Case Report

Triple X Egyptian woman and a Down's syndrome offspring

Faeza Abdel Mogib El-Dahtory

Genetic Unit of Children Hospital, Mansoura University, Mansoura, Egypt

The 47, XXX karyotype (triple X) has a frequency of 1 in 1000 female newborns. However, this karyotype is not usually suspected at birth or childhood. Female patients with a sex chromosome abnormality may be fertile. In patients with a 47, XXX cell line there appears to be an increased risk of a cytogenetically abnormal child but the extent of this risk cannot yet be determined; it is probably lower in the non-mosaic 47, XXX patient than the mosaic 46, XX/47, XXX one. We describe a new rare case of triple X woman and a Down's syndrome offspring. The patient is 26 years of age. She is a housewife, her height is 160 cm and weight is 68 kg and her physical features and mentality are normal. She has had one pregnancy at the age of 25 years resulted in a girl with Down's syndrome. The child had 47 chromosomes with trisomy 21 (47, XX, +21) Figure 1. The patient also has 47 chromosomes with a triple X karyotype (47, XX, +X) Figure 2. The patient's husband (27 years old) is physically and mentally normal. He has 46 chromosomes with a normal XY karyotype (46, XY). There are neither Consanguinity between her parent's nor she and her husband.

Key words: Chromosomal abnormalities, Down's syndrome, triple X

Introduction

The 47, XXX karyotype (triple X) has a frequency of 1 in 1000 female newborns. However, this karyotype is not usually suspected at birth or childhood.[1] Female patients with a sex chromosome abnormality may be fertile. In patients with a 47, XXX cell line there appears to be an increased risk of a cytogenetically abnormal child; however, the extent of this risk cannot yet be determined; it is probably lower in the non-mosaic 47, XXX patient than the mosaic 46, XX/47, XXX one.[2]

Access this article online	
Quick Response Code:	Website: www.ijhg.com
	DOI: 10.4103/0971-6866.112925

Case Report

The patient is 26 years of age. She is a housewife, her height is 160 cm and weight is 68 kg and her physical features and mentality are normal. She has had one pregnancy at the age of 25 years resulted in a girl with Down's syndrome. The child had 47 chromosomes with trisomy 21 (47, XX, +21) Figure 1. The patient also has 47 chromosomes with a triple X karyotype (47, XX, +X) Figure 2. The patient's husband (27-years-old) is physically and mentally normal. He has 46 chromosomes with a normal XY karyotype (46, XY). There are neither consanguinity between the patient's parents nor she and her husband.

Discussion

The present triple X female was drawn to our attention after she gave birth to a child with Down's syndrome, where, there was nothing about this woman's physical appearance that would have indicated that she possessed an extra X chromosome. While many of the reported patients with the triple X syndrome have had reduced intelligence, this finding is probably due to a biased selection of patients (Day *et al*., 1964).[3] However, in (1972) Singer *et al*.[4] have reported a case with the same condition of our case. While many of the reported patients with the triple X syndrome have had reduced intelligence, this finding is probably due to a biased

Address for correspondence: Dr. Faeza Abdel Mogib El-Dahtory, Genetic Unit of Children Hospital, Mansoura University, Mansoura, Egypt. E-mail: fayzass_2002@mans.edu.eg

Figure 1: Karyotype 47, XX + 21 of the daughter of Triple X syndrome

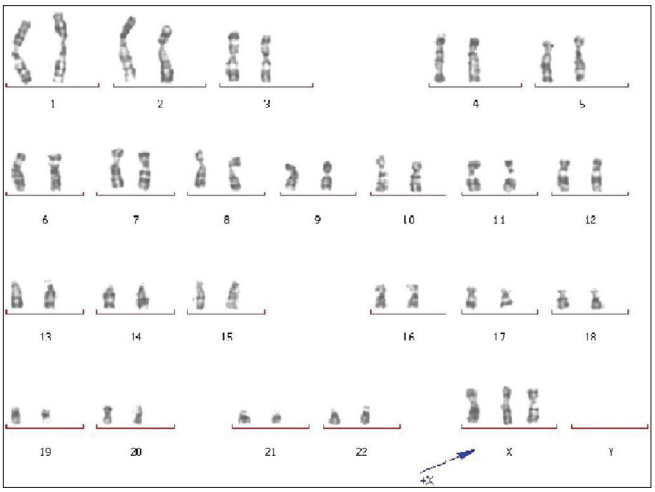

Figure 2: Karyptype 47, XX + X of the Down syndrome's mother

selection of patients (Day *et al*., 1964). Many of the initial reports about these females came from institutions for the mentally retarded where mass screening was performed. Our patient is one of the few observed with normal intelligence and this agrees with Singer's case.

To our knowledge, this is the first example of a triple X female giving birth to a child with Down syndrome in Egypt. It would be of interest to speculate that there is a causal relationship in the fact that this triple X mother produced a trisomy 21 child. If there is a cause and effect relationship between this triple X mother and the trisomic 21 offspring, it is difficult to propose a mechanism.[4]

References

1. Goldschmidt E, Márquez M, Solari A, Ziembar MI, Laudicina A. Phenotypic variability in 47, XXX patients: Clinical report of four new cases. Arch Argent Pediatr 2010;108:e88-91.
2. Dewhurst J. Fertility in 47, XXX and 45, X patients. J Med Genet 1978;15:132-5.
3. Day RW, Larson W, Wright SW. Clinical and cytogenetic studies on a group of females with XXX sex chromosome complements. J Pediatr 1964;64:24-33.
4. Singer J, Sachdeva S, Smith GF, Hsia DY. 2160 S. First Avenue, Maywood. Illinois, USA: Stritch School of Medicine, Loyola University; 1972. p. 60153.

Cite this article as: El-Dahtory FA. Triple X Egyptian woman and a Down's syndrome offspring. Indian J Hum Genet 2013;19:111-2.

Source of Support: Nil, **Conflict of Interest:** None declared.