Maxillonasal dysplasia, mandibular retrognathia and cleft palate

By Peter Mossey, BDS, FDS, M Orth, FFD and John Sandham, BDS, FDS, D Orth, PhD

Hypoplasia of the middle third of the face associated with congenital absence of the anterior nasal spine and depression of the nasal bones with flattened nasal alae was described by Binder[1] in 1962, and Hopkin[2] in 1963. This craniofacial deformity is known as maxillonasal dysplasia; its craniofacial effects have been further defined by McWilliam and Linder-Aronson[3]. The effects of maxillonasal dysplasia on craniofacial morphology include a smaller anterior cranial base; maxillary retrognathism; smaller maxillary length and pharyngeal airway dimension; shorter mandibular ramus and body; greater gonial angle with concave profile and flattened nasal bones together with absence or partial absence of anterior nasal spine (Fig. 1). Binder suggested the facial appearance at birth resembled arhinencephaly and he described frontal sinus hypoplasia and nasal mucosal atrophy in association with the deformity. Hopkin likened the facial appearance to a child with a cleft lip and palate and reported certain similarities. Since both these studies described findings in small samples, the descriptions were subjective assessments.

The embryological origins of first arch derivatives and cranial base and upper cervical vertebrae are similar. Resche[4] and Sandham[5] reported that subjects with maxillonasal dysplasia and subjects with cleft palate are more likely than controls to have cervical vertebral anomalies. Dahl[6] and Sandham and Cheng[7] reported a shorter clivus length in children with cleft lip and palate. Maxillonasal dysplasia may be one of a number of possible expressions of the effect of an etiological agent, and an overlap of anomalies in subjects with craniofacial anomalies would not be unexpected.

Cephalometric studies by Hopkin[2], McWilliam and Linder-Aronson[3], and Olow-Nordenram et al.[8] demonstrate similar morphological characteristics found in subjects with maxillonasal dysplasia. In these studies, the dysplasia sample showed a smaller anterior cranial base (n-s), more retrusive nasal bones (s-n-r), maxillary retrognathism (smaller s-n-ss angle), smaller

Abstract

The present paper describes the craniofacial form of subjects with maxillonasal dysplasia and reports its occurrence in two siblings who also have cleft palate and mandibular retrognathia. Maxillonasal dysplasia and a cleft deformity may be illustrative of a field effect of a teratogen on developing midface components or even suggestive of a possible inherited etiology.

This paper was submitted September, 1988.

Key Words

Maxillonasal dysplasia • Mandibular retrognathia • Cleft palate • Cervical vertebral anomalies • Teratogenic activity

