DYSOSTOSIS MANDIBULO-FACIALIS

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Dysostosis mandibulo-facialis (d.m.f.) is the name of a complex of symptoms described by Franceschetti and Zwahlen in 1944. It is a congenital malformation which manifests itself within the region derived from the first and second branchial arches and the first branchial cleft and pouch. As a result of the abnormal development of the bones of the face, the ears and the orbits, those afflicted have a bird-like appearance. For the patient and for otologists the hearing impairment often present is also of the greatest interest. Franceschetti and Zwahlen were not the first to observe this malformation, but they were the first to make a thorough study of it. It is, therefore, often designated as Franceschetti's syndrome. In the literature one also finds the name Treacher Collins' syndrome, after an English physician who reported two cases in 1900. As early as 1846, however, Thomson had described the malformation in considerably greater detail; hence the name Treacher Collins' syndrome seems hardly appropriate. Thomson described three cases with "local malformation of the ear, considerable imperfection in the shape of the lower part of the face", and with "the lower jaw remarkably short, its position oblique and its angle very obtuse; the malar bone was placed far back on the cheek and the zygomatic arch much shorter than usual". He described the familial occurrence of the malformation and believed that the concurrent difficulty in hearing was "related to the imperfect development of some of the parts connected with the middle and external ear". He regarded the hardness of hearing in one of his cases as an indication of "a very imperfect state of the auditory apparatus". He also described the absence of a zygomatic arch and stressed the great resemblance among the individual cases.

Besides dysostosis mandibulo-facialis, Franceschetti's syndrome, Treacher Collins' syndrome, and the descriptive "bird face", other names have been proposed but not generally accepted. To us the name dysostosis mandibulo-facialis (d.m.f.) seems to be the most appropriate. There are relatively few cases of d.m.f. described. Franceschetti found about 150 cases published up to 1959, the major part of these had been reported during the last 15 years. For this increased interest the Swiss authors

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Franceschetti *et al.*, and Nager *et al.* (1948) are largely responsible. Their papers contain thorough descriptions of many of the cases published earlier.

Symptomatology

Following the original descriptions of Franceschetti *et al.*, the chief signs associated with this syndrome are:

1. Palpebral fissures sloping downward laterally ("antimongoloid"), with a coloboma in the outer portion of the lower lids and more rarely in the upper lids.
2. Hypoplasia of the facial bones, especially the malar bones and the mandible.
3. Malformations of the external ear, and occasionally of the middle and inner ears.
4. Macrostomia, high palate, abnormal position and malocclusion of the teeth.
5. Blind fistulae between the angles of the mouth and the ears.
6. Atypical hair growth in the form of tongue-shaped processes of the hairline extending towards the cheeks.
7. Eventually other associated anomalies, such as facial clefts and skeletal deformities.

Present Investigation

The motivation for this study came from the examination of a patient with typical d.m.f., who came to the E.N.T. department at the Sahlgrenska Hospital in Göteborg because of impaired hearing. Since it is known that the disease sometimes has a hereditary character, a careful study was made of the members of the family. This investigation disclosed several cases of a similar malformation. Two cases from another family were also included in the study. The members of the two families were given a uniform examination including:

A medical history with special reference to abnormal conditions during the pregnancy.

Examination of the face and eyelids.

Oto-rhino-laryngological status.

Audiogram.

Rœntgenogram of the skull and the facial bones. In cases with otitis and infantile, sclerotic mastoids, the study was completed with special X-ray investigations of the ears. In four cases the ears were examined by tomography.

Photographs of face, ears and eyes.

It would have been of interest to make an analysis of the whole material from a psychiatric, ophthalmological and odontological point

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of view, but that was not possible. All of the subjects were asked about their education. All had completed elementary schooling but none had undertaken further studies.

Results

The present material contains ten cases of d.m.f. Of these, eight are members of family "A" and two of family "B" (see Fig. 1).

