

A Rare Case Report of Spontaneous Pregnancy in Mosaic Turner Syndrome

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ABSTRACT

Background: 1 in every 2500 females is born with Turner syndrome (TS) due to chromosomal abnormalities of the X chromosome. A common symptom of this syndrome is infertility due to ovarian dysgenesis. **Methodology:** This case report is regarding a spontaneous pregnancy in a 25-year-old female, Primi, with mosaic Turner syndrome. At 38weeks 2days, patient came with spontaneous labour pains with a past medical history significant for mosaic Turner syndrome diagnosed in her teens after a diagnostic evaluation for short stature. However, she reported regular menstrual cycles beginning at age 14. **Result:** She delivered a healthy female weighing 2760 g. While this patient could conceive successfully without the aid of medical intervention, only 5.6% of Turner Syndrome patients conceive spontaneously. **Conclusion:** This case reveals the importance of discussing fertility-preserving measures and possible pregnancy comorbidities with Turner Syndrome patients and their families at the time of diagnosis.

Keywords: Turner syndrome, X chromosome, pregnancy,

Introduction

Turner syndrome is one of the most common chromosomal disorders. 1 in every 2500 female newborn infants has Turner syndrome. It is the second most common cause of chromosomal abnormalities resulting in miscarriage (Wasserman and Asch, 2012). Turner syndrome patients are classically present with short stature, webbed neck, cardiovascular and renal abnormalities, and gonadal dysgenesis. Most women with Turner syndrome develop accelerated follicular atresia that predisposes them to primary amenorrhea, premature ovarian failure, and infertility later on in their lives. Several structural X chromosome aberrations have been found, including deletion, duplications, rings, and translocations. Fifty percent of Turner syndrome patients have a complete loss of X chromosome, while 25% have a partial deletion of one X chromosome; 20% carry varying degrees of mosaicism, most commonly a 45,X/46,XX karyotype. About 90% of women with Turner syndrome who spontaneously conceive have a mosaic karyotype (Jacqueline *et al.*, 2013). Therefore, timely fertility counseling and exploration of fertility preservation options are crucial for mosaic Turner syndrome patients who experience spontaneous puberty.

Case Report

A 24-year-old female patient, Primi, 38 weeks 2 days, presented to our outpatient clinic with complaints of lower abdominal pain. The patient was worked up for short stature at age 14 when her peripheral blood samples showed the mosaic karyotype of Turner syndrome. The patient also reported regular menstrual cycles that last 4 days with mild to moderate flow and menarche at 14-years-old.

The patient's pregnancy was spontaneous, and all the routine investigations were in the normal range. The 1st-trimester screening was done, which was expected. An anomaly scan showed no anomalies. Growth scan and interval growth scan done were corresponding to gestational age. Fetal echocardiography was done with the maternal Turner syndrome to rule out cardiac anomalies and came back negative. The patient complained of lower abdominal pain and leaking per vaginum and delivered a healthy 2760g female baby with Apgar scores of 8 and 9 at 1 and 5 minutes, respectively.

Discussion

It has been found that infertility is a major concern for patients with Turner Syndrome. Considering the challenging nature of sharing information about the high likelihood of future infertility with the patients themselves and their parents, it is unsurprising that parents often

feel they have inadequate knowledge to discuss fertility with their daughters. This feeling is compounded by the social stigma of infertility, desire for their daughter to have biological children, and their own loss of not having a biological grandchild. These challenges make it even more important for medical providers to partner with parents and facilitate fertility-related discussions as part of TS patients' routine care (Oktay *et al.*, 2016).

There are different fertility preservation techniques, i.e., vitrification of mature oocyte after controlled ovarian stimulation, or ovarian tissue cryopreservation (OTC). Vitrification of mature oocyte is still the preferred method of FP; however, OTC is commonly used in pre-pubertal girls. Hundreds or thousands of follicles and oocytes in OTC are retrieved via a laparoscopic approach and frozen for later transplantation (Viuff *et al.*, 2019).

In vitro fertilization (IVF) is the most commonly used method in patients with TS. However, our patient had spontaneously conceived. Therefore, patients with Turner syndrome who have functional ovaries are considered good candidates for autologous IVF. Autologous IVF is when oocytes are retrieved and transferred back to the same patient's uterus. Mosaic peripheral blood karyotype, normal serum follicular stimulating hormone (FSH) and anti-Mullerian hormone (AMH) levels, and spontaneous puberty are markers for a successful autologous IVF (Reindollar, 2011).

After our patient encounter and the possible premature ovarian failure sequelae, we stress the importance of timely counseling about different fertility preservation modalities in patients with established Turner syndrome diagnosis. However, pregnancy in patients with Turner syndrome is challenging and can be a grave prognosis. TS patients are at great risk of aortic dissection and rupture, such conditions should be monitored closely in those who plan to attempt pregnancy (Folsom and Fuqua, 2015). Regardless of their karyotype, TS patients are at risk of pregnancy-related morbidities and mortality, even if they show normal cardiovascular workup, i.e. echocardiography or cardiac MRI. We encourage an open discussion with TS patients and their support system detailing all appropriate fertility preservation options and counseling on all risks and complications that might develop during pregnancy and after delivery.

Author contribution

Ramya Ravichandar, Rajam Krishna S, Priya Sivashankar and Sujai Anand encouraged and supervised the findings of this work. All authors discussed the results and contributed to the final manuscript.

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Study significance

Regardless of their karyotype, TS patients are at risk of pregnancy-related morbidities and mortality, even if they show normal cardiovascular workup, i.e. echocardiography or cardiac MRI. Therefore, we encourage an open discussion with TS patients and their support system detailing all appropriate fertility preservation options and counseling on all risks and complications that might develop during pregnancy and after delivery.

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