Case Report

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Young Woman with 46 XY and a Hypoplasticuterus: Swyer Syndrome

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Abstract

Women presenting with primary amenorrhoea require extensive work up. Swyer syndrome or pure 46XY gonadal dysgenesis is a condition with external female phenotype, but fail to attain menarche at the usual age with sexual infantilism. Person with this disorder have female external genitalia but the uterus and fallopian tubes are present, though underdeveloped. They lack functional gonads (ovaries or testes), instead, they have streak gonads. We are reporting a case of young female presenting as primary amenorrhoea, detailed work up revealed it to be Swyer syndrome with partially developed breasts, hypoplastic uterus, and absent streak gonads. These cases are very rare, and require early gonadectomy. This case illustrates a rare case of gonadal dysgenesis and demonstrates the importance of counselling on the options of treatment, especially regarding treatment of dysgenetic gonads, need for replacement therapy and future fertility options.

Keywords: Swyer Syndrome; Pure XY Gonadal Dysgenesis; Gonadectomy.

Introduction

Primmary amenorrhoea is a perplexing problem to the gynaecologist due to wide variety of aetiological factors, requiring extensive and systemic workup to reach at the most appropriate diagnosis and formulate a plan of management of these young patients. It is taxing for the patient and their families too as often they find it difficult to accept the diagnosis. Often medical/surgical treatment go hand in hand to psychological help in form of counselling, medications and much more. Swyer's syndrome is a rare entity, incidence being 1:80,000. Swyer's syndrome was first described in 1955 by Jim Swyer. These individuals are usually raised as a female due to female external genitalia. They also have female internal genitalia, but with the XY karyotype. The gonads are usually replaced and represented by fibrous streaks. These patients presents at the time of adolescence when they fail to attain menarche & lack of secondary sexual characters. A high incidence of tumours such as gonadoblastoma and germ cell malignancies have been reported in these streak gonads.

Case Report

A 25 years young married female came to gynecology OPD with primary amenorrhoea. She was married for last 4 years and reported no sexual difficulty. Personal history, Family history and past medical history did not reveal any abnormality. On General physical Examination, she was of average height and weight with BP of 110/70 mmHg. Facial appearance was normal, thyroid was not enlarged. Breast was Tanners stage 2, axillary and pubic hairs were absent. Per abdomen examination was normal. Local external genital revealed normal monspubis, labia majora and minora, though pubic hairs were

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absent. Per speculum examination revealed a normal length vagina, very small cervix. Per vaginal examination revealed a tiny uterus. She was further evaluated with hormonal profile and USG. USG showed a tiny 2x1 cm structure at the vault, between the bladder and rectum; probably a uterus, ovaries could not be identified on USG. No renal or any other abnormality was noted in USG. The same findings were confirmed by CT / MRI scan of abdomen. Her serum TSH & Prolactin was normal. Serum FSH was very high (110 IU/ml). She was advised karyotype and the report was 46XY.

With all these clinical and laboratory findings she was diagnosed as Swyer syndrome. Initially patient was shocked and required counselling, after a few sessions and reassurance she was ready for further treatment. She was counselled forneed for Gonadectomy and hormone replacement therapy. She underwent laproscopicgonadectomy (Fig. 1), histopathology of which revealed it to be streak gonads with fibrous tissue. She was put on conjugated equine estrogen 0.625 mg per day. Future fertility options were also discussed with her in the form of embryo donation or ovum donation followed by ICSI and embryo transfer after the uterus has grown in size. She still in follow up.

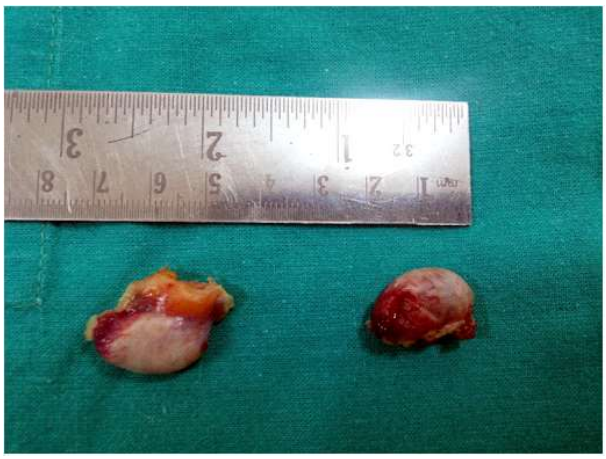

Fig. 1: Gonads of both sides after Gonadectomy

Discussion

Swyer syndrome affects individuals with XY chromosomal make-up; nevertheless they have a female appearance. The exact incidence of this condition is unknown. It has been estimated to occur in 1 in 80,000 births.

Sexual differentiation requires a normal XY fetus to develop into testes. From the second month of gestation early stages of testicular development requires action of several genes, most importantly SRY. Mutations in SRY accounts for most cases of this syndrome, Mutations in the NR5A1 and DHH genes are also known to be associated with this condition.

The gonads fail to develop as testes inspite of XY (genetically male). As testes do not differentiate and produce testosterone and Anti Mullerianhormone[AMH]. The external genitalia then fail to virilise, resulting in a female genitalia. Without AMH, the mullerian duct develops into female internal organs ie uterus and cervix, fallopian tube and vagina.

At birth this child will have female external genitalia will be reared as a female. As the child grows the streak gonads fail to produce hormones required for further development of secondary sexual characters. These individuals are usually detected at the time of adolescence, when they are investigated for primary amenorrhoea. Adrenal gland is not affected and can produce androgens and these persons can develop pubic hair, though it often remains sparse. These patient usually have a normal vagina, so coital function is possible without any problem. Early diagnosis is important for manyreasons: firstly, risk of gonadal malignancy; secondly, early institution of hormonal therapy is vital for the induction of puberty; thirdly, HRT is required to prevent osteoporosis.

In Swyer syndrome there is significant risk of development of malignancy ranging as high as 30%. The most common tumour involved in this condition is Gonadoblastoma,Dysgerminoma and Embryonal carcinoma have also reported. Due to the risk of tumours, search for the rudimentary gonads is needed and bilateral gonadectomy is advisable. The best time for gonadectomy is not well defined as the time needed for tumour formation is not known; probably as early as possible would be the recommendation as there has been case reports in very young children also.

Hormonal supplementation can induce menstruation in these patients. Successful pregnancies have been reported in these patients, Pregnancies were possible through oocyte donation and hormonal treatment. The presence of the XY genotype and the H-Y antigen does not affect the normal uterine and endometrial response.

The treatment of Swyer syndrome should be multi-disciplinary to provide multi-faceted care in terms prevention of malignancy and osteoporosis, induction of puberty, fertility, and psychological support.

How does this case report make a difference-

1. Creating awareness in all gynaecologist and general practitioner.
2. Need for early diagnosis and gonadectomy due to high risk of malignancy, which may be easily accomplished by minimal invasive surgery.
3. Need for psychological and medical support for induction of puberty and breast development, prevention of Osteoporosis and other long term problems.
4. Fertility options are available in spite of XY.

Conclusion

Early diagnosis and thorough workup of females coming with primary amenorrhoea is important. Multi-faceted approach for care of these women is important to address different problems. Early gonadectomy and hormonal therapy is indicated for good outcome. These patients can have normal sexual function and normal life expectancy after gonadectomy. Artificial Reproductive Techniques may help these women to conceive.

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