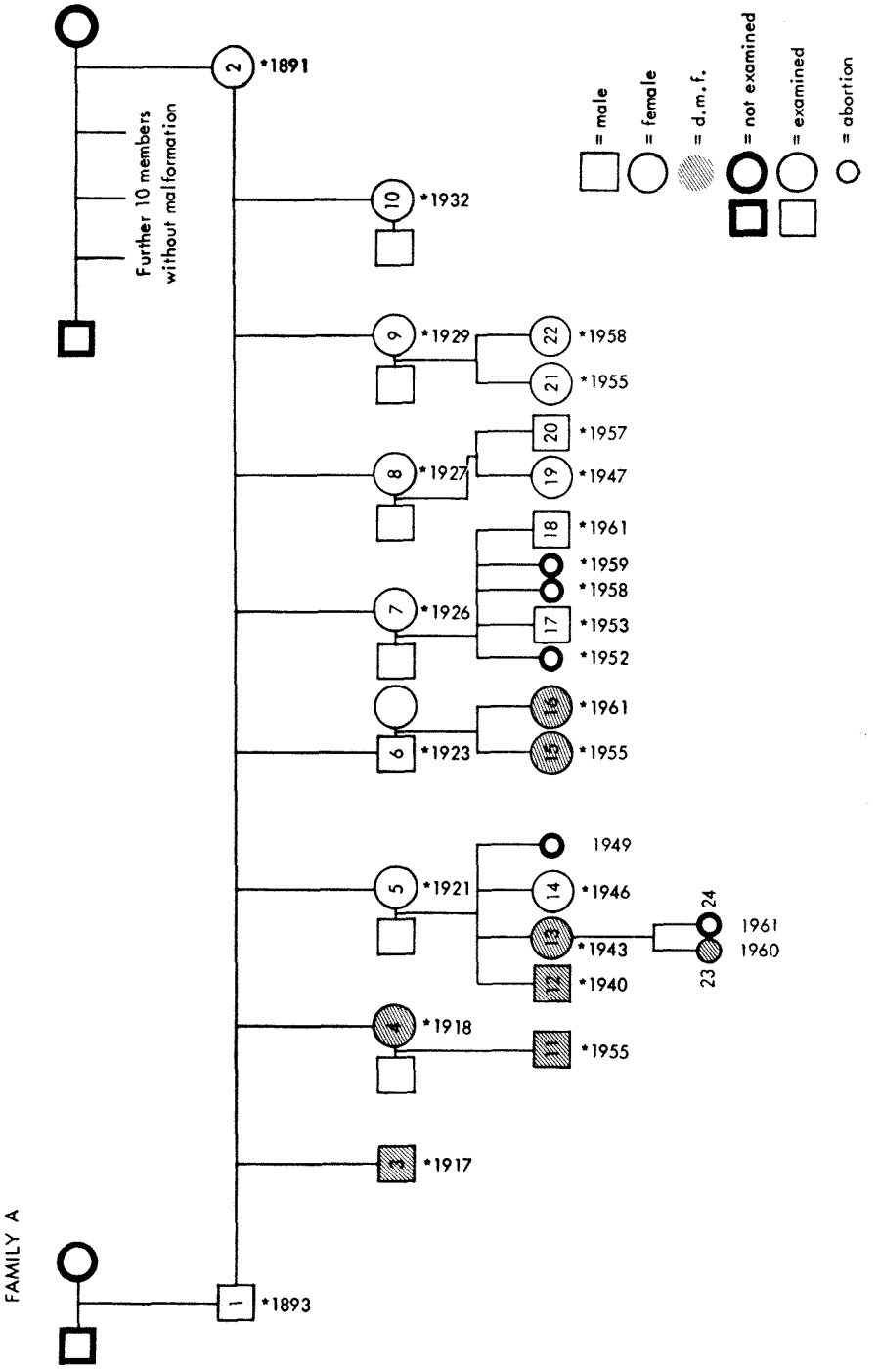
There are few reports concerning the heredity of d.m.f. (Thomson, 1846, Debusmann, 1940, Leopold *et al.*, 1945, Straith *et al.*, 1949, Roussel, 1951, Hunt *et al.*, 1955). The present eight cases in one family must be considered of great interest from this viewpoint. Furthermore, it was possible to get extensive information about the family, and every member of four generations, even the members by marriage, were examined as described above. Thus not less than 30 members of the family were included in the investigation. In the other family d.m.f. was present in two children. As their mother had suffered a recurrent erythema nodosum during the pregnancies as well as other times, this family is also of a certain interest.

An attempt was made to find persons who represented a transitional stage between the "healthy", "normal", members and those with d.m.f. Many slight abnormalities were found among the "normal" members, but no clearly transitional forms. The family-trees of the two families are given in Fig. 1 where the subjects examined, and those with a well-defined syndrome, can easily be recognized.

During the course of the investigation, attempts were made to ascertain whether the malformation was derived from the paternal or the maternal side in the first "healthy" generation. Resemblances between the family-members with d.m.f. and their respective parents were considered. Old photographs of the members of the first generations of each family, their parents and siblings were studied. No conclusions could be drawn from these photos. Nevertheless in family "A" some members of the second generation, e.g. those siblings who were not afflicted with d.m.f., showed a resemblance to each other and to the mother. The distribution of the cases of d.m.f. can be seen in Fig. 1. In the first generation neither of the parents was afflicted. In the second generation, two siblings out of eight had d.m.f., and in the third generation five out of 12. The only representative of the fourth generation, a fœtus, showed the malformation. There seems to be an increase in the incidence of d.m.f. with each generation. It would be of great interest to follow up this family and see what happens to further members of the fourth generation. Until then nothing can be said with certainty about the pattern of heredity.

Since d.m.f. can be the result of traumatic processes occurring during pregnancy, information on this possibility was sought in taking the

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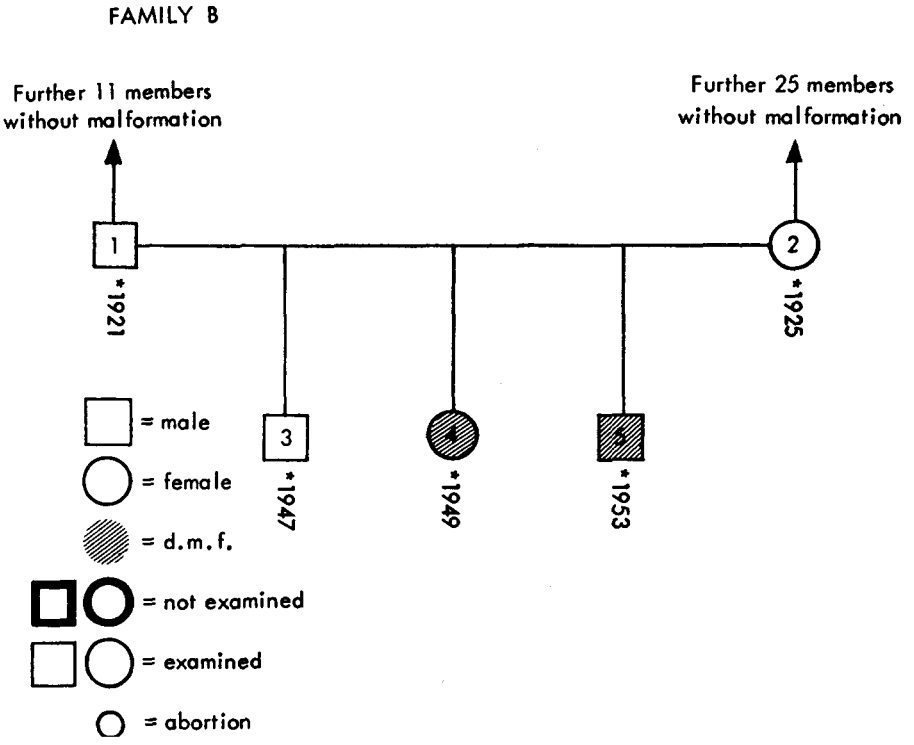


FIG. 1.

