DIFFERENT KORYOTYPIC FINDINGS IN 21 TURNER SYNDROME PATIENTS

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Gazi Medical Journal 1998; 9 : 121-124

SUMMARY :

Purpose : *Turner syndrome is the most common sex chromosome abnormality in females. Turner stigmata lie in a wide spectrum of abnormalities, ranging from short stature to visceral abnormalities. Different karyotypic findings may be determined in these patients. In this study, we aimed to investigate the correlation between karyotypic abnormalities and phenotypes in Turner syndrome patients.* ***Methods :*** *21 Turner syndrome patients were included in the study. Peripheral blood lymphocyte cultures were set up and chromosomes were harvested according to standard protocols. 30 metaphases were analysed from each patient after GTG banding.* ***Results :*** *14 patients had a 45,X chromosome construction, whereas 3 had a 45,X/46,XX mosaicism, 1 had a 45,X/46,i(xq), 1 had a 46,X,i(xq), 1 had a 46,X,del(X)(q21-qter) and 1 had a 46,X,del (X)(p11.2-p11.4) koryotype.* ***Conclusion :*** *Many investigators have been interested in phenotypic findings and their relationship with karyotypes in Turner syndrome patients. There is a common suggestion that lacking segments have the genes responsible for phenotypic findings. However, complex features, such as height, are influenced by many genes in addition to environmental factors.*

Key Words: *Turner Syndrome, Phenotype- Karyotype Abnormalities.*

INTRODUCTION

Turner syndrome is variously estimated to be 1/2000-5000 female births (1). The chromosome complement of Turner syndrome is usually monosomy X. Other structural variations of an X chromosome can also result in this syndrome. The chromosome constitution is clinically significant. Structural abnormalities of the X chromosome may be associated with a clinical picture that ranges from normal phenotypes to phenotypes indistinguishaple from 45,X Turner syndrome (2).

In this report we aimed to find the correlation between karyotypic abnormalities and phenotypes in 21 Turner syndrome patients consulted to our cytogenetics laboratory between March 1992 and June 1996.

MATERIALS AND METHODS

Chromosome analysis was performed on the chromosome spreads of peripheral blood lymphocyte cultures (3). Thirty metaphases obtained by conventional methods and banded according to Seabright's modified GTG banding technique were examined for each patient (4). In cases of mosaicism, 100 metaphases were analysed.

RESULTS

Cases of Turner syndrome exhibit great variation of karyotypes. As shown in the table 1, 14 (66.6 %) of 21 Turner patients showed a classical 45,X karyotype, where as 3 (14.2 %) of them had a 45,X/46,XX mosaicism, 1 (4.7 %) had a 45,X/46,i(Xq) and 1 (4.5 %)had a 45,X/46,XY mosaicism. One (4.7 %) patient had a 46,X,i(Xq) karyotype, 2 (9.5 %) patients had deletions of X chromosome. One had a 46,X,del (X)(q21-qter) and 1 had a 46,X,del (X)(p11.2-p11.4) karyotype.

DISCUSSION

As shown in table 1, 14 of 21 Turner patients had a classical 45,X karyotype. In a 45,X Turner syndrome, short stature, gonadal dysgenesis and Turner stigmata such as web neck, low set ears, low

