Case Report

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Pregnancy in a Patient With Mosaic Turner Syndrome: A Case Report

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ABSTRACT

Turner Syndrome (TS) is a chromosomal sex disorder, phenotypically characterized by short stature, webbed neck, cubitus valgus, and rarely with slight intellectual disability. A majority of TS patients (95%-98%) have infertility due to ovarian failure. Pregnancy in TS patients is an unusual case; however, pregnancy could rarely occur in mosaicism TS patients without any assistance. Pregnancy in such patients is associated with high risks of maternal mortality, spontaneous abortion, as well as the congenital and karyotype abnormalities of the fetus. A 30-year-old pregnant woman has referred to our genetics lab with a history of polyabortivity. Her menarche occurred at the age of 13 years and her menstruation was claimed to be in a regular cycle. The patient's two first pregnancies resulted in stillbirth, whereas the third one was delivered through caesarian surgery, but spoiled after 8 days. Our case was characterized by mosaicism 45, X/45, XX, after referring as a multi-abortion case. The fourth pregnancy has happened at the age of 31 years and a healthy embryo with normal heart function was diagnosed by sonography in 17 weeks of gestation. The result of amniocentesis confirmed a healthy female embryo with 46, XX karyotype. Spontaneous pregnancy is regarded as a precarious situation terrifying by abortion or malignancy; also, chromosomal abnormalities, like trisomy 21 and TS, are prevalent in offspring. Therefore, it is strongly recommended to have cohort studies based on karyotype characterization to decrease the patient's concerns as well as to follow more practical clinical approaches.

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Introduction

Turner Syndrome (TS) is a chromosomal sex disorder with various features, i.e. phenotypically characterized by short stature, webbed neck and cubitus valgus, and rarely with slight intellectual disability; TS occasionally affects internal organs, e.g., the bones and cardiovascular and urinary systems [1, 2]. TS prevalence has been reported as 1 in 2000 females [3]. Complete monosomy of X chromosome or partial monosomy, including ring X, short or long arm deletion, and the isochromosome of the long arm, has been reported as the leading causes of this abnormality as well as a small percentage of mosaicism [4].

The majority of TS patients (95%-98%) have infertility due to ovarian failure [5]; however, in some cases, normal pubescence may occur. Pregnancy in TS patients is unusual; however, some investigators claimed that pregnancy might rarely occur in mosaicism patients without assistance [2, 6, 7]. TS patients' pregnancy is associated with high risks of maternal mortality due to cardiovascular and metabolic complications, spontaneous abortion, as well as the congenital and karyotype abnormalities of the fetus [2, 8]. This study aimed to report a rare case of mosaicism TS with MOS 45, X [52]/46, XX [3] karyotype (Figure 1). The reported case has experienced 4 spontaneous pregnancies, leading to either abortion or infant death three times; she had a healthy and alive child (last pregnancy).

Case Presentation

A 30-year-old pregnant woman has referred to our genetics lab with a history of polyabortivity. The height and weight of the patient were measured as 134 cm and 38 kg, respectively. Her karyotype was determined as mosaicism TS with MOS 45, X[52]/46, XX [3] karyotype. At the time of developing this report, she had an alive and healthy female infant as a result of her fourth pregnancy. Her menarche has occurred at the age of 13 years, and her menstruation has been claimed to be in a regular cycle.

Moreover, she received no particular medicine. Her two first pregnancies resulted in fetal death at 28 weeks, whereas the third outcome was delivered through caesarian surgery but spoiled after 8 days. The last pregnancy resulted in a healthy female infant born at 38 weeks of gestation with cesarean surgery. The patient had one sister and three brothers; no similar medical problems have been reported in her family, as well as her spouse's family.

Discussion

The symptoms of TS patients probably correlate with the haploinsufficiency of X chromosome genes contributing to ovarian function, short stature, as well as gonadal dysgenesis [9]. However, very few reports suggested that, in some patients, spontaneous puberty, menstruation, and pregnancy could be occurred [10]. TS patients have infertility or a very short productivity lifespan. Interestingly, mosaicism patients are more susceptible to spontaneous pregnancy, i.e. usually associated with fetal abnormality and abortion [11].

