

Spontaneous Conception in Mosaic Turner Syndrome

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Abstract

Spontaneous conception in women diagnosed with Mosaic Turner Syndrome has been reported, with increased risk of miscarriages and stillbirths. It is also a well-known fact that there is a high risk of fetal loss, congenital and chromosomal abnormalities, prematurity, and intrauterine growth restriction in the offspring of Turner syndrome.

Routine karyotype for female children has been traditionally been declined citing the reason of absence of dysmorphic features in the newborn.

We feel that offering karyotype to offspring of patients with MTS at a young age may provide advantage of an early intervention.

Keywords: Spontaneous Conception; Mosaic Turner Syndrome

Introduction

Turner's syndrome (TS) is defined as a total or partial absence of one X chromosome that results in ovarian dysgenesis. In women with mosaic Turner syndrome (MTS), some of the body's cells have two X chromosomes and other cells have one. Among women with MTS, spontaneous pregnancy has been reported as 2 - 10% [1]. However, there is an increased risk of miscarriage and stillbirths [1]. We report a case of a young patient with MTS who is in her sixth pregnancy.

Clinical Description

This 29 years old woman was reviewed in our antenatal clinic during her 6th pregnancy.

She was referred to paediatricians at the age of 6 years with a history of short stature. The chromosome analysis revealed a mosaic karyotype with two cell lines present, 46, XX and 45, XO. Clinical examination revealed wide spaced nipples but no other signs of TS (webbed neck, skeletal abnormalities, heart defects, or high blood pressure). She was started on growth hormone treatment.

At 12 years old, follicle stimulating and luteinizing hormone levels were in normal range. Tanner staging showed breast stage 2 and pubic hair at stage 3. At 13 years old she underwent menarche. She was followed up and was deemed to have achieved her height potential. Furthermore, she had a regular menstrual cycle with normal secondary sexual characteristics. At the age of 14, an ultrasound scan of pelvis illustrated normal sized bilateral ovaries and hence growth hormone treatment was stopped. According to her own declaration, she required extra help with her studies and completing her school education.

She has had three normal vaginal deliveries at term (one boy and two girls) and two first trimester miscarriages in the past. None of the children displayed any dysmorphic features at birth. However, the patient says that her eldest daughter has needed extra support in school.

The request for cytogenetic analysis/karyotype for patient's children was declined by the tertiary centre due to lack of any dysmorphic features at birth.

Discussion

Turner Syndrome is the most common sex chromosome disorder affecting 1 in 2000 - 5000 live female births [2]. The presence of a normal cell line in TS, may lead to development beyond puberty, leading to spontaneous pubertal development, regular menses and pregnancy before the onset of premature menopause [3]. However, there is a risk that her female children could have the similar risks (infertility, premature menopause, short stature); in case they have inherited a mosaic pattern [7]. It is a well-known fact that there is a high risk of fetal loss, congenital and chromosomal abnormalities, prematurity, and intrauterine growth restriction in the offspring of Turner syndrome [4-6].

Hence, it seems prudent that offspring of women with MTS/TS should be offered karyotype at the earliest opportunity to plan supportive management and extra help in school.

Conclusion

MTS patients attain normal fertility and growth potential when diagnosed early and managed appropriately. In addition to this, offering karyotype to patients at a young age may help diagnose MTS earlier and can lead to prompt management and possibly to oocyte cryopreservation. Offering karyotype to offspring of patients with MTS at a young age shall lead to early counseling and intervention which may include possibility of oocyte cryopreservation.

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