The family-trees of the two families studied.

medical histories, but in no case was any such evidence brought out. In the second generation of family "A" one woman had a legal abortion and another three spontaneous ones. Another woman in this generation has not become pregnant after 6 years of marriage in spite of an expressed desire for children. The only member of fertile age in the third generation had one legal abortion and one miscarriage. Delivery was usually uneventful but one boy without d.m.f. was born in a breech delivery. Another boy with d.m.f. (A11) was delivered by cæsarean section, performed because of a placenta previa.

The positive cases have shown typical and pronounced malformations. These findings of the positive cases in both families are compiled in Table II. Slight differences in both degree and type of abnormality are seen in individual cases. In one case the eye malformations were not very prominent, but all other signs were clear. As can be seen from Table II, many of the cases showed abnormalities of the lower eyelids. All of the ears that have been operated had a malformation of the stapes. These are two of the symptoms that will be discussed below.

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Table No. 1

Dysostosis mandibulo-facialis

Occurence of malformations in the head

	Obligatory	Frequent	Infrequent
Face and skull	Hypoplasia of malar bones Hypoplasia of mandible	Hypoplasia of maxilla, nasal septum and nasal sinuses Preauricular hair prolongation Preauricular fistulæ Malformation of tempo-romandibular joint Accentuation of digital impressions of skull	Malformation of nose and absence of nasofrontal angle Choanal atresia Hypoplasia of sella turcica Malformation of styloid process Hypertelorism
Mouth		Malformation of teeth and malocclusion Macrostomia ? (see discussion) Arched hard palate	Palatoschisis
Ear		Malformation of auricle, external auditory meatus, middle ear and ossicles Pneumatization of the mastoid small	Malformation of inner ear Hypoplasia of squamous part of temporal bone Malformation of middle ear muscles
Eye	"Antimongoloid" palpebral fissures Anomaly of lower lid: Coloboma Malformation of eyelashes	Atresia of lacrimal puncta of lower lids External ankyloblepharon	Coloboma of upper lids Absence of glands of Meibohm

Since the description of the symptomatology made by Franceschetti, some of the symptoms mentioned by him have come to be regarded as less important, while others have been emphasized. For this reason we have grouped the symptoms in a somewhat different manner (Table I).

There are several members without d.m.f. who have shown minor

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abnormalities in regions usually affected by this syndrome. These findings in family "A" are as follows:

	(□ *man*, O *woman*)
In the first generation: (2) members without d.m.f. Ages 69 and 71 years)	Lobes of the auricles attached (O). Big and low situated auricles (□). Arched narrow hard palate (□). Small palpebral fissures (□). Verrucas in the face (O).
In the second generation: (6 out of 8 family members without d.m.f. Ages 33-45 years)	Lobes of the auricles attached (□OOOO). Pronounced caries or no teeth (OOOOO). Changes of the drum membrane, probably on the basis of middle-ear suppuration. (Only one ear showed hearing impairment) (OOO). Deviation of the nasal septum (O□O). Malformation of the bite (OOO). Arched narrow hard palate (OO). Small and sclerotic mastoids (O□). Malformation of the pharyngopalatine arch on one side (O). Anomaly of the eyelashes (O).
In the third generation: (7 out of 12 family-members without d.m.f. Ages 1-15 years)	Anomalies of the bite (□OOOO). Anomalies of teeth implantation (OO). Malformation of the auricles (□O). A strikingly arched hard palate (O). Pronounced convolutional markings of the vault of the skull (□).

Family "B", consisting of parents and three children, contained two cases of d.m.f. Findings in these patients are also recorded in Table II. Case B5, a boy of nine years, with a very pronounced d.m.f. was the only one who showed any malformation of the eyeball. He had a hyperopia of 4-5 diopters on both eyes. The three members without d.m.f. showed the following recorded abnormalities:

The father had a syndactylia between toes and a deviation of the nasal septum.

The mother had had eight attacks of recurrent erythema nodosum, one of which occurred during the later part of each pregnancy. She showed attached lobes of the auricles, rather narrow external auditory meatuses and a slight prognathia.

The eldest son had a pre-auricular fistula.

There were no malformations known in family "B" in two earlier generations.

Discussion

Nomenclature

A single malformation, independent of its cause, can manifest itself within any of the regions of actual interest for d.m.f. (Hövels, 1953). Several simultaneous malformations within these regions may also appear. When Franceschetti and Klein carefully analysed these combined malformations, they found it possible to distinguish patients comprising a special, rather well-defined group and used for this syndrome the name d.m.f.

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Table No. 2

Present material

Symptoms in 10 cases with d.m.f.

