International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Kavyarani C et al. Int J Reprod Contracept Obstet Gynecol. 2023 Feb;12(2):508-510
www.ijrcog.org
pISSN 2320-1770 | eISSN 2320-1789

DOI: https://dx.doi.org/10.18203/2320-1770.ijrcog20230148

Case Report

A rare case report of Turner's syndrome with Mullerian agenesis

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Received: 21 December 2022
Accepted: 10 January 2023

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ABSTRACT

Amenorrhea is the absence of menstrual blood flow. Primary amenorrhea should be considered in a patient with secondary sex characteristics who has not experienced periodic menstruation by 16 years of age or 5 years after breast development. Patients who have not developed secondary sex characteristics, especially the absence of breast development, and have not established periodic menstruation by age 13 should also be worked up for primary amenorrhea1. Turner's syndrome (TS) also called as Ullrich Turner's syndrome, is a disease of unclear pathogenesis characterized by complete or partial absence of one sex chromosome, with or without cell line mosaicism in a phenotypic female with short stature and primary ovarian insufficiency. However, TS may also involve other complaints including lymphedema, autoimmune diseases, metabolic diseases, cardiac, kidney and bone anomalies, hearing loss, and neurocognitive difficulties which lead to psychosocial and educational inadaptation. Optimizing health care is crucial to allow these individuals succeed in their full potential. In this regard this is a rare case report of Turner's syndrome with Mullerian Agenesis.

Keywords: Primary amenorrhea, Turner's syndrome, Mullerian agenesis, 45Xo

INTRODUCTION

Humans have 46 chromosomes in the cells. Two of them are called sex chromosomes: X and Y. In the early stages of cell division giving rise to an embryo, an erroneous division makes part or all of the X chromosome is lost if the pregnancy continues, the child will have Turner syndrome (TS). TS is a genetic disease with a karyotype of 45, X or 46, XX/45, X (mosaicism) or other structural abnormalities of X chromosomes. It is found in about one in 2,500 female births. The most common age of presentation is between 11 and 15 years, and rarely after 20 years.[2]

These cases are characterized by streak ovaries, primary amenorrhea, short stature, and multiple congenital anomalies in a phenotypic female. One third are recognized at birth by lymphedema, redundant skin or webbed neck. Another third, in childhood, stunting and the remaining third may present with delayed pubertal development or primary infertility. Among its highlights dysmorphic epicanto, pinnae rotated back, wide short neck, low hairline, broad chest shield, separated breasts, cubitus valgus, hands and feet with lymphedema, narrow and concave nails. Psychomotor development and IQ are usually normal. Structural abnormalities that may be present includes kidney 30-40%, 50-85% have middle ear disease: recurrent suppurative otitis media, serous otitis media, chronic suppurative otitis with perforation, conductive hearing loss, cholesteatoma formation, sensorineural hearing loss.[3] Five percent or more of women with Turner syndrome may have abbreviated menstrual function before developing amenorrhea and hypergonadotropic hypogonadism. An estimated 1 to 2% of all patients may become pregnant. Only three patients with TS (and two of them with streak ovaries) have ever been reported to become pregnant after developing amenorrhea and elevated gonadotropin levels.[4]

The development of uterus, fallopian tubes, and vagina are usually normal in TS. We wanted to report this case of 17

year old female with TS, who presented with hypoplastic uterus, cervix and rudimentary vagina, features of Mullerian agenesis; as this association was extremely rare with only few documentations in literature.

CASE REPORT

A 17 year young female presented to our out-patient department with short stature and primary amenorrheae. She had a history of delayed milestones and stunted growth. She was born out of non-consanguineous marriage and her maternal history was insignificant. Her sibling was 20 year old with menarche achieved at the age of 14 years.

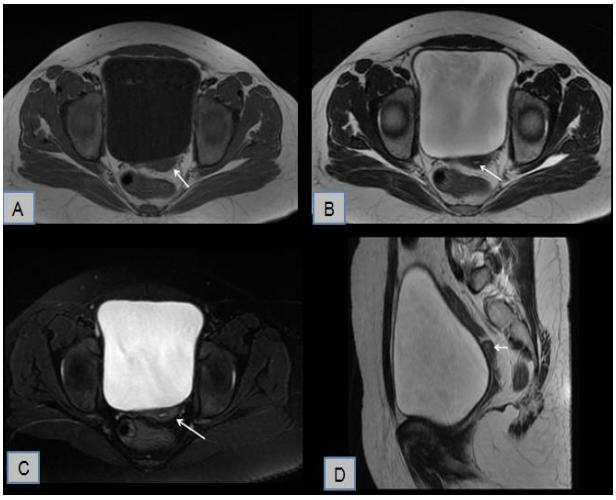

Figure 1: Axial MRI images of pelvis (A) T1W (B) T2W (C) T2 fat saturated and (D) sagittal T2W image showing a atrophic uterus (white arrow) with normal endomyometrial differentiation.

Her height was 130 cm, weight 37 kg with BMI coming to 19.47 kg/m^2 and apparently a normal intelligence. Physical examination revealed a female phenotype with short stature and cubitus valgus. Her breast development, axillary hair and pubic hair development were Tanner's stage I. Examination of the cardiovascular, respiratory, and neurological systems was normal. Gynecological examination revealed small vaginal dimple with no visible cervix. She was evaluated further with a provisional diagnosis of TS.