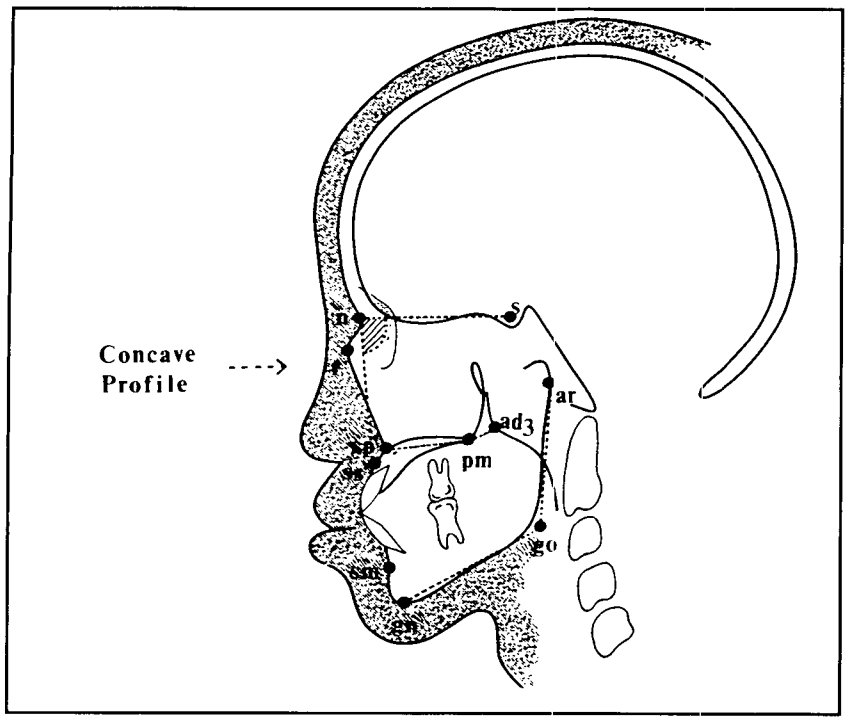


Figure 1

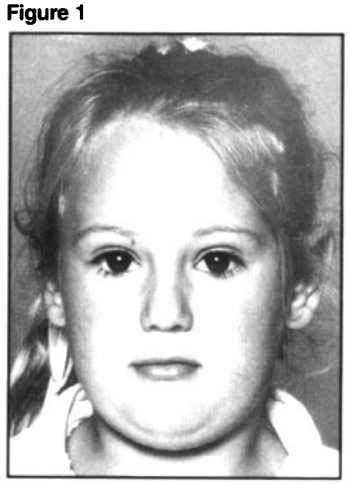


Figure 2A

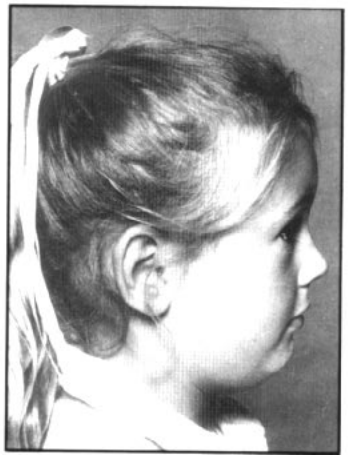


Figure 2B

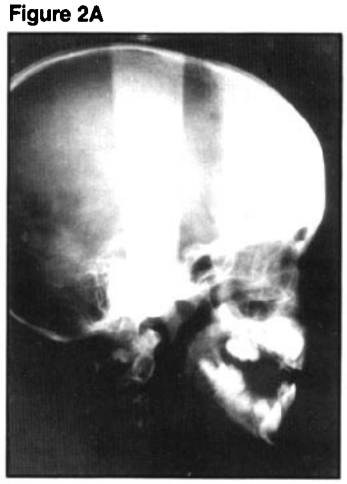


Figure 2C

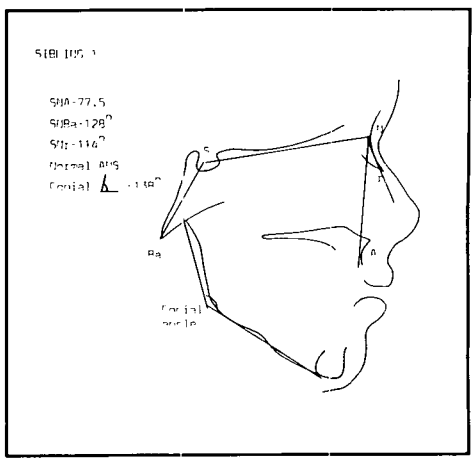


Figure 2D

maxillary length (sp-pm) and reduced pharyngeal airway dimension (pm-ad3) compared to the control group.

The effect on the mandible results in some degree of mandibular prognathism; ramus length (ar-go) and body length (go-gn) are smaller and gonial angle (ar-go-gn) is larger (Fig. 1).

The reduced midface prominence gives the appearance typical of the anomaly with a concave profile. The less protrusive nasal bones are responsible for a broad flat nose and the absent or rudimentary anterior nasal spine with resulting abnormal nasolabial muscle attachments gives rise to the acute nasolabial angle and convex upper lip. The craniofacial effects found on cephalometric radiographs in maxillonasal dysplasia are summarized in Fig. 1.

Materials and methods

The purpose of this paper is to present the facial, oral and cephalometric findings in three siblings, two of whom exhibit similar craniofacial morphology described as maxillonasal dysplasia with cleft palate and retrognathia.