		Family member No.									
		A3	A4	A 11	A 12	A 13	A 15	A 16	A 23	B4	B5
	Mothers age at delivery	26	27	37	19	22	32	38	17	24	28
	Pregnancy abnormal	-	-	-	-	-	-	-	-	+	+
	Delivery abnormal	-	-	+	-	-	-	-	+	-	-
		r. l.	r. l.	r. l.	r. l.	r. l.	r. l.	r. l.	r. l.	r. l.	r. l.
Face and skull	Malformation of malar bone	+ +	+ +	+ +	+ +	+ +	+ +	+ +	+ +	+ +	+ +
	Hypoplasia of mandible	+	(+)	+	+	+	+	+	+	+	+
	Malformation of nose	-	-	-	-	+	-	+	+	-	+
	Deviation of nasal septum	+	-	-	+	+	-	-		+	-
	Hypoplasia of nasal sinuses	-	-	-	-	- -	- -			- -	+ -
	Accentuation of digital impressions of the skull	-	-	(+)	(+)	+	+	-		+	+
	Prolongation of preauric. hair	- -	- (+)	+ +	+ +	+ +	- -	- -		+ +	- +
	Preauric. fistulae	+ -	- -	- -	- -	- -	- -	- +	- -	- +	+ -
	Malformation of temporomandibular joint	- -	- -	- +	+ +	+ +	+ +		- -	- -	- -
Mouth	Malformation of teeth up	-	+	+	-	+	-	0	0	-	+
	down	-	+	+	-	+	-	0	0	-	+
	Malocclusion	-	(+)	+	-	+	+	0		(+)	+
	Arched hard palate	-	-	+	+	-	-	-		+	+
	Malformation of epipharynx	+	-	+	-	+	-	?		+	+
	Dental caries pronounced	(+)	+	+	+	+	+	0		+	+
Ear	Malformation of auricle	- -	- -	+ +	+ +	+ +	- +	+ +	+ +	- -	- -
	Lobe of auricle attached	+ +	+ +	- -	+ +	+ +	+ +		+ +	+ +	- -
	Atresia of external meatus	- -	- -	- (+)	+ +	+ +	(+) +	+ +	+ +	(+) (+)	+ +
	Abnormal drum membrane	+ +	(+) +	+ +	+ +	+ +	- -		+ +	+ +	+ +
	Malformation of middle ear at operation	+			+ +	+ +	+		+ +		
	Malformation of malleus				+	+			+ +	(+) (+)	
	Malformation of incus	+			+ +	+ +	+		+ +		
	Malformation of stapes	+			+ +	+ +	+		+ +		
	Hearing impairment	+ +	- -	+ +	+ +	+ +	+ +	+ +		- -	+ +
	Pneumatization of mastoids small	+ +	+ +	+ +	+ +	+ +	+ +			- +	+ +
Eye	Typical obliquity of palpebral fissures	+ +	- -	(+) +	+ +	+ +	+ +	+ +		+ +	+ +
	Coloboma of lower lids	+ +	- -	- +	+ +	- +	+ +	- +	+ +	- -	+ +
	Malformation of eyelashes	+ +	+ +	+ +	+ +	+ +	+ +	+ +		+ -	- +
	Absence of lacrimal puncta on lower lids	- -	- -	+ +	+ +	+ +	- -	- -		- -	+ -
	External ankyloblepharon	+ +	+ +	+ +	+ +	+ +	+ +			+ +	+ +

In this group seven chief signs could often be recognized. Persons with these signs were called "complete or typical or standard types of the syndrome". These seven signs have been described earlier. Our results support the notion that a diagnosis of d.m.f. should be made only when the afflicted person shows all of the typical signs of the malformation.

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Dysostosis Mandibulo-Facialis

When Franceschetti and Klein introduced the concepts "incomplete", "abortive", "unilateral" and "atypical" forms of d.m.f., great difficulties arose in the differentiation of facial malformations. If we agree upon the nomenclature of typical or standard types of d.m.f., we must also acknowledge that incomplete forms and varieties may occur in which the distinction from other conditions may be difficult. We would recommend, therefore, that d.m.f. should only be used in those cases where a complete syndrome is present. To us it appears extremely difficult to differentiate d.m.f. from other malformations if sub-forms are also accepted. For the sub-forms we find it much more appropriate to use a descriptive anatomical designation as advocated by Hövels and Morlinghaus (1957). We would prefer to differentiate between:

Unilateral	Isolated	malformation of the face
	Combined	,, ,, ,, ,,
Bilateral	Symmetrical isolated	,, ,, ,, ,,
	Symmetrical combined	,, ,, ,, ,,
	Asymmetrical combined	,, ,, ,, ,,

With such a nomenclature, much more precise information is given and vague descriptions such as "abortive", "incomplete", etc., are avoided. The absence of clear definitions has already resulted in the publication of many cases as d.m.f. where the classification is incorrect or faulty. As d.m.f. has received increasing attention in recent years the nomenclature problem seems to be of great importance and uniformity in definition is essential.

Symptomatology

When Franceschetti and Klein described d.m.f. they had at their disposal a rather small number of patients. After a new syndrome such as d.m.f. is reported it is quite natural that some of the "typical" signs may lose, while others may gain, importance. Furthermore, new signs may be detected and added to earlier information. Based upon descriptions given in the literature and our own findings we have grouped the signs of the head regions as shown in Table I.

In cases of d.m.f. additional malformations may occur in other regions (Morlinghaus, 1955). The most frequent of these are located in the vertebral column, in the extremities and in the thorax. Oligophrenia is also often described.

The most frequent signs presented in the literature and observed by us have been brought together in the "obligatory" group.

Combined with these signs several others are frequently present. Some of these need further discussion and they are taken up as they appear in Table I.

First we wish to stress that the malformation of the zygomatic region consists not only of a hypoplasia of the malar bones but also of defects

