A Comparison of Microtia and Temporal Bone Anomalies In Hemifacial Microsomia and Mandibulofacial Dysostosis

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A number of entities can be categorized as *otocraniofacial syndromes*. New clinical and laboratory studies have demonstrated predictable patterns of occurrence, distinct anatomic interrelationships, separate genetic predispositions, and animal models of the varied embryogeneses. These investigations have allowed clinical separation of first and second branchial arch anomalies into syndromes of *hemifacial microsomia* and *mandibulofacial dysostosis*. The present study has established a relationship between the severity of the *microtic auricle* and *middle ear* malformation in those syndromes. Middle ear deformities, while present in both, are more severe when associated with mandibulofacial dysostosis.

Introduction

Recognition of the patterns of occurrence and interrelationships among the deformed structures is necessary for comprehensive clinical management of hemifacial microsomia and mandibulofacial dysostosis. Microtia, a deformity common to both syndromes, is the most clinically apparent malformation, and usually has been studied solely from an otologic, reconstructive, or radiologic perspective, depending upon the area of special interest of the investigator. Consequently, there has been little information about the presence or absence of malformations in structures contiguous to the ear and derived from common embryologic tissue. Furthermore, microtia and its attendant middle ear malformations have been considered devoid of specific syndrome characteristics. Earlier pilot investigations have correlated and catalogued the temporal bone anomalies within each syndrome (Caldarelli, In Press, and Hutchinson, 1977).

This study was done to determine if the temporal bone anomalies in hemifacial microsomia and mandibulofacial dysostosis have specific patterns of occurrence, anatomic characteristics, and degrees of severity common to each syndrome. The first to be considered is hemifacial microsomia.

The hallmark of hemifacial microsomia is its varied clinical expressivity as evidenced by the spectrum of microtia, mandibular deformities, and middle ear malformations (Caldarelli, 1977). The term "hemifacial microsomia" encompasses closely related syndromes that comprise anomalies of the first and second branchial arch derivatives, including otomandibular dysostosis, unilateral mandibulofacial dysostosis, unilateral facial agenesis, and necrotic facial dysplasia. Also included is a probable variant of hemifacial microsomia known as oculoauriculovertebral dysplasia, or Goldenhar's syndrome. This variant is distinguished by associated eye findings such as epibulbar dermoids and colobomata—usually of the upper lid, microphthalmia, microcornea, iris atrophy, and polar cataracts.

Usual manifestations of hemifacial microsomia are a constellation of congenitally anomalous facial structures derived exclusively from the first and second branchial arches and the primordia of the temporal

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bone. In its complete expression, an affected individual manifests unilateral, and infrequently bilateral, mandibular and maxillary hypoplasia, macrostomia, microtia, external auditory canal atresia or stenosis, and various middle ear malformations (Figure 1).

A large number of patients with hemifacial microsomia have been studied clinically and radiographically in order to correlate and catalog the anomalies of first and second branchial arch derivatives (Caldarelli, In Press). Radiologic examination has been essential in delineating these subclinical malformations. In particular, polytomography of the temporal bone and mandible has been paramount in helping to catalog the varied expressivity of these malformations (Petasnick, 1973, and Valvassori, 1963).

To arrive at a rationale for treatment, multidisciplinary investigation of 70 patients with hemifacial microsomia was undertaken to map the variations encountered and develop a pattern of interrelationships among the facial malformations in hemifacial microsomia. Microtia was the clinical marker used to ascertain if the graded severity of the microtic auricle can accurately predict the severity of the mandibular deformity and the status of the ossicular chain and external auditory canal. In addition, the correlation between the severity of the mandibular deformity and the status of the ossicles and its external auditory canal was also determined.

Conventional radiography is limited in evaluating malformations of the temporal bone, except in defining the degree of development of the mastoid. The frequent absence of radiolucencies of the external auditory canal and middle ear cavity, the presence of dense atretic bone obscuring the superimposed structures, and finally the distortion in axis and location of certain temporal bone structures usually make the interpretation of conventional radiography impossible. Therefore, because of these limitations, temporal bone tomography is the ideal tool for assessing subtle temporal bone anatomy.

