

# Seedless orchids: Issues in the anorchid adult

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### ABSTRACT

Disorders of sexual differentiation (DSD) are among the challenging problems in the field of endocrinology. When presenting late in an adult the therapeutic as well as diagnostic issues may be different and difficult. Bilateral anorchia is rare. The desire of future parenting is a real challenge when such patients ask for their own biological generations. This case depicts the late presentation of DSD in an anorchid adult and the management issues.

**Key words:** Adult, anorchia, bilateral, differentiation, 46, XY gonadal dysgenesis

Adult disorders of sexual differentiation (DSD) are on the rise due to the diagnostic advances in laboratory and imaging sciences. Thus, presenting with not only the increase in number of cases but also certain diagnostic as well as therapeutic issues. Future parenting is one of the most desirable wishes of a person and may be the most difficult in adult cases of DSD with infertility. Although advanced scientific options of assisted reproductive techniques are available including surrogacy in few cases, these patients undergo a huge mental toll to their emotions and needs to be tackled gently and empathically. One such problem is when a person does not have or feel the presence of testes in the scrotum, the absent testes, or the vanishing testes syndrome (VTS). This case highlights the diagnostic and therapeutic difficulties faced in a late presentation of an adult anorchid patient.

The causes of non-palpable gonads (testes) in a male child are anorchia, inguinal testes, intra-abdominal testes. Absence of testes in a 46, XY individual with a male phenotype (TRS) or “vanishing testis syndrome” occurs when because of the subsequent atrophy and disappearance in fetal life of an initially present and identified normal testis.

It may occur in 1/20 000 male births, and in approximately 1/177 cases of cryptorchidism.<sup>[1]</sup> It may be impalpable in less than 5% of cryptorchid cases.

### CASE REPORT

A 28-year-old male patient comes with the chief complaints of wishes to get married, worried about his semen. On enquiry, no features suggestive of secondary sexual characteristics (voice cracking, facial hair, body hair). No suggestion of history of atypical genitalia at birth, testicular torsion, trauma, chemotherapy, radiotherapy, infection, or inflammation. No history suggestive of hypospadias. Not associated with any congenital anomaly. Born out of a non-consanguineous marriage, patient was declared a male child at birth. The age of impalpable testes is not known to patient or his family. He has two elder brothers both married and have fathered children, one younger brother, and sister both completed puberty normally. There is no history of atypical genitalia or infertility in the family.

He has poor erections, heterosexual in orientation, but no seminal emissions, which is his prime concern.

This person wants to marry, wishes to lead a normal life.

### EXAMINATION

On examination there is a tall, obese person, body mass index (BMI) of 33 kg/m<sup>2</sup>. Pulse rate 84 beats/min, blood pressure 118/70, 106/70 mm of Hg supine, and standing.

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There was no goiter, but he had acanthosis nigricans. He had poor development of secondary sexual characteristics, no facial hair, scanty axillary hair, bilateral gynecomastia B5, pubic hair present P3, stretched penile length 6 cm, poor girth, scrotum empty bilaterally, rugosity present. No evidence of varicosity or parotid enlargement. He had findings normal on remaining general and full systemic examination.

## INVESTIGATIONS

Laboratory investigations reveal Hb 12.5 g%, serum cholesterol 190 mg/dL, triglyceride 140 mg/dL, high density lipoprotein 40 mg/dL, low density lipoprotein 119 mg/dL, FSH 40 (2-10 iu/mL), LH 28 (3-9 iu/mL), 8 a.m. Testosterone 1.9 ng/mL, SHBG 80, prolactin 10 ng/mL, T3 140, T4 8.9, TSH 1.7 mIU/mL.

8 a.m. Cortisol 20 mcg/dL, FPG 99 mg/dL, PPG 110 mg/dL, SGOT 28, SGPT 30, total bilirubin 0.7, direct bilirubin 0.3, indirect bilirubin 0.4, KARYOTYPE: 46, XY

### Imaging

Ultrasonography: Empty scrotal sacs bilaterally. MRI abdomen and pelvis: *No gonads seen in inguinal/abdomen or pelvic region*

## DIAGNOSTIC DILEMMA

VTS or TRS or embryonic testicular regression (ETR) or anorchia is, or there some another diagnosis possible?

DSD are rare endocrine disorders, where in vanishing testes are only about 9% in one series of DSD of 95 patients.<sup>[2]</sup>

In cases of a newborn baby apparently looking like male with bilateral absent gonad, should not be labeled as simple cryptorchidism. In fact they should be suspected for virilized congenital hyperplasia. However, in the absence of hypoglycemia and presence of normal morphology and size of penis, anorchia is more likely than adrenal etiology.

Microphallus may be present in around 50% of cases of bilateral anorchia, as reported in a recent study, suggesting functionally abnormal gonads before disappearance. While others have a normally developed phallus.<sup>[3]</sup> About 10% of patients have torsion, but the precise mechanism of testicular torsion or vascular insult remains unknown. The testicular infarction may cause hemosiderin deposits and dystrophic calcification, as reported in one study.<sup>[4]</sup>

Certain cases of bilateral anorchia have other members in the family affected. These family members may have

anorchia with genital ambiguity with a 46, XY karyotype and are part of the spectrum of 46, XY gonadal dysgenesis. SF1 (steroidogenic factor 1, also called as NR5A1) mutation are present in the patients with bilateral anorchia and is also the first monogenic cause of bilateral anorchia in humans. There is a mild partial loss of SF1 associated with micropenis and cryptorchidism, suggesting dosage sensitive and domain specific changes can lead in to the spectrum of male reproductive phenotype.<sup>[5]</sup>

Recent times have led us to discover a new term called as acquired cryptorchidism. Here testes have been present at birth but at a later time regresses up reflecting primary failure of total or complete descent.<sup>[6]</sup>

Additionally, in the laboratory serum AMH (anti-mullerian hormone) is proven to be a good biological marker for anorchia along with serum inhibin B. In comparison to 57% predictive value of low testosterone response to human chorionic gonadotropin (HCG) injections, undetectable AMH may have a prediction value of around 92%. Also, Kubini *et al.* reported beneficial values of inhibin B in diagnosis of bilateral impalpable testes. Both these studies had their patients in pre- or peri-pubertal age group. Injection with HCG may not be done if serum AMH and inhibin B are available. Furthermore, serum AMH, and inhibin B may not be available at all places or affordable to all for making a diagnosis.

## THERAPEUTIC DILEMMA

Detecting pre-pubertal, the management remains to induce virilization by injection testosterone. However, very rarely such patients like ours present late with VTS or 46, XY gonadal dysgenesis. We can induce virilization by testosterone injection, but patient is worried about semen. How can we achieve that?

Although infertile, but depending on when they present management may differ. This patient's main concern was to have appropriate seminal emissions. With testosterone injections he can have virilizations and ejaculatory fluid from prostate gland and seminal vesicle. Testicular prosthesis was refused by this patient. He will need extensive counseling regarding parenting issues as the ejaculate will be aspermic in bilateral anorchia. He will also need along with his immediate family and future partner, counseling, and family therapy.<sup>[7]</sup> Sperm donation is generally the option for such patients who need children apart from adopting a child.

## SUMMARY

VTS or anorchia is rare and is included in the spectrum

of 46, XY gonadal dysgenesis. Diagnosed early, this can be managed efficiently from hypogonadism point of view. But when they present late in life, management issues are different. In addition to virilization, fertility, and parenting queries stand prominent in dealing with an anorchid adult. It is a team work of patient, his relatives, assisted reproductive technique, endocrinologist, and the psychiatrist, to tackle all related concerns.

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