Sibling 1

Sibling 1, (Fig. 2a), is a six-year, eight-month-old Caucasian female child, born on 30th December 1980, the eldest child of three in the family. The mother was 23 years old and the father 24 years old at the time of birth. Pregnancy and delivery were uncomplicated and the child was healthy with no general or craniofacial anomalies. Retrospective clinical and radiographic examination following the birth of her younger sisters revealed no abnormalities in craniofacial form. Craniofacial morphology was analyzed and parameters were found to be within the normal range for her age group (Fig. 2b).

Figure 1
Summary of the effects of maxillonasal dysplasia on craniofacial morphology when compared to a control (from results of previous studies).
anterior cranial base is smaller (n-s)
maxillary retrognathism (s-n-ss')
relative mandibular prognathism (ss-n-sm)
smaller maxillary length (sp'-pm)
smaller pharyngeal airway dimension (pm-ad3)
mandibular ramus (ar-go) and body (go-gn) are shorter
gonial angle is greater (ar-go-gn)
concave profile
convex upper lip
flattened nasal bones (s-n-r smaller)
absence or partial absence of anterior nasal spine
acute nasolabial angle with nasal groove at junction of nose and upper lip.

Figure 2
Sibling 1:
A. Full face showing normal appearance
B. Right profile
C. Lateral skull radiograph — anterior nasal spine present and normal craniofacial morphology
D. Cephalometric tracing demonstrating normal craniofacial parameters

Sibling 2

Sibling 2, (Fig. 3), is a four-year-old Caucasian female infant born on the 23rd of June, 1983. She is the second child born to a 26-year-old mother, after 40-plus weeks gestation. The pregnancy was uncomplicated, the only drugs prescribed were iron and folate at eleven weeks. Birth was by non-traumatic spontaneous vertex delivery. The newborn infant experienced some respiratory difficulty and was described as having "an odd-looking face" at birth. The orofacial features described were micrognathia, glossoptosis, cleft of the soft palate and a small nose. She also had a sacral dimple at the base of the spine but on examination no cardiovascular or other congenital abnormalities were noted.

A diagnosis of Pierre Robin syndrome was made and the infant was nursed prone to avoid respiratory complications. With no further notable problems she progressed satisfactorily and was discharged after eight days. Chromosomal analysis using trypsin banding revealed an apparently normal female karyotype (46,XX).

On subsequent review she progressed satisfactorily with apparent mandibular catch-up growth and her cleft palate was repaired at 13 months. Careful monitoring of deciduous dental development revealed maxillary lateral incisor eruption at 10 months, preceding maxillary central incisor eruption by approximately three months. Cephalometric analysis carried out at three years six months is shown in Fig 3b.

Sibling 3

Sibling 3, (Fig. 4), a 15-month-old child, the third child of the same parentage, was born on 12th May 1986 by spontaneous vertex delivery after 38+ weeks gestation. The pregnancy was uncomplicated — the only drugs ingested being iron and folate at 12 weeks. The mother was 29 years old, the father 30 years old at the time of birth.

The child had micrognathia, a U-shaped cleft of the soft palate, a small tongue and oral cavity, and experienced some respiratory difficulty. A diagnosis of Pierre Robin syndrome was made.

