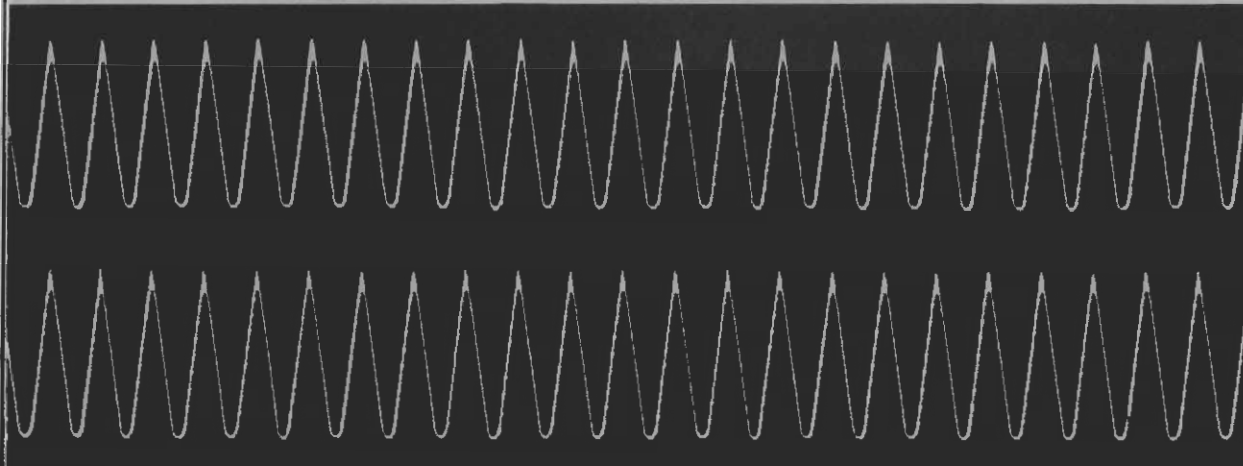


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Orofacial Anomalies: Clinical and Research Implications

Proceedings of the Conference



ASHA REPORTS

NUMBER 8 A PUBLICATION OF THE AMERICAN SPEECH AND HEARING ASSOCIATION

PROCEEDINGS OF THE CONFERENCE
OROFACIAL ANOMALIES:
CLINICAL AND RESEARCH IMPLICATIONS

This Conference was supported in part by
NIH Training Grant DE 210-05
National Institute of Dental Research
U.S. Department of Health, Education, and Welfare

Proceedings of the Conference

**Orofacial Anomalies:
Clinical and Research Implications**

Phoenix, Arizona
April 15-17, 1972

ASHA Reports 8
AMERICAN SPEECH AND HEARING ASSOCIATION
Washington
August 1973

A CONFERENCE ON
OROFACIAL ANOMALIES: CLINICAL AND
RESEARCH IMPLICATIONS

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P R E F A C E

The material in this *Report* represents the proceedings of the third in a series of three annual conferences designed to identify and evaluate clinical and research contributions of mutual value to the professions of dentistry and speech pathology/audiology and to delineate research frontiers of mutual concern. The conferences were sponsored by the Joint Committee on Dentistry and Speech Pathology/Audiology and funded by the National Institute of Dental Research. Since the inception of the Committee in 1966 it has consisted of three members appointed by the American Association of Dental Schools and three members appointed by the American Speech and Hearing Association.

The first conference, held in Ann Arbor, Michigan, in March 1970, was concerned with "Patterns of Orofacial Growth and Development." The papers presented at that conference were published as *ASHA Reports Number 6*. The second conference, "Orofacial Function: Clinical Research in Dentistry and Speech Pathology/Audiology," also was held in Ann Arbor, in March 1971. These conference proceedings were published as *ASHA Reports Number 7*. The papers summarized information from existing research and expressed points of view regarding disciplinary interaction.

The structure of this third conference departs from the format of its predecessors. In this conference the essayists were asked to review clinical reports and findings concerning selected individuals with congenital orofacial anomalies other than cleft lip and palate, and patients who have experienced the effects of ablative surgery to orofacial structures.

The decision to devote an entire conference to persons so afflicted stemmed from the belief that presenting knowledge about these "experiments of nature" can lead to a better understanding of the basic principles of function of the orofacial complex. Two other considerations supported the decision: first, an awareness of how little is known about the functional implications of these anomalies; second, a hope that a review of such materials might suggest relationships between structures and functions that could not be revealed in any other way. Study of behavior before and after treatment may enable the clinician to distinguish merely atypical behavior from that attributable to adaptation to abnormal or inadequate structures, thereby leading to formulation of a basic core of procedures to be used in future investigations.

A second major departure from the previous two conferences is reflected in audience selection. Those attending the earlier conferences were chosen by deans of dental schools or by heads of departments of speech pathology/audiology. For this conference, audience members were selected by the Joint

Committee on the basis of demonstrated active interest and participation in interdisciplinary endeavors.

Implicit in the organization of the conference was an acknowledgment of a primary goal of the Joint Committee—the creation of a greater awareness on the part of each profession of the nature and degree of interdisciplinary interaction that will enhance the educational, service, and research endeavors of the fields of dentistry and speech pathology/audiology.

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WELCOMING REMARKS

DUANE C. SPRIESTERSBACH

University of Iowa, Iowa City, Iowa

On behalf of the Joint Committee on Dentistry and Speech Pathology-Audiology it is my pleasure to welcome you to this conference on *Orofacial Anomalies: Clinical and Research Implications*. This is the third conference sponsored by the committee designed to bring together professional persons who are, in one way or another, directly or indirectly, involved with orofacial anomalies to the end that we can better understand the processes, the issues, and the implications for treatment of persons with these anomalies.

It is trite to observe that the management of orofacial anomalies is complex. We have become increasingly aware of that complexity over time. With that growing awareness has come an appreciation of the parochial nature of our individual training and skills, and the potential value of interacting with persons from a number of different disciplines. As a consequence, it has been one of the major objectives of the Joint Committee, since its organization in 1967, to bring interested professionals from a variety of fields together to examine, in depth, our common interests and to expand our horizons, in research and clinical management, as we gain understanding of the dimensions of the problem and the potential contribution that each of us has to make to the prevention of orofacial anomalies and the treatment of persons who have them. It is our hope that the organization of this conference is consistent with this objective.

I recently heard John Millis, long-time president of Western Reserve University and more recently a scholar of the delivery of health services, observe that we are in the period in our development of health care when we have replaced mortality with morbidity. People are now not only asking "to save my life" but also to "improve the quality of the life that you are saving." Ultimately, that is why we are here. I am confident that both the short- and long-term consequences of this conference will be a contribution to that end.

PSYCHOSOCIAL ASPECTS OF
OROFACIAL ANOMALIES:
SPECULATIONS IN SEARCH OF DATA

EDWARD CLIFFORD

Duke University Medical Center, Durham, North Carolina

An anomaly rarely goes unnoticed, or rather, the person with it rarely is unnoticed. The existence of an anomaly affects family, friends, acquaintances, strangers, professionals, and society itself. The effects are reciprocal, since all of these provide feedback information and, in a sense, a social mirror in which reflections are interpreted by the anomalous person. These reciprocal effects intermesh, become quiescent or intrusive, and may be modified because the person with the anomaly, a dynamic, developing individual with a unique life history, is in constant interaction with those in his environment.

The study of psychological and social effects of facial anomalies and disfigurements, by necessity, not only must deal with the feelings a person has about his face and the meanings he associates to his disfigurement, it must take into account attitudes, feelings, and reactions of all who enter into transactions with him. These may range from the relatively impersonal, abstract values and attitudes of society to the highly personal interaction between parent and child.

We start with complexities. The person is capable of multidimensional behaviors. While transactions and interactions may succeed one another in time, they are taking place simultaneously with other behaviors. Behavior scientists look at a class or domain of behaviors because of their own conceptual needs and abilities (or because of the lack of sophisticated multidimensional research knowledge), often simplifying them in order to explain them. My presentation will also take this exquisitely complex action system and section it, to examine specific behavioral and social processes.

Our specific orientation is toward psychological concomitants of orofacial anomalies and facial disfigurements. A brief overview of the psychological literature specific to these anomalies, exclusive of clefts of the lip and palate, will be presented. I shall then examine facial anomalies and disfigurements from a number of different perspectives.

PSYCHOLOGICAL LITERATURE SPECIFIC
TO OROFACIAL ANOMALIES

The search for relevant social and psychological information ends quickly if the exploration is limited to orofacial anomalies excluding clefts of the lip and palate. A considerable body of knowledge has been accumulated about cleft lip and palate, and this is reflected in several recent reviews concerning psychosocial aspects of this condition such as those by Goodstein (1968), McWilliams (1971), and Wirls (1971). Little of psychological or sociological relevance has been published about other orofacial anomalies. Demb,¹ in a manuscript prepared for the Center for Craniofacial Anomalies at the University of Illinois, came to the same conclusion.

Incidence

Perhaps this dearth of information reflects, and is somewhat proportional to, the incidence of orofacial anomalies in the population. To put things in perspective, Apgar and Stickle (1968) estimate that 15 million persons with one or more congenital malformations exist in the population. They also estimate that of this total approximately 500,000 are blind or have serious loss of vision, 750,000 have congenital hearing problems, and 100,000 have speech defects of "prenatal origin." The incidence of orofacial anomalies, including cleft palate, was not reported. A recent public health survey (Public Health Service, 1971) found that 482,000 children received physicians' services under the Crippled Children's Program and 20% of these were classified as having congenital malformations. Of those with congenital anomalies, approximately 22,000 had clefts of the lip or palate or both; 666 had congenital ptosis of the eyelids; and 1605 had congenital malformations of the ear. No other orofacial anomalies are listed. It appears, based on these surveys, that orofacial malformations constitute a relatively small percentage of the anomalies that mankind suffers.

While cleft lip and palate may have an incidence of about one in 800 live births (Aita, 1969), giving rise to my estimate of approximately 250,000 in the population, other orofacial anomalies are considerably less frequent. For example, craniofacial synostosis (Crouzon's disease) occurs so infrequently that Witkop does not cite incidence figures, merely stating that it is uncommon (Witkop, MacCollum, and Rubin, 1967); and one estimate of the incidence of Apert's syndrome places the prevalence in the living population as one in two million (Warkany²). Since hospitals accumulate more cases and serve as a primary source of subjects for study, admission statistics become of interest. Lindsay (1968) examined 2223 cases admitted to the Hospital for Sick Chil-

¹N. Demb, personal communication.

²Josef Warkany, personal communication.

dren in Toronto over a five-year period and found 25 cases of Crouzon's disease and four cases of Apert's syndrome.

The fact that some orofacial anomalies have relatively low incidence rates should not lessen their importance as vital areas for study and research. The incidence figures, however, do highlight problems that might be encountered in research. Two conclusions may be drawn from the rates cited. First, the difficulty of obtaining enough patients in any one diagnostic category is evident and, in one sense, is also reflected in the number of published studies concerning that anomaly. Second, if substantive studies were to be undertaken, it would probably require the existence of a center for the study of orofacial anomalies—one that would attract many such cases—or, more probably, cooperation between two or more of these centers to obtain sufficient cases.

The Status of Research

A relatively large patient population enabled one team of psychosocial investigators, representing the fields of sociology, cultural anthropology, clinical psychology, and psychiatry, to investigate problems associated with facial deformities (Macgregor et al., 1953). This pioneer work, based mainly on extended interviews with patients, was published approximately 20 years ago and is consistently referred to in works dealing with facial disfigurement.

Later publications concerning orofacial anomalies demonstrate an appalling lack of interest in psychological or sociological approaches. For example, only one psychological paper, dealing with psychosocial aspects of cleft palate (Spriestersbach, 1961), appeared on the program of an international symposium on congenital anomalies of the face (Pruzansky, 1961), held several years after the work by Macgregor and her coworkers appeared. Interestingly, this research was conducted by a speech scientist. The situation improved slightly some two years later with the publication of the proceedings of a conference on facial disfigurement sponsored by the Vocational Rehabilitation Administration (Rogers, 1963). One session considered psychosocial aspects of cleft palate (Spriestersbach, 1963), accompanied by papers on psychiatric (Jeppson, 1963) and employment (Macgregor, 1963) aspects of facial disfigurement. A session was also directed to vocational rehabilitation. The presentations were primarily clinically oriented, with emphasis on case presentations.

At a recent international conference on the diagnosis and treatment of craniofacial anomalies, only one of the papers given might be considered somewhere under the rubric of psychosocial aspects of the problem; this was a nonresearch position paper presented by a pediatrician (Battle, 1971). It is clear that active research is not being carried out, particularly in those behavioral sciences devoted to psychological or sociological adaptation. Perhaps this is best reflected in the membership rolls of the American Cleft Palate Association, where I would guess no more than a half dozen members have a

primary professional identification as psychologists. This may explain why the work of Macgregor and her coworkers assumes the importance that it does.

This paucity of research information, however, can be augmented. First, a body of information about cleft lip and palate is available. Second, considerable research has been conducted examining the general problem of physical disability. We can and should include the work of such investigators as Barker and his colleagues (Barker, 1948; Barker et al., 1953), Meyerson (1948, 1955), and Wright (1960, 1964). These investigators believe they can generalize across all disabilities. They find classes of behaviors arising as a result of being disabled that are true for a variety of physical conditions and that reactions may not be symptom-specific. For example, a wheelchair patient may be just as concerned with appearance as one who is facially disfigured. From this viewpoint, facial anomalies become one of a number of disabilities that are a part of the human condition. Thus, Dembo, Leviton, and Wright (1956) do not differentiate in their subject population between amputees and those with facial disfigurements. It also becomes possible for a chapter on facial disfigurement (Maddan, 1962) to appear in an edited volume on psychological practices with the physically disabled (Garrett and Levine, 1962).

A consistent theme in the literature is the role of physical attraction in our culture. It is to this that I would now like to turn our attention.

PHYSICAL ATTRACTION

Central to estimations of physical attraction is facial appearance. Certainly it is through the face that we are identified, and it is the face that is the primary focus of attention in interpersonal relationships (Meerlo, 1956). We search for meanings of interactions or cues to our relations to others in facial expressions. As Macgregor (1951) points out, looking at one another is a basic form of communication. We look to the face for expressions of emotions as well as for social reactions. There are those who use facial appearance and expression to reach conclusions about the intellectual status or personality of the person. One need only point to certain facial stigmata, as in Down's syndrome, and to the impressions of intellectual functioning that are strongly implied. In a psychiatric sense, mental disorders are reflected in the face (Berndorfer, 1949).

Developmentally, we also focus our attention on the face. Smiling behavior occurs early in the life of the infant and is looked upon as an important milestone in the socialization process (Spitz and Wolf, 1946). This is one of the first social responses of babies. Kagan and his associates have demonstrated that by four months of age infants have built up expectations about the human face. When infants were exposed to three-dimensional models of normal and distorted faces, smiling responses occurred more often in the presence of normal faces (Kagan et al., 1966). By eight months of age, when children are

exposed to normal faces and to faces which have their components rearranged, it is the grotesque (rather than the normal face) that induces anxiety (Richardson, 1969).

The saliency of the face can also be noted in children's drawings. Abercrombie (1971) notes that first drawings are of the human face. Corroboration can be found in the work of Goodenough (1926), where drawings are scored to obtain an index of intellectual functioning. Young children's early drawings are essentially facial drawings to which arms and legs are attached.

Effects of Physical Attraction

Not only is attention paid to the face early in life; there is strong evidence that nursery school children discriminate between attractive and unattractive faces. Berscheid and Walster (1972) studied a group of nursery school children between the ages of four and six whose physical attractiveness had been rated by adults. Less attractive boys were not as well liked by their peers as the more attractive boys, independent of age. Younger girls, however, who were unattractive were popular, but this popularity declined with age. Unattractive boys were rated more aggressive and more antisocial than attractive boys. Unattractive children, regardless of sex, were rated by their classmates as being less independent.

In an interesting study of adult reactions to the physical attractiveness of children, Dion and Berscheid (Berscheid and Walster, 1972) provided young women with reports they presumed to be teachers' descriptions of classroom and playground disturbances. A photograph of a child previously rated for physical attractiveness accompanied each report. The women were asked to rate the severity of the reported disturbance and to evaluate the child's behavior on a typical day. When the disturbance was perceived as mild, the physical attractiveness of the child had no effect. When the misbehavior was perceived as severe, the children who were physically unattractive were presumed to be "chronically antisocial in their everyday behavior." There was also the significant presumption that the misbehavior was likely to be continued in the future, if the child was unattractive.

In a similar study, Clifford and Walster (Berscheid and Walster, 1972) asked 400 fifth-grade teachers to rate a child's record containing information about grades, absences, work habits, and attitudes. Attached to each report was a preselected photograph rated for attractiveness. An equal number of male and female attractive and unattractive pictures were used. Although the records of the attractive and unattractive children were identical in content, the teachers perceived the attractive child to be higher in IQ, to be more likely to go on to college, and to have parents who were more concerned with his education than the unattractive child.

The effects of attractiveness are also in evidence in adulthood. It probably comes as no surprise that physical attractiveness is important in dating be-

havior. One group of investigators (Walster et al., 1966) arranged a computer dance for college students where the students were supposedly matched with their dates on the basis of shared interests. Actually the students were randomly paired, except that the male had to be taller than the female. Physical attractiveness was rated at the time he or she purchased a ticket. In addition, estimates of the students' social skills were obtained. During the intermission, the students were asked to rate liking for, and desire to see their dates again. The primary factor accounting for liking and wanting to see the date again was attractiveness. Similar findings are reported by Stroebe et al. (1971) in their study of some of the parameters of interpersonal attraction.

How does one account for the importance of physical attractiveness, particularly when we are able to utter such homilies as "Beauty is only skin deep" or when people may profess that it is the personal rather than the physical characteristics of the person that are important? Dion, Berscheid, and Walster (Berscheid and Walster, 1972) found that associations between good looks and personality characteristics were positive. Those who were good looking were characterized as more sensitive, kind, interesting, strong, poised, sociable, sexually warm, and sexually responsive than their less attractive peers. Further, in contrast to those whose attractiveness was not as great, those who were physically more appealing were expected to have better jobs and to be happier and more fulfilled in all aspects of life, including marriage.

Influence of Facial Appearance on Judgment

Johnson (Secord, 1958) reversed the procedure by providing personality descriptions and asking her subjects to rate expected facial features. She presented her subjects with brief personality descriptions of two men. One man was characterized as ruthless and brutal, while the other was described as warmhearted and honest. The 21 college freshmen rated facial features on a number of seven-point scales. Twenty-five of the 32 attributes were significantly different from each other. For example, the warmhearted man was described as having an average eye depth, an average distance between the eyes, an average width of nose, an average facial tension, and a direct gaze. The ruthless man was described as having deep or protruding eyes, eyes either too close together or too wide apart, a wide or narrow nose, and an averted gaze.

Cook (1939) found some relationship between facial appearance and judgments of intelligence. Ten judges each rated 150 photographs for IQ. Those whose IQ was judged to be high did not have any facial feature that was out of proportion. Those whose IQ was judged to be low did, however, share some facial characteristics. In this group were photographs of persons with crossed eyes; sunken eyes; lopsided, fat, or narrow faces; and large noses. Cook also determined the veridicality of the perceptions by comparing them with the actual IQs of the persons photographed. The judges were very poor in estimat-

ing IQ. Moreover, they did not do any better in judging the extremes, where presumably some of the depicted characteristics would be of help—strong evidence indeed that these associations do not correspond to reality.

Most studies dealing with the psychology of physical attraction use as subjects people whose appearances fall within normal variational limits. These investigators have avoided using either the extremely beautiful or the facially disfigured. Presumably, reactions might become even more polarized if the facially disfigured were included.

That our culture is oriented toward beauty is almost self-evident. One need only point to the various beauty pageants that are part of the national and international scene. For further evidence, one need only look at the number of products devoted to maintaining or increasing attractiveness. If beauty is the ideal, how do those who markedly deviate from this state fare?

THE DISFIGURED AND SOCIETY

What may differentiate the unattractive from the disfigured is a characteristic of the face that is immediately apparent. Further, this face has persistently intrusive characteristics. Epstein (1958) states that the deformed face cannot help but draw attention to itself. Not only is he noticed, but the person with the facial disfigurement is also labeled. We may, for example, call him handicapped, or disabled, or even disfigured. These labels have the status of socially acceptable categorizations. As soon as labels are applied, however, there is an imputation that the person is different from other people. Moreover, there is an imputation that the difference is undesirable (Freidson, 1965). The person is different because he deviates from a culturally approved norm. In the case of facial anomalies the standard is not an absolute one, because there is a range of acceptable appearances. The person who is disfigured is marked, not because he fails to achieve the ideal state of being beautiful, but because he has failed to achieve an unstated minimal standard of acceptability.

Failure to achieve a societal norm places the person within a subgroup to which a stigma is usually attached. This process results in a "spoiled identity," for, as Goffman (1963) points out, the person becomes tainted or discounted. Presumably, then, society creates groups of discounted persons who are immediately recognizable. It seems reasonable to suppose that the process of stigmatization calls forth behaviors in the observers that are independent of the personal identities of those stigmatized. One would then expect prejudicial and discriminatory behavior in the presence of facial anomalies.

Facial Anomalies and Marginal Status

How does prejudice operate toward those with facial disfigurements? After stressing reactions of disgust and horror on the part of those seeing a severely facially deformed person, Hirschenfang, Goldberg, and Benton (1969), on the

basis of interviews with patients with facial paralysis, found the disfigured believe they are regarded as social inferiors and attribute a marginal or minority status to themselves. This agrees with the findings of Macgregor (Macgregor, 1951; Macgregor et al., 1953). Meyerson (1955) also emphasizes the minority status, stating that the disabled person lives in the world of the physically intact majority, while he also exists in a special world his disability creates for him. Barker (1948) emphasizes the underprivileged status of the disabled and sees them as a minority, equivalent to any other underprivileged minority group. Barker points out that the minority status of the underprivileged and the disabled are not completely comparable. The member of the religious or racial minority group can blame the majority for his frustrations and thus avoid self-blame. The physically disabled cannot transfer his frustrations to others so easily.

The disabled and the underprivileged are not comparable in minority status in another crucial dimension. The typical member of a minority group has others of his group as models for behavior. In the ghetto, for example, members of the minority group have ready access to one another. This is not true of the disabled, particularly if they do not organize themselves into voluntary associations. The model from which there seemingly is no escape at home, in the neighborhood, or in society at large is the standard, nondisabled man.

Minority Status and Prejudice

Following the reasoning that attitudes toward minority groups should be similar, English (1971a) found, in a study of 116 college students, that those expressing ethnocentric attitudes (high rejection of outgroups) also expressed more negative attitudes toward blindness. In a review of the correlates of stigma toward physically disabled persons, English (1971b) reports data indicating a significant relationship between negative attitudes toward deafness and anti-black, as well as authoritarian attitudes. Using both high school and college students, Chesler (1965) examined relationships between ethnocentrism and attitudes toward the disabled. He found those who expressed high ethnocentrism also expressed rejection of the physically disabled.

Despite such evidence, McDaniel (1969) states that the belief that disabled persons are assigned a minority status, and hence become targets for prejudice, may be an oversimplification. He points out that a universal stereotype of the disabled does not exist, making the analogy to minority groups somewhat questionable. Further, ethnocentric attitudes may generalize to include the disabled and therefore may not indicate prejudice toward the disabled per se. Finally, not all attitudes toward the disabled are negative; positive attitudes do exist and, according to McDaniel, the degree of acceptance of the physically disabled varies with the sex, age, maturity, and education of the nondisabled. As a matter of fact, McDaniel asserts, on the average, public, verbalized attitudes toward disabled persons are mildly favorable. To deny the existence of preju-

dice would be foolhardy indeed, however; for, if not for the presence of negative community attitudes, the problem of disability would reduce itself to a medical one and many of the problems the person, his family, and rehabilitative services have would disappear (English, 1971a).

An important distinction can be made between the expression of negative attitudes toward the disabled—which occurs in almost half of the nondisabled population, according to English (1971a)—and active discriminatory behaviors. It is of some value to distinguish between the disability itself and the person who is disabled. For example, McDaniel (1969) takes the position that disabling conditions are perceived to be more inherently undesirable than the persons having them. While the anomalous condition may be a state to be avoided, the handicapped person can be accepted on his personal merit.

The mere existence of a handicap, however, may be a strong barrier to effective interpersonal relations. Certainly, on the basis of first impression, the demonstrated discomfort of the nonhandicapped in the presence of persons with facial anomalies (Macgregor, 1970) would reduce or prevent exchange of information between them. Described in terms of social distance, defined as degrees of social intimacy between people, one might expect that persons with facial anomalies would experience relationships involving greater social distance than the physically normal and that the facially disfigured person would encounter the greatest discrimination in social encounters where the social distance is great and, conversely, would experience the least discrimination in encounters involving the greatest social intimacy.

Social Distance and Disability

Evidence does exist that anomalies are deterrents to establishing more intimate social relations. In an unpublished study, G. Krieger (cited by Sieka, 1970) examined preferred social distances between the nondisabled and the disabled. He found the disabled were more readily accepted as casual acquaintances, but were less likely to be accepted in relationships of greater emotional involvement, such as marriage. This is in keeping with the findings of Meyerson (1948), with a group of 50 college students. Half of his sample would not date and 65% would not marry an amputee, while 72% would not date and 85% would not marry a deaf person. Interviews with facially disfigured adults reveal complaints about establishing relationships that involve varying degrees of social intimacy ranging from difficulties in getting jobs to making friends and getting married (Macgregor et al., 1953). Sieka (1970) stresses that the difficulty the facially disfigured have in establishing close personal relationships restricts them from experiencing and exercising their sex roles. In turn, this prevents the person with a facial anomaly from experiencing true feelings of being accepted.

Bryt (1953) conducted psychiatric interviews with 57 facially disfigured adults: a group of 29 patients who were self-referred for plastic surgery, and

a group of 28 patients who had been referred by others. Of particular interest is that some in the self-referred group expressed strong feelings that the facially disfigured had no free choice in their relationships with members of the opposite sex as the nondisfigured did. They felt they had to content themselves with any willing partner. While the reactions of those who were not self-referred were not as strong, they too felt their prospects for marriage were not good and that opportunities for close relationships with the opposite sex were limited.

Kleck, Ono, and Hastorf (1966) investigated the effects of physical deviance on face-to-face interactions in an experimental situation. They evaluated the behavior of college students under two conditions. Half the students were exposed to a confederate simulating an amputee, while the other half were exposed to him as he appeared normally. Students exposed to the amputee, in contrast to those exposed to the normal-appearing person, were less variable in their behavior, terminated their interactions sooner, and in the expression of opinions not directly related to disability, expressed those which were less representative of their actual beliefs. For the latter finding, an index of opinion distortion was obtained by examining the difference between opinions expressed during the experimental session and responses to the same questions one month later.

Richardson et al. (1961) tested several hundred children, 10 and 11 years of age, to ascertain their reactions to physical anomalies. Over a period of years, several sets of children participated in the studies. More than 350 of the children were in summer camps, some of them in camps for nonhandicapped and handicapped children. Approximately 250 children were obtained from several school systems. The children were asked to rank a series of drawings portraying physical anomalies. They rated the following drawings in order of preference: a child with no physical handicap, a child with crutches and a brace on his left leg, a child sitting in a wheelchair with a blanket covering both legs, a child with his left hand missing, a child with a facial disfigurement on the left side of his mouth, and an obese child. The stated preferences of the nondisabled and disabled children were identical and were in the order given here. These findings were replicated in subsequent investigations by Richardson and his colleagues (Richardson, 1963; Richardson, Hastorf, and Dornbusch, 1964; Richardson and Royce, 1968). These findings were also replicated by Alessi and Anthony (1969), who took the position that the assignment of mean ranks to the pictures does not give the true order of preference of individual children. They detected a number of selection patterns in their sample, and claimed that the pattern resulting from assignment of mean ranks was rarely found in actuality.

Of particular interest here is a further report of Richardson's (1963), examining the preference at the beginning and toward the end of a camp experience. Since both handicapped and nonhandicapped children were in the same camp, it might be assumed that their accessibility to each other might be reflected in

decreased social distance between the two. Contrary to expectations, the drawing of the nonhandicapped child was chosen more frequently, while the number of choices for the handicapped child remained approximately the same. Richardson concluded that first impressions, in the presence of a disability, are not quickly overcome. Similarly, Force (1956) examined the social status of physically handicapped school children by obtaining sociometric choices of 63 handicapped and 361 physically normal children in 14 integrated elementary classes. The mere presence of physically handicapped children among normal peers did not offset the negative effect of being stigmatized by their normal peers.

Social Encounters and Disability

Siller (1966) developed a set of 20 scales, the Disability Factor Scales, to measure attitudes toward amputation, blindness, and a number of cosmetic deformities. These were administered to three groups of approximately 500 each. His subjects expressed distinct uneasiness at the prospect of encountering those with any of the disabilities sampled. Two major types of items accounted for the uneasiness; the first involved expressions of uncertainty of what to do in the presence of a disabled person, while the second involved feelings of emotional distress in encounters with the handicapped.

The belief young adults have that young handicapped adults may be socially rejected and hence are lonely may partly account for the feeling of unease. Initiating a social relationship under these conditions might commit them to a relationship from which it would be difficult to withdraw (Richardson, 1969). Kelley et al. (1960) assert there is a commonly held social value that kindness and consideration should be shown to others, particularly if they are perceived as being less fortunate. In establishing initial contacts with the handicapped, the physically normal person may feel internal and external pressures to maintain a relationship. Because he may experience conflict between the socially correct thing to do and his own reluctance, the normal person may settle for a less satisfying relationship than he usually would. This can give rise to feelings of unease, uncertainty, or ambivalence in the relationship, perhaps because he fears that he will reveal his negative attitudes toward the handicapped person with whom he is interacting (Richardson, 1969). While these feelings would tend to keep the social intimacy at a more distant level, they would reduce overt displays of discrimination. This relationship may be characterized as one of supposed acceptance because the physically normal person attempts to function on the basis of equality while he denies that differences do in fact exist (Davis, 1961).

Davis (1961) would differentiate two further stages in the development of interpersonal relationships between the handicapped and nonhandicapped. The next stage, of necessity involving more social intimacy, is one in which the physically normal person is no longer aware of the disability and is reacting

more to the personal characteristics of the disfigured partner. The final stage for Davis would involve the qualification and amendment of the relationship so that awareness of the disability exists, particularly if it involves a functional impairment, but it does not interfere with the social relationship. With this reduction of social distance, discriminatory attitudes are dissipated and the relationship proceeds on a more equitable basis.

There is evidence that decreasing social distance between the handicapped and the physically normal person can decrease prejudice. Using the Attitude Toward Disabled Persons Scales, Yuker, Block, and Campbell (1960) found that scores on this instrument correlated highly with the amount of contact that subjects had with disabled persons. They further demonstrated that changes of attitudes in a positive direction could come about with increased favorable interaction between the physically normal and the disabled. English (1971a), on the basis of an extensive review of the literature, concludes that the quality of the contact between disabled people and the physically normal affects attitudes. Superficial contact does little to change attitudes in a positive direction. He makes a particular point of emphasizing that the maximum improvement in attitudes comes about when the interpersonal relationship between the physically normal and disabled persons is on an equalitarian basis.

To this point, we have been considering the social forces impinging on the person with a handicap. I would like to turn to more personal considerations—the familial and intrapersonal factors that affect the development of those with facial anomalies.

FAMILY FACTORS AND THE DEVELOPMENT OF CHILDREN WITH FACIAL ANOMALIES

Attention is focused on the birth of a child with a defect because the crisis the birth precipitates, depending on its resolution, is presumed to have profound effects on the future development of the child. The crisis arises because parents expect to give birth to a normal, healthy infant and are not prepared for a disfigured baby. Although the resolution of the crisis is probably more dependent on the parents' ability to cope with the situation, their strengths and weaknesses, the state of the marriage, the adaptive capacities of family members, and the unique personal histories each person brings with him to the crisis, much of the literature focuses on initial reactions of the parents, and, more specifically, that of the mother.

Impact of Giving Birth to a Baby with a Defect

There can be little doubt that giving birth to a baby with a defect gives rise to a number of feelings, for it is rare that anyone is emotionally prepared for this unhappy fact (Blattner, 1964). Parents have been described as having

feelings of acute grief, anger, anxiety, confusion, depression, disappointment, disbelief, frustration, guilt, hurt, inadequacy, rejection, resentment, shock, stigmatization, and withdrawal (Barker, 1948; Castellanos and Stewart, 1964; D'Arcy, 1968; Easson, 1966; Fishman and Fishman, 1971; Koch-Schulte, 1968; Slutsky, 1969; Tisza and Gumpertz, 1962). These feelings, indicating the impact of the crisis on the parents, are presumed to be important, for the assumption is that they are translated into attitudes toward the defect and against the child himself. These attitudes and feelings are transmitted to the child, thus he is affected. Spriestersbach (1963) states that within a few hours of the birth of an infant with cleft lip and palate, the basic attitudes of the parents are already determined.

While there may be highly emotional reactions to the birth of the infant, Jordan (1962) believes the impact of this crisis is a function of the discrepancy between the perception of the event and the resources the family possesses for coping with it. The consequences, however, are presumed to be disorganizing. Winick (1967), for example, giving no data, reports a breakup in more than 50% of the families in one study of the effects of giving birth to children with severe malformations. Farber (1960) and Farber and Jenne (1963) found that the birth of severely mentally retarded children was reflected in increased tendencies toward family disintegration. There is some contradictory evidence, however. Pless and Roghmann (1971), in a large-scale study of chronically ill children, reported a low rate of family disintegration. With cleft-lip and -palate infants, Crocker and I (Clifford, 1968; Clifford and Crocker, 1971) found strong evidence suggesting that the crisis had integrative effects in which husbands and wives independently reported increases in marital satisfaction and changes in themselves in a positive direction.

