CASE REPORT

Rare Presentation of Mosaic Form (45X/46XX) of Turner's Syndrome

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Introduction

Turner syndrome is a genetic condition that only affects females. This condition occurs in about 1 in 2,500 newborn girls worldwide, but it is much more common among pregnancies that do not survive to term (miscarriages and still births) [1].

Approximately, half have monosomy X (45,X) and 5–10 % have duplication (isochromosome) of the long arm of X (46,X,i(Xq)). Most of the rest have mosaicism for 45,X, with one or more additional cell lineages.

TS was named after Dr. Henry Turner, who discovered the condition in 1938. The condition can cause a range of disabilities which can be—physical, emotional, and educational. TS occurs when one of the two chromosomes that are found in females is completely or partially missing [1].

Chromosomes are strands of DNA that are found in all of the cells in the human body. They contain instructions that determine a person's physical and behavioral characteristics [1].

Case of TS where an X chromosome is completely missing are sometimes referred to as “Classical” Turner syndrome, “Mosaic” Turner syndrome is where abnormalities only occur in the X chromosome of some of the

body's cells. In such cases, there may be a few or no symptoms [1].

The 45,X/46,XX chromosomal pattern is the most frequent mosaic type of this disease (36 %) [2].

Mosaic TS is not inherited. It occurs as a random event during cell division in early fetal development. As a result, a few of the affected person's cells have the usual 2 sex chromosome (either 2X chromosomes or 1X chromosome and 1Y chromosome).

Case Report

A 40-year-old lady presented to our unit with the complaint of inguinal mass associated with pain in swelling and fever on and off for the past 8 days. Detailed obstetrics history revealed that she never had a menarche or vaginal spotting. There was no h/o cyclical abdominal pain or nodules on the skin. Patient had c/o inability to conceive after 26 years of marriage. Patient did not report any h/o chemotherapy, radiotherapy, trauma, or surgery to gonads. Past medical history was negative for mumps, TB or any major systemic illness including asthma, malabsorption, celiac disease, cystic fibrosis, renal failure, or HIV infection.

On examination, patient was hemodynamically stable. She had a BP of 110/70 mmHg with no postural drop. Her pulse was 82/min, and regular respiratory rate was 15/min. Findings on her physical examination were as follows:

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1. Height: 5'1".
2. Adequate breast development. Nipples were small.
3. Absence of pubic and axillary hair.

Systemic examination was unremarkable.

Gynecological examination: External genitalia looked normal, vagina was short and blind (Fig. 1), and tender inguinal mass was around 4×4 cm (Fig. 2).

On P/R examination: Uterus was not felt.

Patient was provisionally diagnosed as “testicular feminization syndrome,” and investigations were performed to find out the cause.

- Baseline investigations were all normal.
- Plasma testosterone, estradiol, and LH values were found to be in range.
- Buccal smear showed the presence of barr bodies in 10–20 % of squamous cells.
- Whole abdominal sonography showed—absent uterus and absent B/L tubes and ovaries.
- USG of left inguinal mass was done which showed sonomorphology of ovotestis: Solid lesion was of size 2.5×1.7 cm; adjacent to the above lesion, there was another solid lesion of size 2.3×1.3 cm with cystic areas within it. First described lesion had sonomorphology of testis, while second lesion looked like ovary.
- No renal pathology was seen.
- ECG was normal.
- Thyroid function test, liver function test, serum alkaline phosphatase levels were all normal.

Under diagnosis of incarcerated inguinal hernia preoperatively, surgical exploration was performed through an inguinal incision, which revealed a firm mass lying in inguinal canal. The mass was attached to the fallopian tube and small atrophic ovary. The appearance of the mass was suggestive of atrophic uterus (Fig. 3). These structures

Fig. 2 Inguinal mass of around 4×4 cm

were removed and defect closed. Postoperative recovery was unremarkable. Histopathologic examination shows the organs of atrophic uterus, with single fallopian tube and single ovary instead of ovotestis.

The karyotyping confirmed the diagnosis of Mosaic Turner's SYNDROME, 46XX (96 %)/45X (4 %) (Fig. 4).

Discussion

Occurrence of ovary and fallopian tubes in inguinal hernia has been noted in several case reports in premature infants, and this is related to defects in genital tract, ovarian agenesis, müllerian dysgenesis, and ambiguous genital [3–5]. However, this report entails a rare case of inguinal hernia which contains the organs of the female genital tract in a 40 year old infertile woman.

Spontaneous menstruation and childbirth occur in 2–5 % of patients with TS, which may be explained by substantial 46XX/45X mosaicism, with normal cell populations existing in the ovaries [6].

Fig. 1 External genitalia—normal, vagina—short and blind

Fig. 3 Atrophic uterus with single fallopian tube and single ovary

Fig. 4 Karyotype: MOS 46, XX[48]/45,X [2]

A woman with mosaic TS possibly experiences regular MS until attaining late twenties in age rather than not having any menstrual cycle at all.

The psychosocial impact of Turner's syndrome may be substantial for young girls and women. These effects may be caused by infertility, short stature, and impaired development of sexual characteristics and most importantly lack of libido [7].

However, this is a rare presentation of mosaic Turner's syndrome having no menstruation and history of primary infertility with atrophic genital organs, and absent vagina.

Conclusion

Delayed puberty is not an uncommon clinical problem, and it is often disregarded as constitutional delay. It is stressed that strict clinical vigilance should be maintained to avoid missing a rare diagnosis such as Turner's syndrome and Mosaic Turner's syndrome. Recent advances in the medical science have enabled us to better help patients with Turner's syndrome, especially in the form of growth hormone and

hormone replacement therapy. Furthermore, screening such patients for known association carries the advantage of prophylactic interventions that in many situations may prove to be life saving.

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