

CYTOGENETIC STUDY OF 366 AFFECTED CHILDREN WITH DOWN'S SYNDROME IN IRAN

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Abstract

Down's syndrome, or 21 trisomy, is the most common autosomal abnormality, with incidence of 1 per 815 live births in Iran. Worldwide reports indicate that about 95% are regular trisomy, or nondisjunction, 1% are mosaic and 4% due to translocation. However, these values show a geographical variation.

A cytogenetic study of Down's syndrome, or trisomy 21, was carried out on 366 cases (202 males and 164 females) during a 10 year period (1974-1983) in Akbarabadi maternity hospital in Tehran. The lymphocyte chromosomes were stained with G-banding technique.

Free trisomy 21 constituted 93.44%, D/G translocation 3.55% and G/G translocation 3.01%. Reciprocal translocation and mosaic were not observed.

The values are compared with other studies in some of the countries of the region.

Introduction

Down's syndrome, or 21 trisomy, is the most common autosomal abnormality, with an average incidence of 1 per 600-800 live births in the world and 1 per 815 in Iran (3,4). Worldwide reports indicate that regular trisomy, or nondisjunction, comprises about 95% of cases, 1% of cases are mosaic and about 4% are due to translocation; 45% of the latter are inherited (4) but almost all translocations resulting DS are Robertsonian type and

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usually between the D and G group (5). However, differences have been observed in geographical distribution (3). In about one in three viable offspring of a parent carrying a balanced 21/14 translocation would be a carrier. In these parents the incidence is relatively high for the next offspring (s) (5).

The importance of cytogenetic studies on affected children, in addition to confirming the clinical diagnosis, is to determine the translocation and help in counselling the parents for having further children (2).

The chromosomal state of all children with Down's syndrome is studied in a period of ten consecutive years (1984-1993) in a governmental maternity hospital in Tehran. Being a large city, Tehran has a heterogeneous population and hence consists of mixtures of various communities and ethnic groups of Iran.

Materials and methods

An investigation was carried out on all newborns with Down's syndrome, born in Akbarabadi maternity hospital in Tehran, in the cytogenetics laboratory, during a period of ten years (1984 - 1993). Patients with insufficient data in their records were omitted from the study. 366 patients (202 boys and 164 girls) were investigated first clinically by a pediatrician and then referred to the cytogenetic laboratory. The cell culture was prepared by standard method of peripheral white blood cells and then stained by G-banding method.

The chromosomes were analysed to find out if they were the regular trisomy 21, mosaic or translocated type; if the latter was observed, the type of translocation was determined.

Results and discussion

366 patients (202 boys and 164 girls, with the M/F of 1.23) affected with Down's syndrome, 98% under the age of 5 years, were studied. 75% of the families lived in Tehran, the rest being from various parts of the country.

As the table 1 shows, 342 (93.44%) had regular trisomy and 24 (6.56%) had Robertsonian translocation, of which, 11 cases were G/G type and 13, D/G

type ; mosaicism was not observed. While the sex ratio in G/G translocation shows no difference (M/F = 1/1) , the D/G constelotion shows higher ratio (M/F=2/1). Because of the limited sample size (D/G=13) , an exact interpretation of the male increase in this phenomenon is not possible.

Other studies in the region , close to our findings , include that in India with 93% regular trisomy and 4% Robertsonian translocation (6) , in Kuwait 96.2% regular , 1.9% Robertsonian and 1.4% mosaic (1).

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Table 1- Incidence and percentages of various types of trisomy 21 in 366 children in Tehran.

Chrom. Constitution	Total		Male		Female		M/F
	No	%	No	%	No	%	
G;G	11	3.01	5	-	6	-	1/1
D;G	13	3.55	9	-	4	-	2/1
Total Trans	24	6.56	14	6.93	10	6.10	1.5/1
Regular Tri.21	342	93.44	188	93.07	154	93.90	2/1.5
Total	366	100	202	100	164	100	2/1.5

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