Duration of Impact

In studies by Boles (1959), Farber (1960), and Norval, Larson, and Parshall (1964) there is the implication that the effects of the original impact continue unabated for relatively lengthy periods of time. There is some evidence, however, that these feelings may give way to responses that are more indicative of attempts to cope with the situation. Koch-Schulte (1968), on the basis of interviews with 13 sets of parents of babies with cleft lip and palate, states that the feelings of depression lasted up to four days. Tisza and Gumpertz (1962), on the basis of clinical case studies, report that mothers do achieve a mastery over the painful emotions and repress the original feelings, and that expressions of love and compassion emerge. Based on interviews with 60 pairs of parents with cleft-lip and -palate infants, I reported a relatively rapid dissipation of the effects of the original shock (Clifford, 1968). Similarly, after interviewing 34 families of infants who had combined clefts of the lip and palate, Slutsky (1969) concluded that the mothers recovered from their painful reactions to the birth within a short time.

Mothers do recall, and sometimes quite intensively, the birth of the anomalous baby. Their feelings are affected by the hospital environment. While hospitals may be geared for meeting crises associated with the maintenance of life, they may not be equally equipped for dealing with emotional crises. Richardson (1969) reports that physicians' behavior may upset parents who seek their help because they are often inadequately trained to cope with their own feelings or the parents' feelings. The mothers D'Arcy (1968) interviewed felt there was an abruptness on the part of the professional giving them information about the child. In many cases, there is a delay in telling the mother about her baby (Clifford and Crocker, 1971; Slutsky, 1969). Crocker and I found that the longer the delay in telling the mother about her baby or in showing it to her for the first time, the greater was the recalled impact of the birth up to two years later. In addition, the greater the delay in showing the mother her baby, the greater was the recall of unhappy feelings on first seeing the baby (Clifford and Crocker, 1971).

Maternal Reactions

Giving birth to a baby with cleft lip and palate also affects other maternal reactions. Although, for the most part, the pregnancies of the mothers in our sample were within normal limits, we found that mothers of cleft-lip and -palate babies recalled more difficulties with their pregnancies than did mothers who delivered normal infants (Clifford and Crocker, 1971). I had 60 pairs of mothers and fathers rate a number of characteristics in their cleft-lip and -palate infants. Parental ratings of the infant were related to the perceived severity of the cleft. Children whose condition was perceived to be more severe were more likely to be rated as active-irritable and somewhat less pleasant in personality than infants perceived as less severely impaired (Clifford, 1969b).

Pursuing the effects of maternal reactions on the later development of children, Fishman and Fishman (1971) interviewed 39 mothers of children born with birth defects, including cleft lip and palate. They assessed maternal reactions to the birth and obtained measures of self-esteem and adjustment on the children. They found "no clear, simple, linear relationships" between the children's self-esteem and adjustment measures and the ratings of maternal stress at birth. Perhaps because coping mechanisms in reaction to stress come to the fore and adaptive mechanisms take over, the original strong emotional reactions at the birth may be limited in their subsequent effects. For example, mothers who were concerned about their children, who actively sought information about their children's condition, who encouraged open communication about the anomaly, and who had a positive outlook about the child's future, had children whose self-esteem and adjustment scores were high (Fishman and Fishman, 1971). This, by the way, might be an apt description of any mother whose child develops good self-esteem and is well adjusted.

The existence of a child with a facial anomaly does affect his parents'

behavior. Because of their concern and strenuous efforts to find cures or ameliorations for the disability, available parental energy and family resources may be focused upon the child. Richardson (1969) believes that these uncertainties about the child's status may lead to unrealistic aspirations for him, while Wright (1960) asserts they may lead to parents pressing for accomplishments beyond his capacities. As the parent has also absorbed the values of the culture with regard to physical attraction, he is seen to be ambivalent; this may be reflected in the expression of inconsistent attitudes (Wright, 1960). For example, based on social workers' interviews with parents of disfigured children, Lauer (1953) found parental behaviors varying between expressions of warmth and approval and expressions of rejection and hostility.

Parental Characteristics

There is also an implication that parents of children with anomalies are somehow different, either because they are responsible for having produced such a child or because of the impact of their child on them. Research evidence, however, offers no support for this viewpoint. I have already referred to my work with Crocker in which integrative rather than disintegrative effects following the birth of a baby with a cleft were noted. While mothers of cleft-palate infants indicated fewer positive and more negative changes in themselves as a result of having a cleft-palate baby than mothers having normal babies, other differences were minimal (Clifford and Crocker, 1971). In extensive parallel studies, Goodstein directly attacked the problem by examining personality profiles of 170 mothers and 157 fathers of children with clefts, using the Minnesota Multiphasic Personality Inventory. When these profiles were compared with those of a control sample of 100 mothers and 100 fathers of physically normal children, no significant differences were found (Goodstein, 1960a). A companion study obtained ratings of the social adjustment of the child and the adequacy of parental handling of the child. No relationships were found between these ratings and the personality profiles of the parents (Goodstein, 1960b).

Environmental Characteristics

There are also those who would describe the environments in which children with anomalies are reared as socially restrictive. Castellanos and Stewart (1964), as a result of their clinical experiences, think that parents attempt to avoid social contacts because of the child's appearance. Richardson (1969), citing no evidence, believes that, in contrast to the normal child, the physically handicapped child is exposed to fewer social experiences and is less likely to be taken out on social occasions. In a similar fashion, Richardson et al. (1964) assume the handicapped child has fewer social and interpersonal experiences. These restrictions presumably stem from parents' over-solicitous attitudes and

over-protective behavior. Further, although no evidence is cited, parents are seen as reluctant to talk about the child's anomaly, discouraging the child's questions, providing the child with less responsibility than a normal child, and giving the handicapped child little or no voice in family matters. Further, although parents may deal with the child in a loving manner, they tend to be arbitrary with him (Richardson, 1969; Richardson et al., 1964; Tisza et al., 1958).

The limited research available, however, casts some doubt on these speculations. Based on extensive matched paired observations of seven disabled and seven nondisabled children in everyday activities at school and at home, Schoggen³ could not find evidence of greater over-protectiveness, over-solicitousness, encouragement of dependency, or rejection by mothers of handicapped children compared with mothers of physically normal children. When the behavior of the two groups of mothers was compared, Schoggen found no significant differences in the time they spent with the children, the amount of time they were available to them, their giving unsolicited assistance to the children, and the amount of interpersonal conflict involved in the interactions with their children. There was a slight tendency for mothers to give handicapped children more individual attention, and, contrary to expectations, mothers of handicapped children were more insistent that assigned tasks, such as hanging up a coat, be carried out by their children.

Early Mother-Child Relationships

Personality theorists focus attention on the developing relationship between mother and child. Stress is placed on the very early, emerging mother-child interaction because it is within this relationship that the precursors for future personality development are in operation. Disruptions of these relationships may lead to a variety of personality disturbances (Bowlby, 1951), and may even be a cause of wasting away and death in infants (Spitz, 1945).

One of the earliest points in the interaction between mother and child, particularly from the psychoanalytic point of view, is in the feeding situation, where basic drives seek satisfaction. How these primary needs are met is presumed to have consequence for later personality development. There are those who point out the possibility of emerging personality disturbances because of the lack of adequate sucking opportunities during the feeding of infants with clefts. Further, because feeding the child with cleft palate is slow, frequently interrupted, and unsatisfactory, and because the existence of a cleft may interfere with the primary need for mothering, it is presumed that frustration is introduced into the relationship, the effects of which will be in evidence in later life (Alpert, 1959; Tisza and Gumpertz, 1962; Tisza et al., 1958). Tisza, Irwin, and Zabarenko (1969), for example, found themes of oral

³P. Schoggen, personal communication.

aggression and incorporation in the dramatic play of 11 preschool children with cleft palate. However, in a study of 47 parents of children with clefts who ranged in age from six to 12, I found that the mothers reported low incidences of nonnutritive sucking (Clifford and Sinicrope⁴).

Development of Body Image

The initial relationship between mother and child is primarily expressed through physical contact. The manner in which she holds him, feeds him, and takes care of him is postulated to convey to the child some of her attitudes toward his body. These are later reinforced by verbal expression as well, leading the child to have attitudes toward his body that are reflective of the important people around him (Watson and Johnson, 1958). During the course of development, the child integrates a number of conscious and unconscious attitudes toward his body and its functioning, the totality of which is called the body image (Schilder, 1950). The body image can be compared, in effect, to an unconscious pictorial representation of the physical self. It is presumed to be somewhat resistant to change, particularly at older ages, while, at the same time, it is influenced by unconscious needs and wishes. Discrepancies may arise between the body image and the person's true appearance and functioning, because the model of the body the person holds for himself may not correspond to reality. One of the prime areas of focus for body image considerations is the face, because of its central role in personal identification.

One technique used to measure body image involves asking the child or adult to draw a person. Since this is a relatively unstructured task, it is presumed that the drawing will reflect unconscious aspects of the personality and will thus be a projection of the unconscious body image (Machover, 1949). Interpretations of the drawings lean heavily on the absence as well as the presence of a variety of body parts.

Several studies have examined body image via drawings of persons having facial anomalies. Unfortunately, not only do they suffer from poor experimental design, but there is also some question whether the draw-a-person technique is a valid measure of body image. Corah and Corah (1963) examined the drawings of 12 cleft-lip and -palate children and an equal number of controls, and could find no evidence of facial distortions in the drawings. Ruess (1965) compared the drawings of cleft-palate children and their siblings and found no significant differences between the two sets of drawings. He was of the opinion, however, that had the drawings been confined to the head and face, differences would have been obtained. Palmer and Adams (1962) obtained drawings of whole figures, as well as drawings of the face, from 20 cleft-lip and -palate subjects and two control populations of physically normal children. No sig-

⁴E. Clifford and P. E. Sinicrope, unpublished study on feeding history and fathers' participation in the care of children with clefts. Child Psychiatry Research Laboratory, Durham, North Carolina (1972).

nificant differences in the drawings of the groups were obtained. Contrary to these results are those reported by Abel (1953) and Abel and Weissman (1953). Forty-five patients, primarily adults with facial anomalies, were divided into mild-moderate and severe-gross appearance groups on the basis of ratings of their appearance. All were given the draw-a-person test. Twenty-one of the 26 patients in the mild-moderate group produced drawings without apparent facial disfigurements, while 11 of the 19 in the severe-gross group produced drawings with facial distortions in them. On the basis of these findings, the authors concluded that both groups had a correct body image and that the severely disfigured were more realistic toward their handicap than those less severely disfigured.

In another approach to the study of body image, Crocker, Pope, and I examined the body satisfactions expressed by a group of 98 adults with cleft lip and palate. We found their body satisfaction levels to be high in comparison with a control sample (Crocker, Clifford, and Pope, 1970). Although the expressed body satisfaction levels seemed to indicate little or no concern with the body, Sinicrope and I⁵ were able to demonstrate that the parts of the body and the functions the cleft palate individuals were least satisfied with were mouth, teeth, lips, voice, talking, and speech.

Development of Self-Concepts

The emergence and differentiation of body image parallels the child's establishing himself as an individual and as a family member. The child with an orofacial anomaly, as all children, is involved in many socialization processes ranging from the acquisition of appropriate behaviors to their expression in a socially approved manner. While the psychological literature contains many references to these processes, no studies of them in relation to facial anomalies are in evidence. It is assumed that the facially disfigured child will not only become acculturated in the larger society, he will also become a unique member of his own family, absorbing its traditions, attitudes, and values, as does the normal child. Like the normal child, he will incorporate the values of the physically normal and adopt the cultural standards of attractiveness and the negative attitudes toward the disabled for himself (Goffman, 1963).

During this period of development, the child will be exposed to others in the environment who provide feedback to him about himself. As the child perceives how he is valued by others, and the attitudes they express, his self-concept is affected. For the child with a birth defect, Goffman (1963) would claim that the internalization of generally held negative attitudes toward the disabled, combined with the reactions of others toward a child with a handicap, produces an awareness in him that he does indeed bear a stigma. It is

⁵P. E. Sinicrope and E. Clifford, unpublished study of effects of cleft lip and palate on body satisfaction. Child Psychiatry Research Laboratory, Durham, North Carolina (1972).

this realization that results in a devaluation of the self-concept as distinct from the body image concept.

In partial support of these speculations are the informal observations of Knorr, Hoopes, and Edgerton (1968) and Jabaley et al. (1970) that the facially disfigured child does not perceive himself as different until the age of four or five. They claim that the face becomes an issue and is stigmatized when the child comes in contact with a large number of children, usually in nursery school or first grade (Goffman, 1963; Jabaley et al., 1970; Lauer, 1953). No studies, however, have specifically examined the effects of nursery school or first grade entrance on the behavior of facially disfigured children.

Some researchers claim, either through informal or clinical observations, that the devalued status of those with handicaps is reflected in lowered self-esteem and by self-depreciation (Richardson, 1969; Richardson et al., 1964). Castellanos and Stewart (1964) feel that the facially disfigured have strong fears of rejection which are often expressed by hypersensitivity and increased demands for personal attention. Barron (1955) states the handicapped handle their anxiety by resorting to repression, constriction, and depression. Epstein (1958) believes that those with facial disfigurements suffer from inferiority and shame, are somewhat inhibited in social expression, and may demonstrate antisocial tendencies.

Based on impressions of patients, Hirschenfang et al. (1969) concluded that those with facial paralysis demonstrated tendencies toward paranoid ideation. Lauer (1953), interviewing parents of children with facial anomalies, reports that they characterize their children as quiet, shy, and uncomfortable in the presence of strangers. McWilliams (1966) presents no data, but describes cleft-palate children as somewhat self-effacing, passive, and compliant. More direct evidence comes from a study by Cruickshank (1951), who gave a sentence completion test to 264 handicapped and 264 nonhandicapped children, and found that the handicapped perceived themselves to be more fearful and guilty. Finally, in the series of experiments in which children ranked drawings portraying a nonhandicapped child and five varieties of handicapping, handicapped children, as well as nonhandicapped children, clearly preferred the drawing of the physically normal child (Richardson et al., 1961). Richardson and his coworkers concluded that the results demonstrated that the disabled had internalized the prevalent negative cultural attitudes toward the handicapped.

CLEFT LIP AND PALATE RESEARCH

A body of information about one orofacial anomaly, cleft lip and palate, has been accumulating. Much of the emphasis in the literature has been on personality correlates of cleft palate, with almost uniform results. Hackbush (1951) could not find Rorschach patterns that were unique to a group of cleft-palate patients. In a similar fashion, Wirls and Plotkin (1971) administered an ex-

tensive battery of personality tests to 66 cleft-lip and -palate children and an equal number of their siblings. They failed to find personality differences between the two groups. Watson (1964) evaluated the adjustment of a group of 34 boys with cleft lip and palate, 19 boys with a physical handicap, and a control group of 40 normal boys. He found no differences in adjustment levels of the three groups. Sidney and Matthews (1956) compared the social adjustment of 21 children with cleft palate to that of two control groups and could find no evidence of inferior social adjustment in the cleft-palate group. Schweckendiek and Danzer (1970) also found no differences between their cleft sample and normals. Reuss (1965) compared the performances of 49 cleft-lip and -palate youngsters and their siblings on the Bender Visual Motor Gestalt Test and could find no evidence of discernable pathology in either group. Most of the 60 cleft-lip and -palate patients Billig (1951) studied gave evidence of normal or better adjustment. Five percent of the cleft patients were classified as unsatisfactorily adjusted. I found that cleft-lip and -palate adolescents and adolescents with asthma rated themselves positively. Cleft-lip and -palate adolescents, however, did perceive themselves to have been less accepted at their births than did the adolescents with asthma (Clifford, 1969a). Birch (1952) found no cleft-palate children among the 600 most severely maladjusted children in the Pittsburgh public schools. Finally, a number of reviews concluded no evidence points to the existence of a "cleft-palate personality," confirmation indeed that an anomaly need not imply psychological pathology (Goodstein, 1968; McWilliams, 1971; Spriestersbach, 1963; Reuss, 1967).

To this point I have focused attention on those born with facial anomalies. What can be said about those who acquire disfigurements because of trauma, disease, or ablative surgery?

ACQUIRED FACIAL DISFIGUREMENTS

As the existence of an anomaly does not imply psychological pathology, neither does an acquired facial disfigurement necessarily make a person psychologically maladjusted. Meyerson (1955) asserts that the reaction to disfigurement will lead to maladjustment only if the person feels he has suffered a loss of something required and valued by society, if others knowing about his condition devalue him for it, and if he devalues himself because he accepts the judgment of others.

Accommodations, however, will have to be made if the change in physical status or appearance is sudden. These adjustments are ultimately related to the person's acceptance of himself and his disability. For example, the new, disfigured appearance requires a modification of the body image, if it and reality are to coincide (Watson and Johnson, 1958). As another example, the disfigured person may have difficulty in accepting a new status which is thrust

upon him, particularly if it is defined in terms of stigmas with which he may have characterized other people.

Except for appearance, other characteristics of the facially disfigured person have not changed. He retains his abilities and characteristic ways of reacting and, to prevent a loss of self-esteem, he may emphasize these or deny that he is different in any way (Kelley et al., 1960; Dembo et al., 1956). He may experience difficulty in communicating with the nondisfigured, particularly about his own injury to the face (White, Wright, and Dembo, 1948). Ultimately he will have to accept his difference, if he is to be successfully rehabilitated. Family and friends, as well as those responsible for his rehabilitation, may expect him to conform to role expectations not existing before his disfigurement. Scott (1969), for example, claims that people and organizations devoted to rehabilitation have an unrecognized function of molding the person to the rehabilitative system. In the case of the blind, he states the function of the system "is to teach people who have difficulty seeing how to behave like blind people" (p. 336).

Part of the difficulty in reviewing and assessing the research about facially disfigured persons lies in the various definitions of the subject samples used. These range from those seeking elective cosmetic surgery, such as rhinoplasties and face lifts, to those seeking facial prostheses after surgery or injury. Confusion may arise and conclusions may become suspect when investigators compare patients seeking changes in essentially normal faces with those who have had extensive facial injuries.

Characteristics of Those Seeking Elective Cosmetic Surgery

There have been a number of attempts to assess the motivations and personality characteristics of patients seeking elective cosmetic surgery. Jeppson (1963), using a psychiatric approach, feels that these patients display a greater degree of neurotic behavior than do those with severe facial malformations. Psychiatric evaluations of males seeking cosmetic surgery revealed that all warranted a psychiatric diagnosis. Eighteen of 117 patients were selected for evaluation, and the diagnoses included neurosis, psychosis, and severe personality disorders (Jacobson et al., 1960). Another study of 98 patients seeking cosmetic surgery led Edgerton, Jacobson, and Meyer (1960) to conclude that the typical patient seeking cosmetic surgery had serious personality disorders. Hill and Silver (1950) assessed the motivations for surgery of 48 consecutive patients. While expressing dissatisfaction with their appearances, these patients expressed concerns about their own identities, and an overwhelming number of them displayed neurotic characteristics. There was some indication that the search for surgical solutions to the problem of appearance was part of a neurotic defense mechanism. Druss, Symonds, and Crikelair (1971) were impressed with the number of somatic delusions in patients seeking cosmetic

surgery and stressed the need for psychiatric consultation before plastic surgery.

Behavior of Those With Observable Facial Deformities

How is the behavior of those having observable facial deformities described? Victorin, Hillerstrom, and Sorensen (1969) evaluated 95 patients before surgery for jaw malformations. They reported that these patients dwelled somewhat excessively on their appearance, were somewhat shy in establishing relationships with the opposite sex, and the majority of them had some minor psychological disturbance. Hirschenfang et al. (1969), using a group of personality tests, examined patients with facial paralysis. Patients of both sexes exhibited depressive patterns and preoccupations with illness.

Psychiatric evaluations of 12 cases selected for intensive study from a subject pool of 312 patients undergoing surgery two to five years after facial disfigurement had occurred, revealed three types of patients: those who were normal, those with inadequate personalities who used the handicap as a defense, and those who were prepsychotic or psychotic (Baker and Smith, 1939). Jeppson (1963), using a similar classification, found that adolescents who needed surgery because of peer relationships and patients whose appearance prevented them from achieving realistic vocational and social goals fell into a normal or healthy group. There is evidence that frustrations of vocational goals do exist. Pinner (1963), in a report of employment service practices in New York, reports that 75% of the job placements of the facially disfigured are made in service and unskilled occupations where the person works by himself.

In the 74 patients she interviewed extensively, Macgregor (Macgregor et al., 1953) was able to discriminate three reaction patterns: withdrawal, hostility, and successful coping with the situation. In an earlier study of 115 patients, Macgregor (1951) found withdrawal patterns to range from withdrawal from social activities to psychotic types of behavior, while hostility was manifested in interpersonal conflicts and antisocial behaviors. Coping behavior was manifested by developing excessive charm and friendliness, by making facetious remarks, and, in the severely disfigured, by giving others the opportunity to recover from their shock before proceeding with the relationship. Macgregor (1951, 1970) concludes that the degree of disfigurement is not proportional to the psychic distress experienced by the patient. Perhaps, as Abel and Weissmann (1953) suggest, this is because those with severe deformities repress their fantasies and feelings and avoid close interpersonal contact. Since the facial disfigurement is so apparent, the person is seen as having an obvious social handicap, and he is excused from entering into close interpersonal relationships.

More recently, in a doctoral dissertation, Sieka (1970) investigated the relationship of facial disfigurement to the acceptance of disability and its im-

pact on sex role evaluations. A 55-item acceptance-of-disability scale was given to 40 disfigured patients seeking facial prostheses. These patients, as well as 40 normal controls, were asked to evaluate their own sex roles. In addition, 25 husbands and wives were asked to rate the sex roles of their disfigured mates. In a semantic differential format, the sex role evaluations involved rating the person as a mate, a sexual partner, a companion, and in social relationships with acquaintances of the same and opposite sex. Sieka found that the facially disfigured received significantly poorer sex role evaluations than normal controls. No significant effects for sex or marital status were obtained. Differences between the disfigured patient's rating of his own sex role and his mate's rating of his sex role were examined. When his mate rated the patient higher than he himself did, his acceptance-of-disability score was high. When the mate rated the patient lower than he himself did, no relationship to acceptance of disability was found. Unfortunately, no information concerning the feeling husbands and wives had for each other, or for their marriages, is available. One cannot assume that the history of the patient starts with the disfigurement; information about the previous marital adjustment and the status of the person is necessary if one is going to determine the effects of becoming disfigured.

SUMMARY AND CONCLUSIONS

I would like to briefly recapitulate some of the major themes of this presentation, at the same time redirecting attention to the subtitle, "Speculations in Search of Data."

1. Consensual validation is apparent in efforts to determine the minority or marginal status attributed to the facially disfigured. All pay homage to the notion that our society places a premium on physical attractiveness, and reason, usually by analogy, that the disfigured, violating sensibilities, must be assigned a special status. Case studies do indeed find this to be true for many, if not most of the patients interviewed. One need only point out, however, that the sampling may be biased, since almost all of the cases involve those who are brought or come to hospitals seeking to alleviate a condition about which they are dissatisfied.

2. The classification schema used to describe physical conditions has limited psychological predictive value. The dictum that the presence of an anomaly cannot imply psychological pathology must be repeated. It has also been strongly demonstrated that clusters of personality variables are not associated with any disability. The evidence is overwhelming, for example, that a "cleft palate personality" does not exist.

3. It has been assumed that the existence of a child with a facial anomaly has strong impact on the family and that the effect is long enduring, yet this has not been adequately documented. Further, little attention has been paid to conditions existing in the family before the birth of a child with a defect.

4. Underlying many approaches to the study of the facially disfigured is the

implicit assumption that the condition, in and of itself, must affect the basic personality structure. Attention has been focused primarily on gross effects and on the adaptation of the person to his appearance. Clinical studies stress negative adaptation, yet a surprisingly large number of facially disfigured persons make more than adequate adjustments. At this point, no one can predict who will or will not make an adequate adjustment, and prospective studies are lacking.

5. It is eminently clear that psychological information about many of the orofacial anomalies is nonexistent. The information available, for the most part, consists of case histories and clinical speculations. Further, many of the studies are incredibly naive with respect to both basic theory and scientific methodology. This lack of ideational and methodological sophistication places conclusions drawn from these studies in serious doubt.

ACKNOWLEDGMENT

The preparation of this paper was supported by Research Grant No. DE 01899 from the National Institute of Dental Research. The author is affiliated with the Division of Plastic, Maxillofacial, and Oral Surgery, Division of Child Psychiatry, Duke University Medical Center.

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SURGICAL CONSIDERATIONS:
TEAMWORK IN THE SUCCESSFUL
TREATMENT OF HEAD AND NECK CANCER

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In spite of the intensified efforts of university research scientists for over a decade, and in spite of the dedicated efforts of the National Cancer Institute, the American Cancer Society, and various private foundations, the cause of cancer continues to be nature's well kept secret. Occasionally, the light of discovery flashes momentarily only to dim in spite of repeated attempts to keep it going. Peyton Rous, the famous Rockefeller Institute pathologist, stated it well when he said, "Tumors destroy man in an unique and appalling way, as flesh of his own flesh, which has somehow been rendered proliferative, rampant, predatory and ungovernable." Yet in 1966, four years before his death, when Rous was awarded the Nobel Prize for his viral work on chicken cancers, he said in his acceptance speech, "We have no inkling of what happens when a cell becomes neoplastic, nor how its power is passed, nor when, as it divides" (Eisenberg, 1971).

Gains on the cause of mankind's second greatest killer have been distressingly slow, but data have been compiled and today we have many clues to the mystery, including multiple etiological factors and some epidemiological hints. These have been coupled with a clearer appreciation of molecular cell biology since the discovery of DNA and RNA. Immunology may well have a place in the final solution. Scientists today are forced to recognize that the problem of cancer in mankind will not be easily solved. A complex abnormal cellular growth arises, probably stimulated primarily by two or more inherent factors whose intricacies are compared to double or triple locks. Just what turns RNA on and what turns it off? These are two perplexing questions still to be answered.

While we may already have several keys, important research today is directed toward discovering which combination of keys is necessary to unlock and incite which particular cancer in any particular area of the human body. Once the working combinations are demonstrated repeatedly, we shall be close to answering the questions.

One relevant and more noteworthy discovery came partly from the experi-

ments of Andre Lwoff and later from the work of Renato Dolberco, who studied viruses called phages, which were capable of infecting a bacterium, multiplying on it, and then killing it. One important observation recorded was that frequently when the virus did not kill the bacterium the virus mysteriously disappeared. During this interval, the bacterium grew normally and seemed unaffected by the virus apparently hidden within it. Some time later, the virus abruptly came to life, multiplied, and eventually destroyed the bacterium. When working with polyoma virus, Dolbecco discovered that either the normal cell quickly became neoplastic or the virus seemed to disappear. An "infected cell" probably remains dormant for an unpredicted period of time, but each time the cell replicates, the polynoma DNA is attached so all new cells contain the "time bomb" or first key. The conclusion drawn is that viral DNA can be passed on from generation to generation. It is believed that this "infected" cell, designated the oncogene, normally is "turned off" or locked, but given the proper set of circumstances, the second key, this cell may be switched on or unlocked. According to some authorities, this second key could include cigarettes, chemicals, excessive drinking, trauma, pollutants, hormonal imbalances, or old age—all an answer, in part, to the riddle of cancer's cause. While positive proof is lacking, this modus operandi is currently respected as an explanation. Figure 1 is a schematic representation of the double key theory.

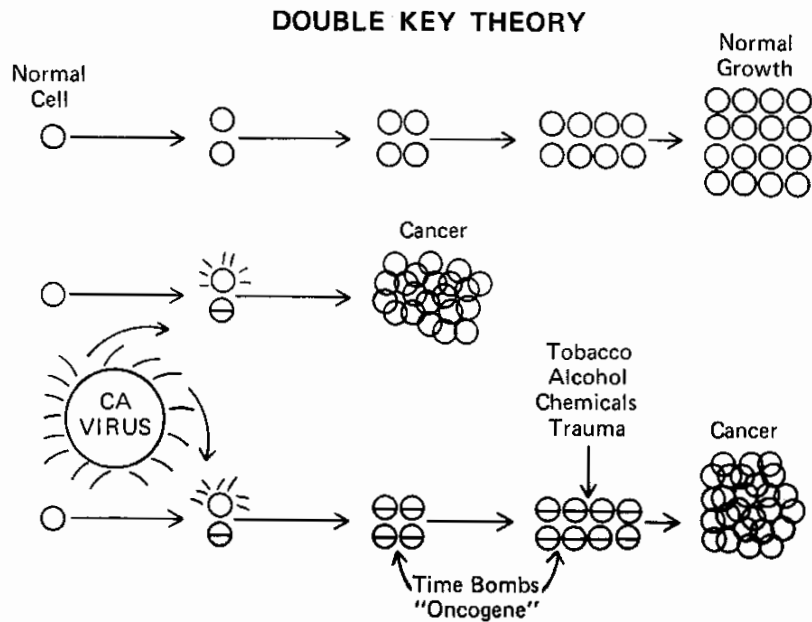


FIGURE 1. Schematic representation of the double key theory, showing how a normal cell may become cancerous.

Good statistical evidence exists today that tobacco has a cause-and-effect relationship to the development of oral cancer. In the Travancore region of India, 46% of all cancer cases are oral malignancies. In this area, tobacco is mixed with betel nut, cloves, and sometimes with camphor and slaked lime, and it is held in the mouth as a quid and chewed. In a study of oral cancers, a definite correlation has been established between the site of cancer within the mouth and the site where tobacco containing a quid has been held (Orr, 1963).

Prolonged exposure to sunlight has long been suspected as a cause of skin cancer. Caucasians with less pigmented skin develop a higher incidence of cancer than blacks with heavily pigmented skin. Farmers with a continual exposure to the sun have a higher incidence of lip cancer than blue-collar workers. Ill-fitting dentures, sharp teeth, and chewing of buccal mucosa are suspected irritants. Excessive alcohol consumption, particularly where cirrhosis of the liver has been demonstrated, is associated with over 50% of all the oral cancers. The theory is that cirrhosis affects the transport of vitamins and results in a vitamin deficiency. Vitamin deficiencies, particularly vitamin B, have long been associated with premalignant conditions such as lingual glossitis, chylosis, and fissures. Since the beginning of medical records, there has been speculation regarding the relationship of diet to the development of cancer. In the oral pharyngeal and upper esophageal regions, signs of anemia related to diet have been found and recognized as relating to Plummer-Vinson syndrome. This complex syndrome is characterized by hypochromic microcytic anemia, hypothyroidism, perioral fissures, glossitis, premature edentia, brittle nail beds, and dysphagia. In Sweden, an association has been noted between Plummer-Vinson syndrome and cancers of the hypopharynx in women.

STATISTICS

In a hypothetical town of 100,000 people, 25,000 are destined to suffer from cancer. Fifteen thousand will die of cancer. Cancer in this community will kill three times as many people as will all accidents, including those caused by automobiles. Forty-five hundred people eventually will develop head and neck cancer and approximately 2000 will die from it (American Cancer Society, 1972).

When one addresses himself to the magnitude of the head and neck cancer problem, it is apparent that statistics can be used to prove that only 4% of all cancers are in the head and neck; however, one could slant them to prove that 20% occur in the region of the head and neck. If only the oral cavity and the larynx cancers are included as head and neck cancers, one comes up with a small though lethal group. If one extrapolates the head and neck cancers from the other more general listings of the American Cancer Society, shown in Table 1, a much higher percentage of head and neck cancers is reached. A complete list should include cancers of the nasal cavity and sinuses, the trachea, facial bones, facial connective tissue, the cervical esophagus, the throat, and

TABLE 1. Incidence and mortality of head and neck cancer (American Cancer Society, 1972).

<i>Site</i>	<i>New Cases</i>	<i>Deaths</i>	<i>Total Cases</i>	<i>% Head and Neck</i>
Oral	15,000	7,500	—	100%
Larynx	6,800	3,050	—	100%
Other and				
Unspecified Respiratory	1,750	900	2,500	75%
Cervical Esophagus	1,890	1,450	6,300	30%
Thyroid	2,600	1,150	—	100%
Facial Bones	380	180	1,900	20%
Connective Tissue	480	300	1,650	30%
Lymphomas	5,000	3,960	19,800	20%
Other	6,800	4,750	34,000	20%
Subtotal—Head and Neck	40,700	23,240		
Skin	88,000	3,900	118,000	75%
Total—Head and Neck	128,700	27,140		
Total Cancer Cases	650,000	345,000	Head and Neck Cancer =	7.8% and 20%

the lymphomas occurring throughout the head and neck. Skin cancers should also be included in this group, since 75% occur in the head and neck. With these recommended additions, we now have a list which accounts for about 20% of all new cancer cases. If one takes the mortality statistics for this revised list, he can conclude that 8% of deaths from cancer are caused by cancer of the head and neck. All statistics should be carefully evaluated before conclusions are drawn or judgments made. This is necessary especially when tackling the complete head and neck cancer problem.

Seventy-five percent of all head and neck cancers occur between the ages of 55 and 85. Most are epidermoid carcinoma. Disease rarely occurs before the age of 40. A ratio of 1:10, dominated by lymphomas, occurs in children. Males are affected more often than females, by a rate of about 4:1. The disease kills by spreading, as do all cancers, and in these cases it usually is by direct extension or by lymphatic invasion into an adjacent region. Metastases into the liver, spine, or brain are rare, but do occur. It continues to be discouraging that head and neck cancers are not recognized earlier, since special examinations are done more easily in this region than in any other internal region of the body, because of the unique anatomy. Seventy-five percent of head and neck cancers actually can be visualized by tongue depression. Eleven percent of head and neck cancers can be palpated, and only 15% require special diagnostic equipment such as x rays, endoscope, nasopharyngoscope, or mirrors. Once suspected, the diagnosis can be made by either an aspiration biopsy, a punch biopsy, an excisional biopsy, or, in some few cases, an incisional biopsy. In our experience, the percentage and types of head and neck cancer are as follows:

- 85% Squamous cell or basal cell from epithelial cells which line oral, nasal, and sinus caviites; nasopharynx; ear; upper esophagus (also cover scalp, forehead, and neck)
- 10% Adenocarcinoma, cylindromas, mucoepidermoid, lymphomas
- 5% Rare: for example, olfactory esthesioneuroblastoma, neurogenic, rhabdomyosarcoma, fibrosarcoma

If the histopathology is positive, a surgeon must decide how to destroy the tumor. If the tumor is radiosensitive, radiotherapy may be the treatment. However, if the tumor is a squamous cell carcinoma, which the greatest percentage of head and neck cancers are, surgery might well be the best treatment. The final decision usually depends on the extent of the tumor and its specific location. If surgery is selected it could be a simple excision, a complicated excision, or a radical or ultraradical excision. If radiotherapy is used, x rays may be used at either low, medium, or supravoltage. In recent years, the gamma rays from Cobalt 60 and Cesium 137 have been used. Occasionally implants using radium, radon seeds, or needles are used. Radioactive isotopes I 131 are useful in thyroid cancer.