Tomography should be performed in at least two projections, frontal and lateral, with sections one or two millimeters apart. Semiaxial sections for study of the oval window and horizontal sections are added when necessary (Valvassori, 1963).

Seventy patients were studied. Eighty-five ears manifested varying degrees of microtia. The external ear deformity was graded as originally described by Meurman (1957). The grading system is as follows (Figure 2): Grade I—The auricle is distinctly malformed and is of smaller size than a normal outer ear but shows most of the characteristic features of the auricle. Grade II—The rudimentary auricle consists of a low, oblong elevation, hook formed at the cranial end corresponding to the helix. Grade III—The auricle is more defective, showing only a part, often a mal-

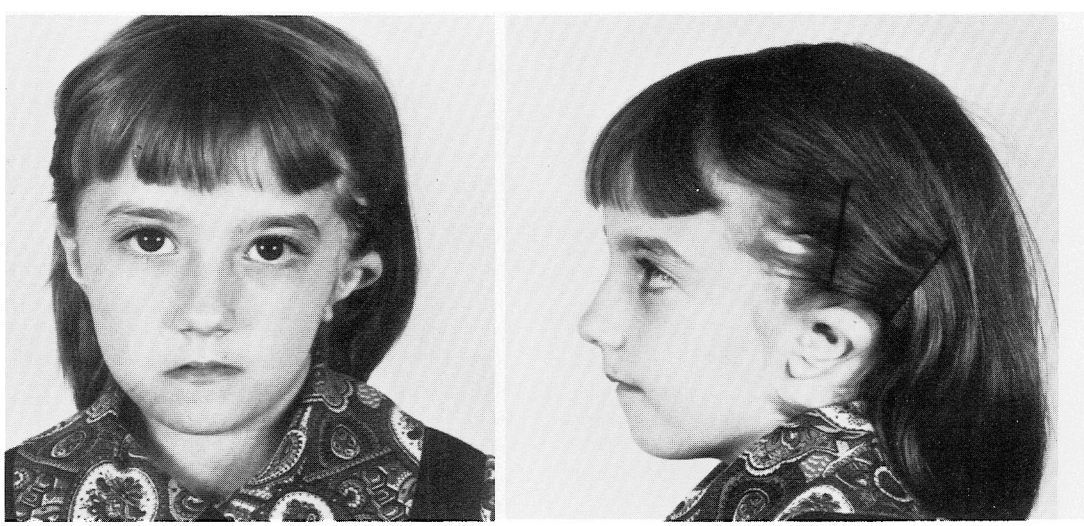

FIGURE 1. Hemifacial microsomia. Note facial asymmetry with hypoplasia of maxilla and mandible, and microtia.

