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DIAGNOSTIC RIDDLE- SECONDARY AMENORRHOEA WITH PRIMRAY INFERTILITY

Obstetrics & Gynaecology

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ABSTRACT

Turner syndrome affects one out of 2000 to 5000 female newuborns. About 55% of them have non-mosaic Monosomy X, whereas 45% have a mosaic chromosomal component. A 30 year old female presented to Gynaecological OP in view of secondary amenorrhea and primary infertility. On examination, Secondary sexual characteristics were well developed. She was given Progesterone challenge and estrogen- progesterone challenge, but no withdrawal bleeding noted. On thorough workup, streak ovary was observed on left side and no ovary on right side. Early detection and intervention can help maintain normal menstrual cycles and potentially improve the rates of spontaneous conception. Counseling and psychological support are necessary.

KEYWORDS

Turner Syndrome, Mosaic Turner syndrome, Streak ovary, Secondary amenorrhoea, Primary infertility

INTRODUCTION

Turner syndrome is a congenital ovarian hypoplastic syndrome, characterized by a woman's X chromosome being partially or fully absent, resulting in short stature, primary ovarian insufficiency, gonadal dysgenesis, and infertility. It affects one out of 2000 to 5000 female newborns. In this syndrome, the absence of Y chromosome resembles the female, but like males they are chromatin negative due to absence of second X chromosome. In approximately 75% of Turner cases, the X chromosome is maternal in origin.[1] This chromosomal disorder may arise from monosomy of the X chromosome (complete loss of one X chromosome), mosaicism involving X chromosome aneuploidy or loss, or structural rearrangements of the X chromosome that result in the loss of syndrome-specific loci on the X chromosome.[2, 3] About 55% of population with Tuner syndrome have non-mosaic Monosomy X (45,XO), and other 45% have a mosaic chromosomal component like monosomy/ disomy, monosomy/ trisomy, monosomy/ marker chromosome, monosomy/ isochormosome, monosomy/ ring chromosome, monosomy/ structural arrangements, Xp or Xq deletion (part of short arm of the X chromosome).

Classic clinical presentation features of turner syndrome include short stature (associated with SHOX gene on X chromosome) and primary ovarian insufficiency.[4] Due to complexity in phenotypic and clinical presentations, the diagnosis is most commonly suspected during adolescence due to delayed secondary sexual characteristics and amenorrhea. According to the available literature, there have been significant efforts for uncovering genotype–phenotype correlations in cohorts of females suffering from Turner's syndrome with special attention to mosaic cases.[5-9]

In this case report, we describe the clinical, laboratory, cytogenomic findings and diagnostic hystero- laparoscopic findings in a rare case of an adult mosaic turner syndrome female who presented with secondary amenorrhoea and primary infertility.

Case Study

A 30 year old female presented to Gynaecological OP in view of amenorrhoea since 13 years and she is anxious to conceive. She is third child of a non- consanguineous couple and her 2 elder sisters and 1 younger sister were married, and having biological children. As per history, she attained menarche at the age of 14 years and her cycles being - 2/30 days, regular, changes 1-2 pads/day, scanty flow (not fully soaked), no clots and no dysmenorrhea. She married at the age of 17 years and since then she did not have menstrual cycles i.e., secondary amenorrhoea since 13 years. She did not have any other significant past history. She did not have a family history of premature ovarian failure. Her mother gave no history of gross motor or intellectual or neurocognitive impairment. She gave history of PCOS 10 years back and was given Tab. Metformin for 2 months. She was also given progesterone challenge 4 months back followed by estrogen-progesterone challenge, but no withdrawal bleeding noted.

On physical examination, she was 158 cms in height and weighs 52 kgs with BMI 20.83 kg/m2. Arm- span length was 158 cms. Her vital signs were normal. Axillary hair (Tanner stage 2), breast (Tanner stage 5) and pubic hair (Tanner stage 5). She had no signs of hirsutism or clitoromegaly. She had no short neck or webbing of neck, no cubitus valgus, high arched palate, or hearing loss. On per speculum examination, cervix was tubular with pin hole os. Vaginal rugosities present. On Bimanual examination, uterus was less than normal size, anteverted, mobile, fornices free and no forniceal tenderness noted.

On laboratory evaluation, follicular- stimulating hormone and leutinizing hormone levels were within the normal range. Estradiol (E2) and Progesterone levels were decreased. Serum Prolactin levels were increased, MRI brain showed no significant abnormalities. Thyroid function tests, liver function tests and renal function tests were within normal limits. Ultrasonography imaging of abdomen and pelvis showed uterus measuring 7.1 x 4.3 x 3.2 cms, ET- 4mm, left ovary normal and right ovary not visualized. Cytogenetic evaluation of peripheral blood revealed a normal female karyotype- 46XX, done by conventional cytogenetic testing method which has 30% detection rate.