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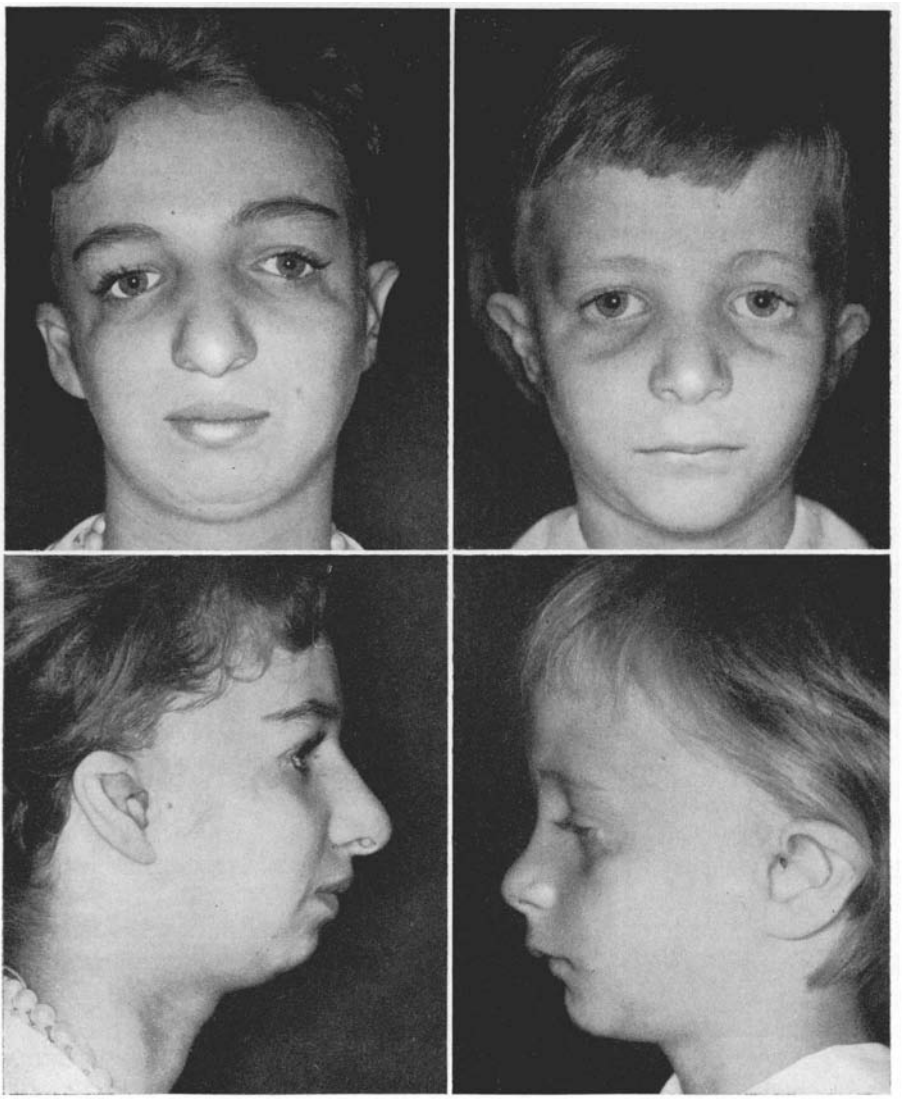


FIG. 2.

Two cases of dysostosis mandibulo-facialis from family A. Note especially malformations of palpebral fissures, auricles, and mandibles, and on the left pre-auricular prolongation of the hair.

in the zygomatic arches. This results from a non-fusion of the temporal process of the malar bone with the zygomatic process of the temporal bone. The hypoplasia of the malar bone gives an oblique position to the whole orbit and thus contributes to the "antimongoloid" palpebral fissures.

Reports on the conditions of the eyelids and particularly the eyelashes

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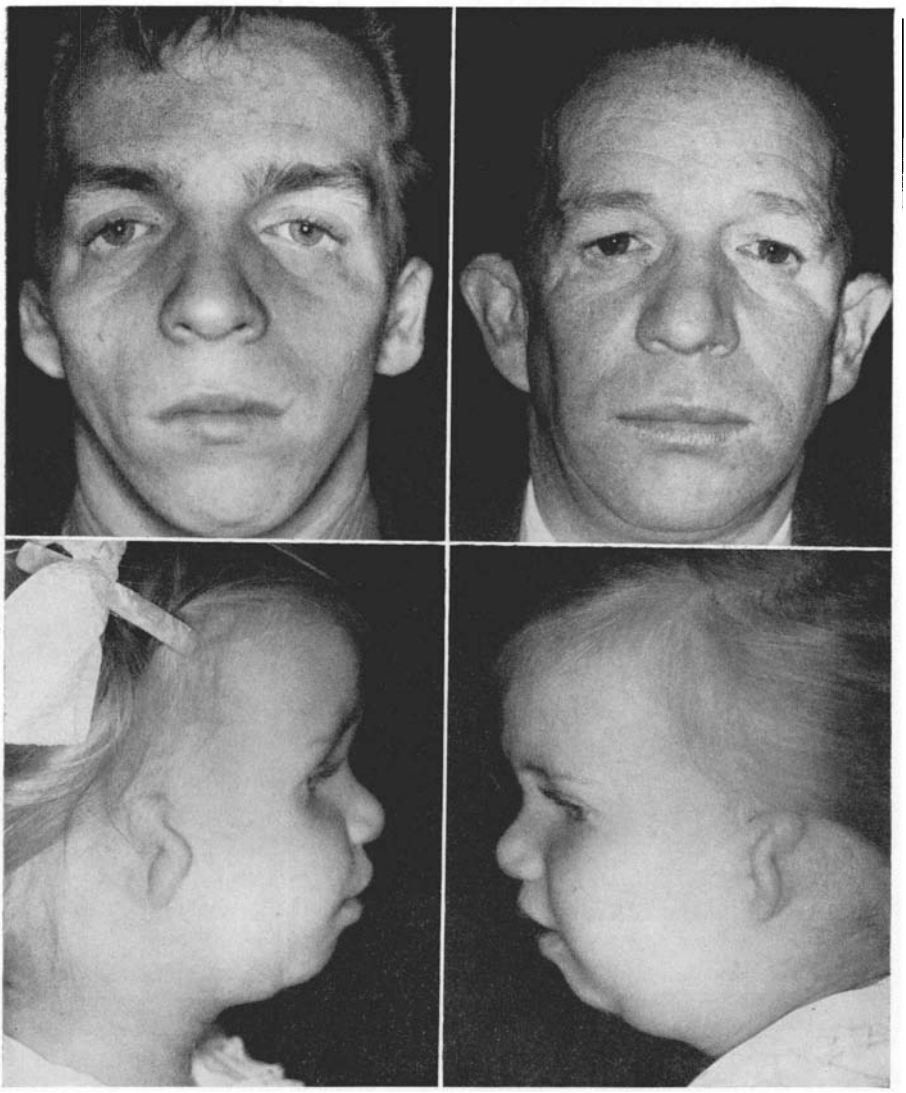


FIG. 3.
Three cases of dysostosis mandibulo-facialis from family A. Note especially the malformation of the auricles on both sides in the child.

have usually been omitted. If mentioned they have been considered to be underdeveloped. In the present material we always found small, irregular, tufty lashes.