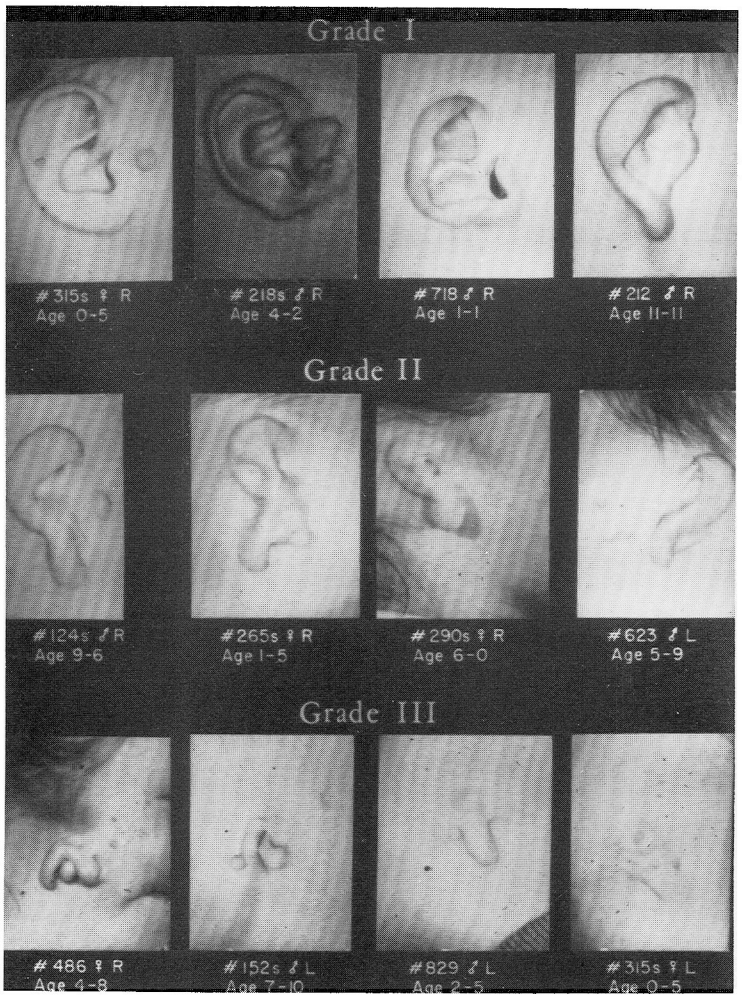

FIGURE 2. Graded spectrum of microtia encountered in hemifacial microsomia.

formed lobule, while the rest of the pinna is totally absent. Three ears were not included because accurate classification was precluded by poor auricular reconstruction.

Polytomographic analyses of frontal and lateral temporal bone sections of each patient were made with particular attention to classification of first branchial arch derivatives as reflected by ossicular (malleus/incus conglomerate) and external auditory canal deformities. The external auditory canal was deemed abnormal when it was either stenotic or absent.

Grading of mandibular deformities was determined by tracings of the cephalometric tomograms of both halves of the patient's mandible. These were superimposed upon each other and graded according to severity from I to III as illustrated in Figure 3 (Pruzansky, 1969). In grade I, the difference was one of size. The grade II mandible retained the essential morphologic characteristics of the ramus but with considerable distortion in form. The most severe grade III ranged from complete loss of the distinguishing features of the ramus to its complete absence.

Ossicular chain anomalies and external auditory canal deformities were individually matched to the severity of microtia and gradation of mandibular deformities. The radiographic representations of temporal bone malformations in this study are by no means a complete inventory of all malformations encountered. Only those structures considered most reliably apparent radiographically and thought to best represent first branchial arch derivatives were tabulated. The stapes suprastructure, a second branchial arch deriva-

