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CASE STUDY

A RARE CASE OF DSD DIAGNOSED AS SWYER SYNDROME

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ABSTRACT

Introduction: Simple 46, XY gonadal dysgenesis syndrome, also called Swyer syndrome, is known as pure gonadal dysgenesis. Individuals with the syndrome are characterized by 46, XY karyotype and phenotypically female with female genital appearance, normal mullerian structures and absent testicular tissue. The condition usually first becomes apparent in adolescence with delayed puberty and primary amenorrhea due to the gonads have no hormonal or reproductive potential.

Case report: A23 years old Indian female with normal female genitalia presented with arrested secondary sexual character (arrested breast development) & absence of regular menstrual cycle. Chromosomal Analysis revealed- 46XY karyotype. So it is a case of 46XY DSD with female external genitalia. She failed to achieve secondary sexual characteristics until five years back when she was treated with OCP &subsequently developed scanty pubic hair along with painful breast enlargement. She had periodic cyclical bleeding which stopped after discontinuing OCP. Further investigation for primary amenorrhea & laparoscopy showed rudimentary uterus & B/L streak gonads. She underwent prophylactic gonadectomy with H/P report showing loose textured fibrous tissue on right side & fibro collagenous tissue with epididymal duct on left side, no palpable gonads in labia majora or in the inguinal region & presence of uterus, female range testosterone & undetectable AMH which excludes CAIS (complete androgen insensitivity syndrome). Our case also didn't have ovarian tissue, thus, the differential diagnosis was made based on true hermaphroditism/ovotesticular DSD can be ruled out.

Conclusion: This patient has bilateral streak gonads that do not secret sex steroids or AMH. Hence serum AMH is undetectable, and rogens (of adrenal origin) are low and do not respond to HCG stimulation and gonadotropin levels are elevated. Consequently, this patient has female external & internal genitalia. All the above mentioned facts led to the diagnosis of Complete Gonadal Dysgenesis or Swyer Syndrome.

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INTRODUCTION

Simple 46, XY gonadal dysgenesis syndrome, also called Swyer syndrome, is known as pure gonadal dysgenesis. Individuals with the syndrome are characterized by 46, XY karyotype and phenotypically female with female genital appearance, normal Mullerian structures and absent testicular tissue. The condition usually first becomes apparent in adolescence with delayed puberty and primary amenorrhea due to the gonads have no hormonal or reproductive potential.The incidence of Swyer syndrome reported in literature is1:100,000. (Coutin *et al*., 1996; Behtash *et al*., 2007) Herein, we report a 23-year-old patient with Swyer syndrome. Doctor Swyer described two women whom had a 46, XY karyotype, tall stature, primary amenorrhea, female external genitalia, normal vagina (albeit hypoestrogenised) and cervix in 1955 (Swyer, 1955). Individuals with Swyer syndrome are phenotypically female with unambiguously female genital appearance from birth and normal Mullerian structures. The patients usually first become apparent in adolescence with delayed puberty and primary amenorrhea due to the fact that the gonads have no hormonal or reproductive potential. Here, we report a case of Swyer syndrome.