There has been intermittent interest in the use of chemotherapy, either in combination with radiotherapy or surgery, or for terminal palliation. Cryosurgery, the freezing of tumors to destroy cancer, is also a useful treatment. Cryosurgery may be the primary treatment in selected cases, but it usually is used in combination with the other more common methods of treatment. The different approaches to destroying tumor are four major types:

Surgical

1. Minor—simple excisional
2. Major—complicated excisional
3. Radical and ultraradical

Radiotherapy

1. X rays—low, medium, mega, supervoltage, linear acceleration, and betatron
2. Gamma rays and CO 60, Cesium 137
3. Implants—radium, radon seeds, and needles
4. Radioactive isotopes—I-131

Chemotherapy

Combinations

1. Surgery
2. Radiotherapy
3. Chemotherapy
4. Cryosurgery

MAXILLOFACIAL TEAM

A patient with head and neck cancer has the best chance for survival and

rehabilitation if a maxillofacial team is provided who can take over the patient's complete care. The work of this group is referred to as the "total head and neck team approach." The members of this maxillofacial team are these:

surgeon	prosthodontist and laboratory
internist	psychologist
speech pathologist	social services
nurses	orthodontist
inhalation	audiologist
intensive care	physical therapist

Surgical Procedures

Simple excisional techniques often are all that are needed to control and cure small cancers. This is true particularly for skin, small lip, and intraoral cancers. In addition, early cancers of the epiglottis and small T₁ laryngeal cordal cancers may be removed by endoscopic techniques. In all of these situations, surgery is so directed that a circumscribed border of normal tissue around the local cancer is removed. If removal is early, the cure rate is high in these cases and the rehabilitation problems are minimal. Complicated excisional techniques are used to remove large localized cancers. These complicated cases involve not only the removal of the cancer and the area of normal tissue around the cancer, but often also necessitate the removal of vital, functioning portions and structures whose removal leads to disfigurement. An example is cancer of the nasal cavity or paranasal sinuses where frequently a part or all of the nose must be removed. Often it is necessary to remove the eye and the contents of the orbital cavity. These procedures leave defects which must be surgically reconstructed or repaired by the use of prosthetic appliances. Sometimes we must remove part or all of the hard or soft palate, or both. This leaves the patient in a particularly crucial state, since he may have difficulty speaking and swallowing. In these cases, the need for rehabilitation by a prosthodontist is imperative. Other examples of complicated head and neck excisional techniques are those used when part or all of the larynx is removed. In the former, although the voice is preserved, a great deal of speech rehabilitation is necessary to produce satisfactory communication. After a total laryngectomy, the patient must be taught how to develop esophageal voice or use an artificial larynx.

Neoplasms of the thyroid necessitate a subtotal or total thyroidectomy. The complicating factor here is that the recurrent laryngeal nerves are near the excision. At least one of these nerves must be preserved; otherwise the patient will have difficulty breathing and will require a tracheostomy. If one nerve is sectioned, the patient may have minimal trouble breathing, but, more important, he will have difficulty speaking and often need the services of a speech pathologist.

Although most tumors of the parotid are benign, they may recur unless adequate tissue around the tumor is removed. This is not always easy to do and often presents a problem, since the facial nerve—which innervates all of the facial muscles—passes through the center of the parotid gland. To be thorough, a surgeon frequently must dissect the neoplasm from the facial nerve. This can result in a prolonged paralysis which requires the services of a physiotherapist, who must instruct the patient in passive and active stimulation of his facial muscles. The physiotherapist also measures the electroexcitability of the nerve, since this is important in determining the prognosis. If the patient has cancer of the ear, it may or may not be possible to preserve his hearing. In cases where hearing is impaired or totally destroyed after surgery, the services of an audiologist are important.

When cancer of the head and neck leaves the primary area, it usually metastasizes into the lymph nodes in the neck in a fairly orderly pattern. Cancer in the cervical lymph nodes is curable by a radical neck dissection. This operation is designed to remove all of the lymph node stations through which cancer might spread from the intraoral cavity or larynx to the other areas of the head and neck. When the surgeon knows that a primary tumor has a high incidence of spreading to the cervical region, an elective neck dissection often is performed at the time the primary tumor is resected. A neck dissection performed to remove a palpable node in the neck is referred to as a therapeutic neck dissection. Neck dissections may be performed on one side or on both sides during the same surgery. When a bilateral neck dissection is performed, the surgeon frequently saves one of the internal jugular veins to alleviate postoperative morbidity. If both jugular veins are resected, the patient's face becomes distorted by venous congestion and generalized edema, and it may take weeks and sometimes months for this to subside. During this period a patient needs reassurance and support from everyone, including nurses, family, and occasionally a psychologist.

When a primary lesion in the tongue, tonsil, mandible, floor of the mouth, buccal succus, pharynx, larynx, esophagus, parotid, ear, or thyroid is combined with radical neck dissection, it is referred to as a composite resection. At times, it is referred to as a "commando" or a combined resection. In selected cases of recurrent cancer, particularly after a laryngectomy and radical neck dissection, the surgeon will remove a previously healed laryngostoma and lower it into the upper chest by removing the clavicles and the sternum. This technique of removing the sternum and clavicle also is used in selected cases for removing large thyroid, cervical esophageal, and advanced laryngeal cancers, for it enables a surgeon to obtain a wider margin of resection.

Rehabilitation and Surgical Reconstruction Procedures

Every effort is made to preserve the vital nerves and arteries necessary for adequate chewing and swallowing. In many advanced cases when vital struc-

tures are removed, the surgeon must employ reconstructive surgical procedures to repair or reconstitute this function. An example of this is arterial grafting to reestablish blood flow to the brain. Autogenous vein or synthetic arterial grafts are used. Large regional flaps from the chest and neck are used to reconstruct the esophagus or pharynx. Autogenous bone grafts are employed to reconstruct the mandible. Nerve grafting is used to reconstruct the facial nerve and the spinal accessory nerve. In the latter case, losing the XIth nerve leads to a shoulder drop which incapacitates the patient because of the partial loss in the use of the upper arm. Although removing part of the face can distort or sometimes greatly disfigure the patient's general appearance, it is possible to replace portions of the nose, cheek, face, and chin with bone grafts or metallic implants. Unfortunately, the surgeon is restricted in his efforts to reconstitute these body parts so that they are acceptable cosmetically and esthetically to the patient and society.

Rehabilitation by Prosthodontistry

The prosthodontist, the orthodontist, the periodontist, and the dentist are vitally important to the successful management of the head and neck patient. They not only render valuable services during treatment by the surgeon, but also often aid the radiotherapist. At the M. D. Anderson Hospital, at the University of Texas, 20% of head and neck cancer patients require the services of the prosthodontist. Frequently, devices are provided that will shield the undiseased area, to protect normal tissue from unnecessary radiation. In addition, prosthodontists construct appliances to apply intracavitary radiation to a diseased area. These appliances are used to hold the diseased tissue in constant contact with the radioactive material. Certain appliances also are used to seal off one cavity from another during the healing process. In the reconstruction of the mandible, appliances are necessary to hold bone grafts in place. If large doses of radiotherapy are contemplated, dentists are consulted about the application of fluoride to the teeth, and about teeth that should be removed, before therapy. Inadequate chewing or the inability to chew can be alleviated by the skillful application of appliances to the remaining teeth. Swallowing can be made tolerable by constructing an appliance which seals off the walls of the nasal pharynx and nasal cavity from the oral cavity. The proper construction and use of these appliances are indispensable to developing intelligible speech. In cases where the cervical esophagus must be temporarily interrupted, the prosthodontist may make an esophageal bypass which directs the saliva from the oral pharynx to the esophagus. This spares a patient the embarrassment of continually facing his friends and family with saliva spread all over his neck and clothes.

If the surgeon fails to provide an acceptable facial contour, the prosthodontist contributes considerably to the patient's psyche when he provides a well-matched synthetic facial piece. These can contain an eye or sometimes a

part or all of a nose. When the prosthodontist creates a nose or an ear, it is always more acceptable in appearance than the one the surgeon might construct. Oversized dentures are valuable for holding out the upper lip or improving the facial contour. The only objection is they are false.

Speech Pathologist

The speech pathologist, an important member of the team, should meet the patient before surgery and should work closely with the surgeon so that he is aware of the problems anticipated during the postoperative rehabilitation. The services of the speech pathologist are not only valuable to the laryngectomized patient because an esophageal voice must be taught, but also, and equally important, because intelligible speech must be acquired by patients who have had partial or total resections of their tongue, palate, or pharynx.

Clinical Psychologist

A patient's psyche is always in jeopardy when his appearance or vital functions are altered or threatened, and he may become depressed, hostile, or even recriminatory after surgery has been performed. A patient must be retrained and taught to live with a residual physical disability. The clinical psychologist is directed toward this retraining, not only so the patient can live within the limits of his disability, but also so he will live to the best of his remaining capabilities.

The clinical psychologist should follow through by initiating the patient's contact with a laryngectomee club, and he should advise the patient of other rehabilitation groups and the services of the American Cancer Society.

Complete success in treating the patient afflicted with cancer of the head and neck should be achieved by the total involvement of a maxillofacial team which necessitates the services of many disciplines.

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PROSTHETIC CONSIDERATIONS IN ORAL ABLATIVE SURGERY

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The prosthetic considerations related to patients undergoing orofacial ablative surgery are many and varied. One of the main considerations is the number of patients who are, or will be, in need of prosthetic rehabilitative procedures. According to the American Cancer Society publication *'72 Cancer Facts and Figures* (1972, p. 4), there are one and a half million Americans alive today who have been cured of cancer. An additional 700,000 cancer patients diagnosed and treated within the last five years will live to enter the ranks of those called cured. This means that there are actually more than two million patients who have been treated for cancer and are alive and free of disease today.

Many patients are treated for their primary tumor, told that they are cured, and discharged. One such patient seen recently in the Regional Maxillofacial Restorative Center in Houston had been free of disease for three years following surgical resection of the mid-third of his face. He had also been a recluse in his home for those three years. I feel that with patients such as this we must consider other factors, such as how the patient performs—not necessarily in relation to others, but in relation to his everyday function. How does he eat? How does he talk and can he be understood? Is his appearance acceptable to the people with whom he should be in contact during daily activities? This patient had difficulty eating. His speech was unintelligible. His appearance was unacceptable even to some members of his own family. Perhaps it is time that we change the definition of *cured* to read, "To free the patient of disease and, as much as possible, to restore him as a functioning member of society."

Reconstructive surgical procedures are certainly the treatment of choice in many instances and should be used as long as the results are as good as or better than prosthetic reconstruction. I do believe, however, that both kinds of treatment should be available to serve the patient better. This is born out, in part, by patients with lesions of the palate treated by surgical resection that results in cleft palates. All of the problems inherent in a congenital cleft are present, except that they are accentuated. The patient has not had long to acclimate and accommodate to this break in the continuity of the palate and,

therefore, finds his problems more difficult. Presurgical treatment planning and discussion by those who deliver all the available treatments can prevent the patient from having many of these problems, or their effect can be diminished.

Comparatively simple and easily constructed prostheses can aid the surgeon in holding packing in the defect and in closing it, do away with the need for a nasogastric feeding tube, prevent the patient from acquiring compensatory speech habits, and reduce the average hospital stay by one third. This last benefit saves the patient money and makes more hospital beds available.

By far the best type of rehabilitation is preventive rehabilitation. If the problem can be prevented, the need for all sorts of heroic procedures can be obviated. Certain problems are not as obvious as the surgically created cleft, and some would not occur until a much later time. Such a situation is demonstrated by the youngest patient treated in our center who, at 10 weeks of age, was treated by radiation for a tumor of the upper lip. A protective radiation stent was used during her treatment to prevent later complications in tooth development and growth in the upper jaw.

This patient also illustrates the wide age range involved in this type of patient care. She is our youngest patient, and our oldest patient is a woman of 101 years of age. In a study by Jesse (1965) of paranasal sinus tumor patients, the average age was 57, a young age at which to be incapacitated by a surgical defect of the palate.

Where do these patients come from, and what types of procedures do they need in their treatment and rehabilitation? The first patient described above came from a state adjacent to Texas and illustrates another problem we are confronted with in prosthetic rehabilitation—the lack of knowledge that maxillofacial prosthetic services are available. Unfortunately, this patient was not referred by the surgeon who had done the primary procedure, probably because he did not know this service was available. The patient was finally discovered by a field vocational rehabilitation counselor after existing in his postsurgical condition for more than three years.

This represents one source of patient referrals. Most patients, however, come directly from physicians, both surgeons and radiation therapists, who are involved in treating the primary disease. We have found at our center over the past several years that approximately 22% of head and neck tumor patients require maxillofacial prosthetic restoration. These prosthetic procedures include three basic types: (1) implant prostheses such as facial bone replacements, cranial bone prostheses, and soft tissue buildup prostheses; (2) removable oral prostheses such as modified complete and partial dentures, removable resection appliances, obturators, and trismus appliances; and (3) facial prostheses for the artificial replacement of lost structures. The fabrication and fitting of these prostheses at times becomes tedious, involved, and fairly expensive. However, regardless of the cost of rehabilitative procedures, they total up to much less than the cost of maintaining patients on welfare. Also, these costs do not include the terrible psychological costs to the patients.

Many factors enter into the decision of whether to use plastic surgery or maxillofacial prosthetics or both: age and medical condition of patient, type and size of defect, and time and cost factors.

Another consideration should be the use of interim prostheses between surgical procedures, such as the one used on a patient who had an angiofibrosarcoma of the nose, an especially vicious tumor. The tumor was removed and the patient was a prime candidate for plastic surgical repair. With the nasal cartilage remaining and nasal septum intact, a forehead flap could have been rotated to reconstruct his nose. In this case, due to a questionable margin, the surgeon elected to leave the area open for visual and digital examination to assure that any recurrence or renewed activity would be seen at the earliest possible time. However, this patient was a physician and could no longer practice his profession in this condition. A facial prosthesis replacing his nose was made for him and he continues to be a productive individual. He returns to the clinic periodically, removes the prosthesis, is examined, and, if no disease is found, returns to his practice. In the seven years since surgery for his primary tumor, this patient has had four recurrences, all of which have been surgically removed. By alterations of his prosthesis during this period, he has continued to function as a contributing member of society.

We all can see the value of returning a physician to society, but should not overlook the value of all individuals. We must include not only people in the working force, but all people. A housewife has the same problems of function and esthetics and must be rehabilitated as well as the patient who is gainfully employed. R. Lee Clark, president of the University of Texas at Houston, M. D. Anderson Hospital and Tumor Institute, has said many times in recent years that we can no longer be satisfied with only increasing the number of patients surviving, but that we must also improve the quality of this survival.

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SPEECH CONSIDERATIONS:
SPEECH DISORDERS ASSOCIATED WITH
ABLATIVE SURGERY OF THE FACE,
MOUTH, AND PHARYNX—ABLATIVE
APPROACHES TO LEARNING

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In his master's thesis at the University of Michigan, Brien Lang (1965) notes that on July 1, 1893, Grover Cleveland underwent surgery for the removal of a cancerous lesion of the left maxillary antrum. A second operation was performed on July 17, 1893, to make certain that no fragment of malignant tissue remained. An immediate appliance constructed of vulcanite rubber was inserted at that second operation. By October the operated area had healed adequately, so that an obturator could be constructed of hard rubber, fused with an upper dental appliance. When he opened the Pan-American Medical Congress in Washington on September 5, 1893, Cleveland's voice was said to be as clear as on March 4, when he had delivered his inaugural address as the twenty-fourth President of the United States. This procedure, carried out in the greatest of secrecy, was unknown to the American public until September 22, 1917, 24 years later.

The foregoing account introduces the topic of this paper—the effects which surgical removal of tissues of the face, mouth, pharynx, and nose may have upon speech. Unfortunately, all subjects thus treated do not have the excellent results reported for President Cleveland. The threat of partial or complete loss of communication by speech is a matter of grave concern for a majority of patients scheduled for ablative surgery.

Speech is a highly complex behavior system which requires accurate and rapid control of musculoskeletal valves of the mouth, pharynx, and larynx. Although normal phonation is dependent on the functioning of the vocal folds and hence must be considered in a total discussion of disorders of speech affected by ablative surgery, this paper is limited to consideration of only those valves involved in speech articulation and resonance above the larynx. The effects of laryngectomy and auditory disorders are thus excluded from discussion, as are dysfunctions resulting from disorders of the central nervous system or sectioned or damaged peripheral nerves.

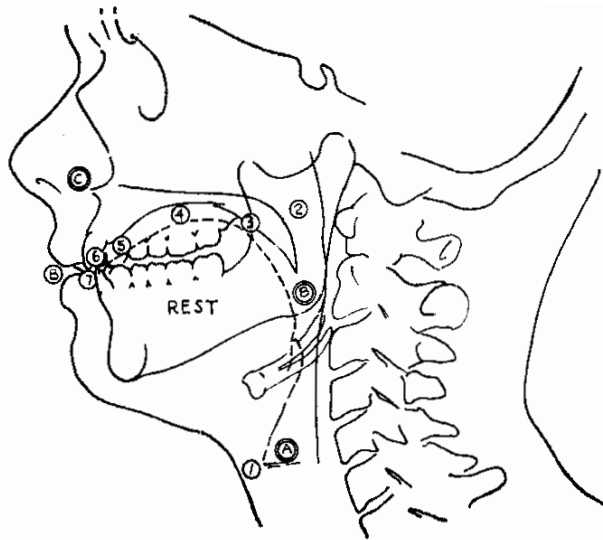


FIGURE 1. Musculoskeletal valves employed in speech and the consonants they form: (1) glottal—*h*; (2) palatopharyngeal—(*m*), (*n*), (*ng*); (3) linguovelar—*k*, *g*, *ng*; (4) linguopalatal—*sh*, *zh*, *ch*, *j*, *r*, *y*; (5) linguoalveolar—*t*, *d*, *n*, *s*, *z*, *l*; (6) linguodental—*th*, *th*; (7) labiodental—*f*, *v*; (8) bilabial—*p*, *b*, *m*, *w*, *wh*. Voiced consonants are in italics, indicating simultaneous action of the laryngeal valve and one or more articulatory valve. The parenthesized listing of (*m*), (*n*), and (*ng*) signifies that the palatopharyngeal valve is relaxed for their production. This valve closes for all other consonants and vowels. A, B, and C represent accessory valves not used in normal speech.

The valves with which we are concerned are depicted in Figure 1. Their respective designations and the consonants they produce are listed in the legend. Note that in the legend /*m*/, /*n*/, and /*ng*/ occur in two places, signifying that they are nasalized consonants which also require articulatory valving of oral structures, with concomitant relaxation of the palatopharyngeal valve. The place at which structures are functionally involved in producing the consonants is illustrated in Figure 2.

The speech mechanism, in simple terms, should be visualized as a tube extending from the laryngopharynx to a bifurcation at the nasopharynx and proceeding thence via its dual oral and nasal pathways. The musculoskeletal valves described above close, constrict, and open this bifurcated tube to regulate the breathstream and the laryngeally generated sound. These valves influence air pressure, modify and direct airflow, and modulate vocal resonance for production of vowels and consonants. In General American English there are 25 consonants and approximately 14 vowels and diphthongs which are recognized as the basic phonemic elements of speech.

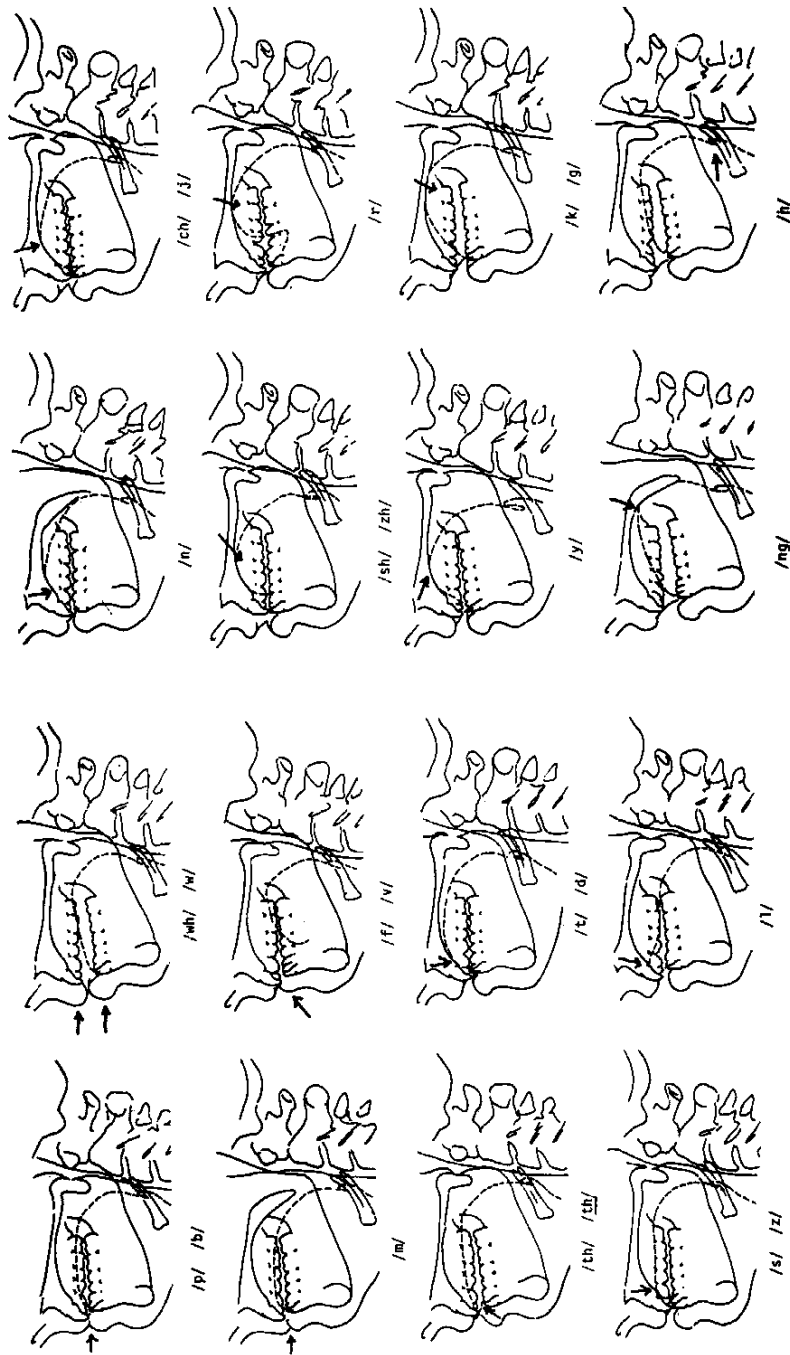


FIGURE 2. Radiographs showing positions of the articulators at a critical moment in consonant production. In each instance where consonants are paired (for example, /ch/ as in church, /j/ as in judge) the illustrative x-ray outline tracing was made of a normal subject enunciating the first consonant of each pair. The articulatory position is approximately the same for each of the voiced and unvoiced pairs. (Adapted from Bloomer, 1971.)

Valve impairment and dysfunction are primary causes of defective speech and poor speech intelligibility. Since ablative surgery may cause irreparable destruction of valve function, it is important for us to identify (1) the sites and types of lesions which may impair or destroy that function; (2) the speech disturbances which may follow tissue destruction, displacement, or removal; and (3) the means available for preventing or at least ameliorating the effects of tissue loss. Furthermore, inasmuch as the qualities of speech can provide significant measures of success in total treatment, the foregoing considerations justify continuing study of functional disturbances resulting from tissue ablation.

Common causes of tissue destruction are trauma, tumor growth, wasting diseases, and surgical removal. We are primarily concerned with the latter, although any of the others may contribute to the need for ablative procedures, and may of themselves impair valve function.

The sites of tissue loss which directly affect speech are those critical points in the mouth and pharynx which either destroy or impair the function of the articulatory valves referred to above, or create abnormal openings in the tube, thereby altering tube resonance characteristics and impairing its ability to modulate sound or to regulate airflow. Damage to the valves may result from cheilotomy, facial excisions, removal of nasal structures, maxillectomy, mandibulectomy, glossectomy, and nasopharyngeal excavations.

The surgical ablation of tissues and its effects on valve function for speech will be considered individually in reference to primary valve mechanisms in the discussion that follows. Some overlapping of dysfunction does occur, however, such as that attributable to combined glossectomy and mandibulectomy.

Facial expression and facial cosmesis are additional factors closely associated with speech. Facial expression and facial form are both important to the understanding of speech, although little is known of the degree to which effectiveness of communication is impaired by tissue destruction that disturbs facial form or expression during speech. It is presumed to have considerable influence, particularly on the communication of emotion.

With reference to cosmesis, Rusk (1963), in his introduction to the *Conference on Facial Disfigurement* at New York University Medical Center, reminds us that the face is "the focus of attention in interpersonal relationships . . . the area most closely identified with the intimate, personal entity that one calls 'himself.'" And further, that "the victims of facial disfigurement are subject to countless indignities and social deprivations" (p. XIII). The importance of such observations to a study of ablative surgery and speech has to do with the intimate relationship believed to exist between the psyche of the individual and the character of his speech. The two are difficult to separate.

ABLATION OF THE LIPS

The lips are involved in articulation of the consonants /p/, /b/, /m/, /w/,

and /hw/ and may contribute qualitatively to the production of /s/, /z/, /t/, /d/, and /r/. They also probably contribute in a secondary way to enunciation of the rounded vowels /ʊ/ (oo), /o/ (o), /ɔ/ (aw), /u/ (oo), /au/ (ow), and /ɔɪ/ (oy). Adequacy of lip function for speech is thus related to the ability of the lips to round, spread, constrict, close, or open rapidly.

Cancer of the lip is a common lesion, accounting for about 25% of oral malignancies. The tumor is usually one that arises on the vermilion margin of the lip, but may extend to involve the skin externally or the mucous membrane internally. The disease is said to involve the lower lip almost exclusively, and nearly all cases occur in men (Freund, 1967).

A quotation from the proceedings of an interprofessional conference on maxillofacial prosthetics (Robinson and Niiranen, 1966) held in Washington, D. C., in 1966 states that "an intact lower lip is more important for speech than the upper lip because of [its] greater potential for movement . . ." (p. 195). Because of tissue adaptability and associated activity of other organs such as the tongue, some patients are able to make "compensatory adjustments, even in the presence of a severe lip deformity."

Whereas swallowing activity is not a subject for discussion in this paper, it is nevertheless of interest that in his paper on "Swallowing Dysfunctions Associated with Radical Surgery of the Head and Neck," John J. Conley (1960) states that partial resection of the lips causes minimal interference; complete resection of the lips corrected by mimetic regional face flaps may cause drooling and make food ingestion more difficult, but does not affect swallowing *per se* (p. 603). It is presumed that Conley refers to a patient who had a "normal swallow" presurgically. If the swallow were abnormal, it is presumed that a lack of mandibular and dental stabilization will make postsurgical swallowing even more difficult.

An example of the functional effects of a shortened and relatively immobilized upper lip occurred in a patient secondary to removal of external and associated internal nasal structures for the treatment of carcinoma (Figure 3). Whereas there was little loss of speech intelligibility from lip deficiency, she was forced to substitute labiodental approximations for all bilabial consonants. These consonants were generally identifiable when embedded in a linguistic context. Consonants created by action of the tongue and structures other than the lips were not affected. She was able to use the tongue successfully in drinking through a straw, even though she was not able to seal the lips around the straw.

The effects of complete removal of the upper lip and the anterior two-thirds of the maxilla (Figure 4) are clearly shown in some movie sequences made nearly 20 years ago with the help of Reed Dingman and the late John Kemper (Bloomer, Dingman, and Kemper, 1952). A description and detailed study of selected sequences from the films was reported in 1953 (Bloomer, 1953). As reported, the patient was able to substitute tongue-tip adaptations for the bilabial and labiodental consonants which normally require lower lip valving



FIGURE 3. 68-year-old-patient who sustained surgery for removal of nose. Shortening of the upper lip from scarring affected lip function for speech and sucking.



FIGURE 4. This patient has lost all nasal structures by surgery, but retains a posterior third of the hard palate and an intact soft palate and nasopharynx. Speech is impaired but intelligible, because of spontaneous articulatory adaptations developed by the patient.

with the upper lip and with maxillary anterior teeth. Thus, when heard in sentences, the consonants /p/, /b/, /m/, /w/, /hw/, /f/, and /v/ were sufficiently well imitated that her speech was moderately intelligible.

It is difficult to state how much or in what ways vowel qualities were changed by her facial deformities. Most of the vowels seemed to be identifiable, but were obviously abnormal. Her nasal resonance was also presumed to be altered from presurgical resonance, but the changes that one heard are difficult to describe. A facial prosthesis designed for her was an attempt at cosmetic aid and to protect her from the weather, but was otherwise nonfunctional.

FACIOBUCCAL EXCISIONS

Faciobuccal tissue removals affecting speech will be considered by site in reference to the hard and soft palate—those above the palate, and those exposing the oral cavity.

Supramaxillary Excisions

Tumors of the paranasal sinuses and nasopharynx are sometimes diagnosed only after considerable delay, and the results of treatment may be inadequate. A majority of tumors are reported to arise from the antrum; however, because of the contiguity of the antrum, ethmoid, and nasal cavities, it is difficult to determine the site of origin or to isolate the effects of lesions (Freund, 1967, p. 291).

Several instances of tissue excisions creating openings in the face above the palate have been reported. An early report of a case was provided by Harrington in 1944. The functional consequences of this radical procedure are illustrated by the second patient (Figure 5) recorded in the film *Palatopharyngeal Action in Speech and Deglutition* (Bloomer, Dingman, and Kemper, 1952).

Several other patients with supramaxillary facial apertures have been observed. The effects on speech intelligibility and vocal resonance in such cases are believed to be relatively minor as long as the maxilla is intact and the palatopharyngeal valve functions satisfactorily.

Maxillectomy

Removal of any part of the maxilla, if not restored surgically or prosthetically, creates a serious problem for the speaker. Labiodental, linguodental, and linguopalatal contacts account for 17 of the 25 consonants used in American speech. These include the labiodentals /f/ and /v/; the linguodentals /θ/ (th) and /ð/ (th); the linguodentoalveolars /t/, /d/, /n/, and /l/; the linguopalatals /s/, /z/, /ʃ/ (sh), /ʒ/ (zh), /tʃ/ (ch), /dʒ/ (j), /j/ (y), and /r/; and the linguopalatovelars /k/, /g/, and /ŋ/ (ng). Without an effective maxillary tissue approximation for these consonants, speech becomes unintelligible.

Fortunately, prosthetic restoration is often available to ameliorate or correct

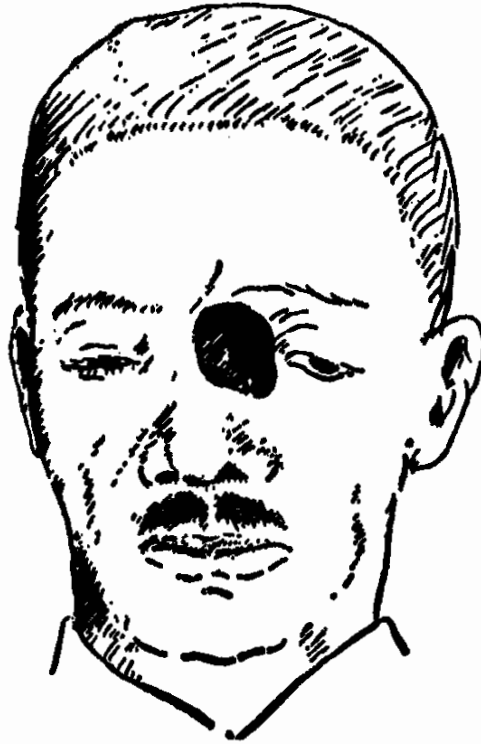


FIGURE 5. Patient with a surgically created inter-orbital defect. The nasal septum and some turbinate tissue have been removed, but the palate and nasopharynx are intact. The patient's speech is only mildly affected.

the problem created by tissue loss. Such cases presumably are of the type which Joe Drane, earlier in this *Report*, describes in reference to immediate postsurgical obturation.

A study of six maxillectomy patients has been reported by Leo Kipfmüller, a speech pathologist, and Brien Lang, a prosthodontist (1972). The patients, who ranged in age from 24 to 59 years and had individually variant degrees and sites of tissue loss, were provided with prostheses immediately after surgery. Three patients were edentulous and three had sufficient maxillary dentition to provide abutment support for a prosthesis. Approximately 10 days postoperatively (or as soon as impression procedures could be tolerated), the prostheses were modified by the addition of bulbs designed to obliterate the defect partially and to improve speech. Speech intelligibility was measured pre- and postoperatively, with and without prosthesis, by administering the Fairbanks Rhyme Test (Prins and Bloomer, 1965). The 50-word recordings of each patient were scored by 30 untrained listeners, thereby yielding 1500

informational items on each of the subjects. The results of the tests are shown in Table 1.

TABLE 1. Perceptual results based on listening to six maxillectomy patients produce the Fairbanks Rhyme Test stimuli. (From Kipfmüller and Lang, 1972.)

Patient Number	Date of Recordings	Condition and Number of Errors		
		Presurgery	With Prosthesis	Without Prosthesis
1	10-09-65	9	-	-
	11-02-65	-	64	733
	12-20-65	-	7	853
2	01-25-66	83	-	-
	04-19-66	-	265	588
	06-15-66	-	302	380
3	03-15-66	29	-	-
	07-11-66	-	435	1058
	12-07-67	-	302	1073
4	06-15-66	18	-	-
	08-01-66	-	154	1054
5	11-01-66	68	-	-
	12-06-66	-	131	1152
	01-11-67	-	124	1170
	07-18-67	-	52	1070
6	01-16-67	84	-	-
	02-16-67	-	389	666
	04-16-67	-	287	552
	12-20-67	-	167	447

Scores in the table show that whereas speech intelligibility is markedly impaired without prosthetic treatment, installation of a prosthesis improved speech intelligibility in all instances. Test-retest scores over elapsed time intervals ranging from two to 11 months show that speech intelligibility tended to improve after the patients became accustomed to the prosthesis. Lang has commented that during a period of two years postoperatively, the prostheses generally can be reduced in size each time the patient returns for consultation. This observation leads to speculation that nature appears to be trying to obliterate the defect in its own way through tissue proliferation and aperture reduction.