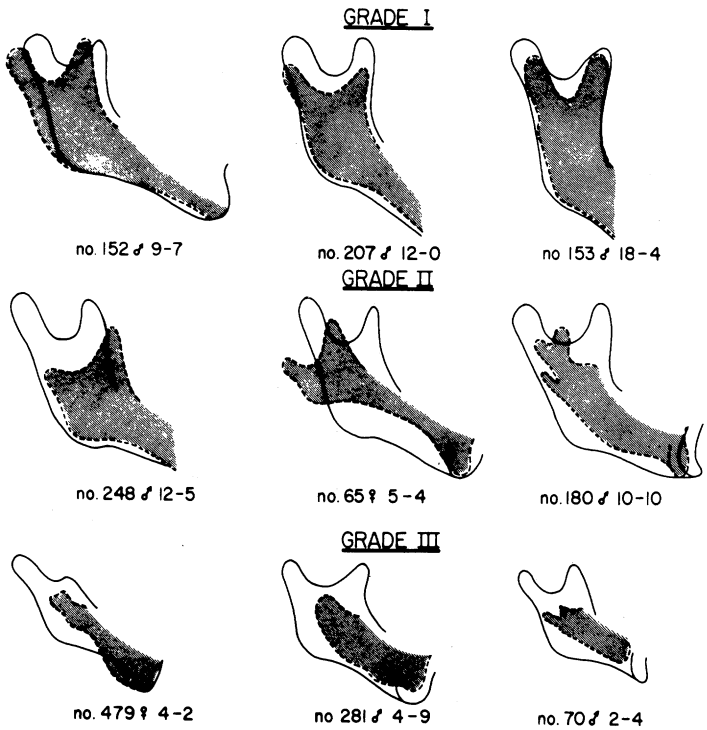

FIGURE 3. Graded spectrum of variation in mandibular deformity encountered in hemifacial microsomia. Tracings of the two halves of the mandible in the same patient were obtained from cephalometric tomograms. The shaded half of the mandible is ipsilateral to the microtic auricle.

tive, could not be accurately assessed radiographically for purposes of correlation with the severity of microtia or mandibular deformity. The following questions were asked in regard to correlation of the range of malformation in hemifacial microsomia:

1. Is there significant correlation between the severity of microtia and the severity of mandibular deformity?
2. Is there significant correlation between ossicular chain deformities and the severity of microtia and severity of mandibular deformity?
3. Is there any correlation between external auditory canal deformity and the severity of microtia or severity of mandibular deformity?

Discussion

The temporal bone, although not, except for the styloid process, a first and second branchial arch derivative, is a factor involved in differentiation of adjacent branchial arch tissue. Based upon the accepted concepts of branchial arch embryogenesis, it is logical to conclude that any sequential or simultaneous teratogenic insult should inevitably result in an orderly interrelationship among auricular, mandibular and ossicular malformations. However, clinical confirmation of this premise has not been forthcoming.

In our series, patients were radiographically evaluated using the microtic auricle as a clinical marker to predict the reliability of the grade of microtia for associated mandibular, ossicular chain, and external auditory canal deformities in hemifacial microsomia. As noted in Table 1, microtia and mandibular anomalies were matched as to the degree of severity. The Pearson correlation coefficient was 0.48, significant at a p-value of 0.001. These data indicate that the severity of microtia parallels the severity of mandibular deformity, but not necessarily in terms of gradation specificity. For example, a grade III microtia does not always imply a grade III mandible, but a severely affected auricle implies a more severely affected mandible.

In Tables 2 and 3, ossicular malformations, as represented by the malleus and incus, paralleled either the severity of microtia or the severity of mandibular deformity. The Pearson correlation coefficient between ossicular deformities and mandibular deformities was 0.60, significant at a p-value of 0.001. In

TABLE 1. Comparison of Gradation of Auricular Deformity to Mandibular Deformity in Hemifacial Microsomia

Mandible		*Auricle*		
Grade	*Normal*	*Grade I*	*Grade II*	*Grade III*
Normal	52	15	1	0
I	0	16	19	8
II	0	6	8	8
III	0	2	0	3

TABLE 2. Correlation between Graded Auricular Deformity and Status of Ossicles (Malleus/Incus) in Hemifacial Microsomia

Ear		*Ossicular Chain*	
Grade	*Number*	*Normal*	*Abnormal*
Normal	52	50 (96.2%)	2 (3.8%)
I	36	17 (47.2%)	19 (52.8%)
II	28	3 (10.7%)	25 (89.3%)
III	21	1 (4.8%)	20 (95.2%)

TABLE 3. Correlation between Graded Mandibular Deformity and Status of Ossicles (Malleus/Incus) in Hemifacial Microsomia

Mandible		*Ossicular chain*	
Grade	*Number*	*Normal*	*Abnormal*
Normal	68	62 (91.2%)	6 (8.8%)
I	43	6 (14.0%)	37 (86.0%)
II	22	1 (4.5%)	21 (95.5%)
III	5	2 (40.0%)	3 (60.0%)

TABLE 4. Comparison between Graded Auricular Deformity and External Auditory Malformation in Hemifacial Microsomia

