
**CHROMOSOME STUDY IN SUSPECTED CASES OF TURNER'S
SYNDROME FROM JAMMU REGION OF JAMMU & KASHMIR**

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ABSTRACT: Turner syndrome is one of the commonest sex chromosomal abnormalities where the leading cause is monosomy of X chromosome. Karyotypic study was carried out in 13 cases of suspected Turner syndrome from the Jammu region of Jammu and Kashmir. Of the 13 cases, abnormal karyotypes were detected in 06 cases (46.1%) and normal karyotype was detected in 07 cases (53.9%). Of the six abnormal cases, three were found to have typical Turner syndrome with 45, XO karyotype and remaining three were found to have mosaic Turner syndrome with 46, XX/45, XO karyotype.

KEYWORDS: Turner Syndrome, Monosomy, Mosaic Turner syndrome, Chromosomal abnormalities, Karyotype.

INTRODUCTION

Turner syndrome is the sex chromosomal disorder resulting due to the monosomy of X chromosome. Henry Turner, an Oklahoma endocrinologist in 1938 first time describes Turner syndrome, when he, described 7 patients between the ages of 15 and 23, who were referred to him for dwarfism and lack of sexual development. He treated them with pituitary extracts, but the treatment was ineffective. In Europe, it is often called Ullrich-Turner syndrome or even Bonnevie-Ulrich-Turner syndrome to acknowledge that earlier cases had also been described by European doctors.

Individuals with Turner syndrome are female and usually have characteristic phenotype. Although short stature is the most common feature of Turner Syndrome, many other

recognizable characteristics like sexual infantilism, pattern of major and minor malformations, triangle shaped face, posteriorly rotated external ears and a broad, webbed neck may also exist. Girls with Turner Syndrome are at increased risk for several disorders that do not manifest as obvious physical features during childhood. An example of one characteristic that commonly occurs in these patients is ovarian failure. In addition, the chest is broad and shield like in shape. Lymphedema of the hands and feet is observable at birth. Many patients with Turner syndrome have congenital heart defects, most commonly obstructive lesions of the left side of the heart. About 50% of individuals with Turner syndrome have structural kidney defects but they usually do not cause medical problem. There is typically some diminution in

spatial perceptual ability, but females with Turner syndrome usually have normal intelligence⁶.

The incidence of Turner syndrome in consecutive neonates has been reported to be 0.04%⁷. Turner syndrome is one of the few chromosomal aberrations that can be recognized clinically during infancy or childhood based on short stature, broad shield chest, lymphoedema of the lower limbs, webbed neck and multiple minor anomalies⁵. However, Karyotyping is necessary to confirm the diagnosis.

MATERIALS AND METHOD

A total of 13 females clinically diagnosed as Turner syndrome were referred for their chromosome study. Some of these females had all the typical features of Turner syndrome while others had only a few of these features. Therefore, all these 13 females were subjected to their chromosome study. Chromosome study was carried out from the whole blood cultures and the prepared slides were G-banded following Seabright, 1971.

RESULTS AND DISCUSSION

The chromosomal abnormalities in females with Turner syndrome are quite variable. Based on the details of their clinical profile and chromosome study, the six females were found to have sex chromosomal abnormalities

and based on the nature of the abnormality, they are categorized in to two groups:

Group 1. Turner syndrome with Monosomy X

Group 2. Mosaic Turner Syndrome (XO/XX)

Details of the clinical profile along with chromosome study of both the groups is described here:

Group 1: Turner syndrome with Monosomy X Clinical Profile

Three of the six females where chromosome study was found abnormal, had the following physical abnormalities namely Short stature, Webbed Neck, Cubitus Valgus, Shield like chest, Infantile external (without pubic and axillary hairs) and internal genitalia, Irregular dermatoglyphic pattern, Poorly developed secondary sex characters. Beside these abnormalities, they had low I.Q., and were infertile due to infantile internal genitalia.

Chromosome Profile

In all the three females, chromosome study was carried out from some of the well spread G-banded metaphase plates. All the G-banded metaphase plates had an abnormal chromosome number of 45 i.e., they had monosomy of one of the chromosome (Fig.1).