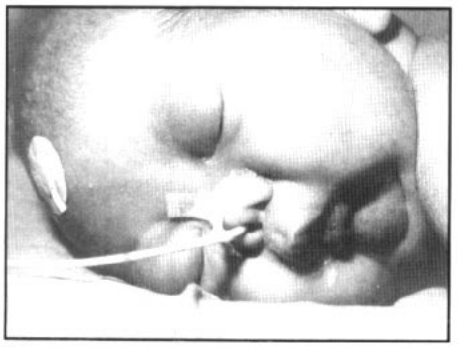


Figure 3A

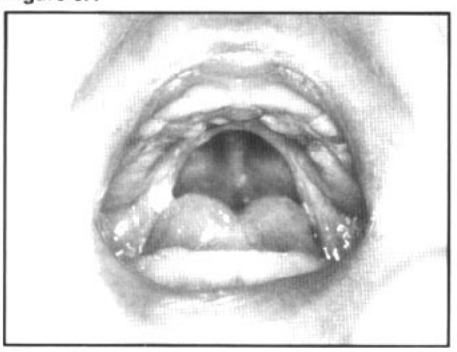
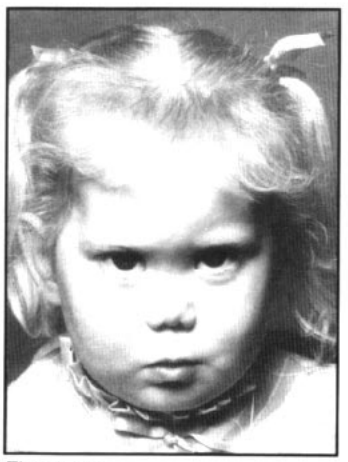


Figure 3B



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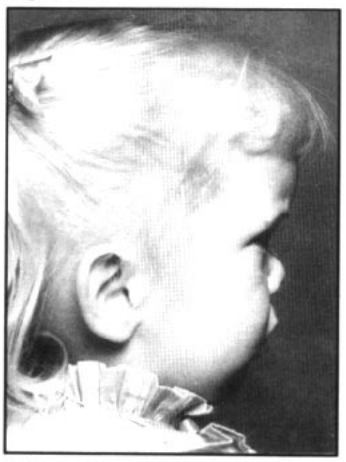

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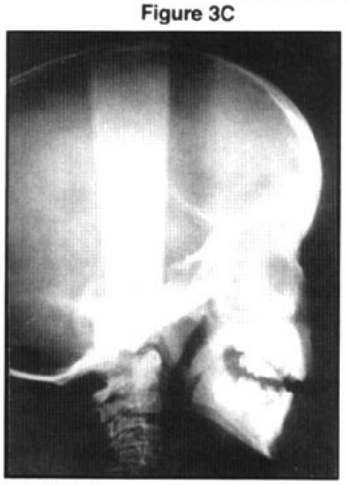

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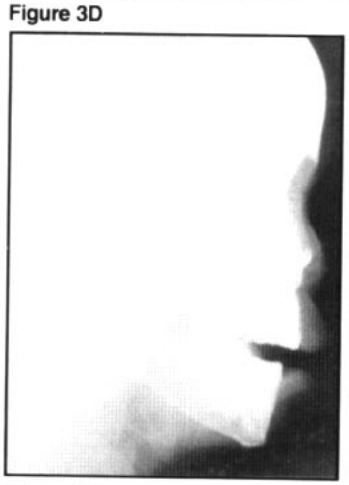


Figure 3F

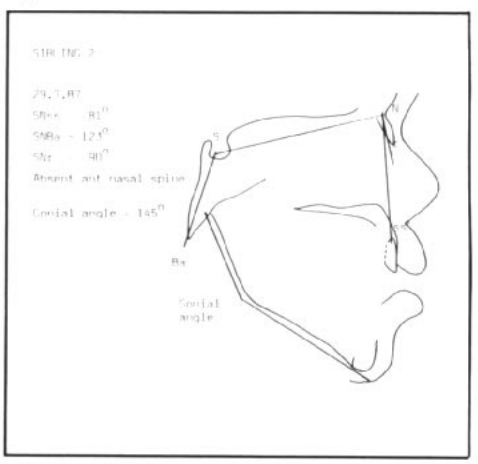


Figure 3G

Figure 3
Sibling 2:
A. Neonate with retrognathia and midface retrusion (a nasogastric tube used for feeding)
B. Palatal cleft at 3 months of age
C. Full face at 15 months
D. Right profile at 15 months showing soft tissue morphology typical of maxillonasal dysplasia
E. Lateral skull radiograph, absence of anterior nasal spine and obtuse naso-frontal angle (s-n-r)
F. Lateral skull radiograph demonstrating soft tissue profile
G. Cephalometric tracing showing salient features of craniofacial morphology

In cases of absence from anterior nasal spine, ss' point represents the point of greatest convexity on the outline of the anterior maxillary alveolus between the nasal floor and the upper central incisors.