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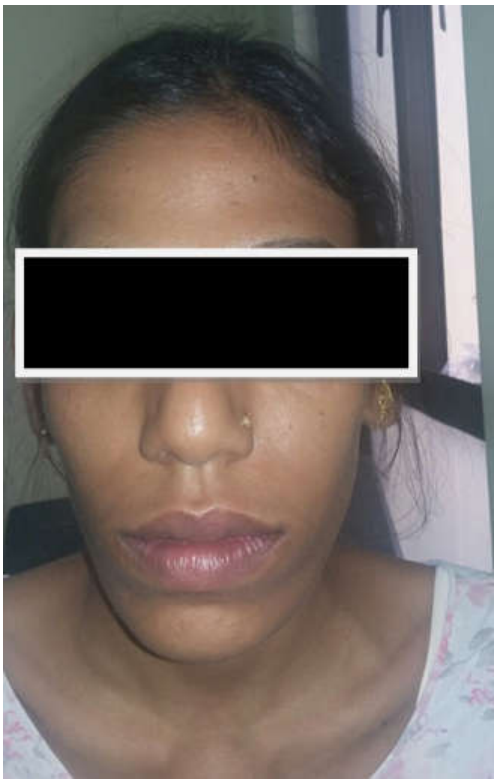
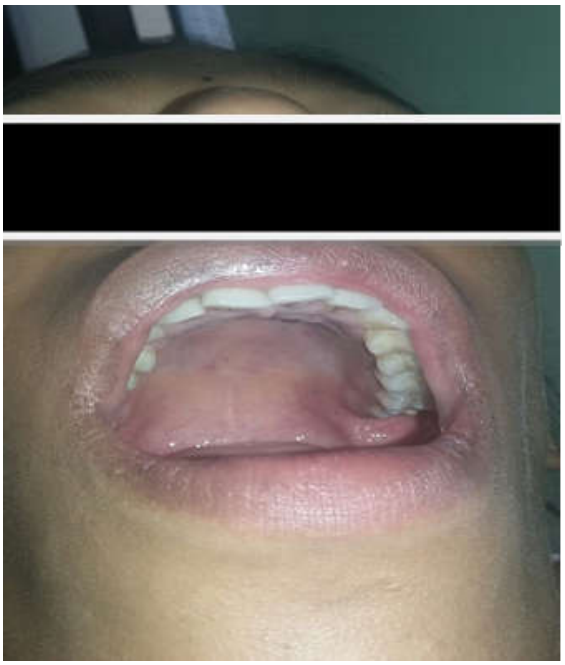
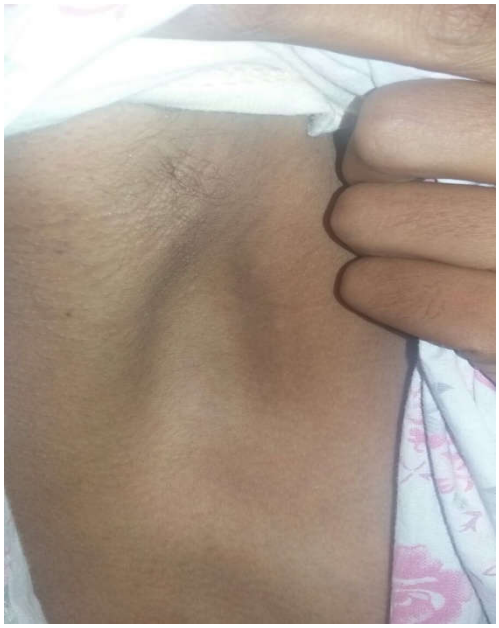
Case Report

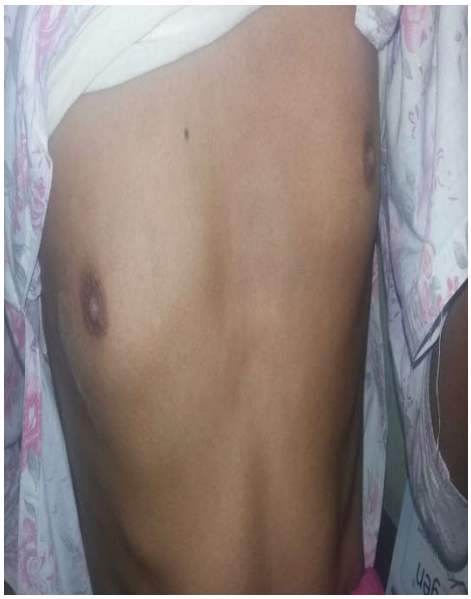
A 23 year old female presented with arrested secondary sexual character & absence of regular menstrual cycle. Patient is born of non-consanguinous marriage, normal delivery without any significant antenatal or postnatal events. Normal developmental milestone followed by normal growth patterns & normal height gain as compared to her peers. No h/o childhood hospitalization or crisis. She failed to achieve