Patient No	Age (years)	Height (percentile)	Turner stigmata	Gonadal function	Karyotype
1	13	113 cm (3 %↓)	slightly webbed neck scapula alata pectus excavatus maxillary hypoplasia genu valgum	2°sex development (-) menarche (-)	45,X/46,XX
2	12	110 cm (3 %↓)	typical	?	45,X
3	10	115 cm (3 %↓)	pectus excavatus	?	45,X
4	13	122 cm (3 %↓)	flat nasal bridge webbed neck	2° sex development (-) menarche (-)	45,X
5	10/12	61 cm (3 %)	Microcephaly short neck high arched palate low posterior hairline flat nasal bridge dorsal edema of the right foot	?	45,X
6	7	105 cm (3 %↓)	low posterior hairline flat nasal bridge pectus excavatus cubitus valgus	?	45,X/46,X,i(Xq)
7	17	152 cm (3 - 10 %)	no stigmata	primary amenorrhea 2° sex development (-)	45,X/46,XX
8	14	136 cm (3 %↓)	high arched palate short neek low posterior hairline	2° sex development (-) menarche (-)	45,X
9	2	75 cm (3 %↓)	no stigmata	?	45,X
10	5 days	52 cm (50 %↑)	high arched palate short neck low posterior hairline widely spaced nipples umblical hernia	?	46,X,i(Xq)
11	16	138 cm (3 %↓)	hirsutismus	primary amenorrhea	45,X
12	$8^{6}/12$	117 cm (3 %↓)	no stigmata	?	46,X,del(X)(q21.1-qter)

Table 1 : Clinical features and karyotypes of the Turner patients.

Patient No	Age (years)	Height (percentile)	Turner stigmata	Gonadal function	Karyotype
13	$15^{3}/12$	129 cm (3 %↓)	mental retardation hypertelorism hearing defect high arched palate short neck low posterior hairline widely spaced nipples cubitus valgus	menarche (-) 2°sex development (-)	46,X,del(X)(p11.2-p11.4)
14	$6^{2}/12$	100 cm (3 %↓)	typical	?	45,X
15	16	140 cm (3 %)	typical	primary amenorrhea	45,X
16	11	120 cm (3 %↓)	low posterior hairline webbed neck	?	45,X
17	$8^{3}/12$	103 cm (3 %↓)	no stigmata	?	45,X/46,XX
18	17	157 cm (3-10 %)	clitoromegaly	primary amenorrhea	45,X
19	$14^{7}/12$	138 cm (3 %↓)	typical	menarche (-) 2° sex development (-)	45,X
20	$9^{5}/12$	120 cm (3 %↓)	typical	?	45,X
21	$15^{2}/12$	145 cm (3 %↓)	mental retardation	menarche (-)	45,X

Table 1 (Continued) : Clinical features and karyotypes of the Turner patients.

posterior hair line have reported in most of the patients (5). Our 14 patients who had 45,X karyotypes showed all of these findings phenotypically.

Mosaic cases are the second most common chromosome constitution in Turner syndrome (5). Most of them are 45,X/46,XX karyotypes (10.9 %) which we observed in 3 of our 22 patients. 46,X,i(Xq) and 45, X/46,i(Xq) mosaic karyotypes constitute 18 % of all Turner patients (6). Patients with an i(Xq) are like classic 45,X patients clinically. One of our cases had a pure 46,X,i(Xq) and one had 45,X/ 46,X,i(Xq) mosaic karyotype. Both of them showed typical Turner features.

Deletions of X chromosome make up 6 % of all Turner patients (6). Patients with terminal Xp deletions have somatic traits characteristic of the Turner syndrome, whereas gonadal function is generally preserved (2). More extensive Xp deletions, extending to the proximal region of p11, are associated in most cases with a complete Turner syndrome phenotype, including gonadal dysgenesis (7). One of the cases, a 14 year old girl, had an interstitial deletion whose breakpoints occured at Xp11.2 and p11.4 with a loss of intervening segment and had ovarian dysgenesis, short stature and other Turner stigmata as a classic Turner case. A large deletion of Xq, with the breakpoint at or proximal to q21, produces gonadal dysgenesis with primary amenorrhea and in half of the cases, somatic Turner features (2). One of our cases had a 46,X,del(X)(q21-qter) karyotype. Phenotypically,the 8 year old girl did not show any Turner stigmata. She had short stature

To sum up , many investigators have been interested on different karyotypic findings and their relation with patients' phenotypes, in Turner

syndrome. If it could be established that a given phenotypic abnormality was consistently associated with a given chromosomal deletion, it would follow that the missing segment carriers the genes responsible for the phenotypic feature in question (8). And finally, it is suggested that complex features studied, such as height, are certainly influenced by many genes and also by environmental factors.

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