Ear		*External Auditory Canal*	
Grade	*Number*	*Normal*	*Abnormal*
Normal	52	51 (98.1%)	1 (1.9%)
I	36	15 (41.7%)	21 (58.3%)
II	28	0 (0.0%)	28 (100.0%)
III	21	3 (14.3%)	18 (85.7%)

TABLE 5. Comparison between Graded Mandibular Deformity and External Auditory Malformation in Hemifacial Microsomia

Mandible		*External Auditory Canal*	
Grade	*Number*	*Normal*	*Abnormal*
Normal	68	63 (92.6%)	5 (7.4%)
I	43	4 (9.3%)	39 (90.7%)
II	22	1 (4.5%)	21 (95.5%)
III	5	1 (20.0%)	4 (80.0%)

Tables 4 and 5, a parallel between the severity of the external auditory canal malformations and the severity of either microtia or mandibular deformity is evident.

Mandibulofacial Dysostosis

Mandibulofacial dysotosis is a well-recognized syndrome first reported by Berry in 1888, then by Treacher and Collins in 1900, and, more fully, by Franceschetti and his associates Zwahlen and Klein in 1944.

Sixteen patients with the phenotypic characteristics of mandibulofacial dysostosis were studied radiographically and otologically. Although incomplete forms of this syndrome have been described, the patients in our study manifested all aspects of the syndrome. Major clinical features included hypoplasia of the malar bones, antimongoloid obliquity of the palpebral fissures, coloboma of the outer third of the lower eyelid, a tongue-shaped progression of hair extending towards the cheek, obliteration of the nasal frontal angle, dysplasia of the mandible, and microtia (Figure 4).

Records of 27 patients with diagnoses of mandibulofacial dysostosis were reviewed. Complete data were available on 16 patients (32 ears).

Otologic examinations were performed on all patients. The microtic auricle was graded in accordance with the classification for hemifacial microsomia (Meurman, 1957). Unlike those in hemifacial microsomia, mandibular deformities in mandibulofacial dysostosis are bilaterally symmetrical and usually show minimal variation among patients. Therefore, degree of microtia and middle ear malformations was not analyzed in relationship to the mandibular deformity.

Temporal bone tomography was obtained on 32 ears. Where feasible, development of a particular temporal bone structure was studied in relation to the degree of microtia.

Bilaterally symmetrical microtic auricles

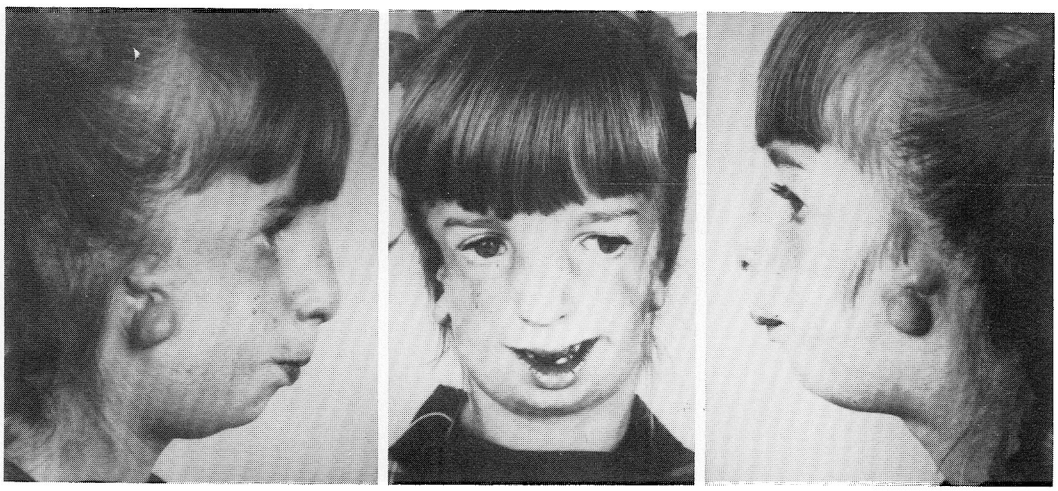

FIGURE 4. Mandibulofacial dysostosis. Note antimongoloid slant.

were observed in 13 patients and were classified as noted in Figure 2. Twenty-one external ears (12 left, and 9 right) were grade I, nine (three left and six right) were grade II, and two were grade III. Our data indicate that the microtic auricle is most often symmetric and grade I.