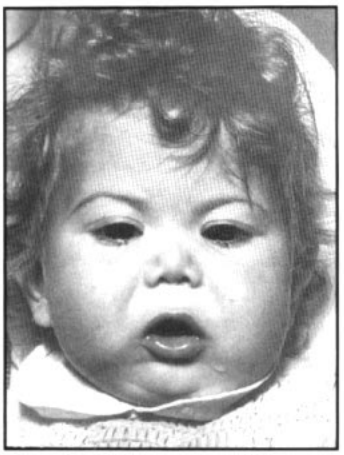


Figure 4A

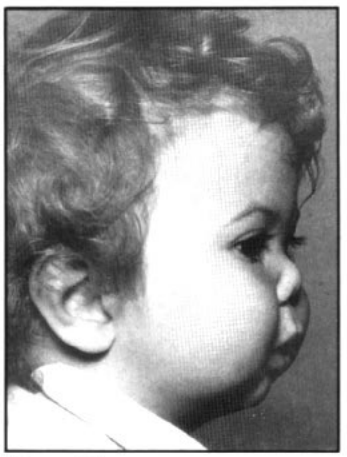
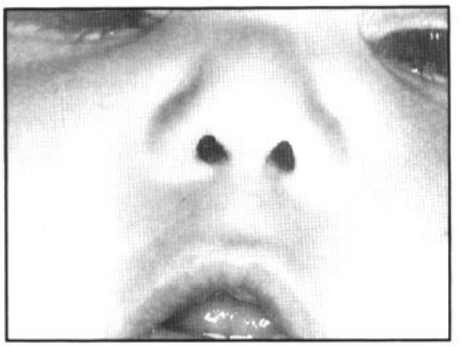


Figure 4B



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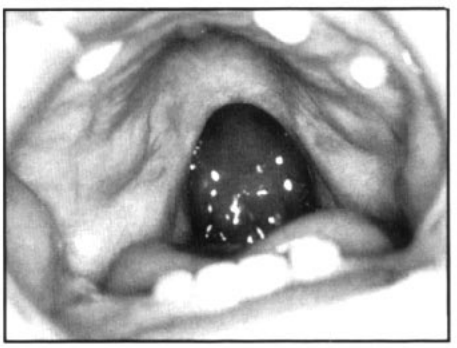
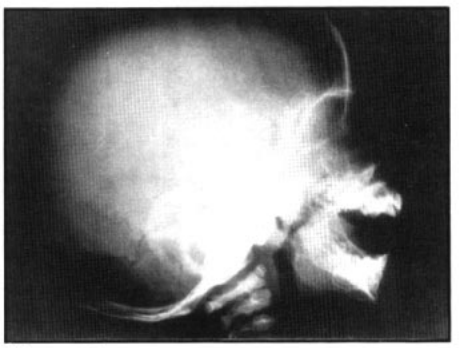


Figure 4D



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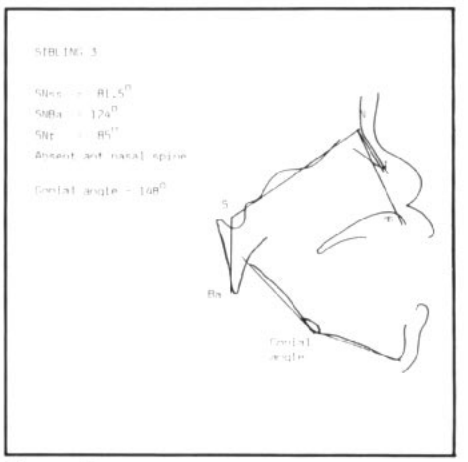

Figure 4F

Figure 4
Sibling 3:
A. Full face at nine months showing soft tissue morphology typical of maxillonasal dysplasia
B. Right profile at nine months
C. Palatal cleft
D. Nasolabial morphology in maxillonasal dysplasia
E. Lateral skull radiograph — absence of anterior nasal spine and obtuse nasofrontal angle (s-n-r)
F. Cephalometric tracing

She also had a wide, full anterior fontanelle and was described as significantly hypotonic with regard to her general musculature. Transient neonatal jaundice was described but no other abnormalities were noted.