Laboratory investigations revealed hypergonadotropic picture raised LH and FSH levels. Her thyroid function test and prolactin levels were within normal limit. Her pelvic ultrasound revealed a hypoplastic uterus and ovaries. CT abdomen and pelvis had a complementary MRI also revealed hypoplastic uterus and absent ovaries on both sides. Other organs like kidneys, pancreas, liver, and adrenals were normal (Figure 1 and 2). Chromosomal analysis report of peripheral blood showed X0 sex chromosomal pattern with normal autosomes confirming the diagnosis of TS (Figure 3). Laparoscopy could not be proceeded because of denial of consent.

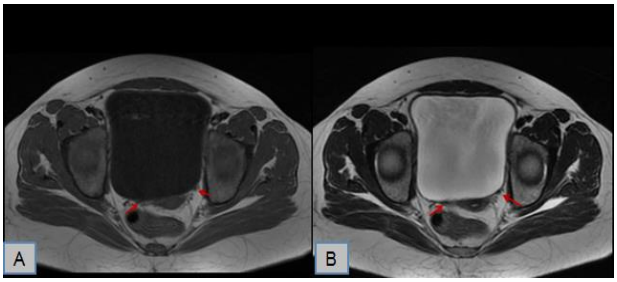

Figure 2: (A) Axial T1W; (B) axial T2W images of pelvis demonstrating bilateral streak ovaries (red arrows).

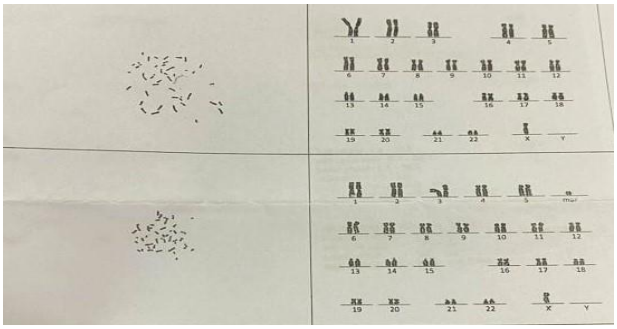

Figure 3: Chromosomal analysis report of peripheral blood showed X0 sex chromosomal pattern with normal autosomes.

DISCUSSION

TS is one among the most common causes of primary amenorrhea.[5] Approximately 98-99% of TS fetuses are spontaneously aborted, and about 20% of all spontaneously aborted fetuses have TS.[6] TS is associated with a constellation of potential abnormalities involving many organ systems, making it a challenging disorder for health care providers and families. In the absence of a functional second X chromosome the oocytes degenerate more rapidly than normal, so that at the time of birth there are few, if any, left and the ovarian tissue resembles fibrotic streaks. One fourth to one-third patients have renal malformation on ultrasonographic examination. Estrogen therapy in these patients can help in sexual development and bone maturation. *In vitro* fertilization using donor oocytes has been successfully employed with good pregnancy outcomes.

Mullerian agenesis on the other hand presents with normal secondary sexual characters and developmental anomalies of female genital tract including absent/malformed uterus and vagina (Mayer Rokitansky Kuster Hauser syndrome). They have normal female genotype 46, XX and they ovulate normally. They usually present with primary amenorrhea or hypomenorrhea, and pelvic pain due to structural abnormality of female genital tract. With appropriate reconstructive surgeries, these patients can have normal sexual life, but pregnancy is unlikely, unless the uterus is structurally compatible.

It is difficult to postulate a common theory for coexistence of these two anomalies in the same patient. It is possible that activating mutation of anti Mullerian hormone (AMH) gene located on q13 on chromosome 12 might be contributory for the regression of Mullerian duct derivatives like fallopian tubes, uterus, cervix, and upper two third of vagina in this patient, who is a Turner's phenotype. This combination is a double blow to the patient with very poor chances of conception. In these patients, estrogen therapy can result in the development of breasts and bone growth, but induction of menstruation and subsequent pregnancy is extremely unlikely.

A multidisciplinary team should oversee management of girls with TS from diagnosis through to adult life. Recombinant human growth hormone replacement therapy with estrogen and psychosocial support are the treatment modalities In general, oestrogen should be commenced around age 12-13, but generally no later than age 14. Gonadotrophin levels should be determined prior to introduction of oestrogens in order to confirm gonadal failure (elevated FSH).

CONCLUSION

Patients with TS may present with primary amenorrhea and lack of secondary sexual characters if ovarian failure develops before the onset of puberty. Patients who present with primary amenorrhea should have a thorough review of their clinical history and a complete physical examination, including external and internal genitalia. Karyotyping should be conducted in all patients with hypergonadotropic hypogonadism and androgenic features. It is also advisable to monitor all individuals with TS for auditory disturbances, thyroid dysfunction, hypertension, diabetes, and dyslipidemia. Clinicians from all specialties should be mindful of the characteristic clinical features of this uncommon entity and should do their best to make the best use of incidental clinical findings. Unusual combination of Turner's and Mullerian agenesis make this case an interesting one, with cases rarely reported in the literature.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Kavyarani C, Rathnamma P, Priya MJ. A rare case report of Turner's syndrome with Mullerian agenesis. Int J Reprod Contracept Obstet Gynecol 2023;12:508-10.