The external auditory canal was atretic in 22 ears. Eight external auditory canals were normal; two were mildly stenotic. A patent external auditory canal was observed in one patient.

Development of the middle ear cavity was classified as normal, minimally or markedly hypoplastic, or absent. Minimal hypoplasia was ascertained by a slight reduction in the size of the middle ear cavity. A narrow slit-like middle ear cavity represented marked hypoplasia.

Five middle ear cavities were normal, and four were considered to be minimally hypoplastic. In 18, the degree of hypoplasia was marked, and in five, there was a complete agenesis (Table 6).

These findings suggest that the middle ear cavity in mandibulofacial dysostosis is either narrow or absent (Figure 5).

The malleus/incus conglomerate was normal in three ears, malformed in 18, and absent in 11 (Table 7). In five of the 11 ears in which the malleus/incus conglomerate was absent, the middle ear cavity was also absent. This means that, in six, there was a middle ear

TABLE 6. Middle Ear Cavity in Mandibulofacial Dysostosis

Graded Deformity Pinna	*Normal*	*Hypoplasia Minimal*	*Hypoplasia Marked*	*Absent*
I	5	4	11	1
II	0	0	7	2
III	0	0	0	2
Total	5	4	18	5

present. Ossicular malformations are frequent cavity, yet the malleus and incus were not in mandibulofacial dysostosis, and in certain instances the ossicles may be absent.

The stapes suprastructure is not visualized by hopocycloidal tomography except in rare instances and, therefore, was not assessed in this study. Numerous authors, however, have reported surgically-verified stapes malformations usually of a columellar type (Caldarelli, 1977; Pruzansky, 1969; and Holbrow, 1961).

Marked disparity is usually present between the grade of microtia and the degree of malformation of the remaining first and second branchial arch derivatives that constitute the external and middle ear. Grade I microtia was the most prevalent finding, with the external auditory canal, although occasionally patent, most often absent.

Agenesis of the middle ear cleft is not infrequent. When present, the middle ear cleft is normal or only slightly deformed. Agenesis or malformation of the malleus and incus is