She experienced some respiratory and feeding difficulties and was nursed prone in a cradle and fed with a naso-gastric tube. She continued to be hypotonic and contracted aspiration pneumonia and remained in hospital care for almost nine weeks. Blood sent for chromosomal analysis showed an apparently normal female karotype (46,XX) using trypsin banding.

A three-month follow-up examination revealed a slightly hypotonic child but apart from her micrognathia and cleft palate her physical condition was otherwise normal.

Mandibular catch-up growth progressed satisfactorily and her cleft palate was repaired at 14 months. Fig. 4b shows a cephalometric tracing from a radiograph taken prior to repair of the cleft palate. Observation at review appointments revealed a similar sequence of eruption of the deciduous dentition to that of her elder sister whereby deciduous central incisor eruption lagged behind the eruption of the lateral incisors.

Discussion

The etiology of maxillofacial dysplasia is unknown, Ferguson and Thompson[9] suggested the evidence of inheritance is inconclusive. The present study of three siblings suggests an inherited factor may be involved, but what is more interesting is the associated cleft palate and mandibular retrognathia. Two subjects in the study had respiratory distress at birth and were diagnosed as Pierre Robin syndrome with glossoptosis.

Morphological studies by Sandham[5] demonstrated an association between a clefting deformity and cervical vertebral anomalies. A study by Resche[4] demonstrated an association between maxillonasal dysplasia and cervical vertebral anomalies. The present association between maxillonasal dysplasia and mandibular retrognathia does seem to indicate that both maxillary and mandibular components derived from the first branchial arch may be affected, although the mechanism is not clear. Cohen[10] postulated that the Pierre Robin syndrome could be more accurately described as a sequence, the cleft palate being secondary to the retrognathic mandible. However, more recent evidence, notably the clinical investigations of Rintala et al.[11] and animal research by Edwards and Newall[12] contradict this, and suggest instead a simultaneous failure of normal development of both maxillary and mandibular components in the Pierre Robin syndrome.

Irrespective of the speculative role of the tongue in the palatal clefting of the cases

presented, the siblings undoubtedly demonstrated hypoplasia of maxillonasal and mandibular components.

The evidence presented here would seem to lend support to the aforementioned work of Rintala[11] and Edwards and Newall[12], in that simultaneous maxillary-mandibular agenesis may be an essential ingredient for the Pierre Robin syndrome; the presentation of two consecutive siblings suggests the possibility of a genetic rather than an environmental etiology.

Embryologic studies show that the cartilagenous cranial base, nasal septum, nasal capsule and upper cervical vertebrae are all derived from upper somites in the embryo; the occipital, prechordal and upper cervical somites, and the induction process in these structures occur simultaneously at the beginning of the fifth week of intra-uterine life. Hence, if subjected to teratogenic activity, the whole midface complex, cranial base and upper cervical vertebral components are vulnerable to anomalous development; the earlier the teratogenic effect on the differentiating embryonic cellular components, the greater may be the possible deformity.

A study of human embryos from six- to 12-weeks post conception by Diewert[13] showed that before elevation of palatal shelves, the rapid amount of facial growth produced a four-fold increase in facial length, and a two-fold increase in height. Teratogenic activity during this time may exert a field effect on developing facial components, with a greater or less morphological defect. An example of this is fetal alcohol syndrome. Alcohol is one of the most commonly known teratogens; exposure may result in microcephaly, hypoplastic maxilla and cleft palate[14]. It is interesting to note that a study of subjects with fetal alcohol syndrome by Tredwell et al.[15] showed that 53 percent had cervical vertebral anomalies.

If the teratogenic activity exerts its effects at an earlier stage, an extreme effect on first and second branchial arches is manifest, as in octocephaly, where mandible, maxilla and tongue may be absent, with extreme micrognathia and ventromedial displacement of external ears.

Although some very valuable work has been done by Olow-Nordenram to investigate the possible role of heredity, further investigation is required and data is at present being collected from subjects with maxillonasal dysplasia to determine whether a true familial incidence can be expected.

Acknowledgement

I should like to thank Dr. Andrew Sandham of Edinburgh University Dental School for his help with the manuscript.

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