The convolutional markings of the vault of the skull were pronounced in the children with d.m.f., a sign of increased intracranial pressure

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which may also be present in normal children. In no case we have found any other such signs as suture diastasis or change in the sella turcica.

As can be seen from the Table I we list hypoplasia of the nasal sinuses among frequent additional malformations. This depends upon the fact that previous authors have reported it as a frequent symptom (Franceschetti and Klein, 1949, Granrud, 1953, Wieland, 1954, Pavsek, 1958, Plester, 1961). In the present material we have only found a hypoplasia in one of the cases with d.m.f. As three of the patients with d.m.f. were below 7 years of age and the "normal" size of the sinuses shows a great variation in children, it is difficult to estimate what is a hypoplasia. No pronounced reduction of size was found.

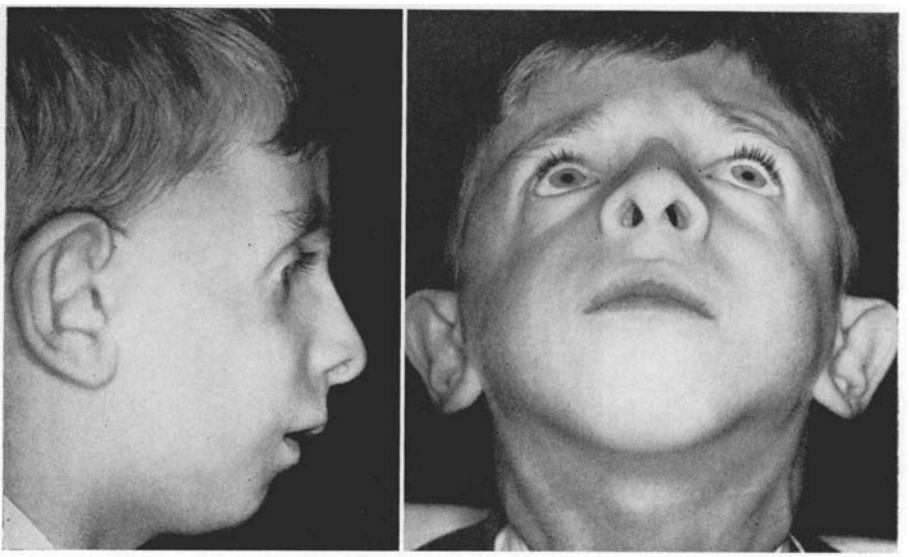


FIG. 4.

A case of dysostosis mandibulo-facialis from family B. Note especially hypoplasia of the malar regions, the external ankyloblepharon, and the pre-auricular fistula on the right.

As shown above, all the ears on which operations were performed in the present material have a malformation of the middle ear and the ossicles. There are few previous reports on the condition of the middle ear in d.m.f. In our material the drum membrane and the middle ear have often been diminished in size. The drum membrane may be replaced by a bony plate. The malleus and the incus are often fused into one single bone. The long process of the incus is hypoplastic and continues in a deformed and hypoplastic stapes, which is fixed in a narrow oval window. A malformation of the stapes is described in a few cases (Rüedi, 1954, Altmann, 1955, Harrison, 1957, Plester, 1961, Herberts, 1962). The edge of the footplate and the margin of the oval window are not so distinct as normally,

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but the appearance is different from that in otosclerosis. Similar findings have been reported earlier. The only normal stapes described was found in one of Herberts' cases, where, however, there was a malformation of both incus and malleus. As a consequence of the malformation of the ossicles the patients have a hearing impairment. On the basis of a narrow epipharynx, which is often present, patients with d.m.f. may suffer a recurrent otosalpingitis. This is another explanation to the existing hearing impairment (Aubry *et al.*, 1953). A malformation of the eustachian tube is seldom reported. Modern methods for the examination of the function of the middle ear will be of great help in revealing different kinds of disorders and will thus be helpful in planning the operation.

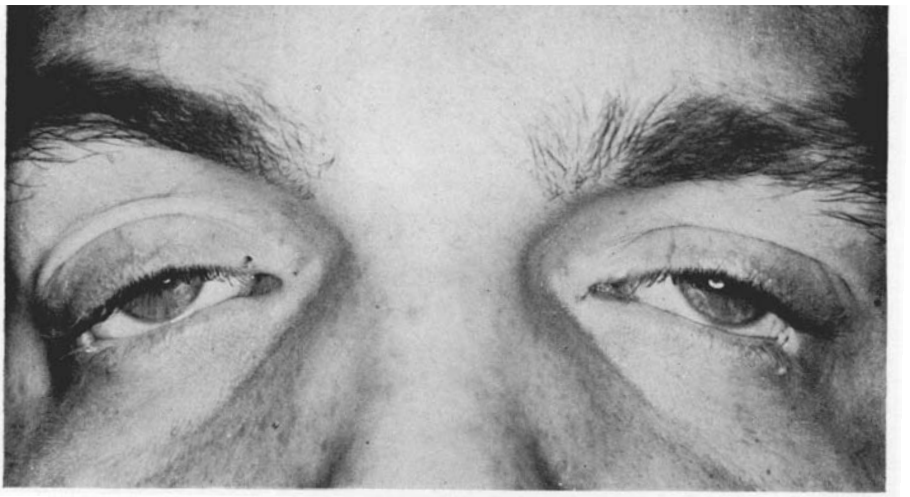


FIG. 5.

