Maxillofacial dysostosis¹

VICTOR ESCOBAR, JOHN EASTMAN, DAVID WEAVER, AND MICHAEL MELNICK

From the Departments of Oral-Facial Genetics and Medical Genetics, Indiana University School of Medicine, Indianapolis, Indiana

SUMMARY Four individuals in a single family affected with maxillofacial dysostosis are reported. Maxillary hypoplasia, delayed onset of speech, and poor development of language skills without associated hearing loss are the main characteristics of the syndrome which is transmitted as an autosomal dominant. Cephalometric analysis and speech and hearing evaluation of our patients confirmed the above findings.

Maxillofacial dysostosis is a rare autosomal dominant disorder characterised by maxillary hypoplasia, delayed onset of speech, and poor development of language skills without associated hearing loss. Occasional findings are down slanting of the palpebral fissures and minor auricle abnormalities. To our knowledge, there have been only three published reports on this condition: Villaret and Desoille (1932) as a 'primary familial hypoplasia of the maxilla', Peters and Hövels (1960) as a variant of mandibulofacial dysostosis (Treacher Collins syndrome), and Melnick and Eastman (1976) who delineated the phenotype as a distinct syndrome.

In this report we present a new family in which the mother and three of her children show the characteristic triad of maxillofacial dysostosis, providing further support that this condition is a distinct entity and needs to be differentiated from other first and second branchial arch syndromes.

Report of a family

The family pedigree is shown in Fig. 1 and the physical findings of the affected individuals (Figs. 2-3) are listed in the Table. All other physical findings are within normal limits, including height, weight, and occipitofrontal circumference. Audiological and speech evaluations showed dysarthria in the absence of hearing loss in all affected individuals. The three affected sibs show a receptive and expressive language development at a level 2 to 3 years below that appropriate for chronological age. The Stanford-Binet

Fig. 1 Family pedigree.

intelligence scale disclosed normal IQ for all family members.

As one might expect with an autosomal dominant trait, the cephalo-metric findings showed some variation from affected person to affected person. Nevertheless, all affected individuals in this family had maxillary hypoplasia (Table) and the proband (Fig. 1, II-8) also had a small anterior cranial fossa (4.0 SD below mean for age and sex).

An older sib (Fig. 1, II-5), age 12, of the proband and her father, age 44, were also examined. They did not show any features of maxillofacial dysostosis. Further evaluation of other kindred members was not possible because of lack of co-operation.

Discussion

A review of individual cases of inherited defects has shown that many clinical varieties of any given defect

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Fig. 2 (A) Proband, notice low set ear and ptosis of eyelids. (B) II.7, Fig. 1. (C) II.4, Fig. 1. (D) Mother of the proband. Notice maxillary hypoplasia in all of these individuals.

Table Clinical findings of affected individuals

	II.8	II.7	II.4	I.2
Sex	F	F	M	F
Age (y)	5	7	14	37
Small anterior cranial fossa	+	—	—	—
Flat occiput	+	+	—	+
Maxillary hypoplasia	36.0 (48.9 ± 2.3)	37.0 (50.5 ± 2.9)	44.6 (58.7 ± 3.6)	45.9 (57.0 ± 4.4)
Flat nasal bridge	+	+	—	—
Ptosis eyelids	+	—	+	+
Down slanting palpebral fissures	—	—	+	+
Nystagmus	+	—	—	+
Strabismus	—	—	—	+
Pectus excavatum	—	+	+	—
Hypoplastic nipples	—	+	—	—
Hearing loss	—	—	—	—
Delayed onset of speech	+	+	+	+
Dysarthria	+	+	+	+
Mental retardation	—	—	—	—
Dermatoglyphic abnormalities	—	—	—	—
Chromosomes	46,XX	ND	ND	ND
Metabolic screen	—	ND	ND	ND

ND = Not done.
Numbers in parentheses represent mean for sex and age with standard deviation (Riolo *et al.*, 1974) measured from anterior nasal spine to posterior nasal spine in mm.

Fig. 3 (A) X-ray of proband. (B) X-ray of II.7. (C) X-ray of II.4. (D) X-ray of the mother of the proband. Notice maxillary hypoplasia in all of these individuals.

can occur. It has also been shown that a combination of apparently unrelated symptoms is sometimes present in those cases, for example distichiasis and chronic lymphatic oedema of the lower extremities (Smith, 1976). Of course, this combination of symptoms might only be the result of chance association, but when more than one case occurs in a family, the repetition of the combination of defects in all affected subjects rules out that possibility. Furthermore, even if the cases are sporadic, the reappearance of a similar combination of symptoms in unrelated individuals is suggestive of a distinct entity. In the family with maxillofacial dysostosis reported here a recognisable pattern of abnormalities of the face and head was observed in 4 of 7 family members in two generations. The findings included down slanting of the palpebral fissures, severe maxillary hypoplasia, and delayed onset of speech with dysarthria which was not associated with a hearing deficit. These features, with the exception of the nystagmus in two family members, are identical to those present in a father and his son

who were reported by Peters and Hovels (1960) and in three family members in three generations as reported by Villaret and Desoilles (1932). In this latter family only the father and the son were fully described; the grandfather was thought to be affected but was not examined. Similar findings were also noted in the two individuals reported by Melnick and Eastman (1977).

A number of conditions should be considered in the differential diagnosis of maxillofacial dysostosis including Goldenhar syndrome, mandibulofacial dysostosis, acrodysostosis, and acrofacial dysostosis (Nager syndrome). The epibulbar dermoids seen in Goldenhar syndrome, and the eyelid colobomas and severe mandibular hypoplasia of mandibulofacial dysostosis clearly separate these conditions from the one reported here. Acrodysostosis includes peripheral dysostosis, premature skeletal maturation in the hands, feet, and elbows, and often abnormalities of the spine (Robinow *et al.*, 1971), while acrofacial dysostosis is an autosomal recessive trait that has

absent or hypoplastic radii and/or thumbs (Gorlin *et al.*, 1976). Furthermore, speech difficulties have not been observed in any of the above conditions except for maxillofacial dysostosis.

Melnick and Eastman (1977) have recently suggested that the minimal diagnostic criteria for maxillofacial dysostosis should include maxillary hypoplasia, delayed development of speech and language skills without hearing loss, a normal or near normal intelligence, and an autosomal dominant mode of inheritance. The family reported here supports this contention.

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Requests for reprints to Dr Victor Escobar, Department of Oral-Facial Genetics, Indiana University School of Dentistry, 1121 West Michigan Street, Indianapolis, Indiana 46202, U.S.A.