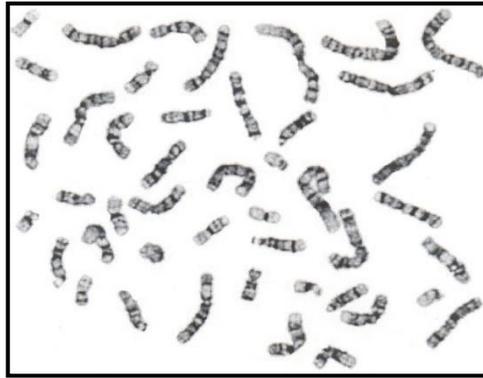


Fig.1: Complement of Turner Syndrome with Monosomy X (2n=45).

Karyotypic Analysis

To find out the nature of the Monosomy some of these G-banded metaphase plates were selected for the preparation of their karyotypes. Every Karyotype possessed 44 autosomes and a single X-chromosome i.e. these

Karyotypes had XO sex chromosome constitution. XO sex chromosome constitution detected in these three females was held responsible for Turner syndrome features in these females (Fig.2).

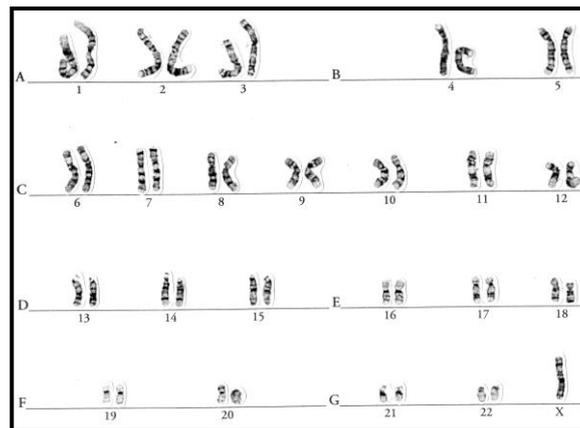


Fig.2: Karyotype of Turner Syndrome with Monosomy X (45, XO).

Chromosome study, therefore, confirmed the clinical diagnosis in all the females of this group.

Group 2: Mosaic Turner Syndrome Clinical Profile

Clinically, three females were having the following features: Short

stature, Webbing of neck was not prominent, Cubitus valgus was not prominent. All these females had mild degree of mental retardation with poorly developed secondary sexual characteristics. All these females besides having other abnormalities had

primary amenorrhea- the failure of the mensuration in a female at puberty. Because of primary Amenorrhea and some features of Turner syndrome, they were advised chromosome study to know the status of both the X-chromosomes.

Chromosome Profile

All the three females were found possessing two cell lines:

- a) Cells with 46 chromosomes
- b) Cells with 45 chromosomes

Both the cell lines were present in 1:1 ratio. With respect to chromosome numbers, they were therefore, mosaics.

Karyotypic Analysis

Both the cell lines were selected for the preparation of karyotypes. Karyotypes prepared from the G-banded metaphase plates containing 46 chromosomes had 44 autosomes and a pair of homomorphic sex chromosomes-XX. Hence these were the karyotypes of a normal female.

Karyotypes prepared from the cells containing 45 chromosomes had 44 autosomes and a single sex chromosome- X. These were the karyotypes of a Turner syndrome, 45, XO. Two cell lines in all these females were 46, XX/46, XO and the clinical abnormalities especially Primary Amenorrhea was caused due to XO cell line.

Chromosome study in these three females was therefore abnormal and it was held responsible for clinical abnormalities.

The incidence of 45, X is approximately 1 in 2000 to 1 in 5000 live female births³. The cause for Turner syndrome is monosomy of X chromosome which may arise from meiotic non-disjunction or anaphase lagging during spermatogenesis, oogenesis or from postzygotic error. Most reports of Turner syndrome cite its incidence as 1 in 2000 to 2500 live female births⁹.

Monosomy for short arm of X leads to short stature and patients usually have the stigmata of Turner². Some researchers believe that all live born females with Turner Syndrome have cell line containing two sex chromosomes that may be present at a low level of mosaicism^{1,4}.

Molecular studies have shown that approximately 60 to 80% of monosomy X cases are caused by the absence of a paternally derived sex chromosome, occurring either during early meiosis in the embryo or during meiosis in the father i.e., the offspring receives an X chromosome only from the mother⁶.

However, in the present study all the three females had 45, XO and the remaining three females had mosaicism of X chromosome, 45, XO/46, XX.

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