One of the patients, the oral defect, the prosthesis, and the prosthesis in place are shown in Figures 6, 7, 8, and 9. His intelligibility scores are Number 6 in Table 1. A recording of his voice and articulation demonstrated his hypernasal voice quality while wearing the appliance, and the altered resonance which distinguished his nonobtured speech. Examination of the patient showed that he also had deficient palatopharyngeal valve function, probably due to denervation and scarring of the soft palate. Not all of his speech defects, therefore, are due merely to the maxillectomy, but they reflect palatal dysfunction as well.

SOFT PALATE

Removal of posterior portions of the maxilla may affect palatopharyngeal valve function (1) by destroying the points of attachment for the palate on one or both sides, (2) through coincident denervation of palatal muscles, and (3) from relative shrinkage and immobilization of the palate through formation of scar tissue associated with the maxillectomy. The patient presented in Figures 6 through 9 exemplified a case in which nerve impairment and scar tissue formation disturbed palatal function. Obturation corrected the right unilateral maxillary defect, but left the patient hypernasal and with impaired intelligibility resulting from an incompetent palatopharyngeal valve. Observations of the patient's speech when unobturated revealed that the vocal reson-



FIGURE 6. Maxillectomy patient without prosthetic restoration.

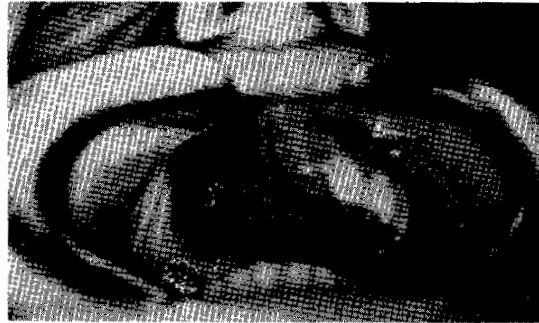
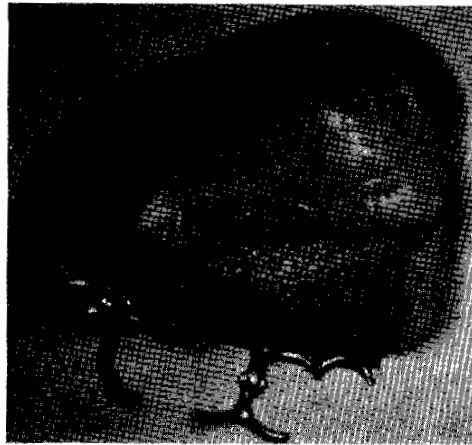


FIGURE 7. Maxillary defect.

FIGURE 8. Superior view of obturator.



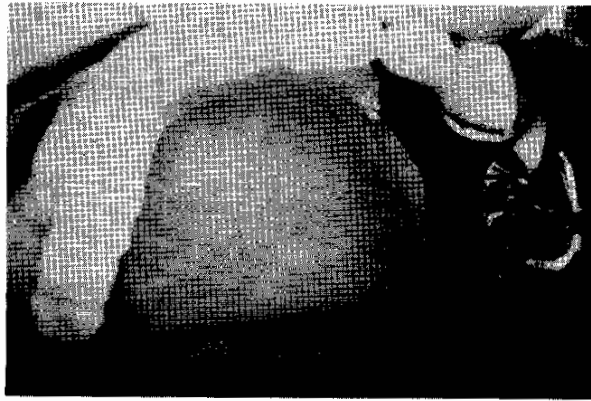


FIGURE 9. Oral view of prosthesis in place.

ances were further dampened and exhibited "hyponasality" in addition to hypernasality. Manual elevation of the palate by a dental mirror improved the voice during sustained phonation of "ah" after installation of the hard palate prosthesis. Presumably the patient's speech could therefore be further improved by construction of a maxillary prosthesis incorporating a lift appliance or a combined lift and bulb to assist palatopharyngeal closure.

Scarring of the palate as a result of an ineptly performed adenoidectomy may create a handicapping hypernasality. I observed such a case some years ago—a 28-year-old female for whom an adenoidectomy had been performed by a physician who practiced "finger surgery" for removal of adenoid tissues. The subsequent scarring of the palate left it taut and virtually immobilized. Treatment required removal of some of the badly scarred tissues and the development of a palatal prosthesis. Speech intelligibility and voice quality were thereby restored to near-normal condition.

Palatal carcinoma is one of the causes for removal of all or a portion of the palate. Bradley (1971) reports, although without documentation, that restoration of "missing structures with functional prostheses usually results in normal speech" and indicates that "rarely speech training may be required" (p. 662).

Wise and Baker (1968) describe a procedure using a tongue flap for palatal repair for the treatment of velar tissue loss. They provide no information about the nature or severity of speech disorder or the functional results of this method of treatment for the speech of patients who have sustained this procedure. It is presumed that the flap is nonfunctional except insofar as it provides tissue coverage. My own clinical experience leads me to believe that a posterior palatal prosthesis might be a preferable method of treatment for speech in such a case.

PHARYNGEAL EXCAVATION

Surgical deepening of the pharynx in connection with adenoidectomy or

surgical removal of a retropharyngeal tumor may impair palatopharyngeal valve closure and thus cause hypernasality. Bradley (1971) cites references attributing palatal malfunction to adenoid removal. Fletcher notes that adenoidectomy may unmask a potentially inadequate palate. Instances of this type are not rare. Figure 10 is a drawing adapted from Subtelny and Koeppe-

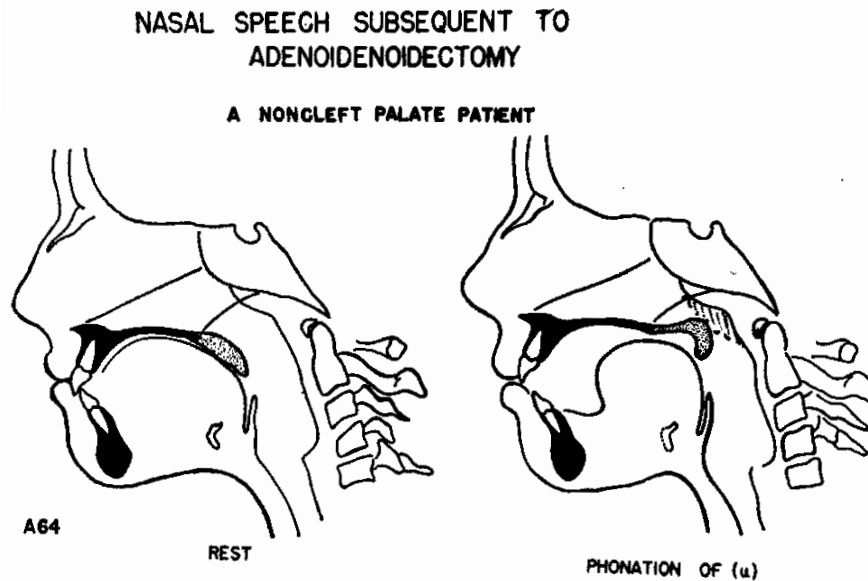


FIGURE 10. Views of palatal elevation at rest and for sustained / \bar{oo} /. The shading in the epipharynx indicates approximate position of the former adenoid pad. Obviously post-operative palatopharyngeal closure does not take place. (From Subtelny and Koeppe-Baker, 1956.)

Baker (1956) illustrating such a case and the potential effect of removal of the adenoid mass.

Diagnosis of the speech disorders of such patients should employ methods applicable to those used in the study of cleft palate. Some patients have been observed to respond successfully to pharyngeal flap surgery, and some have been able to use a palatal lift appliance (Gibbons and Bloomer, 1958; Kipfmueller and Lang, 1972) with excellent results.

Speech therapy for impairment of palatopharyngeal valve function stresses exercises designed to improve valve closure and valve control in much the same way that valve function is taught to patients with congenital deficiencies of the palate. The one big advantage that ablative patients have over congenital cases is that they retain an auditory self-image of "normal" speech, to which they will attempt to return as soon as an adequate mechanism is available to them, whereas the congenital subjects must acquire a new self-image,

and oftentimes must overcome firmly established habit patterns in order to improve speech.

INFRAMAXILLARY LEVEL EXCISIONS

The exact location of malignant growths of the inframaxillary region of the oral cavity is often poorly defined, since these tumors commonly invade multiple sites by direct extension. Erich and Kragh (1959), analyzing lesions of the anterior two thirds of the tongue, found that 17.1% invaded the floor of the mouth, and over 13% extended to the anterior pillar, tonsil, base of the tongue, or hypopharynx. Malignancies involving the buccal mucosa are encountered less frequently, although in some cases removal of buccal tissues has created an opening to the oral cavity. When such lesions are unobturated they affect vowel qualities, and, if the dental arches are not intact, will disturb consonant articulation, since the cheeks normally assist in sealing the lateral walls of the mouth cavity against escape of air over the sides of the tongue. If the aperture is obturated, the effects on speech are believed to be only moderately handicapping, although I have found no clinical reports which comment on the speech adequacy of such patients.

A film made some years ago at the University of Michigan demonstrates a patient with extensive tissue removal on one side of the face. The quality of speech was, of course, markedly impaired, but the patient was an excellent subject for the intraoral viewing of lingual and palatal action during speech.

GLOSSECTOMY

Intraoral cancer is reported to account for 5% of all malignant diseases, with about 15,000 patients per million developing cancer of the tongue or floor of the mouth each year (Skelly et al., 1971). Surgical or accidental extirpation of all or parts of the tongue and associated tissues have long been known to impair speech, but it has also been demonstrated repeatedly that such ablations do not necessarily prevent intelligible speech. The late Max Goldstein, founder and former director of Central Institute for the Deaf, in St. Louis, presented films of two such patients, one with a partial glossectomy involving about two-thirds of the tongue and one with removal of all lingual tissues above the level of the hyoid bone. Recordings demonstrated that both men could be understood. The second patient's voice quality seemed to be altered, and, if my memory can be trusted nearly 30 years later, had a sort of "Donald Duck" quality; but his speech was intelligible, and he was able to return to work and to talk on the telephone.

Over 30 years ago, Backus (1940) reported on a 10-year-old boy who had undergone anterolateral excision of approximately one-third of his tongue after radiation treatments. The phonetic profile of this youngster after surgery demonstrated numerous articulatory errors, most of which were related to sub-

stitution of labial consonants for tongue-tip phonemes. Backus reports that, with speech therapy, tongue-blade substitutions acceptably corrected articulation errors, with the *th* production being the most difficult to achieve. She suggests that tongue exercise seemed to help in regaining speech function and may also have aided the youngster's tongue to regain near normal size after a period of time.

The speech of a girl who sustained a hemiglossectomy was observed several years ago (Figure 11). When she spoke carefully, her speech was not defective

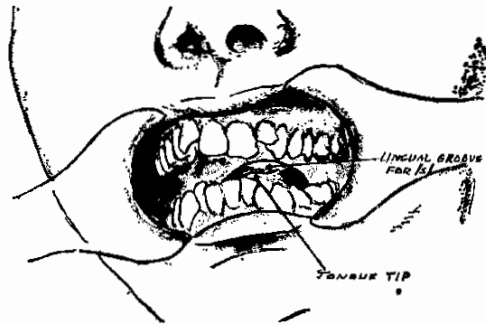


FIGURE 11. Patient after right hemiglossectomy. The lateral grooving of the tongue is adaptive, and permitted the patient to enunciate distinct sibilants.

and, although she no longer had capability of producing a midline lingual grooving, she could produce quite acceptable sibilants. She developed a linguoalveolar constriction between the tongue tip and tongue blade, which although resembling in form a left anterolateral "lisp," sounded nearly normal.

Duguay (1964) reports a case of a 52-year-old woman who lost part of the left side of the tongue and adjacent floor of the mouth and sustained a left radical neck dissection, although the mandible was preserved. Her speech eventually became intelligible and was said to be "well articulated." He cites his observation of two patients with total glossectomy to support Goldstein's assertion that fluent speech can be produced without a tongue. Compensatory articulatory adjustments which he described are to be found in Table 2.

A doctoral dissertation by Kalfuss (1968) reports on the speech of 22 patients, 18 male and four female, subsequent to surgical removal of lingual tissue. The age range was 42 to 74 years, 19 were white, and 3 were black. The basic pathology was generally attributable to squamous cell carcinoma.

Grouping of the patients by site of lesion, extent of excision, and lingual mobility is shown in Figure 12, regrouped from diagrams provided in Kalfuss's dissertation.

Information about the speech of these patients indicates that, as a group, their articulation of 12 phonemes (11 consonants and one vowel) was impaired. These phonemes were, respectively, the vowel /i/ (ee) and consonants /l/, /v/, /k/, /g/, /θ/, /ð/, /s/, /z/, /ʃ/, /tʃ/, and /dʒ/. All except /v/ normally require linguoalveolar or linguopalatal contacts. It is somewhat puzzling that

TABLE 2. Acceptable articulatory approximations for speech after glossectomy. (From Duguay, 1964.)

<i>Consonants</i>	<i>Possible Substitutes*</i>
/p/, /b/, /f/, /v/, /m/	generally unaffected by glossectomy
/k/, /g/, /ng/	(1) contact of buccinator muscles with molars, or (2) movements approximating palatal arches, or (3) replacement of /k/ and /g/ with glottal stop and /ng/ with nasalization of preceding vowel
/t/, /d/, /n/, /l/	(1) floor of mouth elevated to make contact with upper incisors, or (2) lower lip drawn back and elevated to a position on upper incisors
/th/ (voiceless and voiced)	position for /f/ and /v/ modified to give acoustic approximation to /th/
/s/, /z/, /ʃ/, /zh/	air blown through teeth, lips protruded†
/ch/, /j/	glottal (or oral) stop plus /sh/ or /zh/, respectively
/r/	(1) vocal cords trilled ("laryngeal /r/") so as to give acoustic approximation to /r/, or (2) uvular (guttural) /r/

*Alternatives are given in order of preference.

†If the tip of the tongue is lacking, /s/ will generally come out close to /sh/, and /z/ close to /zh/.

/v/ was included, and not /f/, and that any problem was encountered with a labiodental consonant, unless there was concomitant impairment of lip movements.

Analysis of the results of speech intelligibility tests comparing Groups I, II, III, and IV showed that intelligibility correlated positively with the degree of tissue loss. That is, Group I was most intelligible and Group IV least intelligible. However, although Groups I, II, and III showed progressive deterioration in speech, in that order, the group differences appear to have been slight.

The recommended treatment for such cases may be plastic reconstruction or prosthetic installation, if radio-osteonecrosis does not contraindicate. Speech therapy includes tongue exercises and more or less standard approaches to improvement of speech through phrasing, rate controls, and development of compensatory articulatory movements.

Herberman (1958) reported on the rehabilitation of two patients after glossectomy. The complaints following tongue section included continued and uncontrolled salivation, inability to swallow, regurgitation of food into the nose, and unintelligible speech. After pharmaceutical control of salivation, he reported successful amelioration of remaining problems through a regular schedule of muscle stretching, massage, and resistive exercise.

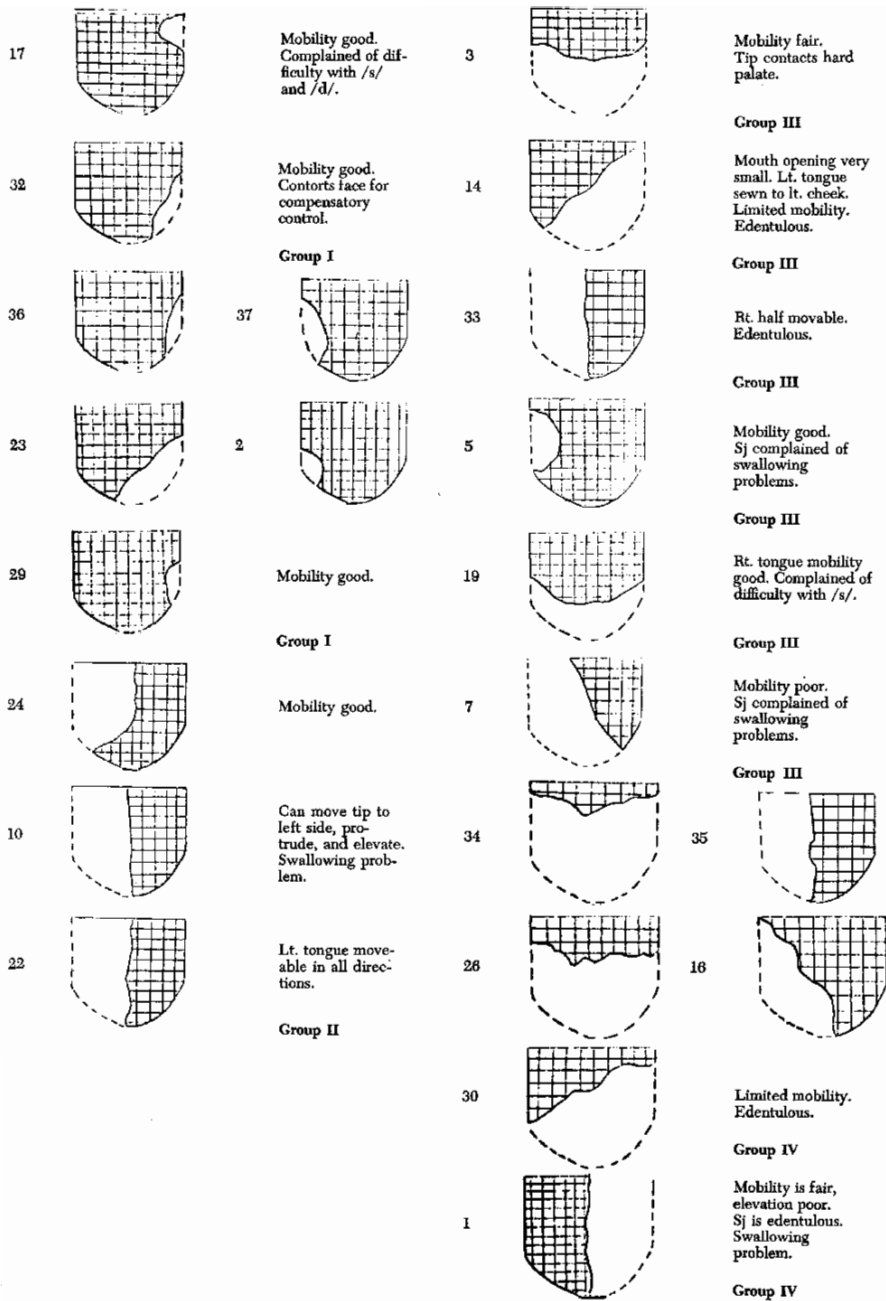


FIGURE 12. Classifications of lingual ablations by degrees of lingual excision and mobility. (From Kalfuss, 1968.)

MANDIBULECTOMY

Mandibulectomy may occur in association with the surgical treatment of lingual cancer. A patient of James Hayward, an oral surgeon at the University of Michigan, sustained a right hemimandibulectomy, with a subsequent surgical anastomosis of the tongue and cheek on that side. Although I have not examined the patient, I have talked with him by telephone. The patient claims that he now has some difficulty in chewing his food and must exercise care in speaking, but otherwise has no particular difficulty functionally. I found his telephone speech to be normal in articulation and voice quality.

William Grabb, a clinical associate professor of surgery at the University of Michigan Medical Center, has stated that he finds little dysfunction as a result of hemimandibulectomy if bone resection does not pass the midline of symphysis.¹

COLLECTING DATA ON SPEECH AFTER ABLATIVE SURGERY OF THE FACE, MOUTH, AND PHARYNX

Research on the speech problems associated with cleft lip and palate has added significantly to our understanding and rehabilitation of this group, but there are few data on speech disorders related to extensive maxillofacial defects. Surgical procedures and techniques for managing orofacial disease in the past 15 years have moved faster than our understanding of their functional results. The paucity of specific information on the communicative disorders attributable to ablative surgery of the maxillofacial complex suggests the desirability of accumulating such data.

Studies are needed, based on established methods of data collection and analysis, for the identification and description of communication disorders accompanying ablative defects of the face, mouth, and pharynx.

A minimal effort in this direction could be effected by the routine collection of materials which should include audio and visual recordings. The speech samples should demonstrate the extent of speech defectiveness according to criteria of intelligibility. The parameters of articulation should be described according to place and manner of articulation. A differential diagnosis should seek to identify those speech characteristics attributable to ablative surgery and those attributable to other causes, such as hearing loss, congenital disabilities, and emotional problems.

Speech samples should be brief, but a relatively standard protocol of data collection should include speech samples, descriptions, and test findings. A suggested procedure is provided below.

Voice Recordings Specifications

First, high-fidelity equipment should be used to record the patient's name,

¹Personal communication.

date of examination, a speech sample of the patient counting from one to 10, the "mini-test" of speech articulation evaluated according to place of articulation and manner of articulation (see below), and a brief speech sample specially designed to show the patient's speech peculiarities. These should be made pre- and postoperatively and at significant stages in treatment and recovery. The mini-test of speech articulation (Bloomer, 1973) is presented in the following sentences.

Place:

We bought my father two new sun lamps.
You should choose a red coat hanger.

Manner:

Bobby pulled down two go-carts.
The thing is very full.
Send his shoe measure to Charlie Jones.
Why won't you let her run?
Mary never sang.

Oralized sentences:

Zippers are easy to close.
Why whisper everywhere.
Violets cover the grove.
Wrap the carrots with paper.
Keep the pocketbook.
Go get a bigger egg.
Buy baby a bib.

Second, word intelligibility tests, such as the Fairbanks Rhyme Tests (see Table 3), should be recorded at intervals of three seconds, to allow for subsequent scoring by a panel of listeners.

TABLE 3. Fairbanks Rhyme Test (Fairbanks, 1958) recommended for evaluating word intelligibility.

<i>Sample List</i>					
	A	B	C	D	E
1	not	wire	pen	din	nail
2	say	male	lark	just	light
3	mop	sent	boil	mine	worn
4	reel	noon	dig	wink	cod
5	cake	pick	sage	sold	lock
6	paw	game	past	sit	pump
7	vile	ride	vain	bed	late
8	heat	hip	nest	lend	sell
9	look	bore	run	did	yet
10	kill	bang	deal	lack	suck

Pictorial Records of Patient Performances

1. Preoperative and postoperative orofacial photographs.
2. Pre- and postrestoration (surgical or prosthetic) movies or videotapes of orofacial structures at rest, speaking, chewing, and swallowing.
3. X rays and cine or video radiography where illustrative of the functions listed above.

SUMMARY

Speech problems, sites of tissue ablation associated with them, approaches to treatment, and suggestions for data collection in future studies of patients have been presented. The current deficiency of appropriate reports of such cases has been noted. From this introduction it may be possible to assemble future reference materials that will lead to better methods of dealing with the functional consequences of ablative surgery. The heroic people who sustain and survive ablative procedures are in truth our teachers. We should acknowledge our debt to them by a careful and systematic notation of their experiences and the results of our attempts to diagnose and treat their attendant communication disorders.

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Congenital Orofacial Anomalies

CLINICAL INVESTIGATION OF THE EXPERIMENTS OF NATURE

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Throughout the history of medicine we encounter the advice to study the "accidents of nature." McQuarrie (1944), in a series of lectures on the subject, wrote that the experiments which nature makes upon our fellow creatures are often unique in that they cannot be duplicated in the laboratory or reproduced at will in the clinic. Such experiments of nature, properly considered, permit acquisition of new and useful knowledge applicable far beyond the studies of patients with the anomalies. From the study of these "experiments" we may anticipate advances in the knowledge of organ, tissue, or cellular function, physiologic interrelationships, and the nature of pathologic processes.

In a collection of essays dedicated to the proposition that fundamental advances in basic science can be accelerated through the study of clinical disease, Paul Weiss (1961), the eminent experimental biologist, wrote about deformities as cues to understanding development of form. Weiss, the laboratory scientist, echoed the words of McQuarrie, the clinical investigator, by drawing attention to the extraordinary insights and the suggestions for laboratory investigation that can be gained through the study of "experiments of nature" in man. One is reminded in this connection that clinical observations by Gregg (1941) led to the discovery of the teratogenic effects of the rubella virus. There are, of course, numerous other examples. Much of what we have learned about the vitamin deficiencies was first motivated by controlled observations in human populations. Similarly, clinical observations led to laboratory studies on the physiology of the endocrines.

Many books on human anomalies carry similar quotations, as if to justify what may seem to be an undue preoccupation with the esoterica of human disease. For example, McKusick (1964), in his introduction to Gorlin and Pindborg's book, *Syndromes of the Head and Neck*, quoted the geneticist Bateson, who said, "Treasure your exceptions!" He also drew attention to William Harvey's eloquent statement written in 1657:

Nature is nowhere accustomed more openly to display her secret mysteries than in cases where she shows tracings of her workings apart from the beaten path; nor is there any better way to advance the proper practice of medicine than to give our minds to the discovery of the usual law of Nature by careful investigation of cases of rarer forms of disease. For it has been found, in almost all things, that what they contain of useful or applicable nature is hardly perceived unless we are deprived of them, or they become deranged in some way.

Smith (1970), in the introduction to his book *Recognizable Patterns of Human Malformation*, drew attention to a similar statement by Paget, written in 1882:

We ought not to set aside with idle thoughts or idle words about "curiosities or chances." Not one of them is without meaning; not one that might not become the beginning of excellent knowledge, if only we could answer the question—why is it rare? or being rare, why did it in this instance happen?

Garn (1968), in a foreword to a book on human growth, also recognized the investigative advantages inherent in the study of the abnormal:

Differences and interrelationships, barely discernible even at the fifth or ninety-fifth percentile of normal growth become more clearly delineated. . . . Growth retardations hold potential for the understanding of normal growth when the cause of the retardation is known. . . .

There are, of course, some practical reasons for the study of human anomalies. Epidemiologic evidence indicates that in the developed countries of the world birth defects now loom as a major cause of mortality and morbidity in infants and children. This is probably due to the conquest of nutritional and infectious diseases which have exposed birth defects as a major factor in childhood disease. Epidemiologists have pointed out that the true frequency of such disease is generally underestimated because of inadequate recognition and poor reporting. Also, certain types of anomalies are not apparent at birth and do not emerge until the child is older. Consequently, incidence data compiled from birth certificates or hospital records of births for late-appearing anomalies are not available.

Among the various congenital anomalies, those affecting craniofacial structures constitute a special category. The face of man is his window to the world. It contains the organs of sight, vision, and speech with which he communicates with his environment, receiving information and responding to it. The face reflects the state of health, emotion, and character. It is the facade by which others perceive and judge the individual. That which affects the face of man and its organs, strikes at the most visible part of his body and his most human functions, such as speech.

Clinicians identify disease entities and syndromes by recognizable related elements of the face that correspond with previously learned experience. Demyer, Zeman, and Palmer (1964) demonstrated that median faciocerebral anomalies appear in various lawfully related gradations and combinations. In

such cases, the facies are diagnostic of the type of brain malformation so that the "face predicts the brain." Indeed, it is a curious phenomenon that patients with the same syndrome, irrespective of their ethnic origin, look more like each other than they resemble their own kin. It can be said, with some qualification, that if you have seen one patient with progeria, one with mandibulo-facial dysostosis, one with Apert's, and so on, you have seen them all.

The skull is a community of bones and organ systems of diverse phylogenetic origin and variable patterns of development, altogether relating to several functions vital to the life and well-being of the organism. If in the course of development one member of this community is affected adversely, inevitably other parts will suffer. Thus, if the maxilla, which contributes to the bony orbit, is arrested in its growth, then the development of the eye and its adnexa may become secondarily involved. Vision dependent in part on a precise geometric interrelationship of the globes and their extraocular muscles can be affected by a primary defect in the skeletal system that forms the bony orbits. Conversely, if the eye should fail to develop, or requires enucleation in infancy because of retinoblastoma, then the surrounding skeletal framework will reflect the arrested development as well (Figures 1-4).

The head contains the portals for respiration and feeding. Structural defects within the head of the newborn can be life-threatening if such vital functions are impaired. Arrest of growth of the neurocranium, if undetected and untreated during the period of rapid brain growth, can cause serious damage to the central nervous system.

Acquired malformations due to trauma, particularly those resulting from craniofacial injuries incurred in automobile accidents or those resulting from war injuries, comprise a significant group of patients.

The number of patients reported to have undergone ablative surgery of the face and oral cavity for the treatment of cancer has increased significantly.



FIGURE 1. CCFA number 1754, female, age one year, 10 months. Congenital defect of the left side of the face including microphthalmia, displacement of middle cranial fossa, and arrest of growth of midface. Intelligence and general somatic development are normal.

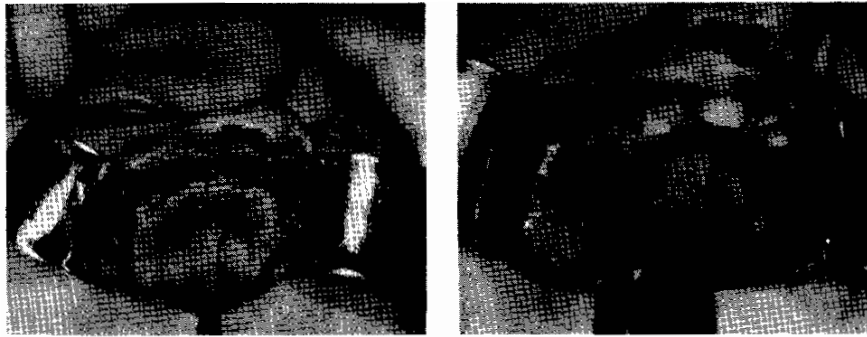


FIGURE 2. Intraoral photos of child in Figure 1 at five months and 13 months of age. Note asymmetry of palate and dental development.



FIGURE 3. CCFA number 3473, male age 12 years, four months. Craniofacial dysostosis with gross exophthalmos, strabismus, and arrest of growth of midface.

This increase may be attributed to an increased population, increased longevity, and improved recognition of disease with referral of patients to medical centers where tumor registries are maintained.

As problems in clinical management, the congenital, developmental, and acquired malformations have one thing in common. Each requires prolonged supervision for optimal treatment by an interdisciplinary team that includes specialists in medicine, dentistry, speech and hearing, behavioral science, genetics, and educational and vocational counselling.

It is the thesis of this presentation that the professional audience served by these publications has the potential, individually and collectively through existing interdisciplinary units, to contribute to the clinical management of such patients. At the same time, the opportunity to study cause and effect relationships by controlled, systematic study during clinical management needs to be recognized and used.

To demonstrate these opportunities, a selected series of congenital and de-

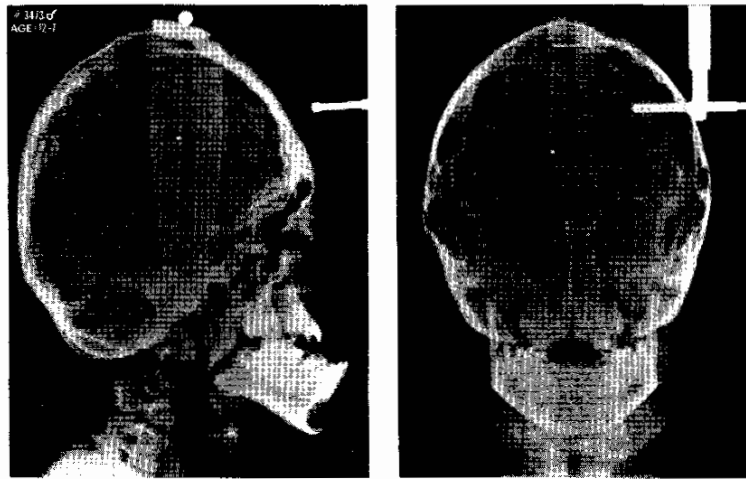


FIGURE 4. Cephalometric radiographs of patient illustrated in Figure 3, at age 12 years, seven months. Note shallow orbit due to underdevelopment of the maxilla, frontalization of the orbital roof, forward displacement of the great wing of the sphenoid, and lateral expansion of the ethmoid. Occlusion of the epipharynx, partial occipitalization of the atlas, and multiple fusions of the cervical vertebrae are apparent, among other deformities of the neurocranium and facial skeleton.

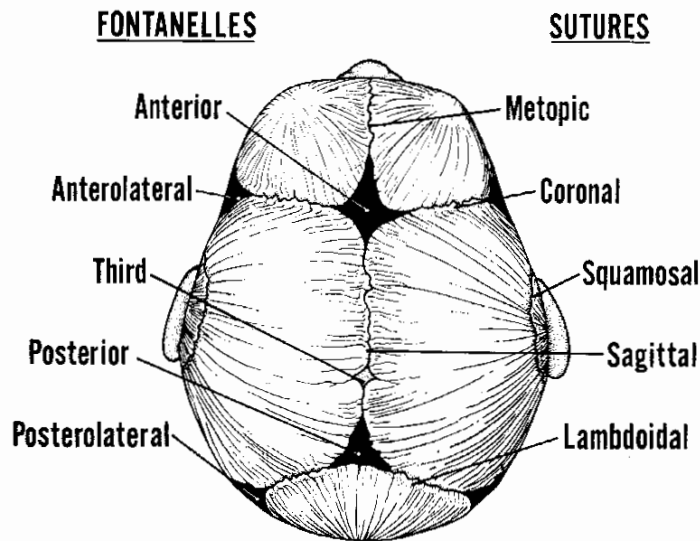


FIGURE 5. Schematic drawing of the superior view of a normal infant skull. The membrane bones are separated by open sutures and fontanelles.

velopmental malformations will be illustrated. The selections were made to illustrate the following variations:

1. Disorders in facial-oral-pharyngeal physiology that are probably secondary to defects in the craniofacial skeleton wherein the severity of the pathology may be age-dependent, relating to developmental changes in contiguous structures. Because of the severity of the defects, the time of onset, and the organ systems involved, primary referral is generally made to the neurosurgeon or plastic surgeon. Such cases do not ordinarily come within the purview of the speech pathologist, audiologist, or dental specialist.
2. Disorders in which the facial-oral-pharyngeal structures are commonly and characteristically malformed from birth. This group differs from the preceding in that the malformation remains essentially unchanged as the child grows older. In this category, as in all the others, an appreciation of the regional pathology comprising contiguous organ systems is essential as a basis for multidisciplinary treatment-planning with appropriate priorities in proper sequence.
3. Samples from a wide range of disorders which represent high risks for speech and audiologic problems. Such cases can be found with increasing frequency in birth defect clinics and in departments of clinical genetics.