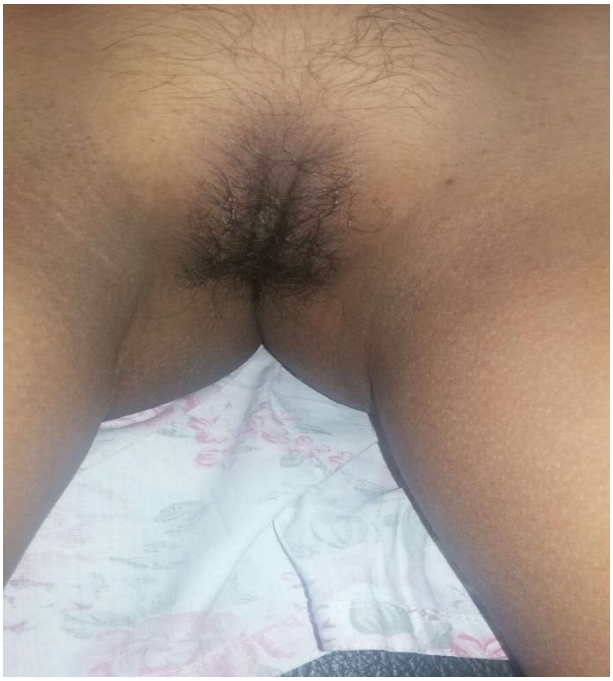
secondary sexual charecterstics until 6 years back when she was treated with OCP & she developed scanty pubic hair, painful breast enlargement. She had periodic cyclical bleeding. She stopped OCP for last 4 years after which there is no cyclical bleeding. 6 years back she underwent investigation for primary amenorrhoea & laparoscopy showed rudimentary uterus & B/L streak gonads and she underwent prophylactic gonadectomy (21.04.2011) with H/P report showing loose textured fibrous tissue on right side & fibrocollagenous tissue with epididymal duct on left side. There is no h/o asthenia, mental retardation, anosmia or hyposmia, synkinesia, headache, visual disturbance, convulsion. No family h/o similar case of amenorrhea or infertility.

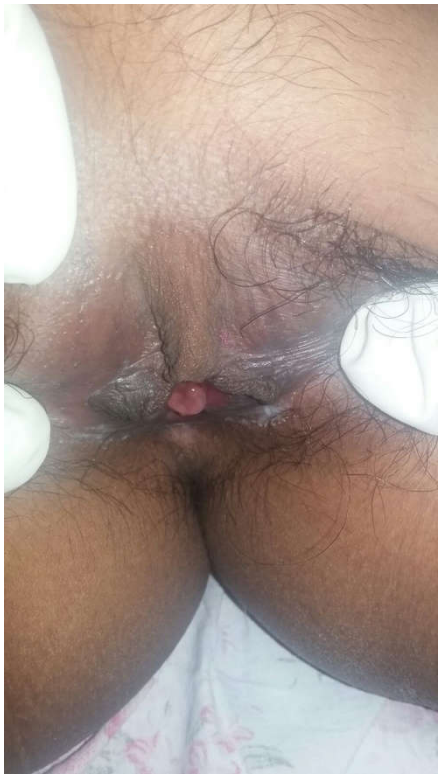












On examination

Ht.-168.9 cm (>97th %tile), Ht.SDS +2.3
Arm span 180.6 cm,
US-78.7 cm, LS-90.2 cm, US/LS-0.87
Scoliosis –Neg, Marfanoid habitus present, high arched palate.
SMR-B2, P3, Scanty axillary hair, external genitalia female with normal clitoris & hymenal orifice, poorly developed labia minora. No palpable gonads in labia majora or in inguinal region.
Pulse-84/min, regular, equally palpable in all limbs, no radio-radial or radio-femoral delay.BP-110/70(supine) & 108/70 mm Hg(standing). Other system examination unremarkable.

