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Case Report

Swyer Syndrome in a Woman with Pure 46, XY Gonadal Dysgenesis, A Rare Disorder, Late Presentation: Case Report

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A 22-year-old nulliparous woman was consulted for primary amenorrhoea at the Gynaecological out-patient department of the tertiary academic hospital. The initial referral diagnosis was testicular feminization syndrome. On physical examination, the patient was phenotypically female (height: 187cm; weight: 82kg) with normal secondary sexual characteristics (pubic and axillary hair present, breast development, stage iv tunner system). Normal external genitalia with normal clitoris. On speculum examination normal vagina length, cervix appeared normal and pap smear was normal.

On bi-manual examination the uterus was found to be of small size and no adnexal masses were palpable. Investigations revealed the presence of elevated gonadotropins (FSH, LH) and low levels of estrogens. A Karyotype study reveals that she had 46, XY chromosome (male).

Vaginal ultrasound confirmed the clinical findings that the uterus is of normal shape but hypoplastic measuring 4.1 x 2 cm. The ovaries cannot be visualized.

The patient underwent diagnostic and operative laparoscopy where no ovaries were present; instead, fibrous tissue (streak gonads) was evident.

Bilateral gonadectomy and salpingectomy was performed and the histology confirmed the features of both gonads consistent with fibrous tissue (streak ovaries), and there was no neoplasia within the tissue submitted. The patient is on combined oral contraceptive (COC) with good evolution.

Discussion

Since the first description of Swyer syndrome in 1955, a number of cases have been reported. Pure Gonadal dysgenesis (or Swyer syndrome) is characterized by 46, XY karyotype in a phenotypically female patient. Despite having the XY chromosome, girls with Swyer syndrome look female and have functional female genitalia and structures, including a vagina, uterus and fallopian tubes [1].

Pathogenesis had been described during embryogenesis, where the early stages of genital development are similar in the male and female. Normal development in the female consists in the growth of Mullerian elements and atrophy of Wolffian ones. Moreover the gonad itself, in its early stage of genital development remains, for a short period of time, in a neutral stage (indifferent gonads) during which, it is capable of developing into a testis or an ovary. In Swyer syndrome the indifferent gonads fail to differentiate into testes in a XY (genetically male) fetus. In the absence of testes, no testosterone or anti-müllerian hormone (AMH) is produced. Without testosterone, the external genitalia fail to virilize, resulting in normal female genitalia. The Wolffian ducts fail to develop, so no internal male organs are present. Without AMH, the Mullerian ducts develop into normal internal female organs (uterus, fallopian tubes, cervix and vagina).

As in this case, having normal female external genitalia was considered to be normal except that she was having non-functional streak gonads instead of ovaries or testes. Before puberty (even in normal females) the ovaries play little or no role in bodily changes. The problem manifests at puberty due to an inability of the streak gonads to produce sex hormones (both estrogens and androgens). Most of the secondary sexual characteristics do not develop, as well as a lack of menses in the majority of phenotypically female patients with pure gonadal dysgenesis.

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In this case, the secondary sexual characteristics have been developed as pubic and axillary hair were present. This is attributed to the limited amount of androgen produced by adrenal glands which are not affected by this syndrome. The breasts were not fully developed, as the main source of estrogens was the peripheral aromatization of androgens. The main complaint of this patient was the primary amenorrhoea. Being sexually active for 3 years, primary infertility was also one of the concerns that were expressed at first consultation, as the patient was not on any form of contraception at the time.

Diagnosis was based on clinical examination. Elevated gonadotropins indicated that pituitary was normal but there was a failure of response from the gonads. The karyotype reveals 46 XY (male) chromosome. Vaginal ultrasound demonstrated the presence of the uterus but no ovaries. Laparoscopy demonstrated that the ovaries bilaterally have been replaced by fibrous tissue streak gonads, but also the presence of uterus and normal tubes (Figure 1).