THE PREMATURE CRANIOFACIAL SYNOSTOSES

Two syndromes, bearing the eponyms of Apert and Crouzon, are considered under this heading. The common denominator is arrest of growth of several sutures of the neurocranium and midface affecting the various organ systems and functions subserved by this skeletal framework.

The development of the neurocranium is partly influenced by the development of the brain. From birth to two years of age, intracranial volume is nearly tripled, closely paralleling the increase in brain weight (Vignaud, 1966; Coppoletta and Wolbach, 1933). Premature closure of one or more sutures of the neurocranium leads to arrest of growth with compensatory growth in the sutures that remain open. Presumably this compensation is due to the intracranial pressure of the growing brain. This compensatory growth leads to alterations in shape, size, and position of cranial structures (Figures 5-7).

The secondary effects of distorted head growth can be demonstrated in the form and function of several organs. The exophthalmos is clearly due to an imbalance between the size of the bony orbits and that of the eyeballs. Reduction in the size of the bony orbit can be ascribed to several factors. The increased intracranial pressure of a growing brain accommodating to a restricted intracranial capacity leads to displacement of parts of the cranial base. The great wings of the sphenoid are projected forward as a result of the expansion of the middle cranial fossae thereby impinging on the volume of the bony orbit. Vertical sloping of the orbital plate of the frontal bone, readily

seen on radiographic examination (Figures 4 and 8), also serves to reduce the anteroposterior dimension of the bony orbit. Bulging of the ethmoidal walls medially, due to increased intracranial pressure, contributes to the hypertelorism and adds to the reduction of orbital capacity. Arrest of growth in the mid-face, particularly in the infraorbital region, accentuates the exophthalmos.

Abnormal ocular alignment and motility are common findings (Figures 9 and 10). Theoretically, this can be explained by the abnormal vectors of action on the part of the extraocular muscles. Five of these muscles originate in the bony apex of the orbit and one (inferior oblique) originates from the medial wall. Aberrations in the architecture of the bony orbit could affect the mechanical action of these muscles. Since the relationship between the orbits is altered

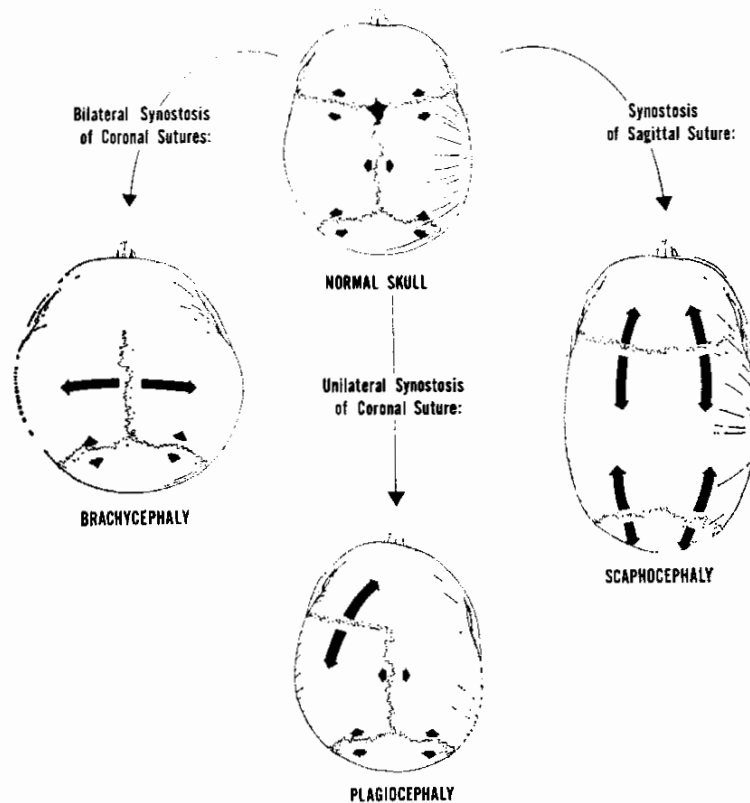


FIGURE 6. Premature synostosis: superior view. Directional contribution of sutural growth to skull form is illustrated by arrows. Arrest of growth due to premature stenosis of selected sutures, combined with compensatory growth in remaining open sutures, leads to distortion of skull shape.

in distance and in angulation, the correlated movements of the eyes might be expected to be disturbed.

Occasionally, patients with Apert and Crouzon syndromes present severe forms of exorbitism resulting in forward dislocation of the eyeball. Tarsorrhaphy may be required to protect the cornea and conjunctiva.

Because of the basilar kyphosis and arrested maxillary growth, the epipharynx may be occluded, leading to respiratory distress. Baldwin (1968) reported the death of a five-month-old infant with craniofacial dysostosis in which cor pulmonale was a finding. He did not associate this observation with epipharyngeal obstruction. More recently, Don and Siggers (1971) recognized the cause and effect relationship between postnasal airway obstruction and cor

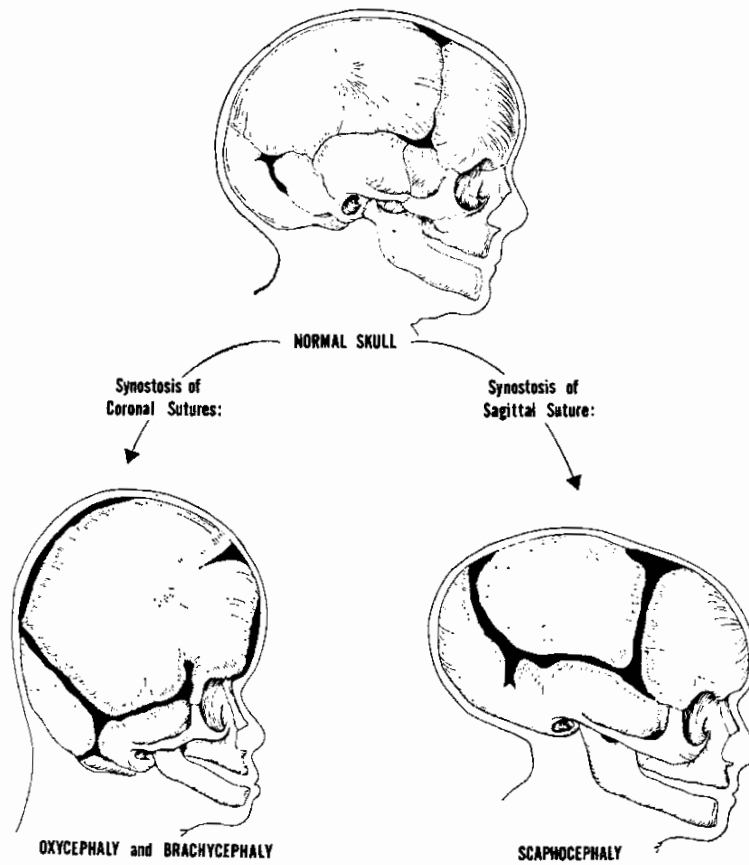


FIGURE 7. Premature synostosis: lateral view. Effect of cranio- and cranio-facial synostosis on skull shape.

pulmonale in a patient with Crouzon's disease. Previously, Cox et al. (1965) and Menashe, Farrehi, and Miller (1965) described the relationship between obstructive adenoids and cor pulmonale. Jersaty, Huszar, and Basu (1969) and Shah, Pruzansky, and Harris (1970) reported a similar relationship between Pierre Robin syndrome and cor pulmonale.

Patients with the syndromes of Apert and Crouzon frequently have middle ear disease. Since cleft palate is a commonly associated finding, dysfunction of the middle ear can be related to the cleft. However, as Baldwin (1968) re-

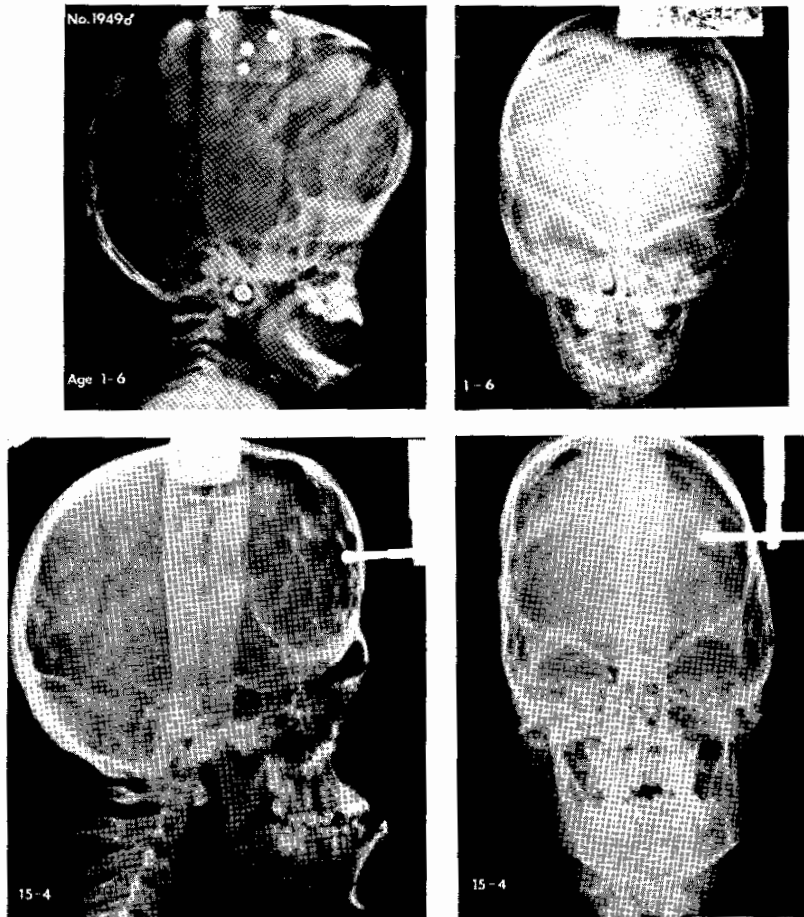


FIGURE 8. Serial cephalometric radiographs of a patient with Apert's syndrome. The initial films, obtained at one year and six months of age, are contrasted with the most recent films taken at 15 years and four months. Maxillary-mandibular disharmony has increased because of the retarded growth of the midface.

ported, otologic disease may be due to a multiplicity of causes. Atresia and other deformities of the external canal are readily apparent. Tomography has revealed deformities of the conducting mechanisms with occasional abnormalities of the inner ear. Susceptibility to recurrent middle ear inflammation might be related to tubal dysfunction compounded by the obstructed epi-pharyngeal space.



FIGURE 9. CCFA number 2033, female, age 12 years, two months. Craniofacial dysostosis (Crouzon's disease). Note exophthalmos and strabismus.

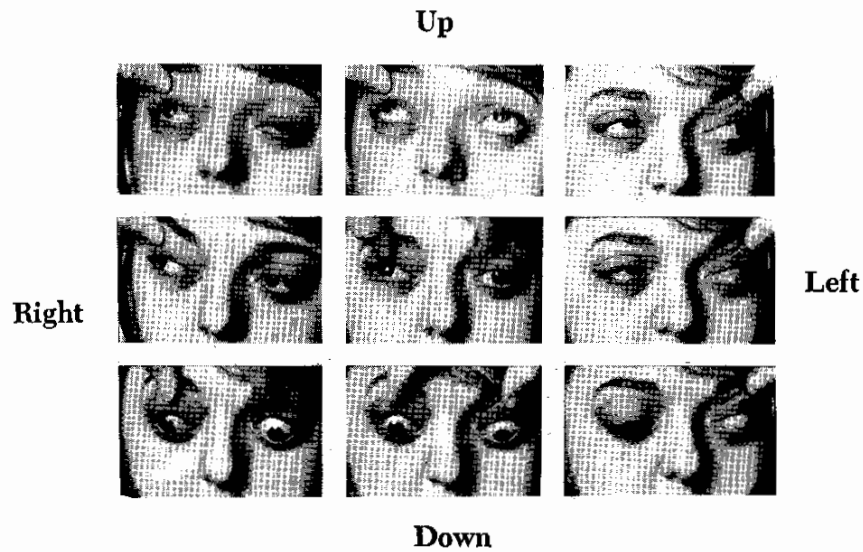


FIGURE 10. Ocular deviations in the patient illustrated in Figure 9, at age 19 years, six months.



FIGURE 11. CCFA number 1013. Acrocephalosyndactyly (Apert's syndrome) in a female, age seven years and 11 months.



FIGURE 12. Patient illustrated in Figure 11 at age 13 years and four months. Note the severe acne.



FIGURE 13. Severe malocclusion in patient with Apert's syndrome illustrated in Figure 12, at same age. Note palatal swellings producing pseudocleft in midline.

The natural history of these syndromes merits attention because the severity of the problems presented are age dependent. With increasing age, the disparity in the facial skeleton increases as a result of unequal growth in the upper and lower jaw (Figures 8, 11, and 12).

The widespread and unusual distribution of acne vulgaris in Apert's syndrome reported by Solomon, Fretzin, and Pruzansky (1970), as well as the accumulation of mucopolysaccharides in the palatal swellings (Figures 13 and 20) reported by Solomon et al. (in press), evoke interesting speculations regarding possible biochemical defects in this syndrome.

The major factor compelling a resurgence of clinical interest in these syndromes can be ascribed to the remarkable success of a small group of plastic surgeons in Europe and the United States. Foremost among these, the French surgeon Paul Tessier (1971) demonstrated the capacity to reorient the osseous architecture of the midface so as to transpose the orbits medially to correct hypertelorism; shift the maxilla forward to increase orbital capacity and reduce the exophthalmos; realign the jaws to improve facial esthetics, correct the severe malocclusion, and increase the dimensions of the epipharyngeal airway (Figures 14-16).



FIGURE 14. CCFA number 1909, female. A child with Apert's syndrome at age six years and three months.



FIGURE 15. The patient illustrated in Figure 14 at age 16 years and nine months, one month before midfacial osteotomy.

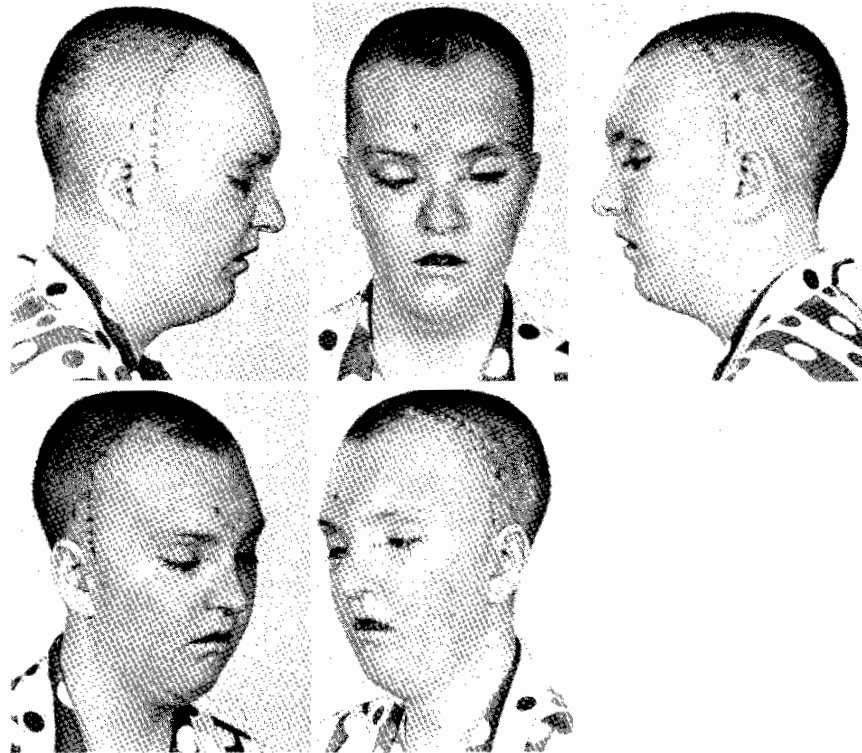


FIGURE 16. The patient illustrated in Figures 14 and 15, 26 days after surgery by Paul Tessier. Note the reduction in exophthalmos and improvement in facial balance.

This surgery is remarkable not only for the cosmetic and functional improvement achieved for the patient but also for the opportunity presented to the clinical investigator. By this surgery, the experiment of nature is reversed and a new form is created. This allows the clinical investigator to restudy form and function under a new set of conditions in the same patient—a remarkable experimental opportunity which can hardly be duplicated in the laboratory. There is an urgent need to document the effect of this surgery on vision, eye motility, palatopharyngeal function, middle ear and eustachian tube physiology, speech, olfaction, taste, deglutition, mastication, oral sensory perception, and behavior. Such documentation is critical for the assessment of the surgery and for the opportunity to gain insight into the causes and mechanisms of the disease.

Although the frequency of such malformations is relatively rare, the multiple problems presented (Figures 17-21) warrant referral to special centers capable

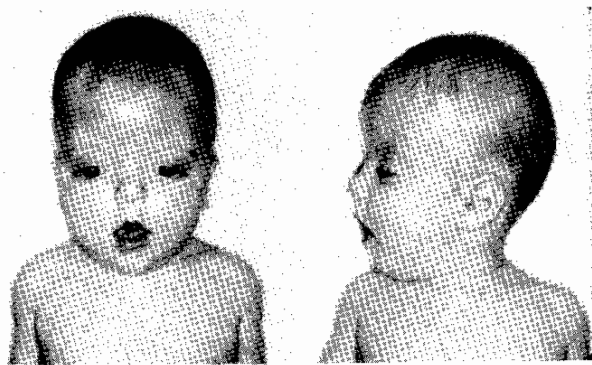


FIGURE 17. CCFA number 3695. A five-month-old male with Apert's syndrome, acrocephalosyndactyly.

of providing long-term interdisciplinary care for optimal habilitation. It is important that the potential for habilitation be recognized, lest ignorance and revulsion lead to unwarranted relegation of infants with such malformations to custodial institutions for the mentally retarded.

OTOCRANIOFACIAL SYNDROMES

There are several syndromes in which it may be useful to consider the ear as a frame of reference. The ear is a compound organ with a phylogenetic history that is both ancient and relatively recent in the evolutionary scale. The development of the inner ear is nearly complete by the end of the first trimester of pregnancy, while the external ear and parts of the middle ear continue to differentiate into later stages of fetal development. The interaction of the development of the ear and contiguous organs, in space and in time, deserves attention as a basis for understanding the pathogenesis of the syndromes under consideration. As an initial step toward such understanding, a comprehensive diagnosis with a cataloging of the affected structures and gradation of the severity of the malformation would be helpful.

Two such syndromes will be considered. The first, mandibulofacial dysostosis, is a well-defined entity, having been described by several authors including Thomson, Berry, Treacher-Collins, Franceschetti and his associates, and Zwahlen and Klein (Gorlin and Pindborg, 1964). The major features include hypoplasia of the malar bones, antimongoloid slant of the palpebral fissures due to downward displacement of the lateral canthus, coloboma of the outer third of the lower lid, a tongue-shaped process of hair that extends toward the cheek, malformations of the ear, obliteration of the nasofrontal angle, elevation of the bridge of the nose, and dysplasia of the mandible, among other malformations (Figure 22).

The deformity of the external ears is variable, as illustrated by the range of variation in four classic cases of mandibulofacial dysostosis shown in Figure

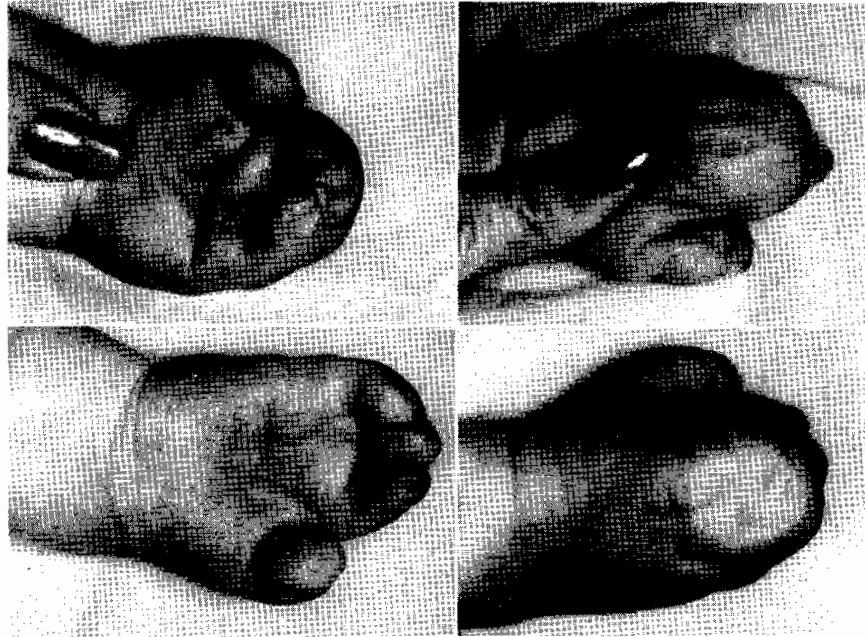


FIGURE 18. Mitten-glove hand deformity of patient in Figure 17.

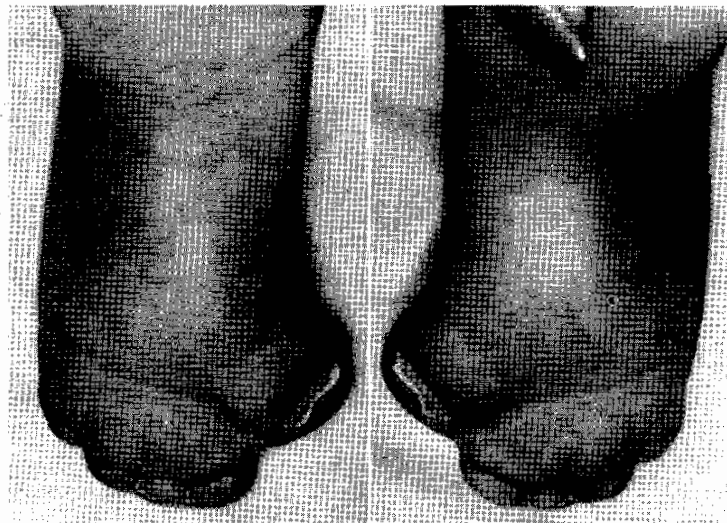


FIGURE 19. Stocking-foot deformity of patient in Figure 17.



FIGURE 20. Palatal swelling with midline pseudocleft in patient illustrated in Figure 17. The palatal swellings increase in mass as the patient grows older.

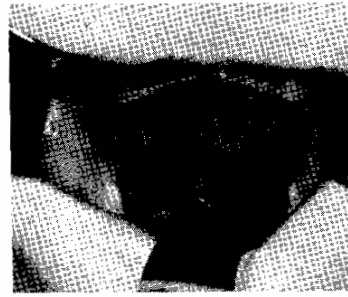


FIGURE 21. Bifid uvula in the patient with Apert's syndrome illustrated in Figure 17. Clefts of the secondary palate are a commonly associated malformation in this syndrome.



FIGURE 22. CCFA number 2022, female, age eight years, six months. Mandibulofacial dysostosis. Note the malformations of the external ears, malar hypoplasia, antimongoloid slant of palpebral fissures, colobomata of the outer third of the lower lid, preauricular tongue-shaped process of hair extending toward the cheek, elevated bridge of nose with obliteration of nasofrontal angle, and mandibular hypoplasia.

23. Middle ear deformities are common. Tomographic examination of the temporal bones is essential in a comprehensive survey of the hearing mechanism.

The micrognathia exhibited in mandibulofacial dysostosis is unique by virtue of the characteristic curvature of the lower border of the mandible (Figure 24) which is maintained throughout the growth of the individual (Figure 25). The shape and growth pattern of the mandible are held to be pathognomonic for the syndrome (Pruzansky, 1969). Anterior open bite and severe malocclusion are common (Figure 26).

Considerable evidence indicates that the syndrome is inherited as an autosomal dominant trait with incomplete penetrance and variable expressivity. Sporadic cases are not uncommon.

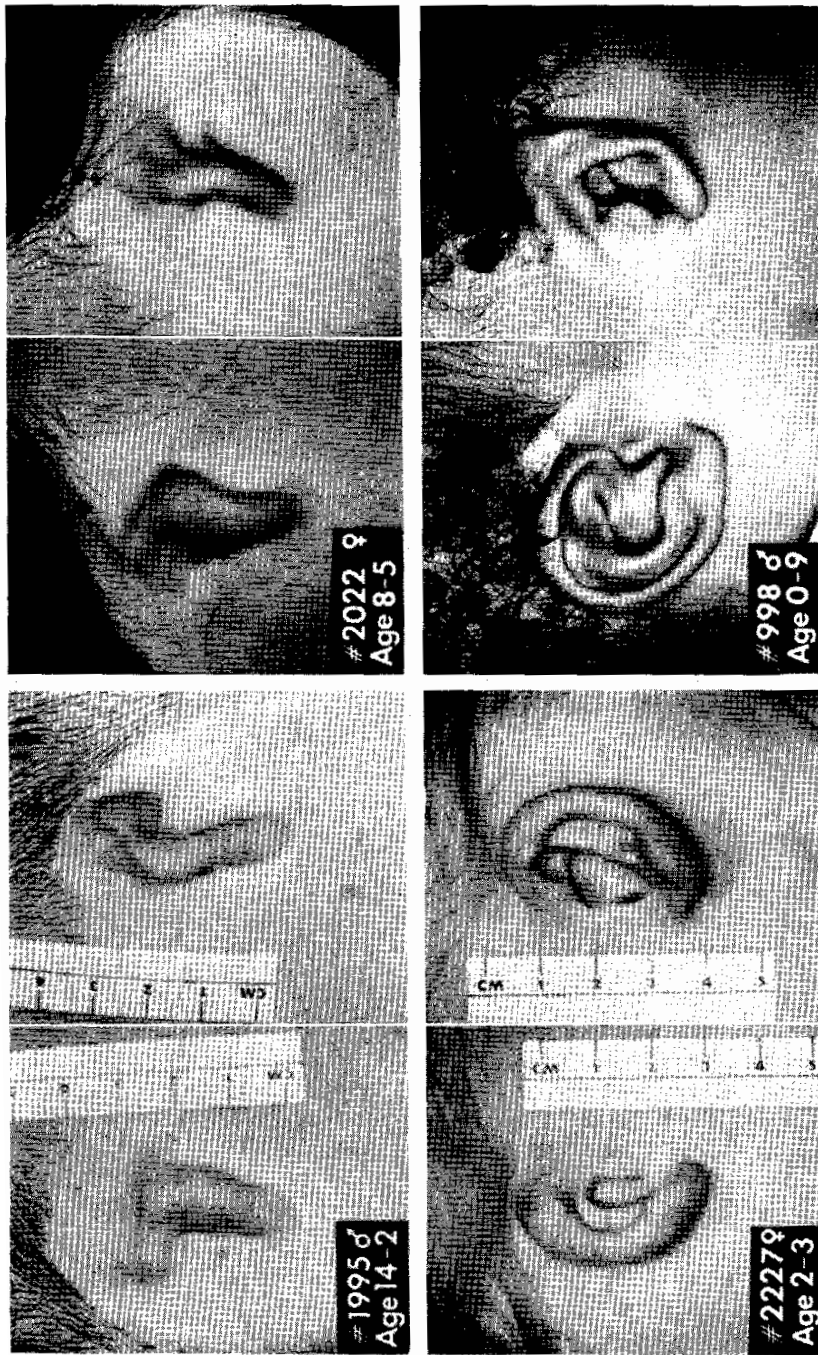


FIGURE 23. Range of variation in deformity of the external ear depicted in four cases of mandibulofacial dysostosis. Note the lack of symmetry in two cases.

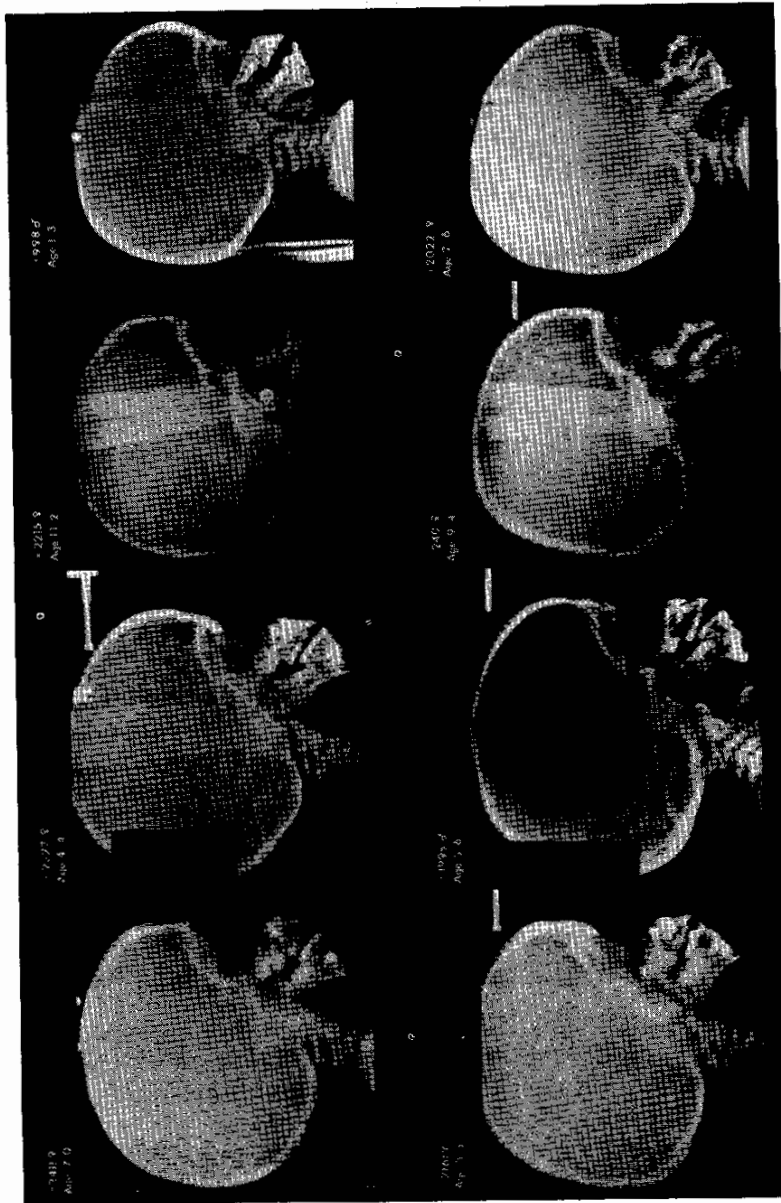


FIGURE 24. Lateral cephalometric radiographs of eight patients with mandibulofacial dysostosis, ranging in age from one year and three months to 11 years and two months. The curvature of the lower border of the mandible and the bowing of the body of the lower jaw are characteristic for the syndrome.

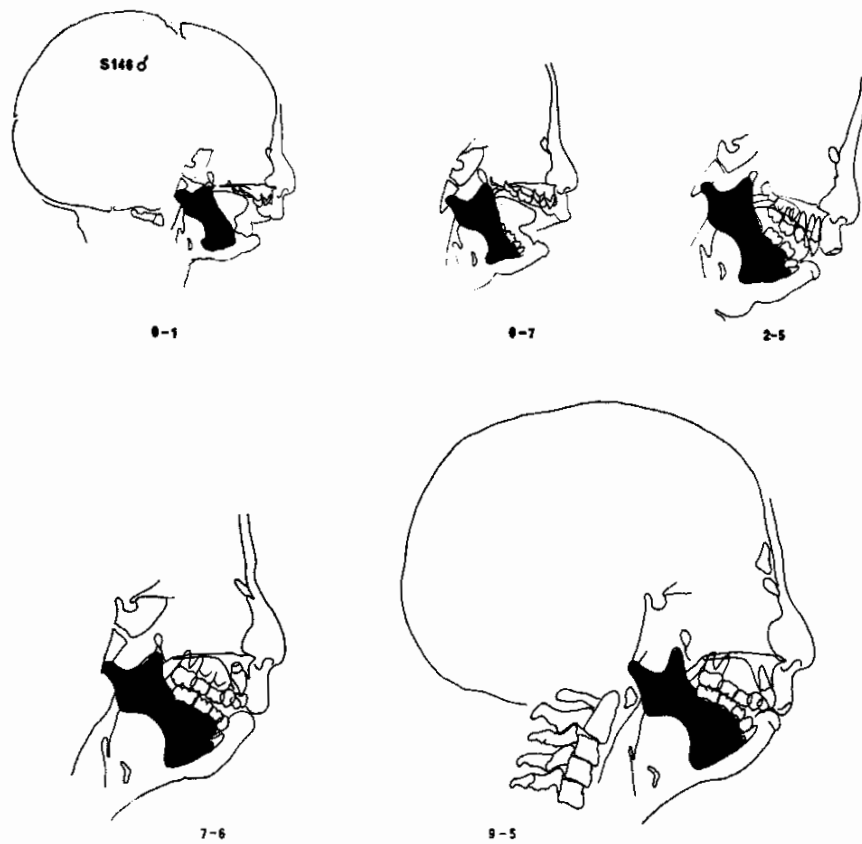


FIGURE 25. Tracings of serial cephalometric radiographs, in the lateral projection, of a patient with mandibulofacial dysostosis from one month to nine years and five months of age. The peculiar form of the mandible is retained from infancy into adulthood.

The second syndrome to be considered is known by many names, including hemifacial microsomia (Gorlin and Pindborg, 1964), first and second branchial arch syndrome, and otomandibular dysostosis, among others. It includes variants such as Goldenhar's syndrome and oculoauriculovertebral dysplasia. Commonly associated eye findings include epibulbar dermoids, colobomata—generally of the upper lids, microphthalmia, microcornea, choroidal or iridial coloboma, iris atrophy, and polar cataract.

