Indian Journal of Plastic Surgery (1986), 19(1), pp. 8-10

CLEFT LIP : THE THREE AFFECTED SIBLINGS

J. L. Srivastava and R. P. Narayan

SUMMARY

Three siblings having cleft lip in a family are being reported. The probable mode of hereditary transmission has been discussed. The severity of deformity in the last affected sibling was found to be less than the 1st and 2nd sibling.

Small family norm is in vogue today. With limitation of family size to 2 or 3 siblings, families with more than one child with cleft lip and or palate is rare. In developing countries where family size is big, two or three affected siblings are not uncommon (Boo-chai, 1970).

Case Report

A. A., 24 years old muslim male presented to our outpatient department with his three sons, M. A. aged 4 years, S. A. aged $2\frac{1}{2}$ years, and I. A. aged 8 months, with right sided group I partial cleft, left sided group I partial cleft and left sided group I partial cleft respectively (Fig. 1). Ante-natal history was not contributory. Family history and examination revealed that maternal aunt Miss S. aged 18 years, had partial group I right sided cleft. Pedigree of the family has been shown in Fig. 2.

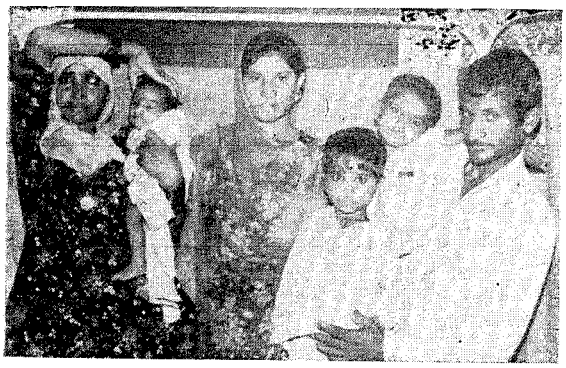

Fig. 1. Showing the three siblings and the maternal aunt having cleft lip.

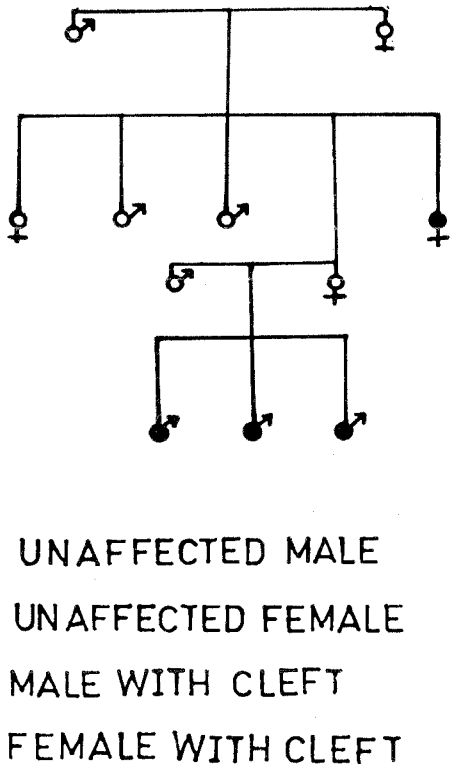

Fig. 2. Pedigree of the family.

Discussion

In a big family two or three siblings with cleft of the lip and palate are not uncommon. Pitanguy (1968) in Brazil reported a family of seven siblings in which father and five children had clefts of lip and palate. Champion (1964) reviewed 800 cases of cleft lip and palate. Eleven families had two affected siblings and three families had three affected siblings. Dave (1980)

reported a family where all the three siblings were affected. Sanui (1962) reviewed 2,532 cases of cleft lip and palate and reported eleven families having three siblings with cleft of lip and palate. The exact cause of cleft lip and palate is unknown. Hereditary and environmental factors have been blamed for it. Few environmental factors have been experimentally proved in animals. Hereditary factors also play a major role in causation of cleft lip and palate. Precise mode of inheritance of cleft lip and palate is incompletely understood but siblings having positive family history show increased susceptibility. In some families a major dominant gene may manifest itself by producing cleft lip, isolated cleft palate and fistulae of the lower lip or any combination of two, one or none of these traits (Woolf et al., 1964).

Mode of inheritance of cleft lip trait can be explained in the present case by polygenic inheritance hypothesis of Woolf et al. (1964). According to the above hypothesis a minimum number of genes (threshold) carrying traits (carrier genes) should be passed to offsprings for expression of the trait. Threshold number of carrier genes needed for expression of trait is lower in males than females. An affected female possesses more of these carrier genes on average than an affected male. The risk of transmission of carrier genes increases if the affected parent is female and likewise the risk that a couple will produce a second affected child is increased if the first affected child is a female.

Contrary to the view held by Woolf et al. (1964), Mathews (1979) expressed that hereditary transmission is usually through male sex linked recessive genes. Great concern has been expressed regarding the severity of cleft lip and palate in last affected sibling. Twenty-two cases collected from literature revealed that only in 2 families severity of expression increased in the last affected siblings, while in 11 families its severity decreased and it remained unchanged in 8 families (Table I).

Table I

Author	Year	No. of families with 3 affected siblings	Severity in last affected siblings		
			De-creased	Un-changed	In-creased
Sanui	1962	11	5	5	1
Champion	1964	3	3	0	0
Boo-chai	1969	6	3	2	1
Dave	1978	1	0	0	1
Present study	1985	1	0	1	0
Total		22	11	8	3

Conclusion

We firmly endorse the above hypothesis. In the reported family carrier genes were contributed from maternal side (Maternal aunt of child had group I cleft of lip). The unaffected mother of siblings could not pass on the trait due to less number of carrier genes than threshold, but passed it to her offsprings. All the offsprings being male, expressed the trait due to lower threshold of expression in males.

REFERENCES

1. Boo-chai, K. : Cleft of the lip and palate—third affected sibling. Brit. J. Plast. Surg., 1970; 23 : 50.
2. Champion, R. : In "Early treatment of cleft lip and palate". Ed. Horz, R., Berne : Huber, 1964; 234.
3. Dave, S. : Cleft of the lip and palate : The third affected sibling. Ind. J. Plast. Surg. 1978; 11 : 77.
4. Mathews, D. N. : In "Plastic surgery in infancy and childhood". Ed. Mustarde, J. C., Churchill, Livingstone, 1979; 1.
5. Pitanguy, I. : In Craniofacial anomalies. Ed. Longacre, J. J., Philadelphia: Lippincott, 1968; 167.
6. Woolf, C. M., Woolf, R. M. and Broadbent, T. R. : Cleft lip and hereditary. Pl. Reconstr. Surg., 1964; 34 : 11.

The Authors

DR. J. L. SRIVASTAVA, M.S., M.Ch. *Head*, Deptt. of Burns, Plastic & Maxillofacial Surgery, Safdarjang Hospital, New Delhi.

DR. R. P. NARAYAN, M.S., M.Ch., *Senior Resident*, Deptt. of Burns, Plastic & Maxillofacial Surgery, Safdarjang Hospital, New Delhi.

Request for Reprints

Dr. R. P. Narayan, 1878, Laxmi Bai Nagar, New Delhi-110 023.