Chromosomal Studies in Cleft Lip And/Or Palate (*)

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Cleft lip and/or palate is an entity created by both hereditary and non-hereditary factors, thus constituting a heterogenous group in nature. It is commonly associated with other congenital malformations, and is seen in such conditions as D- and E-group trisomy syndromes.

In this report findings are given in a group of patients with the condition with special attention to the chromosome constitutions.

Materials and Methods

There were 20 patients with cleft lip and/or palate from different parts of the Country. Their distribution with respect to the age, sex, and other relevant clinical findings are shown in the Tables. Standard 3-day lympochyte cultures were set up for each case, and the karyotypes analysed by the routine Giemsa technique.

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Observations

For relevant data pertaining to the patients were summarized in the Tables, only chromosome studies are to be considered.

TABLE : I

No. of Cases	Age	Sex	Clinical Finding
1	3	F	CLP
2	5	M	CLP
3	3	F	CLP
4	4	F	CLP
5	2	M	CLP
6	2	M	CLP
7	6	M	CLP
8	6	M	CP
9	4	M	CP
10	2	M	CP + Other relevant clinical findings
11	1	M	CP + Other relevant clinical findings
12	5	M	CP
13	3	F	CP
14	2	F	CP
15	4	F	CL
16	4	F	CL
17	5	M	CL
18	6	M	CL
19	2	F	CL
20	1m.	F	Median CLP and other relevant clinical findings

TABLE : II

No. of Cases	Cohromosomal Finding	Clinical Finding
1	46, xx, inv (2) - Pericentric inversion No: 2	CLP
2	46, xx, i (2p) - Isochromosome of No: 2 for short arm.	CLP
3	46, xy, - abnormalities of No: 2 and 12	CLP
4	47, xy, + 13	CP+other relevent clinical findings
5	46, xy, / 47, xy, + 13	CP+other relevent clinical findings
6	45, xx, - 18/47, xx, + 21	Median Cleft Lip and Palate + other relevant clinical findings

Out of 20 patients 14 revealed no numerical or structural chromosom aberrations. In the remaining 6 there were both structural and numerical abnormalities as fellows :

1. In one case with cleft lip and palate, modal chromosome number was reported to be 46 and sex chromosome constitution as XX. In about 50 percent of the cells so far analysed, there were 3 No. 1 chromosomes indisguishable from each other; therefore, it has been considered that a pericentric inversion of No. 2 was involved.

2. In the second patient with again cleft lip and palate, 46, XX. 2pi abnormality was found. The incidence of abnormal karyotypes was about % 50.

3. The third case with cleft lip and palate was a male with 46, XY constitution, but assymmetrical constellations of Nos. 2 and 12 were noted.

Numerical anomalies were in the following :

In 2 patients with cleft palate there occured D-trisomy in about 40 percent of the cells. These 2 cases were described as D1-trisomy mosaics. One of theme, in addition to cleft palate, showed a low-birth weight, hypertelorism, epicanthal folds, micrognathia and an abnormal ear helix.

The last case with the condition exhibited a 45, XX,-E/47, XX, + G mosaicism. In this case too in addition to the median cleft lip and palate other congenital malformations were observed: mainly, microcephaly, a low-brith weight, closed crainial sutures, a prominent occiput, short neck, epicanthus, microphthalmy on the left an abnormal tissue of about 4 to 5 cm in size on the left back, a peculiar dermatoglyphic pattern and spasticity. Her body measurements were as follows while she was 1-month old: head circumference, 27 cm; chest circumference, 31; height, 47.5 cm, and weight, 2.8 kg.

Discussion and Result

The results indicate that, although in an appreciable number of cases there are chromosome abnormalities and that the relatively homogenous clinical picture, apart from D-trisomy syndrome cases, chromosome abnormalities are not consistent in the cleft lip and/or palate. It has also been shown that chromosomal abnormalities in the condition are quite heterogenous, a result supported by different works.

It seems clear that more studies are required to establish the chromosomal basis of the cleft lip and/or palate.