Unilateral hypoplasia of the mandible, maxilla, orbits, and cranial basis is manifest in a graded spectrum of severity (Figures 27 and 28). The classification of ear deformities illustrated is incomplete in failing to code low-set or tilted ears (Figure 29).

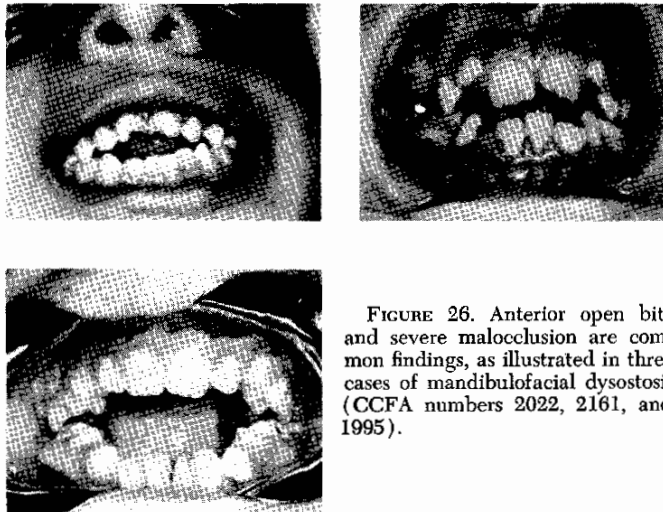


FIGURE 26. Anterior open bite and severe malocclusion are common findings, as illustrated in three cases of mandibulofacial dysostosis (CCFA numbers 2022, 2161, and 1995).

The current status of treatment of such syndromes is in a state of disorganization similar to that which prevailed for children with oral-facial clefts a generation ago. As a first step, and to provide a rationale for their treatment, a multidisciplinary investigation of a large series of cases was undertaken to map the range of variation encountered, the interrelationship of deformed parts, and the changes incident to growth.

As is evident from the sampling of cases illustrated, the range of variation encountered within the syndrome is of sufficient magnitude to warrant a highly individualized approach to treatment planning. For example, there is no correlation between the severity of the deformity of the external ear and the rest of the face (Figures 30-36).

Since the mandible is derived from the first branchial arch and the auricle from the first and second branchial arches, one should not expect to find a direct correlation in the severity of their deformities. Indeed, this is not the case, as Table 1 illustrates. On the other hand, a more direct correlation was found between the severity of the mandibular deformity and that of the malleus and incus, but not with the stapes. This might be expected since the malleus, incus, and mandible are derived from Meckel's cartilage, while the stapes originates from Reichert's cartilage.

The severity of the malocclusion in this syndrome is proportional to the dysmorphogenesis of the maxilla and mandible. Absence of the parotid duct and velar paralysis ipsilateral to the microtia are common findings. The concurrence of oral-facial clefts and hemifacial microsomia has been noted. In our series, we have not observed that the side on which the microtia is found

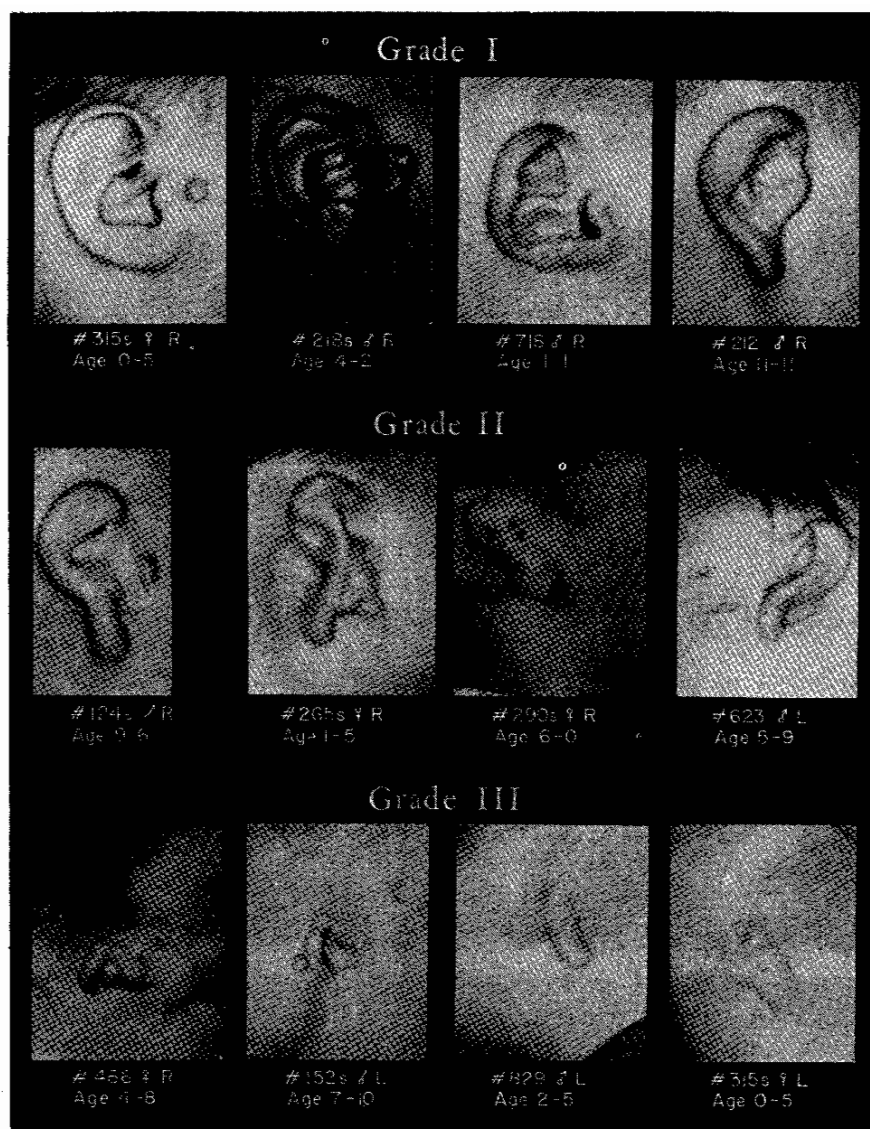


FIGURE 27. Graded spectrum of auricular deformity encountered in hemifacial microsomia ranging from preauricular tags and sinuses to anotia.

is always identical to the side of the unilateral cleft (Table 2). Unilateral hypoplasia of the tongue has also been observed (Figures 37 and 38).

To guide the clinician in assessing the factors contributing to the maxillary-mandibular disharmony and in planning a coordinated surgical-orthodontic approach to treatment, a schematic diagram of the interaction of the various affected organ systems was developed based on our survey of a large series of cases (Figure 39).

In the moderately to severely affected cases, facial asymmetry increases with increasing age. Serial cephalometric radiographs have revealed that the angular

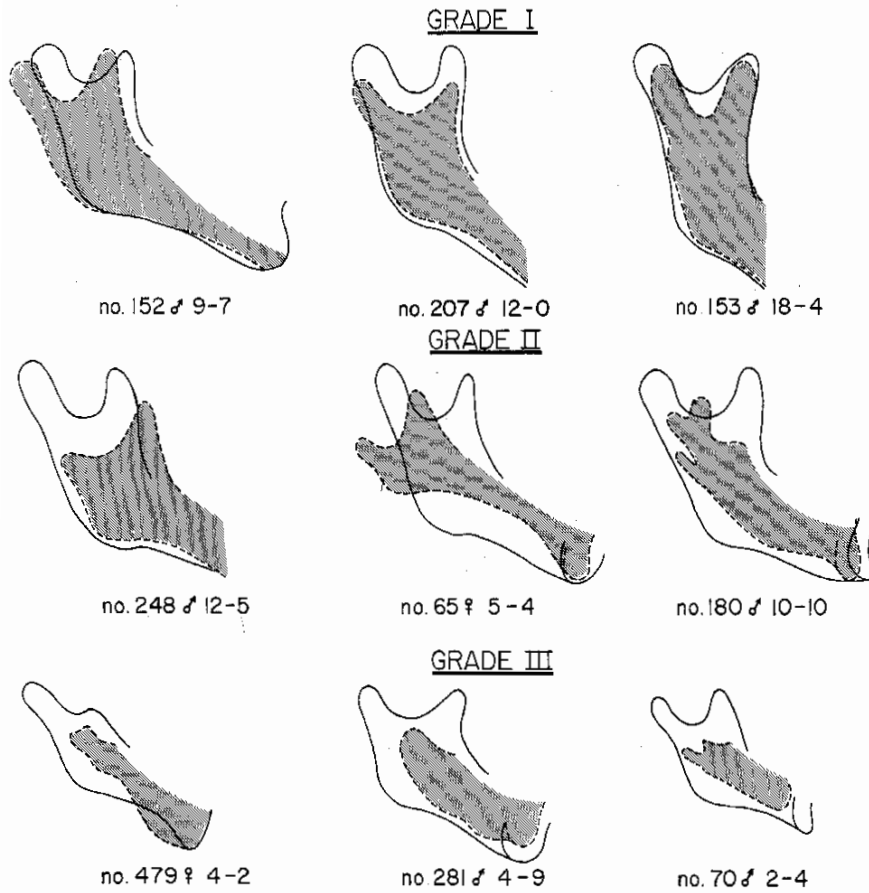


FIGURE 28. Graded spectrum of variation in mandibular deformity encountered in hemifacial microsomia. Tracings of the two halves of the mandible in the same patient were made from cephalometric tomograms. The shaded half of the mandible is homolateral to the microtia.

TABLE 1. Comparison of severity of auricular deformity to ipsilateral jaw deformity ($n = 91$); both sides rated separately for each patient.

Grade	Ear		Jaw			Not Graded
	Number	Normal	I	II	III	
Normal	71	67	4	-	-	-
I	37	17	11	6	3	-
II	30	8	16	4	2	-
III	34	6	14	8	5	1
Not Graded	10	4	4	2	-	-
Total	182	102	49	20	10	1

TABLE 2. Combinations of facial clefts, microtia, and jaw deformities. N = normal, - = not available.

Type of Cleft in Relation to Microtia	Number of Cases	Sex	Gradation of Defect			
			R. Ear	L. Ear	R. Jaw	L. Jaw
<i>Bilateral Microtia</i>						
Unilateral Cleft Lip and Palate	3	2M, 1F	III	I	II	I
			I	I	I	I
Bilateral Cleft Lip and Palate	1	M	II	III	I	II
			I	I	N	N
Cleft Palate	3	1M, 2F	III	III	I	I
			II	I	I	N
			I	I	III	I
<i>Unilateral Microtia</i>						
Ipsilateral to Unilateral Cleft	4	4M	N	-	N	II
			I	N	I	N
			II	N	I	N
Contralateral to Unilateral Cleft	2	2M	II	N	II	I
			I	N	I	N
Cleft Palate, Including One Case of Submucous Cleft Palate	8	5M, 3F	III	N	I	N
			I	N	II	I
			III	N	II	N
			-	N	N	N
			II	N	I	N
			III	N	II	N
I	N	I	I			

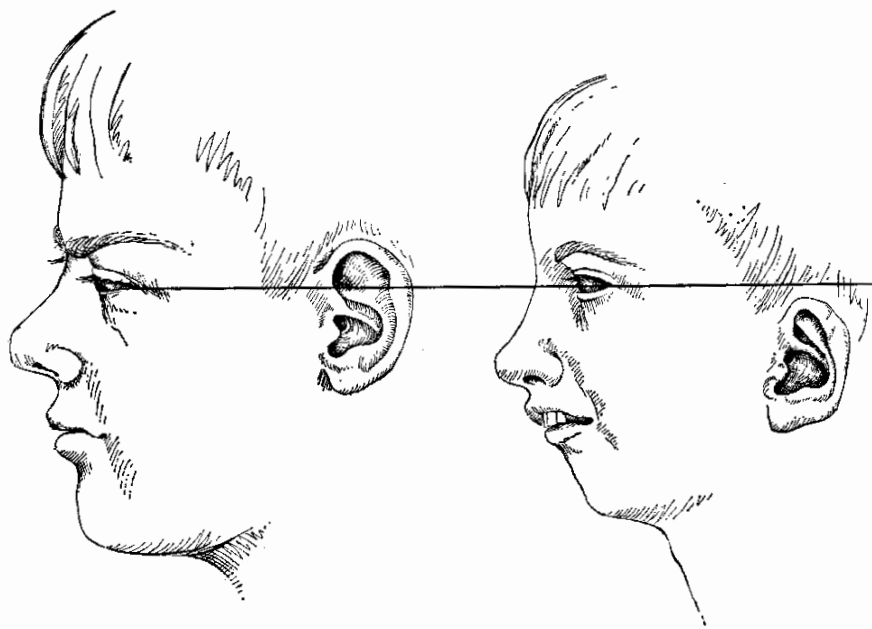


FIGURE 29. Relation of helix of external ear to eye. Left, normal. Right, low-set ear.



FIGURE 30. CCFA number 2196, male, age five years, seven months. Hemifacial microsomia characterized by Grade III deformity of the external ear and minimal facial asymmetry.



FIGURE 31. CCFA number 2130, male, age three years, two months. Hemifacial microsomia with Grade I deformity of the external ear, Grade III deformity of the mandible, severe malocclusion, and mild facial asymmetry.

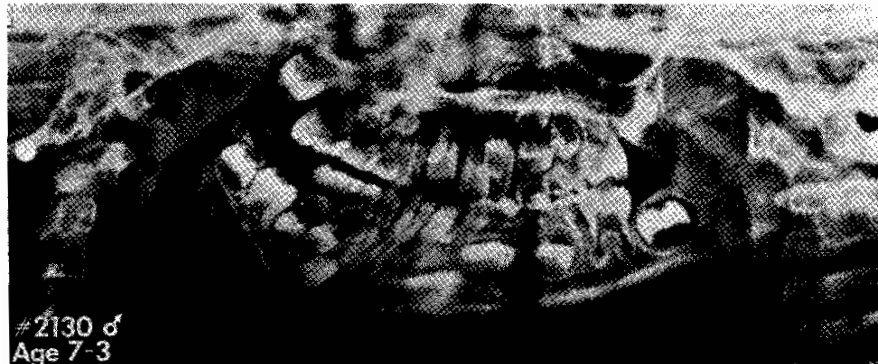


FIGURE 32. Panoramic radiographic of mandible of patient illustrated in Figure 31. Note absence of ramus on affected side. Grade III deformity of mandible.



FIGURE 33. Cephalometric radiographs of patient in Figure 31. Note relative symmetry of midface.

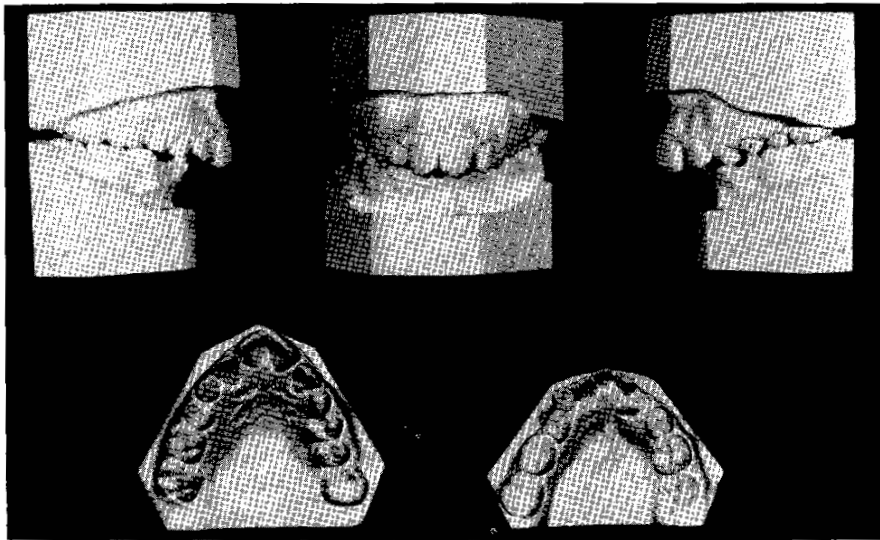


FIGURE 34. Dental casts illustrating malocclusion in patient illustrated in Figure 31, at age 10 years, four months.



FIGURE 35. CCFA number 2155, female, age six years, 10 months. Hemifacial microsomia with gross facial asymmetry.

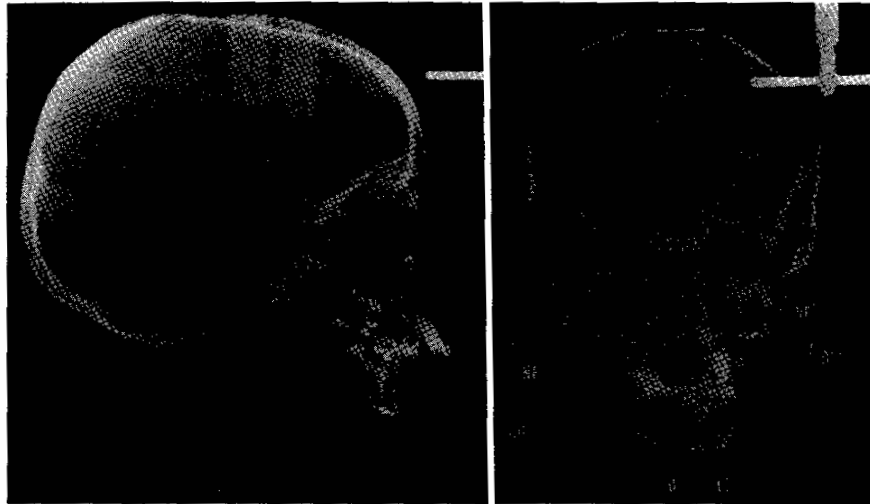


FIGURE 36. Cephalometric radiographs of patient illustrated in Figure 35, at age 10 years, two months. Note asymmetry in nasal floor as depicted in tilt of the plane $NF - NF^1$.



FIGURE 37. CCFA number 2171, female, age eight years, two months. Hemifacial microsomia with Grade II ear deformity and moderate facial asymmetry.

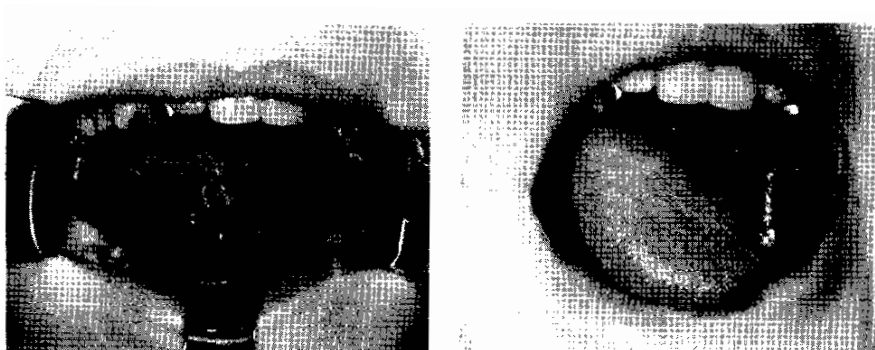


FIGURE 38. Unilateral hypoplasia of the tongue and bifid uvula in patient illustrated in Figure 37, at age nine years, three months.

Pathogenesis of Maxillary-Mandibular Disharmony

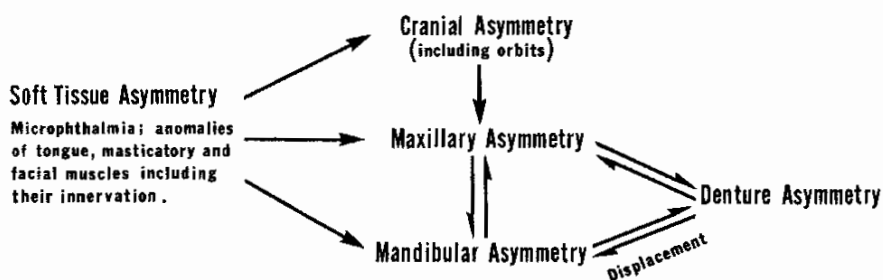


FIGURE 39. Schematic illustration of the interaction of various organ systems in hemifacial microsomia in terms of their contribution to maxillary-mandibular disharmony.

deviation does not increase. However, with increasing vertical height of the face the linear deviation of the chin-point from the constructed midline of the face enlarges, thereby accentuating the asymmetry (Figure 40).

Although familial incidence of this syndrome has been encountered, the mode of inheritance and recurrence risks have not been clearly delineated because of the paucity of reported family studies. The importance of noting microforms of the syndrome is illustrated in one of the pedigrees Pashayan and this author (unpublished data) have studied. Two out of three siblings have obvious deformities of the external ear (Figures 41-43), and the mother presents a microform manifest in a preauricular node and bifid tragus of the right ear (Figure 44).

From the foregoing, it should be clear that the professional resources, skills, and experiences developed in major cleft palate centers are uniquely suited to provide comprehensive diagnosis and treatment for patients with otocranio-facial syndromes.

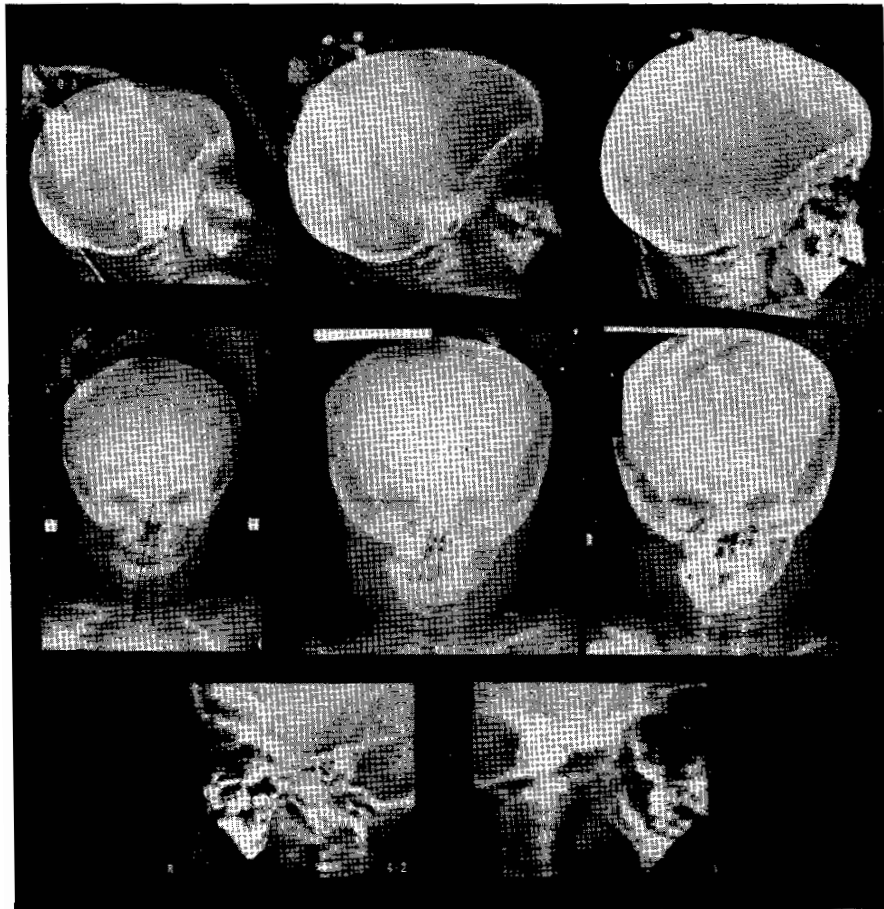


FIGURE 40. Plotting changes in facial symmetry. Serial cephalometric radiographs in hemifacial microsomia from age three months to two years and six months. Lower face asymmetry increases with increasing face height. Although the angular deviation remains relatively constant, the linear deviation of the chin-point from the constructed midline increases as the face grows longer.

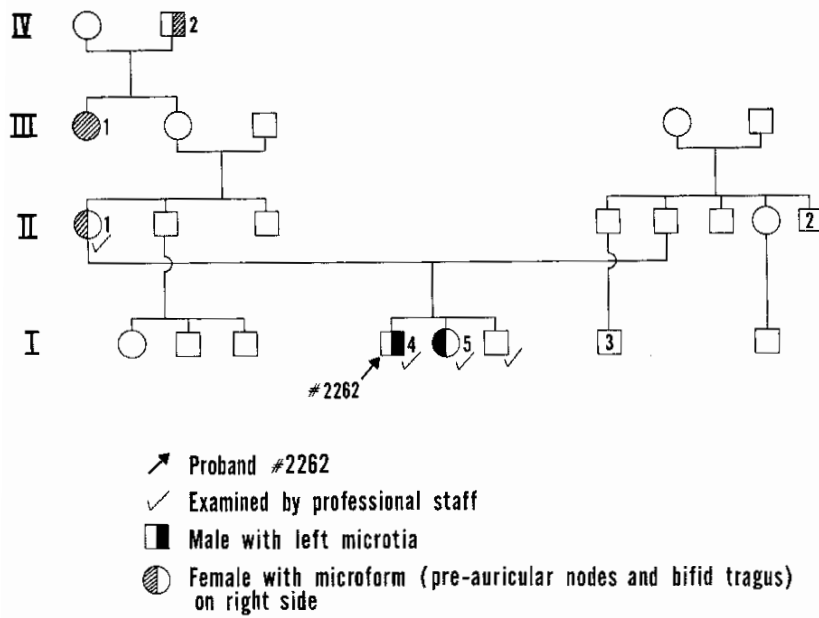


FIGURE 41. Pedigree of a family presenting two children with hemifacial microsomia and microforms in three previous generations on the maternal side.



FIGURE 42. Grade I ear deformity in the proband, CCFA number 2262, male, age eight years, nine months.



FIGURE 43. Grade III ear deformity in a sister of the proband, CCFA number 2263, age six years, six months.

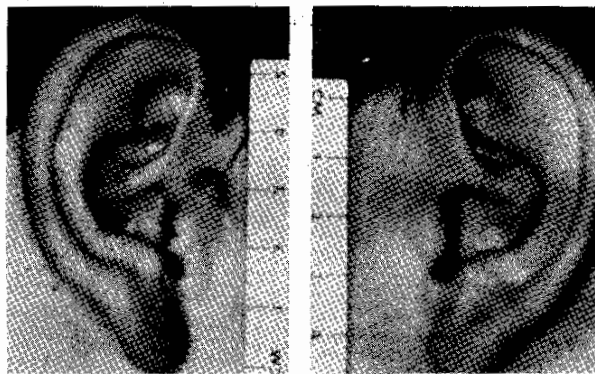


FIGURE 44. Mother of the proband represented in Figure 41. Note the bifid tragus and preauricular node. CCFA number 2267, age 30.

DEVELOPMENTAL AND ACQUIRED DEFECTS

Developmental defects due to infection (such as juvenile rheumatoid arthritis), traumatic injuries to the face, or ablative surgery for cancer have in common the production of oral-facial deformities that require multidisciplinary treatment in major medical centers to maximize the potential for habilitation.

Since the treatment of acquired defects is discussed elsewhere in this *Report*, the reader should apply the intellectual constructions suggested herein to the consideration of such experiments of nature as well.

CONCLUSIONS

In considering the experiments of nature that affect the structures of the head, our purpose was twofold. First, to reiterate Feinstein's (1970) suggestions for more effective clinical research. This would include reducing observer variability, developing new systems of taxonomy, establishing criteria for diverse clinical judgments, and improving quantification for "normality," prognosis, and therapy. The methodologies for doing so were illustrated from our long-term studies of craniofacial anomalies.

The second purpose would follow from the implementation of the first. By appropriate documentation of the phenomena observed in our patients and the results achieved through various therapies, we may presume that insights will be gained into causes and mechanisms. This approach should also yield more effective strategies of intervention to prevent deleterious phenomena and to provide improved therapeutic actions to remedy that which cannot be prevented.

ACKNOWLEDGMENT

The preparation of this paper was supported in part by grant Number DE-02872 from the National Institutes of Health and the Maternal and Child Health Services, Department of Health, Education, and Welfare.

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HEARING DISORDERS IN CHILDREN WITH OTOCRANIOFACIAL SYNDROMES

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The number of syndromes involving defects of hearing is so great that it would be presumptuous to attempt to cover the entire field. Instead, we have chosen to limit discussion to three clinical entities, omitting entirely a review of the extensive and more widely known work on hearing problems in cleft palate. These three groups are the premature craniosynostoses, hemifacial microsomia, and mandibulofacial dysostosis. The selection of these syndromes was dictated by a number of considerations. Each presents a separate and unique pathological basis for hearing disorders. In the premature craniosynostoses, we may assume that the otologic problems are generally secondary to developmental derangements in structures contiguous to the ear. On the other hand, in the otocraniofacial syndromes, the ear is malformed as part of a primary defect involving both the ear and contiguous structures. A more direct reason for consideration of these syndromes is that we have accumulated sufficient data on an adequate number of cases to permit us to unravel some of the complex interrelationships between structural malformation, clinical history, and audiologic and radiographic findings.

PREMATURE CRANIOSYNOSTOSIS

Among the various classifications of the premature synostoses, we prefer that of Bertelsen (1958). His system refers to skull shape, that is, the clinical picture presented by the patient. By inference, the involved suture is suggested. Thus, oxycephaly implies closure of the coronal suture, scaphocephaly implies sagittal closure, trigonocephaly implies metopic suture closure, and plagiocephaly implies unilateral closure, usually of the coronal suture. For most purposes, these types may be grouped together and called the simple forms of craniosynostosis. In complex forms, such as Crouzon's disease, Apert's syndrome, and related atypical cases, multiple sutures are involved with malformation in the midface as well as the neurocranium.

The literature on the simple forms of craniosynostosis is concerned mainly with the advisability, timing, and technique of surgical treatment of the

cranial sutures (Anderson and Geiger, 1965; Monnet and Pugeut, 1962). There is scant mention of associated ear malformations or hearing defects. While the paucity of literature may imply that the problem has been overlooked or considered of little interest, our own experience indicates that there simply are few otologic problems in this group. Of the 10 patients for whom we have complete records, eight have shown consistently normal hearing and appeared normal on otoscopic examination. One patient has shown occasional mild conductive loss associated with upper respiratory infections, and one older patient presented surgically confirmed otosclerosis. Hearing status in simple craniosynostosis is summarized in Table 1. From the standpoint of the

TABLE 1. Hearing status in simple craniosynostosis (20 ears in 10 patients).

<i>Number of Ears</i>	<i>Description</i>	<i>Hearing Loss in dB</i>
16	Normal Acuity	0-15
2	Mild Loss	15-30
2	Severe Loss	70-90

audiologist and otologist, this group may be considered as a "normal" population with no increase in frequency of a particular malformation or dysfunction of the hearing apparatus than might be encountered in the general population.

Crouzon's Disease, Craniofacial Dysostosis (CFD)

The situation is quite different for the complex forms of craniosynostosis. The literature is extensive, particularly in the French journals, dealing specifically with audiologic and otologic problems presented by patients with craniofacial dysostosis. While Crouzon (1912) made no mention of the state of hearing in his original description of craniofacial dysostosis in 1912, he subsequently described a patient with marked hearing loss of a progressive nature. Of 10 patients with Crouzon's disease studied by Aubry (1935), three showed atresia or stenosis of the external canal and four had unusually positioned tympanic membranes. All of his patients showed conductive hearing loss as determined by tuning fork tests. He attributed the loss, for cases without atresia, to ankylosis of the stapes or other ossicular fixation. Radiologic examination of these patients revealed acellular mastoids and areas of decalcification in the petrous pyramid.

The following year, Nager (1936) reported on sections of two sets of temporal bones from patients with Crouzon's disease. Both sets showed normalized middle ear cavities containing a normal malleus and incus. The stapes was malformed, the crura being oblique rather than perpendicular to the footplate, and was inserted into a narrowed oval window niche. The incus and

malleus were ankylosed. In contrast to the normal-sized middle ear space, the mastoids were small and showed unusual arrangement of the air cell walls. The bony and membranous labyrinths appeared normal, although the general orientation of the labyrinth was altered. Nager reported a general pattern of deficiency in structures dependent on periosteal ossification, while the endochondral parts of the labyrinth were normally developed. He also found two patients with stenosis of the external canal who showed hearing loss related to otitis media.

More recently, Boedts (1967) pointed out that the midfacial hypoplasia associated with craniofacial dysostosis creates chronic nasal and nasopharyngeal obstruction which may affect eustachian tube function. Baldwin (1968) reported that malformations of the eustachian tube have been seen in post-mortem examination of patients with Crouzon's disease, but did not elaborate on the findings.

Sensorineural hearing loss has also been observed, with Boedts (1967) and Gouzy et al. (1966) each reporting single cases. Several other authors have reported similar cases (Lafon, Collot, and Jakubowicz, 1964), but the loss was either poorly documented or contributory factors in the patient's history were ignored, so the validity of the findings is questionable.

The foregoing may give the impression that otologic problems are common to all patients with Crouzon's disease. Before reaching such a conclusion, let us examine the reasons such patients seek medical attention and the avenues for their referral. Gaudier et al. (1967), reviewing hospital records, observed that until recently, few patients with craniofacial dysostosis were referred for general evaluation on the basis of skull shape or facial appearance. Instead, they came to medical attention for specific symptoms, one of which was loss of hearing. Patients without such problems were seen by other specialists, such as ophthalmologists and plastic surgeons, according to their chief complaint, and were not studied and reported from an otologic point of view. Considering this, Clerc and Deumier (1958) estimated that one-third of patients with craniofacial dysostosis exhibit otologic problems.

In submitting our own experience for comparison, the question arises whether our sample is representative of the population of affected individuals. We are aware that some of our early referrals were made on the basis that the patient had a cleft palate. Thus, it may be assumed that our identification with a cleft palate clinic would skew our sample so that a greater percentage of the syndromes seen in our clinic would be associated with cleft palate. However, because of our primary identification as a center for craniofacial anomalies, most cases with craniofacial birth defects are now referred in the absence of cleft palate.

For these reasons, we should note that of nine cases of Crouzon's disease selected from our clinic for discussion, only one exhibited a repaired cleft of the soft and posterior third of the hard palate. Concurrently, we note that Gorlin and Pindborg (1964) reported, on the basis of their review of the

literature, that clefts of the secondary palate were a relatively commonly associated finding in this syndrome.

