ORIGINAL RESEARCH PAPER

Obstetrics & Gynaecology

KEY WORDS: Turner Syndrome, genetic, monosomy, Mosaic Turner

SUCCESSFUL PREGNANCY OUTCOME IN A CASE OF MOSAIC TURNER SYNDROME USING ICSI

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ABSTRACT

Turners syndrome is a genetic condition that affects only females . Occurs in 1 in 2500 newborn girls but it is much more common among pregnancies that do not survive to term . Half have monosomy X (45 X) , 10% have duplication of the long arm of X . Most of the rest have mosaicism for 45 X with one or more additional cell lineages . In contrast to classical Turner syndrome (X is completely missing), Mosaic Turner syndrome is where abnormalities occur in the X chromosome of some of the body cells . In such cases there may be few or no symptoms .

INTRODUCTION

Turner syndrome (TS), also known as 45,X, or 45,XO, is a genetic condition in which a female is partially or completely missing an X chromosome. It is not usually inherited; rather, it occurs during formation of the reproductive cells in a parent or in early cell division during development. People with Turner syndrome usually have 45 in some or all cells. The chromosomal abnormality is often present in just some cells, in which case it is known as Turner syndrome with mosaicism. In these cases the symptoms are usually fewer, and possibly none occur at all. Diagnosis is based on physical signs and genetic testing. It is the most common sex chromosome abnormality in females and occurs in approximately 1 in 2000 to 1 in 2500 live female births Mosaic Turner syndrome is defined as a partial loss of the second X chromosome. Symptoms for people with mosaic Turner syndrome tend to be less severe than symptoms for people with complete Turner syndrome. This is because the number of cells that are affected by the missing or deficient X chromosome is fewer with mosaicism.

CASE STUDY

The patient is a 31-year-old female with a history of Turner Syndrome, diagnosed in 2015 on the basis of medical history such as short stature, amenorrhea, infertility, and congenital heart disease, difficulties with spatial awareness, low hairline on the back of the head, narrow upturned fingernails, webbing skin on the neck, broad "shield" chest with wide-set nipples, puffy hands and feet (lymphedema) and pigmented moles (nevi). She presented to the OPD with the desire to conceive for the past year and amenorrhea for the past three years. She had h/o hypothyroidism for which she was taking medications intermittently. She had previously visited the clinic in 2015 with irregular cycles, which were regularized with medication. Her menstrual history indicated that she had menarche at 13 years of age with regular cycles lasting 2-3 days. Her cycle length gradually increased from the age of 25 years to every 3-4 months, and then to every 6-7 months and 8-9 months. The flow of menses reduced from 2-3 pads a day to 1 pad/day. The patient was diagnosed with premature ovarian failure (POF) based on FSH levels of 63 mIU/mL and an ultrasound revealing streak ovaries in 2015. She was put on oral contraceptive pills to regularize her cycles.

She was born of a full term vaginal delivery. Very low birth weight of 1.9 kg attributed to maternal malnutrition- NICU stay uneventful. Developmental milestones achieved at age appropriate time.

In 2015, we did a USG for her which was s/o Normal sized uterus and Streak ovaries. We further evaluated her with a battery of blood tests reveled FSH 63miu/ml S/o premature ovarian failure. On karyotyping Mosaic Turner Syndrome 45X, 46 XX (80:20) was seen. We ruled out other problems related to Turner Syndrome like cardiac disorders, DM, HTN. At this time patient was recently married and was not planning to conceive anytime soon. Patient and husband were counselled about her condition, and the need for fertility treatment for when they planned to conceive in the future. Patient was medically managed and was put on OC pills to regularize her cycles.

The patient followed up in 2019 with the desire to conceive. Her husband was evaluated and had no comorbidities or history of addictions. His semen analysis was within normal limits. The couple was counseled on IVF ET using donor oocytes. The patient underwent hysteroscopy, which showed scanty endometrium, Normal lateral walls and fundus, positive bilateral ostia and UCL 6.5cm. The patient was started on T estradiol valerate to thicken the endometrium, and the thickness was checked with TVS. The patient was then given T medroxyprogesterone to induce withdrawal bleeding, and this cycle was repeated two more times, resulting in a good 10mm triple-line endometrium.

The patient underwent ICSI using her husband's semen and donor oocytes. The donor was stimulated with recombinant FSH 225Units daily for a total of 11 days, and Inj cetrorelix was added on D5 of stimulation. The final trigger was done with inj decapeptil, and oocyte retrieval was performed after 35 hours. Ten oocytes were retrieved, of which eight were M2 and two were G1. Of the eight mature oocytes, five were fertilized, and four made it to the blastocyst stage. Two blastocysts were transferred. During preparation for embryo transfer we gave her tab estradiol 2 mg (2-2-2) for a period 12 days. By then she had a endometrial thickness of 10 mm , triple line , and had a good subendometrial flow. We added progesterone (on D13 of estradiol) –tab Duphaston 1-1-1 and vaginal progesterone 200 mg cap 1-1-1 and after 6 days progesterone we transferred 2 blastocysts under sonographic guidance. Two blastocysts of grade A were transferred , one of which was fully expanded and the other was an early blastocyst. We continued estradiol and progesterone till BHCG was positive (14 days after transfer). resulting Successful pregnancy was observed. Patient was regular in her ANC visits with us. Elective LSCS was done in June 2021 in view of Primi with IVF conception with short stature and maternal request. LSCS was uneventful and Healthy baby born.

DISCUSSION

Turner syndrome is a condition that can lead to various developmental, endocrine, cardiovascular, psychosocial, and

reproductive problems, often resulting in infertility. While spontaneous pregnancies have been reported in some cases, they are rare and associated with a high risk of miscarriage and chromosomal abnormalities. Oocyte donation has been successfully used for over 20 years as a treatment option for patients with Turner syndrome who experience premature ovarian failure. The success rates of pregnancy for Turner syndrome patients after oocyte donation are similar to those of other women. However, the number of reported cases is still relatively small.

While natural pregnancies are uncommon in women with TS, those who do become pregnant have a higher risk of adverse outcomes such as miscarriage, stillbirth, and chromosomal abnormalities. It is important for couples diagnosed with TS to be informed of the high probability of gonadal failure and infertility in their future children, and offering preimplantation genetic diagnosis (PGD) may be a solution for decreasing the high risk for the fetus. Additionally, women with TS have an increased risk of multiple organ system anomalies, making cautious pregnancy and regular screening essential.

CONLCLUSION

This case demonstrates the importance of proper diagnostic and follow-up procedures when a chromosomal abnormality is detected in an infertile couple. As technology continues to advance, there is growing interest in fertility options for women with Turner syndrome. Current parenting options include adoption, surrogacy, and both spontaneous and assisted reproduction.

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