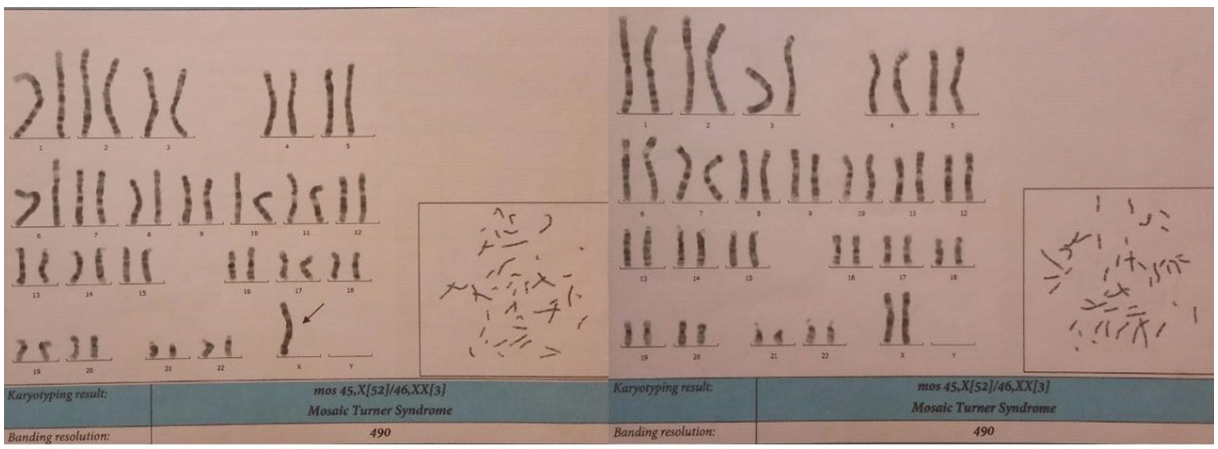

Figure 1. The karyotype of the reported patient with mosaic Turner syndrome

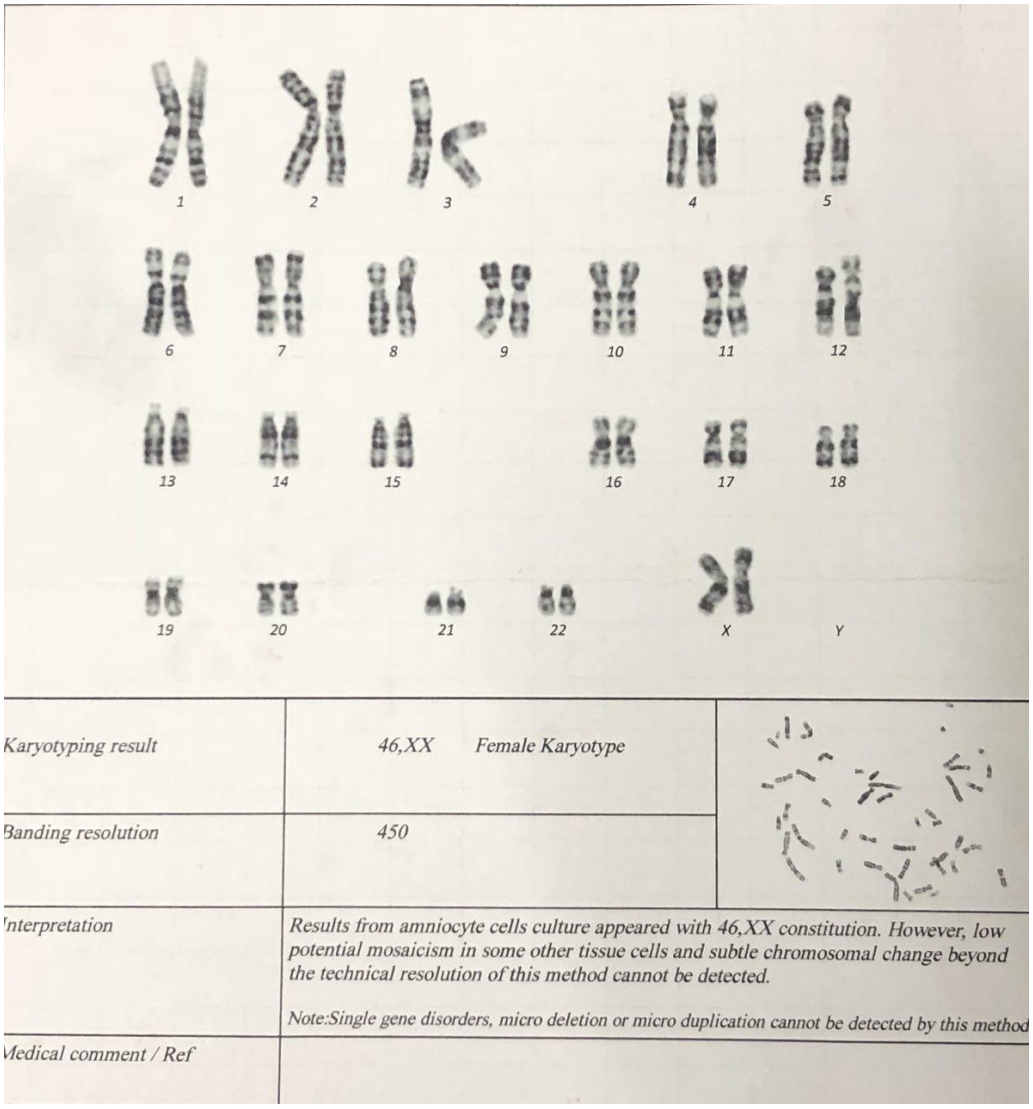

Figure 2. The normal karyotype of reported Turner patient's fetus

The rate of spontaneous pregnancy in TS was reported as 7.6% in a Danish TS population; however, the clinical and phenotypic indications for karyotyping were not illustrated in detail. However, at the same time, this rate has been evaluated as 4.8% and 1.8% in two independent studies in Sweden and the United States, respectively [12-14]. In a large cohort study on a population of 480 French TS women, only 27 patients experienced 52 spontaneous pregnancies (5.6 %), i.e. involved 30 successful deliveries (57.7%), including two newborn TS girls. Importantly, it has been suggested that menarche occurrence (25/27; 92.6 %) and 45,X/46,XX mosaicism (19/27; 70.4 %) are predictive factors of spontaneous pregnancy in TS patients [15].

One of the most concerns in TS women is either infertility or the consequence issues, like cardiovascular complications upon pregnancy; the same clinical practice guideline is applied for all different TS karyotypic subtypes. Moreover, there is no reported cardiovascular or blood pressure complication during pregnancy in mosaic TS patients, with more than 20% of cells represented as two X chromosomes. However, in the presented case, the patient illustrated approximately 5% cell lines with two X chromosomes. This low percentage might be the cause of her impaired reproductively. As previously discussed, the spontaneous pregnancy occurrence is significantly higher in TS patients with the 45, X/46, XX mosaicism, compared with the other TS karyotypes. Moreover, the presence of 45, X cells would not severely affect the reproductive lifespan as long as there are >20% of cells with two X chromosomes in blood cells [16].