Of the nine patients we studied, four had normal hearing; four, including the patient with a repaired cleft, had conductive hearing loss ranging from mild to moderately severe, in association with otitis media; and the ninth patient exhibited atresia of the external auditory canal with moderately severe conductive loss. None of our cases had sensorineural loss. The hearing status of these patients is summarized in Table 2.

TABLE 2. Hearing status in Crouzon's disease (18 ears in nine patients).

<i>Number of Ears</i>	<i>Description</i>	<i>Hearing Loss in dB</i>
10	Normal Acuity	0-15
2	Mild Loss	15-30
3	Moderate Loss	30-50
3	Moderate to Severe Loss	50-70

The basis for hearing loss in these patients is varied. Loss may be primary, arising from malformations of the hearing apparatus, or hearing loss may develop secondarily as result of chronic ear disease. While chronic ear disease in these patients is well documented, their hypothesized predisposition to middle ear infection has not been subjected to any systematic study.

Our roentgencephalometric data support the hypothesis that the configuration of the nasopharyngeal area may predispose to middle ear disease. Lack of forward and downward growth of the midface reduces the vertical height of the nasopharynx. Basilar kyphosis further reduces the diameter of the nasopharyngeal port to the extent that many of these patients are forced to become mouth-breathers. It is also apparent that the petrous portion of the temporal bone is displaced upward and medially as part of the general distortion of the cranial base observed in the complex forms of craniosynostosis. In view of these findings, it seems reasonable to speculate that the eustachian tube physiology may be altered. However, direct proof of dysfunction remains to be demonstrated. To confound this thesis, we have observed patients with equivalent severity of midfacial hypoplasia who did not display chronic middle ear infection.

Acellularity of the mastoids has been reported as a common finding in craniofacial dysostosis, and as a contributing factor to chronic otitis media. In our own series, only one patient showed acellular mastoids, and one additional patient had a poorly developed mastoids system. Both showed signs of previous otitis media. The relationship of mastoids aeration and middle ear disease remains an unsettled question, with some investigators maintaining that lesser aeration of the mastoid predisposes to otitis media, and others claiming that poor aeration is itself a result of early infections.

No convincing explanation for the reports of sensorineural hearing loss has been offered. Gouzy et al. (1966) have suggested that changes in cochlear position and form secondary to the synostosis may account for sensorineural loss. Polytomographs in our series include cases exhibiting distortion and displacement of the cochlea without impairment of function. Baldwin (1968) hypothesized that hyperostosis of the inner table of the skull may narrow the internal auditory canal, compressing the nerve and interrupting blood supply to the labyrinth. Radiologic confirmation of a narrowed internal canal has not been reported for the published cases of sensorineural hearing loss. Coincidentally, studies of patients with osteopetrosis have demonstrated that the internal canal may be markedly constricted without incurring sensorineural hearing loss (Jones and Mulcahy, 1968).

Interruption of blood supply to the cochlea due to increased intracranial pressure has also been postulated. However, it seems unlikely that such pressures would affect the relatively protected auditory nerve while sparing the more vulnerable optic pathways. Patients with sensorineural hearing loss reported to this time have not been described as showing signs of optic atrophy. To our knowledge, central nervous system dysfunctions involving auditory perception have not been reported for this syndrome.

Apert's Syndrome, Acrocephalosyndactyly (ACS)

In contrast to craniofacial dysostosis, there is practically no literature on the hearing status in Apert's syndrome. Blank (1960) estimated an incidence of one in 160,000 live births, which, when adjusted for a high infant mortality rate, yields a figure of one in two million for the general population. Despite the relative infrequency of its occurrence, the syndrome is of such interest that at least 150 cases of Apert's syndrome have been reported in the world literature. Since our own experience suggests that otologic problems are common to this syndrome, we can only suggest that such pathology has been overlooked in the presence of the more dramatic neurologic, ophthalmologic, and orthopedic problems.

We have studied nine patients with Apert's syndrome (ACS Type I, McKusick), none of whom were referred specifically for otologic problems. Of the nine, one had a bifid uvula and another had a repaired cleft of the hard and soft palates. One child was so profoundly retarded that reliable examination was impossible. The remaining eight patients showed chronic otitis media. Seven patients, including the one with the repaired cleft palate, showed at least moderate hearing loss and required the insertion of myringotomy tubes. The hearing loss, while fluctuating according to the status of the frequently inserted myringotomy tubes, did not approach normal. The eighth child, with the bifid uvula, is as yet too young to respond reliably to play audiometry. Three older patients showed the effects of the chronic infection as manifest by clouding of the mastoid air cells and thickening of the air cell

walls. One five-year-old patient exhibited central perforation of one tympanic membrane and erosion of the ossicles in the other ear. The hearing status of seven of our nine patients is summarized in Table 3, and audiograms of four patients are shown in Figure 1.

TABLE 3. Hearing status in Apert's syndrome (14 ears in seven patients).

<i>Number of Ears</i>	<i>Description</i>	<i>Hearing Loss in dB</i>
1	Normal Acuity	0-15
1	Mild Loss	15-30
11	Moderate Loss	30-50
1	Moderate to Severe Loss	50-70

All factors previously discussed as predisposing to middle ear disease in craniofacial dysostosis also apply to patients with Apert's syndrome. This is not surprising, since a recent review of the roentgencephalometric data, in which Crouzon's disease was compared with Apert's syndrome, did not provide a foolproof basis for distinguishing one syndrome from the other solely on the examination of the skull films. Nevertheless, it was clear that, as a group, the patients with Apert's syndrome exhibited craniofacial deformities more severe than those with Crouzon's disease (Kreiborg and Pruzansky, in press).

The demonstration of dermatological findings in patients with Apert's syndrome, even though speculative, may have relevance to the occurrence of middle ear disease. The oral mucosae contained abnormal accumulations of mucopolysaccharides (Solomon et al., 1972¹) the skin was unusually thick, and abnormally distributed acneiform lesions were universally present in post-pubertal patients (Solomon, Fretzen, and Pruzansky, 1970). These findings suggest widespread disorders of the skin and its adnexa. Perhaps the ciliated epithelium which lines the middle ear spaces is also abnormal and either may tend to form exudate more easily than normal tissue or may fail to help clear the middle ear cavity.

Considering craniofacial dysostosis and Apert's syndrome together, the most apparent need is to establish accurate estimates of the prevalence of otologic disorders in these populations. This is not merely an academic question, but has direct relevance to patient care. If otologic disorders are a common finding in these syndromes, then it follows that the initial data base for such patients must include otologic and audiometric examinations with provision made for follow-up care and appropriate counseling of parents.

It is conceivable that unrecognized and untreated ear disease has frustrated rehabilitative efforts for some patients. Forming accurate estimates will necessitate more complete examination and description of individual cases, as well

¹L. M. Solomon, personal communication (1972).

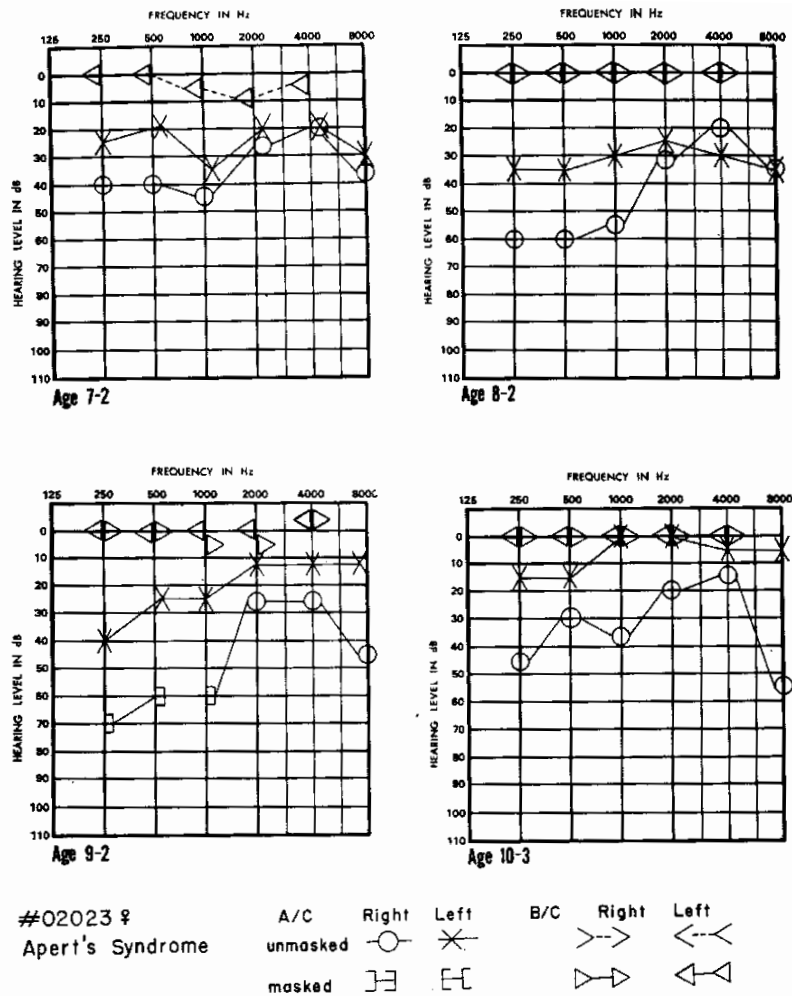


FIGURE 1. Audiograms on four patients with Apert's syndrome.

as surveys of larger clinical groups. Since these patients are not commonly seen in otologic clinics, it would seem useful to survey institutions. Many of these patients are institutionalized, either for the neurologic sequelae of their condition or because their unesthetic physical appearance caused them to be rejected early in life. However, health care, particularly otologic care, is often inadequate in these settings, and it would be impossible, particularly in otitis media, to separate the effects of the syndrome from the effects of placement.

It would be desirable to evaluate eustachian tube position and function on a longitudinal basis in these patients. If Holmquist's (1969) technique for using impedance audiometry to assess eustachian tube function becomes standardized, it will be possible to test patients without requiring a preexisting perforation or the performance of myringotomy.

The effect of periods of raised intracranial pressure on the hearing of these patients is also unknown. Saxena and his coworkers (1969) have documented hearing loss in adults who showed signs of raised intracranial pressure for reasons other than craniosynostosis. In 11 of 16 cases, hearing returned to normal levels after surgical intervention to relieve pressure, and the remaining five cases showed threshold improvement. Assuming that patients with premature craniosynostosis are recognized and tested promptly, initial periods of raised intracranial pressure, if present, will occur early, and the technical difficulties of studying hearing in infants are obvious. A study of the development of auditory skills such as localization, for comparison to normals, might be more in order. Since increases in intracranial pressure sometimes occur in older patients with craniosynostosis, hearing acuity could be studied directly in this group.

Otocraniofacial Syndromes

The otologic problems we have discussed here are, for the most part, secondary to developmental defects in contiguous structures which influence the function of the ear. In contrast, hemifacial microsomia (HFM) and mandibulofacial dysostosis (MFD) are entities in which the ear as well as contiguous structures are malformed early in intrauterine life.

Although the ear is but one of several observable malformations in patients with these syndromes, it holds a special interest for the investigator. The ear is a compound organ arising from several sources and developing over a long period in intrauterine life. Thus it may serve as a marker to indicate the timing of teratogenic factors. Since the malformed external ear is obvious to the clinician, it is tempting to ask to what extent the deformity of the outer structure might predict the status of the inner structures and their function.

Most investigators engaged in the study of the malformed ear have written about the ear in isolation, without delineating the presence or absence of malformations in structures contiguous to the ear. Presumably, such research designs assume that the type and severity of ear malformations in different syndromes are not significantly different. By lumping together all malformed ears, irrespective of the specific syndrome, we run the risk of obscuring possible patterns of malformation specific to a given syndrome. Because the features of mandibulofacial dysostosis have become well known, and the appearance of patients with the syndrome is distinctive, a separate literature concerning otologic problems occurring in mandibulofacial dysostosis has developed. (Axelsson et al., 1963; Clerc and Deumier, 1958; Fernandez and Ronis, 1964;

Herberts, 1962; Holborow, 1961; Ombredanne, 1970; Sando, Hemenway, and Morgan, 1968). The delineation of many other syndromes in which the ear is involved, and their classification into groups, is a relatively recent interest. Consequently, no separate otologic literature has evolved as yet for each of these newly emerged entities.

Even within carefully selected populations, the study of the ear presents problems. What is a malformed auricle, and where is the line of demarcation between a "variant" auricle and an abnormal auricle? There are no widely accepted standards in these matters, although some individual norms such as auricular height and angle of insertion have been studied (Bean, 1915; Linder, 1948). Instead, much work has proceeded on the assumption that an observer will "know" when an auricle is malformed and that he and the next observer will agree. While general agreement probably exists for markedly deviant conditions, it cannot be relied on as a basis for reporting less severe changes. The latter, as formes frustes or microforms, may be highly significant in genetic studies. In the absence of generally accepted standards, we must rely on explicit description and definition of terms as the basis for accumulating meaningful observation. In this connection, we have relied on photography, casts, and standardized radiographs to facilitate documentation.

Not only definition, but also classification of the malformed ear varies in the literature, usually according to the author's field of specialization. Two basic approaches are evident. In one, patients are classified according to middle ear malformation as disclosed by radiography or through surgical exploration. The philosophy behind this approach has been that "an attempt to produce reasonable classification by a study of the external ear alone is doomed to failure. The deformed external ear is only the tip of a submerged iceberg of malformation" (Gill, 1969). In the other approach, the malformed external ear is classified according to its structure, and middle and inner ear findings are related to the degree of external malformation. We prefer the latter approach, since it is based on features readily observed and constitutes the immediate frame of reference for the clinician. The feasibility of a meaningful classification based on the auricle cannot be ruled out until further study is carried out using homogeneous patient samples and interrelating clinical findings with audiometry, tomography of the temporal bone, and evaluation of contiguous structures.

We have retained the outside-in approach employed by Marx (1926) and Meurman (1957), in which the degree of deformity of the auricle serves as the basis for classification, and to which radiographic and audiometric observations are related.

For patients with hemifacial microsomia, the mandible was graded by a parallel system. Grading of the mandible in hemifacial microsomia, however, was not applicable to patients with mandibulofacial dysostosis, since these patients show a constant and distinctive mandibular configuration.

Tomograms were taken in the frontal and lateral views at 2-mm intervals

using a Sieman's multiplanograph with elliptical motion. Results were recorded on a standard form covering mastoid development, the external canal, middle ear cavity, ossicles, inner ear structures, and temporomandibular joint. Standard pure-tone audiometry was performed, with equipment calibrated to ISO standards. Other malformations, such as congenital heart disease or cleft of the lip or palate, were noted for each patient.

HEMIFACIAL MICROSOMIA

As previously noted, this is a syndrome known by many names. In deference to the influence of Gorlin and Pindborg's book (1964) on oral-facial syndromes, the term *hemifacial microsomia* is retained in this paper. However, the term is not entirely satisfactory. It does not specify the focal relationship of ear and mandible; the condition is not always unilateral, and the degree of facial asymmetry is highly variable.

Fifty-seven patients ranging in age from four to 22 years formed the hemifacial microsomia group. Forty-four patients had unilateral involvement, and 13 had bilateral involvement of the ears, altogether comprising 70 malformed auricles. Insofar as we could determine, the sample was typical for the syndrome. There was a greater frequency of right-sided malformations, and a greater number of males than females, in concordance with previously published reports (Meurman, 1957). Within the entire group, severity was evenly distributed, with a predominance of mild malformations in the bilateral group. The results are summarized in Table 4.

TABLE 4. Distribution of microtic ears (70 ears in 57 patients, 40 males and 17 females).

Grade of Ear Deformity	Male		Female		Totals
	Right	Left	Right	Left	
I	10	4	3	6	23
II	10	6	4	3	23
III	10	7	6	1	24
Totals	30	17	13	10	-

Hearing loss was primarily conductive for all degrees of auricular malformation. However, 18 of the 70 malformed cases showed mixed loss (defined as two or more b/c responses of 20 dB or greater), and among the Grade I group, the mildest deformity, we found cases with normal acuity and with sensorineural loss.

Extent of hearing loss was tabulated with response to low, mid, and high frequencies individually grouped. The results are shown in Table 5. The trend is for degree of loss to become greater as auricular deformity becomes greater

TABLE 5. Extent of hearing loss related to different grades of microtic ear deformity (70 ears in 57 patients).

Extent of Hearing Loss (dB ISO)	Grade of Ear Deformity								
	I (23)			II (23)			III (24)		
	low	mid	high	low	mid	high	low	mid	high
0-15	3	5	5	-	-	1	-	-	-
15-30	7	8	6	-	-	-	-	-	-
30-50	2	2	3	2	3	2	-	-	-
50-70	6	4	4	10	14	12	4	7	15
70-90	3	3	2	5	6	8	12	12	5
90-110	-	-	1	5	-	-	6	4	1
110 +	2	1	2	1	-	-	2	1	3

(Pearson $r = 0.86$). However, acuity was unpredictable for Grade I malformation of the auricle. This distribution pattern remained constant when patients were divided into unilateral and bilateral groups. Patients in any group who showed mildly affected mandibles tended to have less severe hearing impairment than similar cases with marked mandibular involvement.

A review of ear structures by tomography showed the same trend toward increased severity of involvement with increased auricular malformation, but with the same reservation that conditions in Grade I auricular malformation cannot be predicted. For example, among Grade I ears the external canal was normal in two-fifths of the patients, stenotic in two-fifths, and atretic in one-fifth. By contrast, only three patients in each of the remaining groups failed to show atresia. Of these six patients, five showed marked stenosis of the canal.

Tables 6 and 7 show that the malleus and incus, reflecting their common origin, were affected as a unit and were malformed more frequently than the

TABLE 6. Relationship between the status of the mandible and the status of the malleus and incus (normal/abnormal) shown as a function of the degree of auricular deformity.

Status of Mandible	Status of Malleus and Incus (Degree of Auricular Deformity)			Totals
	I	II	III	
Normal	8/2	2/4	0/3	10/9
I	2/1	3/9	0/11	5/21
II	1/7	0/4	0/7	1/18
III	1/1	0/1	0/3	1/5
Totals	12/11	5/18	0/24	

TABLE 7. Relationship between the status of the mandible and the status of the stapes (normal/abnormal) shown as a function of the degree of auricular deformity.

Status of Mandible	Status of Stapes (Degree of Auricular Deformity)			Totals
	I	II	III	
Normal	10/0	6/0	1/2	17/2
I	2/1	5/7	1/10	8/18
II	5/3	1/3	2/5	8/11
III	2/0	1/0	0/3	3/3
Totals	19/4	13/10	4/20	

stapes. Again, the parallel between auricular malformation and middle ear structure was evident.

The data presented are a compressed version of our original tabulations which specified the type of ossicular abnormality rather than using a normal-abnormal division. For such a small number of patients, a display of subgroupings of abnormality was useful neither for illustration nor for statistical purposes. As more patients are studied, it will become possible to use the auricle and mandible as axes and to fit a plane of graded ossicular deformities to these combined axes. At that time, more precise association of ossicular deformity to clinical signs may emerge.

Tomography revealed bony malformations of the inner ear structures in 10 of 70 malformed auricles. These included the following malformations: two cases of cochlear hypoplasia, five cases of abnormal internal auditory canal (three cases of marked constriction and two cases of unusual width), and three cases in which the vestibule was enlarged or the semicircular canals shortened and noncanalized. None of these 10 ears presented sensorineural loss, nor was the degree of their loss greater than usual. On the other hand, the two patients demonstrating sensorineural loss showed no inner ear malformation on tomography.

Although patients with clefts accounted for only 13 of the 57 studied, five of the 10 cases of inner ear malformation occurred in patients with clefts.

MANDIBULOFACIAL DYSOSTOSIS

A much smaller group of 10 patients with mandibulofacial dysostosis was available for study. Selection was based on the presence of the obligatory findings for diagnosis prescribed by Axelsson et al. (1963), that is, malar and mandibular hypoplasia, antimongoloid obliquity of the palpebral fissures, and anomalies of the lower eyelids. The patients ranged from three and one-half to seven years of age. Our sample included two males and eight females, but since the literature would indicate an approximately equal number of affected males and females, the sample may not be representative. The group was atypi-

cal in a second sense, in that all had microtic auricles—which is not considered an obligatory finding for diagnosis, since the auricle may be normal in mandibulofacial dysostosis. The literature reveals the external ear may be affected in other ways, such as low placement or unusual configuration of the auricle. As similar patients come within our purview, we shall need to expand our classification. At present, we can only report on patients with mandibulofacial dysostosis who also have microtia, and we will not attempt to generalize beyond this subgroup.

The 10 patients with mandibulofacial dysostosis, coded for each ear, presented the following grades of microtia: 11 Grade I, four Grade II, and five Grade III. The severity of external ear deformity is somewhat less than that encountered in the larger hemifacial microsomia series. As in the hemifacial microsomia series, the type of hearing loss was mainly conductive and did not change for different grades of auricular deformity.

The parallelism between the degree of malformation of the auricle and hearing loss observed in hemifacial microsomia did not seem to prevail for mandibulofacial dysostosis. Only four ears in the mandibulofacial dysostosis series showed thresholds above the 50- to 70-dB range—this despite the large proportion of mildly affected auricles. These data are shown in Table 8.

TABLE 8. Extent of hearing loss shown as a function of microtic deformity (20 ears in 10 patients).

Extent of Hearing Loss (dB ISO)	Grade of Ear Deformity								
	I (11)			II (4)			III (5)		
	low	mid	high	low	mid	high	low	mid	high
0-15	2	2	—	—	—	—	—	—	—
15-30	—	—	—	—	—	—	—	—	—
30-50	—	—	—	2	2	2	—	—	—
50-70	7	8	8	—	—	—	5	3	4
70-90	2	1	3	2	2	2	—	2	1
90-110	—	—	—	—	—	—	—	—	—
110 +	—	—	—	—	—	—	—	—	—

Tomography was available on only seven of the patients. Those studied revealed a greater severity of malformation compared with that of patients with hemifacial microsomia. Nine of the 14 ears showed complete agenesis of the external canal, three were atretic, and two normal.

The middle ear cavity was completely absent in six ears, severely hypoplasia was present in another six, and only two ears exhibited a middle ear space within normal limits. In the eight existing middle ear cavities, only one contained a set of normal ossicles. There was one case of a disarticulated ossicular chain, four cases of a single ossicular mass, and two cases of individual but malformed ossicles.

Since both syndromes have malformed structures in common, it is tempting to draw some comparisons even though our sample sizes are uneven. As a group, patients with mandibulofacial dysostosis had external ears that were less severely involved than the larger hemifacial microsomia series. On the other hand, the middle ear was more profoundly and more constantly affected in mandibulofacial dysostosis than in hemifacial microsomia.

The parallelism between the external ear and the middle ear malformation observed in hemifacial microsomia was obviously absent in mandibulofacial dysostosis. How can this be explained? In hemifacial microsomia, the mandible is affected variably, whereas in mandibulofacial dysostosis the mandibular deformity is uniform and relatively severe in all cases. Since we have already shown that in hemifacial microsomia there is a parallelism between the severity of the mandible and that of the malleus and incus, both derivatives of Meckel's cartilage, it is not altogether surprising to find that the ossicular changes in mandibulofacial dysostosis are nearly always severe, since the mandible is also uniformly affected.

The audiologic dysfunction encountered in hemifacial microsomia and mandibulofacial dysostosis merits further investigation. The severe conductive loss shown by some of these patients exceeds the limits once considered the maximum conductive loss possible. The nature of the ear malformation that might account for this extensive conductive loss is unknown. Our knowledge of the mechanisms of bone conduction does not explain the occasional cases of mixed loss in patients with malformations limited to the conductive mechanism. Perhaps mixed loss represents a cochlear defect not visible on radiographic examination, but it may also represent an artifact of testing. The ossicular chain, generally considered solely a transmitter of air-conducted sound, does affect the bone-conduction threshold obtained in audiometric testing. The contribution of individual structures within the normal ossicular chain has been studied, but no model for the congenitally malformed ear has been attempted.

In a more clinical vein, studies of auditory skills in cases of unilateral malformation and hearing impairment are in order. Although we have been impressed that the patient with one normal ear has no problems in daily functioning, many of our patients report having been urged to obtain expensive hearing aids or to undergo middle ear reconstruction so that they might have the "benefit" of binaural hearing. It is clear that we require better functional assessments of patients with unilateral hearing deficits, before and after specific therapeutic intervention. Concurrently, we require more explicit criteria for surgical reconstruction of the middle ear. Only through such documentation will we be able to weigh the apparent deficit against the promise of improvement and the risks to be incurred by specific surgical treatment.

COMMENTS

As was recently pointed out in a monograph on congenital deafness (Black

et al., 1971), there is a growing interest in congenital ear pathology and the need to facilitate identification of the hearing-impaired infant. The present literature contains nonvalidated pronouncements on the hearing states of a variety of entities and errors of omission where useful data might have been anticipated.

The clinical investigations reported by our own group illustrate the value of the interdisciplinary approach directed toward a description of the organism rather than the organ. In this way, developmental and functional interrelationships between contiguous organs may be revealed.

The investigation of the seemingly rare and unusual case is not only important for the management of the patient affected, but also may yield information about basic processes of hearing and stimulate further research into these processes.

ACKNOWLEDGMENT

The research described here reflects the contributions of many individuals. The overall direction of this project has been the special interest of S. Pruzansky from its inception. G. Valvassori has contributed his outstanding expertise in tomography and has consulted with us on many of the findings and interpretations. From 1965-1967, Sally Peterson served as the audiologist on the project and contributed to systematizing the data. Leslie Rodman performed a similar function as audiologist from 1968-1969. David Caldarelli otologist at the center, has examined many of these patients and has advised on several aspects of the study. The investigations were supported in part by grant number DE 02872 from the National Institutes of Health and the Maternal and Child Health Services, Department of Health, Education, and Welfare.

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SPEECH PATHOLOGY IN CRANIOFACIAL MALFORMATIONS OTHER THAN CLEFT LIP AND PALATE

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Patients with the kinds of craniofacial malformations to be discussed in this paper do not ordinarily come within the purview of community speech and hearing facilities or even university speech clinics. For a majority of speech pathologists, experience with these patients is limited or nonexistent. There are several interrelated reasons for this. First, the incidence of these malformations is relatively low. Second, the severity of the malformations is often so great as to obscure the need for the services provided by the speech pathologist. These two factors have, in turn, led to a third: having had so little experience with patients presenting craniofacial malformations other than clefts of the lip or palate, speech pathologists have come to view the study of such patients as an exercise in esoterica. This picture, however, is changing.

Low Incidence of Craniofacial Malformations. There is evidence that greater numbers of these patients are being identified and referred as the result of increased diagnostic sophistication (see Pruzansky, elsewhere in this *Report*). In a presentation before the recently concluded thirtieth annual meeting of the American Cleft Palate Association, Robert Gorlin (1972) estimated that new syndromes affecting the craniofacial region are being identified at the rate of several a month.

In addition to increased diagnostic sophistication, several other factors are operating to alter the "low incidence" status of many of the craniofacial malformation syndromes. As Pruzansky points out in this *Report*, referrals of patients with such malformations increase when it becomes known that something can be done for them. For example, the newly reported surgical techniques developed by Tessier (1971) can be expected to result in increased referral of patients with premature cranial synostosis. Advances in medical science also mean that more children with congenital defects now survive. An additional factor in changing incidence figures is the variety of "early identification" programs which have been initiated as a result of recent legislation in health care and education for the handicapped, and which will almost certainly lead to heavier clinical populations in virtually all health services.

Interdisciplinary Care. A logical development in the diagnosis and treatment of patients with craniofacial malformations has been that those multidisciplinary clinics or centers which have served cleft-palate patients in the past are now diversifying to include patients with more complex anomalies. Speech pathologists who are members of these interdisciplinary teams are among the first to have the opportunity to examine these patients. The participation of the speech pathologist is critical, since undiagnosed deficits in speech and hearing have too often added to the "hopeless" image of such patients and led to the institutionalization of children who might have benefitted from treatment. Further, the opportunity and motivation for significant improvement in speech (as well as other functions) increases with advances in surgical and prosthetic treatment. One result of improvements and innovations in physical management should be the eventual referral of patients to speech pathologists in community and university speech clinics for therapy. This has been the case in the care of cleft-palate patients.

Esoterica or Basic Science? What are the possibilities that the knowledge gained from more systematic study of patients with craniofacial malformations will add insights into fundamental processes and therefore prove to be of general value, ultimately benefitting other types of patients as well? The history of medicine is replete with examples wherein the pathologic state first provided clues about the normal mechanism. Not until something goes profoundly wrong are we compelled to learn how the machinery really works in the first place. Corollary examples are present in the work of the early neurologists in aphasia. Admittedly, many of the early conclusions had to be altered when it was realized that inferences from the pathologic to the normal mechanism must be made with extreme caution. Nevertheless, the practice of studying normal development and function through observation of, and comparison to, the abnormal is still widespread in medicine today. For example, Reichel, Garcia-Bunuel, and Dilallo (1971) proposed Werner's syndrome and progeria as models for the study of normal human aging.

Indeed, a major justification for the conference leading to this *Report* is the realization that the clinical problem is an "experiment of nature" which, if properly considered, may yield basic information in speech science extending beyond the problem under immediate consideration.

A foray into the literature on craniofacial syndromes, in search of information on speech and language disabilities in these patients, was notably short-lived. As Bloomer (1971) repeatedly points out, the specific effects the various syndromes have on speech are seldom reported. The majority of published case reports do not even mention speech.

Bloomer's effort (1971) is the only known attempt to compile what little information has been published regarding speech, hearing, and language problems in craniofacial anomalies other than, or in addition to, cleft palate. Bloomer approaches his topic primarily from the standpoint of dental and occlusal anomalies as they occur in these syndromes, although he does not

neglect other clinical findings or such equally important factors as intelligence, motivation, and emotional adjustment. In fact, he stresses the need to guard against overemphasis on morphologic findings, stating, "It would be an oversimplification and a misrepresentation of the interrelationship of defective speech and orofacial abnormalities to ascribe all manifestations of the disordered speech to anatomical causes," and "orofacial structures are not necessarily to be considered prime causes of defective articulation. Speakers have demonstrated that the oral mechanisms are capable of many compensatory adaptations by which intelligible speech can be produced even though the patients are deformed."

Bloomer (1971) takes up a variety of orofacial anomaly syndromes, several of which he discusses in the context of abnormal mandibular/maxillary relationship or abnormal development of one or both jaws. In addition, he draws attention to syndromes such as Melkersson-Rosenthal syndrome, Sturge-Weber syndrome, and "orodigitofacial dysostosis," in which oral findings would predict speech difficulties but in which speech characteristics have not been described or documented. Bloomer supplements what little information is given in the literature with descriptions of his own cases, which in some instances constitute the only published descriptions of speech findings in these syndromes (see Bloomer's discussion of Melkersson-Rosenthal syndrome, hemifacial hypertrophy, hemifacial atrophy, and Klippel-Feil). As mentioned earlier, the majority of published case reports dealing with specific craniofacial syndromes do not even mention speech. The articles that do mention speech usually devote no more than one or two sentences to the topic. Those syndromes in which speech has been given at least passing consideration in the literature include Pierre Robin (Bloomer, 1971; Dennison, 1965), Cornelia de Lange (Jervis and Stimson, 1963; McArthur and Edwards, 1967; McIntyre and Eisen, 1965; Nicholson and Goldberg, 1966; Ptacek et al., 1963; Schuster and Johnson, 1966; Silver, 1964), Treacher-Collins (Bloomer, 1971; Massengill et al., 1971), Sturge-Weber (Chao, 1969), Turner's (Jackson and Sougin-Mibashan, 1953), Moebius (Bonnar and Owens, 1929; Butlin, 1885; Everbusch and Nadoleczny, 1914; Fry, 1920; Gordon and Brown, 1933; Henderson, 1939; Merz and Wojtowicz, 1967; Sproffkin and Hillman, 1956), Klippel-Feil (Bloomer, 1971), congenital aglossia (Bloomer, 1971; Eskew and Shepard, 1949; Kelln, 1968; Merson, 1967; Pettersson, 1961; Sulzmann and Seide, 1962; Weinberg et al., 1969; Weinberg and Paras, 1970), ectodermal dysplasia (Massengill et al., 1969), hemifacial microsomia (Massengill et al., 1971), Apert's (Massengill et al., 1971), and Laurence-Moon-Biedl syndrome (Burns, 1950; Ciccarelli and Vesell, 1961; Garstecki, Borton, and Stark, 1972; McCulloch and Ryan, 1941; Roth, 1947; Serejski, 1929). Since no additional references could be found on Sturge-Weber, Moebius, or Turner's syndromes beyond those Bloomer already reviewed, his review will not be repeated here.

It may be useful to focus specific attention on a review of some studies on Pierre Robin syndrome, mandibulofacial dysostosis, hemifacial microsomia,

congenital aglossia, Cornelia de Lange syndrome, the Laurence-Moon-Biedl syndrome, and the premature craniofacial synostoses, and to add information from our own observations of patients with these and other syndromes.

FOUR "PITFALLS" IN REVIEWING THE LITERATURE

Critical appraisal of what little has been reported in the literature about speech in these syndromes is made difficult by at least four recurrent, seemingly unrelenting problems. (1) Patients have too often been misdiagnosed, the speech findings thus being essentially "misreported" insofar as the patient had not really one syndrome but another. (2) There has been a disturbing tendency toward lumping together all the examined cases of one syndrome in a homogeneous group, without recognizing important individual differences and ranges of variability. The most blatant example of this is the failure to point out that some patients exhibiting a syndrome whose findings are being reported in an article actually had cleft palate, while the remaining patients did not. (3) Speech findings have, in some instances, been viewed only in relation to one or two morphologic findings, completely ignoring other critical factors. For instance, speech problems in syndromes involving mandibular prognathism or cleft have been related exclusively to the prognathism or to the cleft with no recognition of other pertinent variables. (4) Mental retardation is often present to a significant degree in these syndromes. Our task would be much easier and our research much "cleaner" if we were not faced with deciding to what degree a speech or language deficit may be due to a child's limited intellectual development and to what degree the child's overall intellectual development may be hindered by a specific language or motor speech deficit. Similarly, the contributing effects of significant hearing loss and adverse psychosocial factors operating in so many of these patients have often escaped the attention of investigators.