Table 1: Clinical and Laboratory Parameters

S.No	Parameters	Values
1	Height (cms)	158
2	Weight (Kgs)	52
3	BMI (Kg/m2)	20.83
4	Hemoglobin (Gm%)	11.8
5	FSH (RR: 2.5- 10.2 ng.mL)	9.44
6	LH (RR: 1.9- 12.5 IU/L)	3.58
7	Estrogen (RR: 12.5- 498 pg/mL)	1.26
8	Progesterone (RR: .15- 28 ng/mL)	0.04
9	Serum AMH (RR: 0.256- 9.72 ng/mL)	1.39
10	Serum Prolactin (RR: 3- 24 ng/mL)	26.57
11	TSH (RR: 0.55- 4.78 uIU/mL)	3.414

She was posted for diagnostic hysterolaparoscopy. On Hysteroscopy- Utero-cervical length- 7cms, endometrium was pale and very thin. Bilateral ostia visualized, but no fluid movement movement seen into ostia. Minimal endometrial scrapings obtained and sample sent for HPE (HPE report- No endometrial tissue can be identified) and CBNAAT (CBNAAT report- Mycobacterial TB not detected). On laparoscopy, uterus visualized, congested and small. Bilateral round ligaments were normal. Bilateral fallopian tubes were very short and blind ended. No fimbiral ends were seen. Right sided ovary and infundibulo- pelvic ligament were absent. Left side yellow colour

streak seen behind the left fallopian tube- ?streaked ovary. No infundibulo-pelvic ligament was seen on left side. On chromopertubation, no dye is visualized passing through the fallopian tubes into the peritoneal cavity. Accidental finding was Fitz- Hugh Curtis Syndrome.

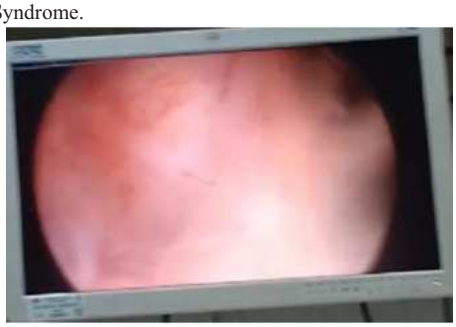

Figure 1: Hysteroscopy image showing pale and thin endometrium

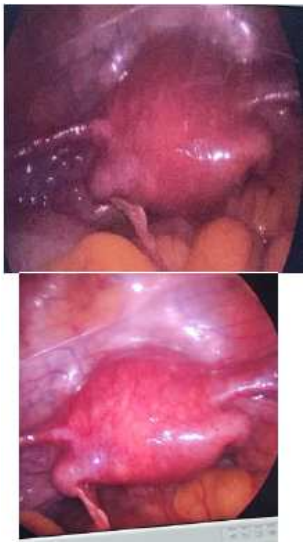

Figure 2: Laparoscopy image showing uterus, blind ended fallopian tubes and streak left ovary

Figure 3: Laparoscopy image showing Fitz- Hugh- Curtis Syndrome

DISCUSSION

Gonadal dysgenesis in women with Turner syndrome may result from a failure of chromosome pairing during meiotic prophase. [10] The majority of germ cells, which trigger spontaneous puberty in 10-30% of cases and support pubertal development, begin to diminish by the third month of intrauterine life. Consequently, only 5-10% of affected patients can menstruate regularly.[11- 14] POF (Premature Ovarian Failure) is another frequent clinical feature of Turner syndrome and mean age of menopause was reported to be 29.3 years. [12]

However, the extent of gonadal dysgenesis depends on the size of the impaired regions of homologous chromosomes. Severe pairing failures cause the degeneration of all oocytes before puberty and are associated with primary amenorrhea and poor sexual development. In contrast, mild pairing failures allow a significant number of oocytes to survive until puberty, resulting in secondary amenorrhea and secondary impaired sexual development. [15] Compared to the classical form, cases of Turner syndrome with a mosaic karyotype are more likely to experience spontaneous puberty, have normal levels of serum sex steroids and gonadotropins, and show follicles in ovarian biopsies.[16]

In the present case, she attained menarche and had regular hypomenorrheic cycles for 3 years. If workup was done in early days of her marriage, she could have been given hormonal supplementation and could be advised assisted reproductive techniques if spontaneous conception fails.

CONCLUSIONS

Counseling and psychological support are necessary for the patient, partner, and family. Early detection and intervention can help maintain normal menstrual cycles and potentially improve the rates of spontaneous conception. If spontaneous conception fails, assisted reproductive techniques should be considered earlier than in the normal female. Oocyte retrieval and preservation can facilitate future fertilization techniques.

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