially if testes is not palpable even in inguinal region

Malignancy potential of remnant tissue of TRS is not well established.

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