Our case was characterized by mosaicism 45, X[52]/45, XX[3] after referring as a multi-abortion case. Interestingly, the TS was almost diagnosed late, i.e. because of healthy body development, regular menstruation cycle, and spontaneous pregnancy. The last pregnancy occurred at the age of 31 years, and a normal embryo with a healthy heartbeat and regular activity was diagnosed upon sonography examination at 17 weeks and 3 days. Moreover, no anomaly was reported for the embryo, and echocardiography results suggested no abnormality. Although amniocentesis was requested based on the TS mother, the result confirmed a healthy female embryo by 46, XX karyotype (Figure 2).

Conclusion

This paper reported a case of mosaic TS. Spontaneous pregnancy in TS is regarded as a precarious situation aggravated by abortion or malignancy, and chromosomal abnormalities in offspring, although the situation could vary based on the karyotype [2, 17]. Therefore, studies based on different TS karyotypes could help to decrease the patients' concerns and facilitate to follow more practical clinical approaches.

Ethical Considerations

Compliance with ethical guidelines

All ethical principles were considered in this article. The participants were informed about the purpose of the research and its implementation stages; they were also assured about the confidentiality of their information. Moreover, they were allowed to leave the study whenever they wish, and if desired, the results of the research would be available to them.

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Conflict of interest

The authors declared no conflict of interest.

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