

CYTOGENETIC STUDY OF TURNER SYNDROME IN IRAN

S.Y. Seyedna¹, PhD; R. Zakikhani², MD

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Abstract

Turner Syndrome is one of the best known chromosome anomalies in human being, by an approximate incidence of 1/2500 female at birth. The cause is a chromosomal aberration, mainly with the karyotype 45,X. Ninety six patients aged 6 to 26 years with short stature were studied for chromosomal anomalies. Out of these, 82 were phenotypically female and 14 phenotypically male. Twenty seven showed abnormal karyotypes, 15 were pure Turner with a chromosome complement of 45, X. Seven showed mosaicism of 45, X/46, XX and 5 showed 45, X/46, XY cell lines. Most of the features of Turner's syndrome were manifested in the fifteen 45, X Turners while the 45, X/46, XX did not show all the features due to the presence of a normal cell line. Out of the five mosaics 45, X/46,XY, four had female phenotypes with normal genitalia while one had a male phenotype with poorly developed male genitalia.

Introduction

Turner Syndrome was first clinically described in 1938(5). It was suggested that a chromosome constitution XO (2). The main clinical features of Turner syndrome are small stature, webbing of the neck, cubitus valgus, shield-shaped thorax, widely spaced nipples, rudimentary ovaries, gonadal streak, primary amenorrhoea, and poor breast development. The majority of Turner's syndrome patients show absence of one X chromosome, the karyotype being 45,X. Variations such as isochromosome X,X chromosome mosaicism, deletion of one arm of X and ring X, have been also reported. These variations are most frequently found in the first trimester abortions (1).

1- Dept. of Biology, University of Shahid Beheshti, Tehran, Iran.

2- Iran Hospital, Tehran, Iran

Materials and methods

Data from 96 patients were collected and analysed for Turner's syndrome. Clinical and cytogenetic data for each liveborne patient was obtained from four hospitals in Tehran by a through investigation of the clinical and cytogenetic history from 1989 to 1996.

The patients investigated belonged to different parts of Iran, mostly from the south-west regions.

Results and discussion

Clinical and cytogenetic analysis performed on the historic data of 96 patients suspected for being involved with Turner's syndrome, having short statures and primary amenorrhoea showed that 82 were phenotypically female and 14 male. Twenty seven showed abnormal karyotypes with the following categories:

Fifteen (55.5%) were pure Turners with short stature and a chromosome complement of 45,X. All were phenotypically female and had primary amenorrhoea. According to their ultrasonography reports, the uterus was hypoplastic and ovaries not well visualised. In individuals of pubertal age, breast development was scanty or absent.

Seven (25.9%) showed mosaicisms of 45,X/46,XX, who were phenotypically female. Of these, five had primary amenorrhoea with hypoplastic uterus and ovaries not well visualised and two had irregular menstrual periods. Their ultrasonography reports showed normal uterus. The number of cells showing 45,X abnormal chromosome complement varied in all five patients. The number of cells showing 45, X abnormal chromosome complement also varied in the two 45,X/46,XX mosaics who had irregular periods.

Five (18.5%) showed mosaicism of 45,X/46,XY. Four of these were phenotypically female and had short stature, normal female external genitalia, with poor breast development and primary amenorrhoea. Their chromosome complements were 45,X/46,XY. On average, 61% of the metaphases had chromosome

complement of 45,X and 39% had 46,XY. One patient was phenotypically male, with short stature and wide shield. He also had cryptorchidism with hypospadias. The genitals were poorly developed. Scrotum with both testicles were not palpable.

Buccal mucosa cells of the four phenotypically female patients stained with thionin showed that X chromatin was absent, while staining with Quinacrine revealed 3% of cells had Y chromatin. Buccal mucosa cells of the phenotypically male patient revealed that X chromatin was absent and on average 6% of the cells had Y chromatin. The patient's chromosome composition was 45,X/46XY mosaicism. (38% with 45,X and 62% with 46,XY chromosome composition). Thus, the number of 46, XY cells is more than that of 45,X cells in this patient, while in the phenotypically female patients the number of 46, XY cells are less.

Other somatic malformations of Turner syndrome were not observed. Turner's syndrome patients tend to have a normal life expectancy and have fairly normal IQ (4). With better medical facilities, for an infant or young child diagnosed as having Turner syndrome, some kind of therapy like surgical correction of webbing of the neck is useful. Therapy with male hormone may cause height increment to some extent. If the diagnosis is made early at 10-14 years of age, hormones can be administered, and menstruation will occur, but ceases immediately upon withdrawal (3). There is no known treatment to enhance the chance of ovulation or fertility. In the present study, of the total of 96 patients, two were 45,X/46,XX mosaics who had irregular periods. But whether they could lead a normal reproductive life or not is not known.

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