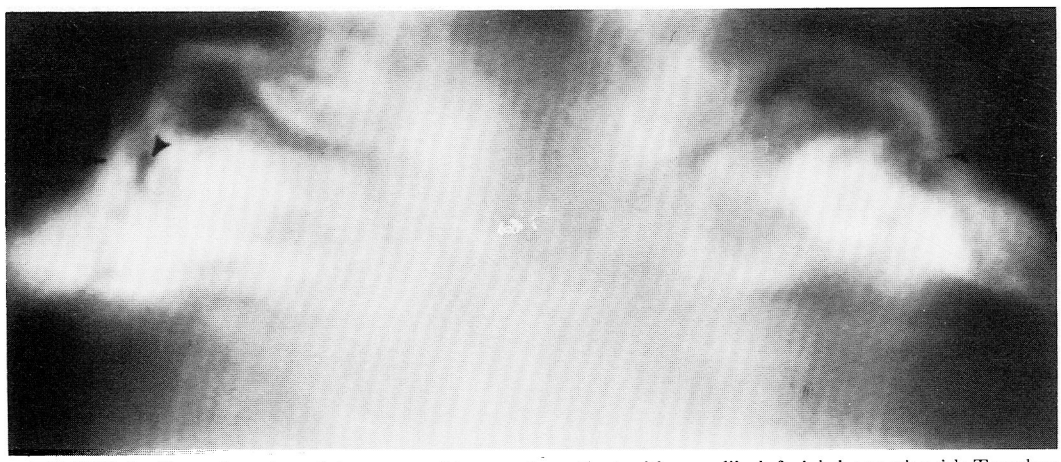

FIGURE 5. Coronal section of the temporal bones of a patient with mandibulofacial dysostosis with Treacher-Collins Syndrome. Note bilateral agenesis of external auditory canal. The horizontal arrows point to the atretic plates, closing the lateral wall of the middle ear cavities. There is a hypoplastic middle ear on the left and no middle ear cavity on the right. The arrow head points to hypoplastic left middle ear cavity. No ossicular chain is seen.

TABLE 7. Malleus and Incus in Mandibulofacial Dysostosis

Graded Deformity Pinna	*Normal*	*Dysplastic*	*Conglomerate Mass*	*Absent*
I	3	2	11	5
II	0	0	5	4
III	0	0	0	2
Total	3	2	16	11

frequent. Stapedial malformations usually occur.

Otologic manifestations of mandibulofacial dysostosis found to be characteristic in this study are as follows.

1. Microtia is most often mild and symmetrical.
2. The external auditory canal is frequently absent.
3. The middle ear cleft is markedly narrow or absent.
4. The malleus and incus are severely malformed or absent.
5. There is marked disparity between the degree of the auricular deformity (mild; grade I microtia) and that of the remaining first and second branchial arch derivatives represented by the external auditory canal, middle ear cavity, and ossicles.

Conclusions

Two factors figure prominently in the recent upsurge of clinical interest in craniofacial anomalies. First is the comprehensive multidisciplinary approach to diagnosis and classification; and second is the recent development of innovative facial reconstructive surgery. As with the numerous facial rehabilitative efforts, otologic rehabilitation of patients with syndrome-related middle ear anomalies necessitates a precise classification and diagnosis of this syndrome-related otopathology. Prerequisite to this goal is a precise definition of the range of temporal bone anomalies and syndrome-specific characteristics of such malformations. Furthermore, recognition of the sequential and simultaneous developmental interaction of the ear and contiguous structures is equally important in understanding the pathogenesis and varied clinical manifestations of these syndromes.

Significant insight into this clinical enigma has evolved from the elucidation of the pathogenesis of the first and second branchial arch malformations in the animal model with subsequent careful extrapolation of the findings to similar malformations in the human (Poswillo, 1973, and 1975). Poswillo's investigations have provided a basis for understanding the random clinical expressivity of various facial and otologic anomalies. Using teratogens in animals, he produced a phenocopy of hemifacial microsomia and mandibulofacial dysostosis and postulated that a separate pathogenesis accounted for their varied clinical manifestations.

In hemifacial microsomia, a parallel existed between the severity of the microtic auricle and that of the middle ear malformation, whereas in mandibulofacial dysostosis this was not the case. Emerging from this study was the concept that grade I microtia in hemifacial microsomia significantly correlated with moderately severe middle ear malformations, whereas grade I microtia in mandibulofacial dysostosis was frequently associated with severe middle ear deformities. This observation suggested a different etiopathogenesis in each syndrome.

Microtia in hemifacial microsomia and mandibulofacial dysostosis may define the anatomic status and predict the potential for reconstruction of the middle ear. Findings in this study suggest that middle ear malformations associated with hemifacial microsomia tend to be less severe than those associated with mandibulofacial dysostosis, irrespective of grade of microtia.

It is evident from this investigation that recognition and understanding of the developmental interaction of the ear with contiguous structures is a prerequisite for diagnosis and therapeutic endeavor directed toward syndrome-related otopathology.

The craniofacial syndromes provide us with experiments of nature affording possibilities to study the pathologic interrelationship between the temporal bone and its contiguous structures. In terms of clinical problem-solving, it is a unique opportunity to contribute significantly, both individually and through an interdisciplinary approach to the total clinical management of such patients.

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