Investigations

Chromosomal Analysis revealed- 46XY karyotype.
USG- Uterine size 4.6 cm*1.2 cm* 2.4cm
Uterine volume-7.15cc
No obvious gonad like structure noted in pelvis
Echocardiography-Normal study
LH-55.23 U/l, FSH-138.36 U/L
Estradiol-<10 pg/ml,
17 hydroxy progesterone-0.6ng/ml
AMH-0.2 ng/ml
Testosterone-0.83nmol/L(22.41 ng/dl)
Post hCG stimulation test (1500 IU hCG IM on three consecutive days, testosterone measured 72 hours after last injection)-1.02 nmol/L(27.54 ng/dl)
Hb-12.2g/dl,MCV-93fl,TLC-4400,Bilirubin-0.9,SGPT-55,SGOT-36,Ca-8.5,PO_4-3.7,Na-144,K-3.9, TG-95, Chol-144.
PD-Pure gonadal dysgenesis

DISCUSSION

This patient presented with normal female genitalia, primary amenorrhea, arrested breast development & on karryotyping it is 46XY. So it is a case of 46XY DSD with female external genitalia. No gonads are palpable in labia majora or in the inguinal region & presence of uterus, female range testosterone & undectable AMH excludes CAIS(complete androgen insensitivity syndrome. CAIS is a/w male range testosterone & AMH& normal breast development. Leydig cell aplasia or severe steroidogenic defects at the level of StAR or P450scc are a/w severe hypoandrogenism, female range testosterone, male range AMH, low Na^+, high K^+. In true hermaphroditism, gonads containing both ovarian and testicular tissues (ovotestis) are observed (Jones *et al*., 1965; Kim *et al*., 2002), usually have XX karyotype or may show mosaicism, such as XX/XY, XX/XO or XY/XO. Our case did not have ovarian tissue, thus, the differential diagnosis was made based on true hermaphroditism/ ovotesticular DSD can be ruled out. This patient have bilateral streak gonads that do not secret sex steroids or AMH. So serum AMH is undectable, androgens (of adrenal origin) are low and do not respond to hCG stimulation and gonadotrophin levels are elevated. Consequently, these patient have female external & internal genitalia. So the patient is diagnosed as Complete Gonadal Dysgenesis or Swyer Syndrome.

Conclusion

Swyer syndrome is caused by an error in sex determination during the course of embryogenesis. Patients with Swyer syndrome present with an incomplete masculinization due to deficiencies in the production of testosterone and Müllerian-inhibiting factors that result in the failure of gonadal progression (Freitas *et al*., 2001). The streak gonads don't produce androgen or AMH. As testosterone is absent wollfian duct fails to develop internal genitalia. As AMH is absent, the mullerian ducts develop into uterus, fallopian tube, cervix and vagina. Uterus is usually present and is hypoplastic.Molecular and genetic abnormalities associated with this condition include mutations in the ARX, ATRX, CBX2, DHH, DMRT1, GATA4, MAMLD1, MAP3K1, NR0B1 (which relates to DAX1 expression and congenital adrenal hypoplasia), NR5A1 (which encodes steroidogenic factor 1), SOX9, WNT4, WT1, WWOX, SRY, and WNT4 genes. The SRY gene is deleted in approximately 10-15% of patients with Swyer syndrome and mutated in an additional 10-15% of Swyer syndrome patients (King and Conway, 2014; Lipay *et al*., 2005). Individuals with Swyer syndrome exhibit female phenotypes and are typically raised as girls; these individuals are generally diagnosed in adolescence when they seek medical assistance for amenorrhea and the absence of secondary sex characteristics. Patients with Swyer syndrome should be subjected to surgery for gonad removal as soon as the diagnosis has been established because the gonads are at high risk for gonadal tumors, which are typically gonadoblastomas and/or dysgerminomas (Freitas *et al*., 2001; Lipay *et al*., 2005). These patients can get married, have normal sexual life and can get pregnant through in-vitro fertilization with donor oocytes if desired. In fact, several cases of pregnancy among Swyer syndrome patients have been described since 1988; the prognoses for these pregnancies is similar to the prognoses for the pregnancies of 46,XX patients with ovarian failure (Plante and Fritz, 2008).

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