The typical outward-slanting eyes ("antimongoloid" palpebral fissures) and the malformation of the lower lid in a case of dysostosis mandibulo-facialis.

Franceschetti and Klein reported "occasional" malformations of the inner ear. In the present material the investigation has not disclosed any definite cases of inner-ear defect. Deafness has been described in the literature in very few cases (Holm, 1948, Campbell, 1954). Most authors have found a coloboma of the lower lid in the complete form of d.m.f. However, in some of the cases reported this symptom was not present (Roussel, 1951, Briggs, 1953, Plester, 1961). Even in the present material the sign was missing in a few cases. We have therefore recorded the coloboma of the lower lid as one possibility under the obligatory category: anomaly of the lower lid. In our opinion coloboma of the upper lid is a very rare finding in d.m.f. Previous authors often recorded a symblepharon in d.m.f. This term means an attachment between the eye-

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lids and the eyeball. The condition in d.m.f., however, corresponds to an external ankyloblepharon which designates an abnormal adhesion between the upper and lower lid laterally. It is possible that previous authors used the term symblepharon to refer to adhesion between the lids. In our opinion the abnormalities of the eyelids are definitely more pronounced on the lower lids and are more frequent than previously pointed out. Of the chief signs mentioned by Franceschetti and Klein, the macrostomia has not been very prominent in our material. Indeed, it is very

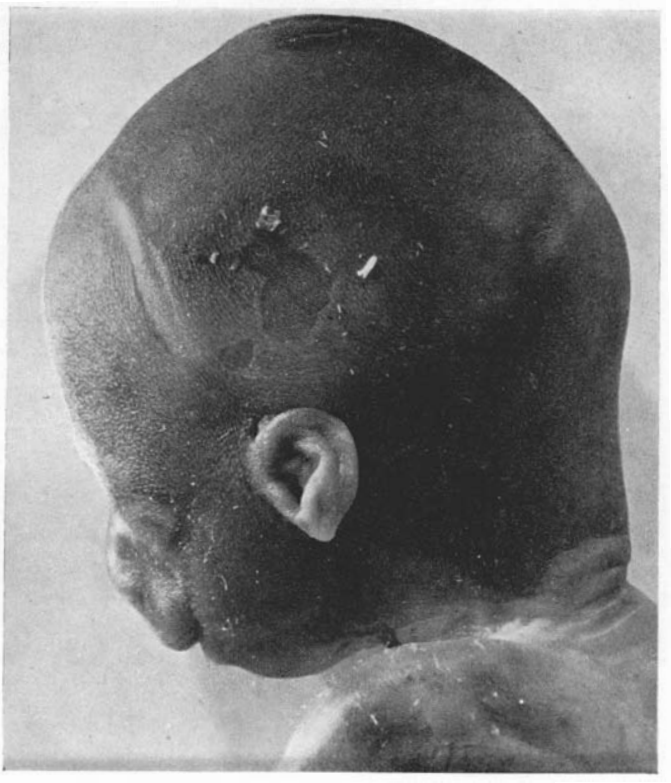


FIG. 6.
Fœtus (25 cm.) showing clear signs of dysostosis mandibulo-facialis.

difficult to define the word macrostomia. If there is an underdevelopment of the lower part of the face, a normal-sized mouth may appear very large. Individual variations in mouth size may also be pronounced.

Among the symptoms of d.m.f., the absence of a naso-frontal angle was not very often described but was indicated by some authors. (Van Lint and Hennebert, 1936, Franceschetti and Klein, 1949, Roussel, 1951, Hurvitz, 1954). After surveying the literature and the present material we have not found that this symptom belongs to the more common ones.

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The present material shows a very high incidence of caries among persons afflicted with d.m.f. There can be several explanations for this. Endogenic factors such as malformation of teeth, of dentition or a malocclusion may make them more vulnerable to caries. An exogenic cause for the high incidence of caries may be that children with d.m.f. are given too many sweets as consolation. Of some interest is the fact that several of the "normal" members of the families presented minor malformations. Whether these different abnormalities could be related to d.m.f. can not be decided. Another interesting observation is that the frequency of

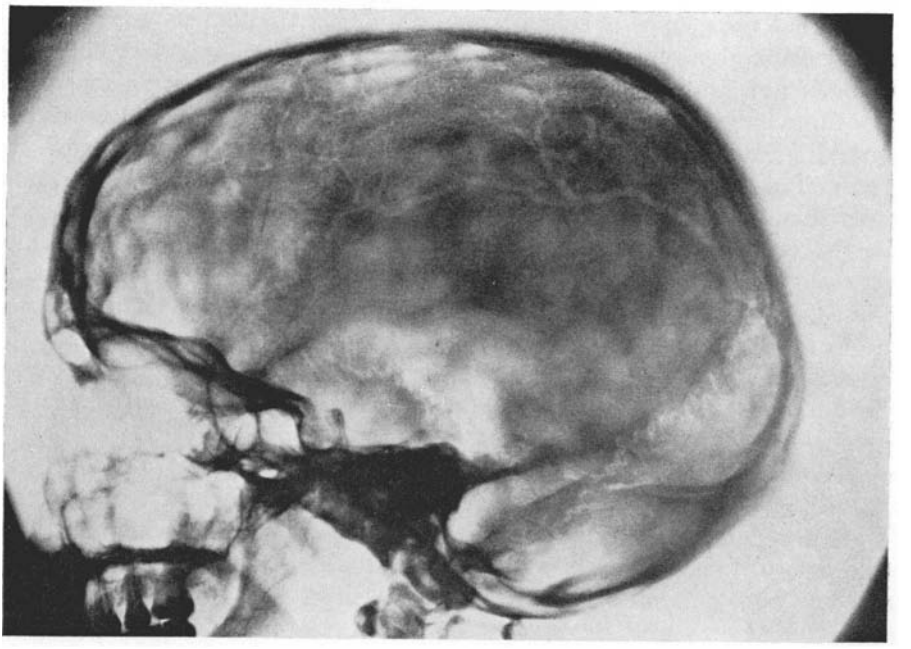


FIG. 7.
The convolutional markings of the skull in a case of dysostosis mandibulo-facialis, 18 years of age.

d.m.f. seems to be increasing with every generation in family "A". A suggestion could be made that in a hereditary malformation such as d.m.f. the frequency and the severity of the disease may increase until a level is reached when the embryos can no longer survive.