The age of diagnosis is important in the management of Swyer syndrome as the gonads of XY pure gonadal dysgenesis have a high risk of gonadal malignancy such as a gonadoblastoma and germ cell tumor [2,3]. In the absence of an ovarian mass, tumour markers that are used to detect the presence of malignancies (AFP; CA125; bHCG) were not performed. Once diagnosed in the presenting patient, a laparoscopic bilateral gonadectomy and salpingectomy was performed (Figure 2). The histology confirmed the presence of bilateral streak gonads and no malignancy was found in the specimen examined. The post-operative evolution was uneventful. The diagnosis of Swyer syndrome is usually made around the time of puberty, when the child who has been reared as a female fails to achieve menarche. This case showed that the diagnosis was delayed, as a diagnosis was only confirmed at the age of 22. The average age at diagnosis in the series of Michala, et al. [4] was at the age of 17.2 years. A case of a delayed diagnosis of Swyer syndrome, 6 years after the initial consultation to the general practitioner, was reported previously by this institution [5].

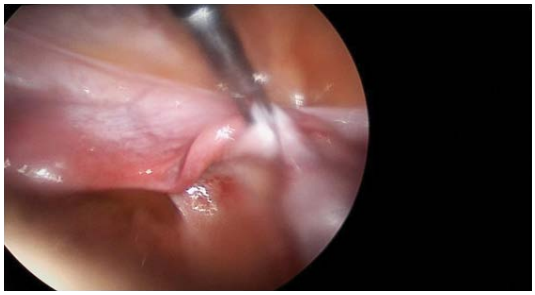

Figure 1: Laparoscopic findings of the uterus, tube and a streak gonad.

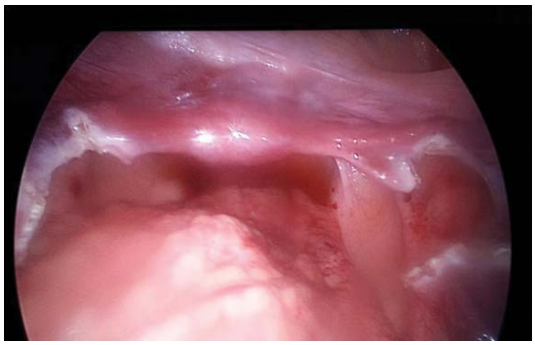

Figure 2: Bilateral gonadectomy and salpingectomy performed.

There is a necessity to increase scientific knowledge and awareness of the disorder among health professionals. Early diagnosis is important, not only because is a risk of gonadal malignancy, but also early institution of hormonal therapy is vital for the induction of puberty. Hormone Replacement Therapy (HRT) may be required to improve bone mineral density and to prevent osteoporosis.

Following removal of the gonads, COC therapy was promptly initiated. The estrogen-progestin sequential therapy supports female secondary sexual characteristics. The COC can lead not only to menstruation but also to an improvement of the uterine size and shape. Fertility can be achieved by using donor oocyte and literature has described successful pregnancies in patients with pure gonadal dysgenesis [4,6].

Upon diagnosis, the presenting patient was referred to a genetic counseling centre. This was done in order to evaluate the risk factors associated with the condition, whether the condition is sporadic or hereditary, as well as to determine whether a pattern of inheritance can be found (recessive or dominant). To do so, an assessment was performed on the immediate family members of the presenting patient (with emphasis placed on the mother).

Additionally, the patient received counseling and support regarding her diagnosis, complications, and outcomes.

Despite having the XY chromosomal makeup, girls with Swyer Syndrome are phenotypically female and have functional female genitalia and associated structures (vagina, uterus, fallopian tubes), as well as a normal sexual life.

Conclusion

This case highlights the role of laparoscopy not only in the diagnosis but also in the management of Swyer syndrome. The primary care physician needs to be aware of this condition, as early referral to tertiary centers is necessary for appropriate management of this condition.

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