Ætiology

Families have been described with a high frequency of d.m.f., as well as single cases without known heredity. When Franceschetti and Klein surveyed the literature in 1949 they found seven cases in which the family history showed d.m.f. to have been present in several genera-

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tions. Since then further cases have been reported (Roussel, 1951; Grönvall, 1953; Hunt *et al.*, 1955; Harrison, 1957; Herberts, 1962). The present material includes eight cases in one family with a complete d.m.f. syndrome, and thus supports the theory that the malformation is sometimes hereditary and appears to follow an irregular form of dominant transmission (Leopold *et al.*, 1945; Pavsek, 1958). Furthermore, there are some cases described where an exogenous trauma during the first stage of pregnancy is clearly established (Granrud, 1953). It is well known that exogenous noxious influences during the first 3 months of the pregnancy can give rise to malformations in the fœtus. It is also recognized that the groups of cells which are under actual development are the most vulnerable ones. Different exogenous injuries applied at the same stage of fœtal development can give rise to the same malformations while the same exogenous trauma applied at different developmental levels can give different malformations. Such potentially traumatic factors include: thermal and mechanical injury, irradiation, anoxia, chemical poisoning, nutritional and hormonal changes, infections and circulatory disturbances in the embryo. Granrud presented six cases in which he was able to demonstrate such exogenous factors in five and heredity in only one. In family "A" no such factors are known. In family "B" in which there was no history of d.m.f. the mother showed an erythema nodosum during the later part of the pregnancies. Here the possibility of an exogenous factor cannot be excluded as the cause of the malformation.

Treatment

D.m.f. is of great importance for the social adaptability of a person. By his peculiar appearance with the small mandible, his deformed ears and outward slanting eyes, the patient will already attract attention as a child. The combination of the malformations with a possible hearing impairment may give the impression of a lack of intelligence. It is, therefore of great importance that these patients should be provided with the benefits of plastic surgery and reconstructive ear surgery as early as possible (Beck, 1925). The most feasible plastic operations are those aimed at correcting higher degrees of underdevelopment in the mandible and the auricles. In cases with total aplasia of the auricles in the male sex, probably the greatest improvement is obtained with prosthesis of the auricle, but corrective surgery may also give good results. Plastic procedures aimed at correcting colobomas and the antimongoloid palpebral fissures, as well as the external ankyloblepharon, may give considerable improvement in appearance.

Abnormalities in the shape of the nose and the nasal septum can be readily corrected, but such operations are usually not performed until late adolescence. Dental malformations and malocclusions should be taken care of in early childhood.

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Other malformations, e.g the hypoplasia of the malar bones, pre-auricular fistula and hair prolongations etc., seldom require surgical intervention. In cases of d.m.f., malformations of the middle ear and hearing impairment are frequent. The malformed middle ear requires, on the whole the same kind of surgical intervention as other microtic ears. On this subject there is a rich literature (Pattee, 1947; Ombrédanne, 1951; House, 1953; Rüedi, 1954; Theobald, 1954; Holmes, 1954).

In spite of the fact that patients with d.m.f. generally seem to have a well-functioning inner ear, there are very few reports of good hearing results after operation (Rüedi, 1954; Wieland, 1954; Harrison, 1957; Plester, 1961; Herberts, 1962). In our material a middle-ear exploration has been the first step in the operations planned. It has disclosed a malformation of the ossicles, including the stapes, in six ears of four patients, who all belonged to family "A". In this family autopsy of a fœtus also showed a bilateral malformation of the stapes. In several of our cases a hypoplasia of the long process of the incus has been found. It is almost lacking or replaced by a rudimentary fibrous strand. In such cases it is necessary to build a columella between the malleus and the oval window. In one of our cases this has been done following stapedectomy with a considerable improvement in hearing. In this case of bilateral severe hearing loss, the post-operative hearing reached the 20 db. level.

We therefore consider it advisable to explore the middle ear in order to determine how the hearing loss may be corrected.

In those cases where the exploration of the middle ear shows anatomical conditions not permitting satisfactory sound transmission even after middle-ear surgery, a fenestration operation should be the method of choice. Such an operation may, however, be both difficult and sometimes unrewarding because of complicated anatomical conditions.

As a concluding remark it may be stated that d.m.f. forms a considerable handicap for the afflicted person. By the use of modern plastic and middle-ear surgery some of this handicap can be reduced, but many problems concerning the heredity and ætiology of the disease are still unsolved. To prevent the disease it will be necessary to solve these and other related problems.

Summary

Ten cases of dysostosis mandibulo-facialis are presented. The findings are grouped according to their frequency and described individually. The familial nature of the disease is considered and family trees are shown for the two families in which the ten cases were found. Since d.m.f. often results not only in a deformity of the face, but also in a malformation of the middle ear combined with hearing impairment, the possibility of improving the hearing by surgery is discussed. Patients with

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malformations of the face who are also hard of hearing are in great need of both plastic reconstruction and middle-ear surgery.

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