Pierre Robin Syndrome

This syndrome is characterized by cleft palate, glossoptosis, and mandibular micrognathia. Respiratory embarrassment and feeding difficulties in the neonatal period may be severe enough to be life threatening or may be relatively mild. The severely affected cases may require tracheostomy, suturing of the tongue to the lower lip, or other procedures to control the glossoptosis.

From an a priori standpoint, we may assume that persisting speech problems are primarily related to incompletely successful treatment of the velopharyngeal incompetency resulting from the isolated cleft of the palate. Indeed, Dennison (1965) has described the speech of Pierre Robin patients as being characterized by hypernasality and articulatory defects. Bloomer (1971) describes two patients in whom defective articulation was ascribed to significant hearing loss, and goes on to mention several patients whose tongue-tip

mobility was impaired (with consequent effect on tongue-tip consonants) as a result of the Beverly-Douglas procedure. However, no one to date has documented the existence of speech defects in Pierre Robin syndrome that were not ascribable to hearing loss or the cleft condition.

One other, time-related variable that may or may not have an effect on speech performance is related to the micrognathia that is a pathognomonic feature of this syndrome. Serial roentgencephalometric studies have revealed that for most cases the micrognathia diminishes in time, thereby alleviating the maxillary-mandibular structural disharmony (Pruzansky, 1969; Pruzansky and Richmond, 1954). The effect of the micrognathia and the resultant malocclusion on speech production would therefore appear to depend on the individual patient's state and pattern of growth.

Mandibulofacial Dysostosis (Berry, Treacher-Collins, Zwahlen, and Franceschetti-Klein Syndromes)

The clinical findings in this syndrome have been summarized by Axelsson et al. (1963). The morphologic features which may have particular bearing on speech and language include hearing loss, hypoplasia of the maxilla and mandible, high-arched or cleft palate, and dental and occlusal anomalies. The mandibular hypoplasia results in a peculiar curvature of the lower border of the mandible, with consequent malocclusion, usually involving an open bite that becomes worse in time. The curvature of the mandible also results in a retracted and elevated posture of the posterior part of the tongue. Cleft palate occurs in approximately 40% of the cases (Gorlin and Pindborg, 1964).

Bloomer (1971) summarizes the speech and language disturbances in this syndrome: delayed language development, articulatory defects, hypernasality, and "central language disorder." Bloomer's descriptions of two of his own patients, a brother and sister aged 10 and 12, reflect the effects of the characteristic mandibular deformity. In the boy, Bloomer states, "the micrognathia interferes with articulatory adaptations of the tongue." In the girl, "restricted mobility of the mandible interferes with mouth opening and hence affects voice quality and articulation to some extent, but its effects are slight." The "hyponasal" voice quality noted in both children may be ascribed to crowding of the nasopharyngeal airway resulting from lack of growth of the maxilla and mandible, as well as from the peculiar curvature of the mandible with resultant retraction of the body of the tongue.

Massengill and associates (1971) published an article entitled "Documentation of Syndactyly and Treacher Collins Syndrome for Possible Concomitant Speech Disorders." (In discussing the cases these authors presented, I will reverse the order, returning to their findings on the syndactyly patients after discussing the speech findings on those patients identified as having Treacher-Collins syndrome.) Unfortunately, this is one of those instances in which the accuracy of diagnosis is subject to serious question. The published article

contains pictures of two of the five patients reported to have Treacher-Collins syndrome. Neither picture is consonant with such a diagnosis, as neither shows the obligatory bilateral antimongoloid slant of the palpebral fissures or the bilateral defects of the lower lids. Both pictures appear, rather, to depict cases of hemifacial microsomia. In addition, the description of at least one other patient does not substantiate a diagnosis of Treacher-Collins syndrome. The only physical description of this child reads, "a 13 year old white male who has been treated for unilateral facial agenesis. In addition to this problem he has a bilateral facial paralysis" (Massengill et al., 1971).

Treacher-Collins patients, like most with congenital malformation syndromes, show a range of physical findings. The overlap between syndromes has puzzled many diagnosticians and geneticists. Nevertheless, in the article under discussion (Massengill et al., 1971), the scanty physical documentation of the presence of the Treacher-Collins syndrome and the two cases of direct pictorial contradiction of the diagnosis make it difficult to accept the reported speech findings as truly representative or "typical" of such patients.

Consider Massengill's findings in the two cases where a diagnosis of Treacher-Collins syndrome is asserted in the descriptions of the patients with no contradictory pictorial evidence. One patient (Subject S4 in the published report) is described as "an 11 year old negro male who was born with a mild Treacher-Collins syndrome, including hypodevelopment of the left side of the face and partial agenesis of the left ear." This patient had a conductive hearing loss unilaterally, associated with the ear deformity. His articulation and voice quality were within normal limits, with diadochokinetic rate indicating adequate control and mobility of the speech musculature. A lateral headplate taken during phonation of /i/ indicated good velopharyngeal closure. A second patient (Subject S2) is described as having Treacher-Collins syndrome and other congenital anomalies, including a cleft palate and left dorsal kyphoscoliosis. The cleft had apparently been treated with unsatisfactory results, since the authors state that her many sound distortions were due chiefly to nasal emission, and cinefluorographic analysis indicated a large velopharyngeal gap with poor palatal and pharyngeal mobility. Mobility of the tongue, lips, and jaw is described as "adequate" based on diadochokinetic rate. Therefore, the article by Massengill and his associates tells us that speech findings were satisfactory in one patient studied with a mild manifestation of the Treacher-Collins syndrome and unsatisfactory in a patient in whom the physical findings included a cleft palate with resultant velopharyngeal inadequacy. Neither of these findings is particularly surprising or informative.

Bloomer's case descriptions (1971) reflect the effects of the abnormalities in mandibular shape and size on the motor speech mechanism. However, no "hard" data (cinefluorographic studies, growth records, speech ratings, articulation test results) are available in the literature which might allow study of the relationship between morphologic findings and motor speech deficits. No one has attempted systematic description (and quantification, if possible) of the

“abnormal” tongue posture. I put the word *abnormal* in quotes here because we may be presumptuous in applying a normal-abnormal continuum to a tongue sitting in a severely bowed mandible. Further, we do not have descriptive data on lingual mobility or the possible effects of deviant tongue posture on the function of anatomically related musculature, including that of the velopharyngeal port.

Hemifacial Microsomia

Returning to the article by Massengill and associates (1971), let us consider their discussion of two patients whom they identify as having Treacher-Collins syndrome, but who appear to exhibit hemifacial microsomia. Speech findings in hemifacial microsomia are not discussed elsewhere in the literature. Both patients described by Massengill and his associates appear, from their pictures, to have been mildly affected. The findings on the first patient (designated as Subject 1 in the report) included right microtia with no visible canal and bilateral conductive hearing loss. Articulation was described as good, and control and mobility of the tongue, lips, and jaw were judged adequate based on diadochokinetic rates. Interestingly, voice quality was judged hypernasal, although there was no mention that the patient had a cleft or palatopharyngeal incompetency. Cinefluorography reportedly showed that the palate moved “well.” There was no indication in the report of how voice quality was judged hypernasal, or what the authors felt accounted for the hypernasality in the apparent presence of adequate palatal mobility.

The second patient (Subject 5) was reported to have excellent speech with good mobility and control of the speech musculature and adequate velopharyngeal closure as demonstrated on lateral head x ray. No data are given on either patient regarding function of facial musculature. Even though both patients appear to have been mildly affected, it would be interesting to know



FIGURE 1. Case 2171, Girl, age eight years, two months, with hemifacial microsomia. Note left microtia and facial asymmetry.

the findings regarding symmetry of function on phonation of /a/, speech tasks, and nonspeech movements of the oral musculature.

Figures 1, 2, and 3 illustrate one of our own patients who presents a mild manifestation of hemifacial microsomia except for marked atrophy of the left side of the tongue and Grade II microtia of the left ear. Figure 3 demonstrates some of the effects of the paralysis of the lower branch of the facial nerve. The palate elevates asymmetrically on phonation, showing a levator dimpling on the right. There is a bifid uvula (Figure 2). In addition to the hypoplasia of

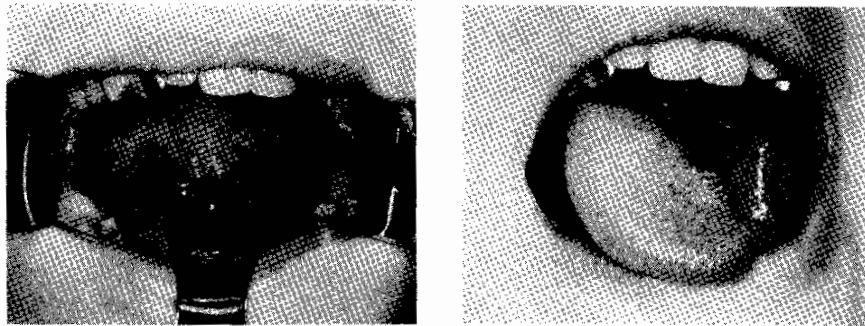


FIGURE 2. Same patient in Figure 1, at age nine years, three months. Left, note bifid uvula. Right, note left hypoplasia of tongue.

the left side of the tongue (shown in Figure 2), the posterior part of the tongue body appears to be rather tightly "anchored," producing a marked depression or central grooving in this area.

This patient's speech in a one-to-one conversational situation is intelligible, with evidence of mild, intermittent hypernasality. The hypernasality increases in a situation requiring increased loudness or in a group setting, although her intelligibility remains satisfactory. On lateral cephalometric films, this patient appears to make closure on /s/ but not on /u/, demonstrating a gap of 1.5 to 2.0 mm on the vowel. Interestingly, this patient performed within the normal range on oral stereognosis testing using the NIH-20 series of oral form perception. Regrettably, we do not have sound cinefluorographic films, which would contribute to our study of such patients, as would electromyographic data.

Congenital Aglossia

Congenital aglossia and aglossia following ablative surgery are discussed at some length elsewhere in this *Report*. The summary impression from this discussion is similar to the consensus derived from prior descriptions of such cases, that is, a great number of these patients do extraordinarily well without a tongue. For an intriguing look at how patients without a tongue manage as

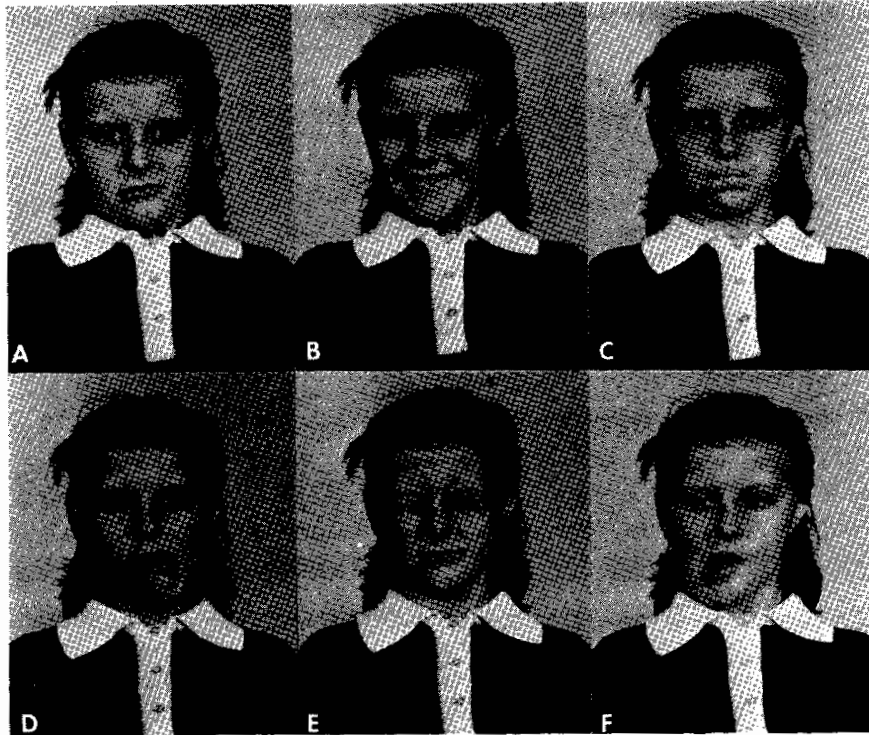


FIGURE 3. Case 2171 at same age as in Figure 2. Pictorial representation of clinical assessment of functional symmetry of mimetic musculature. A. In repose. B. Smile. C. Inflation of cheeks. D. Abduction of mandible to left. E. Closure of eyelids. F. Abduction of mandible to right.

well as they do in oral communication, I refer you to a description by Eskew and Shepard (1949) of a 22-year-old Chinese man with congenital aglossia: "In speaking, the buccinator muscles were very noticeable in their movement, as were the muscles of the floor of the mouth." The floor of the mouth was smooth, but could be elevated in a tongue-like structure which could contact the incisal edges of the maxillary anterior teeth. For /k/, this patient used contact of the buccinators with the molars; /t/ was made similarly, with some difference in breathstream control to differentiate it from /k/. All vowels were made clearly except for /e/ and /i/.

Bloomer (1971) describes the speech of one of his patients who had a "small tongue" as intelligible but characterized by some articulatory distortion of sibilants. Articulation and resonance were affected by velopharyngeal incompetency, with consequent hypernasality and slightly reduced oral air pressure. Weinberg and Paras (1970) have described the speech of a seven-year-old girl with severe congenital hypoplasia of the tongue. They found her

overall intelligibility to be approximately 86%. Voiceless consonants were more intelligible than voiced consonants, and fricatives, affricates, and glides were more intelligible than plosives and nasals. The authors state, "The general patterning of errors was similar to those of a normal talker." This patient had had no speech therapy.

To add to this brief review of aglossia, I will mention one of our patients who exhibits the aglossia-adactylia syndrome. Unfortunately, at age two and one-half this child is too young for detailed articulation tests or sound cinefluorographic films. Our only "hard" data consist of cephalometric films at rest. The surgeon has estimated that this child has no more than the posterior seventh or eighth of a tongue. Yet the family pet is named Caesar, and the boy can say the name intelligibly.

Cornelia de Lange Syndrome

This syndrome has several characteristic craniofacial features, including marked hirsutism, synophris, long lashes, microcephaly, long philtrum, upturned nose, carp-like mouth, and a high-arched or cleft palate (Gorlin and Pindborg, 1964). Patients generally show severe mental retardation. As would be expected, severe delays and deficits in speech and language have been found in children showing this syndrome (McIntyre and Eisen, 1965; McArthur and Edwards, 1967; Moore, 1970; Ptacek et al., 1963). Significant hearing defect has also been noted (Jervis and Stimson, 1963). There has been little interest in the distinctive vocal quality of these children. The quality is characteristically described as feeble, low-pitched, and animal-like (Nicholson and Goldberg, 1966; Ptacek et al., 1963; Schuster and Johnson, 1966; Silver, 1964). The cry heard in the nursery is so characteristic in this syndrome as to be diagnostic. Moore (1970) has wondered whether the low-pitched, growling cry may be secondary to malfunctioning thyroid glands. Definitive studies of these patients by laryngologists and speech scientists are long past due.

Laurence-Moon-Biedl Syndrome

The characteristic findings in the Laurence-Moon-Biedl syndrome, which has been attracting recent interest from speech pathologists, include obesity, hypogenitalism, polydactylism, ocular defect, and mental deficiency. Descriptions of speech deficits in these children have included "slow speech" (Cicarelli and Vesell, 1961), "delayed speech" (Serejski, 1929), "scanning speech" (Roth, 1947), and "difficult speech and tongue deviations" (Burn, 1950). Burn (1950) has also described deafness in this syndrome.

In a recent study by Garstecki, Borton, and Stark (1972), three siblings affected to varying degrees and two unaffected siblings (all from the same family) were assessed for speech, hearing, and language deficits. Audiometric results were not particularly impressive. One affected child had a mild-to-moderate loss unilaterally, while another had a mild bilateral loss. All three

affected children had articulation disorders of varying degrees, hypernasal voices, significantly retarded language, and neuromuscular deviations of the speech mechanism which the authors felt to be the primary reason for the poor articulation. The neuromuscular deviations were of particular interest to the authors, in that they found such deviations had been reported in the literature but not as features of the syndrome. While Garstecki et al. (1972) noted that their subjects were indeed retarded, as assessed by typical school achievement tests, they felt that the significance of the neuromuscular findings had been consistently overlooked in the previous reports. In their own patients, poor velar movement was noted in only two of the affected children, although all three sounded hypernasal. Interestingly, the two with poor velar movement were intelligible (despite poor articulation and poor voluntary tongue and lip control), while the third sibling was unintelligible although velar movement appeared adequate. These complex, puzzling findings certainly point to a need for further definitive research.

In evaluating the literature on this syndrome, we must return to our original caution that relates to the validation of the diagnosis. In this connection, Warkany (1971) has recently reconsidered the difficulties involved in establishing a diagnosis in this syndrome. In fact, he notes it has been asserted that Laurence and Moon's original patients did not have the Laurence-Moon-Biedl syndrome!

Premature Craniofacial Synostosis—Apert's Syndrome

The clinical features of the various types of premature craniofacial synostoses have been delineated by Bertelsen (1958). I will focus on the complex forms involving the craniofacial structures, including two well-known entities, Apert's syndrome and Crouzon's disease.

Although relatively rare, Apert's syndrome has been well reported. The salient characteristics of the syndrome include premature closure of several cranial sutures with resultant abnormalities in growth and shape of the skull, syndactylism of the hands and feet, and midfacial hypoplasia resulting in exophthalmus and severe reduction in growth of the maxilla in comparison with the mandible, leading to severe malocclusions. In addition, and of particular interest to speech pathologists, patients with this syndrome characteristically have a high-arched or Byzantine-shaped palate which is filled with hyperplastic soft tissue. Some patients show overt clefts, although it is more common to find that a palate which has been described as cleft is actually a pseudo cleft. Recently, surgery has been undertaken with some of these patients to remove the excessive soft tissue from the palate to restore a more normal contour. We do not yet know the effects of such surgery on changes in speech over time.

The only reference to speech in the literature on craniofacial synostoses has been almost accidental. In their article on syndactyly and concomitant

speech disorders, Massengill and associates (1971) discuss four syndactyly patients, one of whom was identified as having Apert's syndrome. This patient was not described in detail, and we have no estimate of the severity of the manifestation of the syndrome except that there was bilateral syndactyly and a cleft palate. This patient was found to have a "mild" articulation disorder and profound retardation of language—actually retardation of auditory receptive vocabulary, as measured on the Peabody Picture Vocabulary Test (Dunn, 1959).

In our clinic population, we have cases of Apert's syndrome ranging in severity from mild (in both physical and speech findings, exhibiting only mild articulatory distortions) to severely affected patients in whom the speech and psychologic or emotional status is complex.

I have selected two patients for discussion who more or less "bracket" the continuum of patients in our clinical experience. One is a 15-year-old male (Figure 4) with a hard palate typical of Apert's syndrome but with good



FIGURE 4. Case 1949. Male, age 15 years, four months, with Apert's syndrome.

velar elevation and no evidence of velopharyngeal inadequacy. The only notable speech findings on his most recent examination consisted of slight distortion of sibilants and affricates. Of necessity, /f/ is produced as a linguallabial and /v/ as a bilabial fricative; /t/ and /d/ are produced with the anterior portion of the tongue blade held against the lower border of the upper incisors. None of these four phonemes is defective auditorily, only visually.

In contrast, a second patient whose physical appearance has grown markedly worse with time (see Figure 5) exhibits multiple consonantal distortions and substitutions and disfluency behaviors which tend to occur in conjunction with those consonants she has difficulty producing. Some of this patient's articulatory behaviors appear to be directly related to her physical morphology, while others appear to reflect faulty learning. Among the latter are consistent substitution of /w/ for /r/, /t/ for /k/, and /d/ for /g/. Although it may be



FIGURE 5. Case 1013. A teen-age patient with Apert's syndrome whose appearance has grown markedly worse with time.

argued that the crowding of the nasopharyngeal airway, typical in Apert's syndrome because of the midfacial hypoplasia, may account for this patient's tendency to use frontal stops for velar consonants, the presence of a "functional" or learning component is substantiated by the fact that she is stimulable for correct production of these consonants in isolated words. Her consistent distortion of all sibilants and affricates, however, could be considered "obligatory"—reflecting the severe Class III malocclusion and unavoidable aberrations in tongue placement for these consonants. While we have no sound-cinefluorographic films of this patient, the interrelation of the skeletal dysplasia, the severe malocclusion, and the aberrant tongue placement is revealed in serial cephalometric x rays. This patient's mental retardation (WISC Full Scale score of 84) is not considered sufficiently severe to account for the articulation pattern.

In summary, it might be appropriate to list those features of Apert's syndrome which could be considered etiological factors in speech and language deficits. These include mental retardation (present in approximately 50% of the population with Apert's syndrome), significant conductive hearing loss, Byzantine-shaped palate which may or may not be cleft, and the maxillary hypoplasia which results in severe Class III malocclusion with open bite and which also causes crowding of the nasopharyngeal airway.

In addition, consideration should be given to the psychological effects (which may in turn affect communication and other functions) of having a gross physical deformity. It is quite possible that the opportunity for intellectual and emotional growth in many of these children is "functionally" limited by such factors as overprotection or rejection by parents, adverse reaction of peers to the facial and limb deformities, motor speech difficulties related to the facial deformity, and difficulties in manipulating objects due to the limb deformities. The extreme example of "functional" limitation is the child who has Apert's

syndrome but is erroneously categorized and treated as a retarded child simply because of his appearance.

The need for thorough, interdisciplinary study of children manifesting Apert's syndrome cannot be overemphasized. We have been remiss not only by our failure to investigate communicative disabilities in these patients but also in not recognizing and studying intellectual and psychosocial factors. For the study of motor speech difficulties, as related to the structural malformations listed previously, helpful information might be gathered through use of air-flow, cinefluorographic, and electromyographic armamentaria in addition to some of the more standard assessment techniques such as cephalometric radiography and articulation testing.

Premature Craniofacial Synostosis—Crouzon's Disease

The craniofacial features in Crouzon's disease are similar to those of Apert's, though generally less severe. In a recent study in our center, Kreiborg and Pruzansky (in press) found that distinction between Apert's and Crouzon's syndromes on the basis of skull shape alone, as seen on lateral cephalometrics, was not definitive. The major clinical distinction is that patients with Crouzon's disease do not show the syndactyly characteristic of Apert's. However, they may be similar to Apert patients with regard to physical traits that might affect speech.

In general, the impression gained from our experience is that, as a group, patients with Crouzon's syndrome are less severely handicapped in physical appearance and function than patients with Apert's syndrome.

Again, however, there is a range of severity. In one family (Figures 6 and 7) in which a mother and three children are affected, the mother is only mildly affected and was unaware of her status until her children became our patients. She is asymptomatic in speech except for slightly distorted sibilant production, and this appears to be related to a missing left lower incisor. The three children



FIGURE 6. Case 3476, age 31. Patient with Crouzon's disease (mother of siblings pictured in Figure 7).

are also mildly affected, although each has had at least two craniectomies. The youngest affected child, aged three (Case 3477), shows a number of oral distortions in his speech which are typical in a three-year-old. However, he does show a curious pattern of consistent nasal emission only on /s/ blends.



FIGURE 7. Cases 3475, male, age seven years (top); 3478, female, age five years, one month (center); and 3477, male, age three years, six months (bottom). Three siblings (offspring of patient in Figure 6) with Crouzon's disease. Note craniectomy scars.

This aberrant pattern appears to represent a learned behavior, although its origin is obscure. This child shows a slight Class II malocclusion and an open bite anteriorly. He has a high-arched palate, but no evidence of velopharyngeal incompetency. The second affected child (Case 3478), aged six, is problem-free in speech with the exception of an extraordinarily low habitual pitch. The results of indirect laryngoscopic examination were negative. The oldest affected child (Case 3475), aged seven, has a Class III malocclusion and a bilateral crossbite. The hard palate is high-arched, but the soft palate elevates well on phonation and there is no evidence of velopharyngeal incompetency. This boy's speech is characterized by oral distortions and substitutions which are infantile in pattern but which do not appear to be directly related to his oral morphology.

At the other end of the continuum (Figure 8), the midfacial hypoplasia has resulted in a severe Class III malocclusion and a necessity to carry the head in an extended position to protect the airway. The hard palate is high, with a narrow vault (Figure 9). There is a bifid uvula, but the soft palate



FIGURE 8. Case 3493. A patient with Crouzon's disease. Note obligatory mouth-open posture at rest.



FIGURE 9. Palate of patient with Crouzon's disease who is pictured in Figure 8. Note hyperplastic soft tissue filling palatal vault.

elevates well on phonation. Both mobility and structure of the tongue appear to be normal. This boy's speech is characterized by consistent oral distortions of the sibilants, fricatives, and affricates and inconsistent distortions of /r/ and /l/. Most of his misarticulations appear to be related to abnormal tongue placement, which is not related to faulty tongue movement but to the abnormal mandible-tongue-maxilla relationship. Again, cinefluorographic evaluation of tongue movement and head-neck posture in these patients, in addition to evaluation of other pertinent variables, could be profitable.

Oral-Facial-Digital Syndrome (OFD—Type I)

In this syndrome, the clinical findings related to the speech mechanism *per se* include multiple buccal frena, multilobulated tongue which is incompletely differentiated from the floor of the mouth, alveolar clefts, tripartite submucous cleft palate or irregular cleft palate, congenital absence of teeth, duplicated maxillary canines, malposition of teeth and enamel hypoplasia, variable median cleft of the upper lip, and short philtrum. We should also note the variable central nervous system findings, which include median defects, cysts, hydrocephalus, and mental retardation.

Our cases exhibit a range of severity. Among the severe cases is a 16 year old (Figures 10, 11, and 12) with significant mental retardation who functions at a level several years below her chronological age. She presents a severe articulation defect, some mild nasal emission, and a deviant voice quality which



FIGURE 10. Case 490. Patient with OFD, Type I.

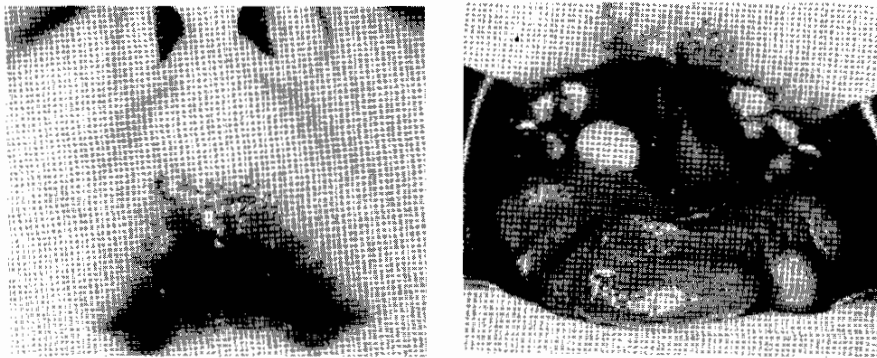


FIGURE 11. Same patient shown in Figure 10. Left, note median defect of upper lip. Right, note multilobulated tongue.

appears to be related to a retracted tongue posture. Intelligibility would be estimated at 70%. Her primary articulation errors are substitution of /k/ for /t/ and /g/ for /d/, gross distortion of affricates, and substitution of pharyngeal fricatives for sibilants. She shows a deviant although auditorily acceptable "shovel-like" production of /l/.

She was born with a cleft palate (Figure 12), which was repaired in conjunction with a superior-based flap when she was eight. Her hard palate is



FIGURE 12. Soft palate of same patient shown in Figures 10 and 11. Note irregular cleft.

markedly high-arched and narrow, with a fistula anterior to the juncture of the hard and soft palates. The lateral ports appear quite wide, and no movement of the palate or posterior or lateral pharyngeal walls is evident on phonation. There is mild hypernasality, although the primary handicap is the articulation.

The speech picture in this patient is complex and warrants thorough and sophisticated investigation. Again, crucial information could be obtained from cinefluorographic and electromyographic data. In addition, spectrographic analysis should prove useful in determining the acoustic features that lead us to apply the nebulous adjective *muffled* to the voice quality of many of the children with OFD, as well as many of those with mandibulofacial dysostosis and hemifacial microsomia.

THREE CRITICAL NEEDS

Bloomer (1971) frequently restates the theme cited earlier:

If the examination of the orofacial complex is pursued independently and without reference to a general examination of the patient's communication abilities, the validity of the findings will be open to serious question. . . . the etiologic significance of orofacial abnormality in an individual instance of defective speech must be interpreted with due regard to the total dynamics of speech and the multiple causes of speech disorders.

This is one critical need we must bear in mind as we study communicative disabilities in patients with craniofacial malformations—the need to study the

entire communicative picture, not just isolated aspects such as velopharyngeal closure or malocclusion. While we look forward to the day when cinefluorographic, electromyographic, and other types of data will be gathered on these patients, we cannot amputate the motor speech mechanism from the person. The novelty of accumulating physiologic data on these patients could lead us to forget Bloomer's warning.

This brings us to a second critical need—to give full consideration to psychological factors when we are evaluating communication abilities. This need takes on particular importance when rehabilitative procedures (either therapy or physical management) are being considered for a patient. All too often we assume that the patient sees himself precisely as we see him. Most of us have had the experience of eagerly embarking on a treatment program for a patient, only to be brought up short when we realize we have failed to ask the patient what he thinks.

Consideration of rehabilitative procedures brings us to the third critical need—to carefully document response to specific treatments, whether the treatments are physical or behavioral. This is particularly crucial with respect to the surgical procedures devised by Tessier, which represent remarkable experimental situations for relating form to function.

There can be little doubt that craniofacial malformation syndromes have constituted an investigative backwater in both speech pathology and the dental sciences. With the startling advances being made in surgical rehabilitation of such severe malformations as those found in the premature cranial synostoses (Converse and Pruzansky, in press), we realize how late we are in answering even the most basic questions. How does tongue posture at rest and in function in patients with mandibulofacial dysostosis, or Pierre Robin, Crouzon's, or Apert's syndromes differ from that of the normal population? To what extent is the deviation in tongue posture a function of the degree of bowing of the mandible in MFD? How does the deviant tongue posture affect articulation and voice quality? How do the variations in tongue and other supralaryngeal musculature in these and other patients interrelate with muscle function at the laryngeal level? How may we quantify the effects of unilateral anomalies such as those of the mandible and tongue in hemifacial microsomia? In Apert's and Crouzon's syndromes, how may we quantify and study the effects of the dental and occlusal anomalies, the crowded nasopharyngeal airway, and the uniquely shaped palate? These are only a few of the questions that come to mind. And while we are deciding what types of physiologic and behavioral data are needed, we must remember that it is not just the unanswered questions that await our belated attention, but the patients themselves.

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GENETIC CONSIDERATIONS IN HEARING DISORDERS

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The information necessary for continuity of a species, for orderly development, and for physiological homeostasis of the individual is carried on the chromosomes in the nuclei of cells and transmitted in the male and female gametes.

Chromosomes are thread-like or rod-like in form, depending on the stage of the cell in the division cycle; and in man, as in most mammals, they are paired, so that there is an even chromosome number in each cell. In normal individuals the number is 46, the diploid number.

In the process of formation of spermatozoa and ova from the diploid germ cells only one of each pair of chromosomes passes into each gamete, so that the chromosome number is 23, the haploid number. Finally, when an ovum is fertilized by a spermatozoon, the haploid nuclei of the two gametes fuse to form the single diploid nucleus of the zygote, the first somatic cell of a new generation. Subsequent cell divisions of the zygote (mitosis) result in diploid daughter cells, which, by repeated divisions and differentiation, form the new individual.

Conventionally, 22 of the chromosome pairs are termed autosomes and are the same in the two sexes. The twenty-third pair in the female consists of two medium-sized chromosomes, the X chromosomes. In the male the pair consists of one such X chromosome and a smaller one called the Y chromosome.

Any cell whose chromosome number is a simple multiple of the haploid number 23 is termed euploid. Any cell whose chromosome number is not a simple multiple of 23 is termed aneuploid. Thus, 23 chromosome gametes are euploid, as are the cells of normal individuals having 46 chromosomes. Those with 47 chromosomes, as in the trisomy type of Down's syndrome, or 45 chromosomes, as in many cases of Turner's syndrome, are aneuploid.

Genes

The traditional concept of genes is that they are ultimate particles of inheritance which are arranged linearly on chromosomes and occupy specific

positions or loci. It follows that each gene locus has its homologue in the same position on the other chromosome of the pair. Alternative forms of genes which can occupy the same gene locus (or pair of loci) are termed alleles.

The gene was conceived of as the ultimate unit of heredity in that it was the smallest unit which could exert any individual function. It was the smallest unit at which mutation could occur, and crossing over in meiosis (exchange in genetic material between chromatids from the opposite chromosomes of a pair) could occur between genes but not within genes. With the new developments in bacterial genetics and a better understanding of the chemical nature of the hereditary material, there is ample evidence that a finite length of the double-stranded, spirally arranged deoxyribose nucleic acid molecule (DNA) does not correspond to a gene in the traditional sense. Thus, a length of DNA may ultimately be responsible for specification of a polypeptide chain or a protein, but changes or mutations at any point in that length determine specification of a different amino acid in the chain. In other words, mutation can take place within what would correspond to a functional gene. Again, crossing over with exchange of material can take place within such lengths of DNA strands in chromatids, that is, within a functional gene.

Each individual results from a zygote formed by the coming together of a paternal and a maternal haploid gamete. He receives one of each chromosome pair from each parent. Therefore, he receives two genes which occupy a given pair of gene loci.

For example, suppose that on a particular pair of autosomes at a particular locus, the father has genes A and B, and the mother, genes C and D. Chromosomes of a pair pass into gametes at random (1) in respect of the paternal or maternal origin of the chromosome, and (2) relative to all the other chromosomes. It follows that, in the given example, on the average one-half of the paternal gametes carry gene A (or allele A) and one half allele B. Similarly, half of the maternal ova carry allele C and half allele D. Marriages of such individuals with alleles A and B, and C and D, will result in equal frequencies of offspring who carry A and C, A and D, B and C, and B and D alleles (Figure 1).

Thus, segregations of gene combinations at loci follow from the segregations of the chromosomes on which the loci are situated.

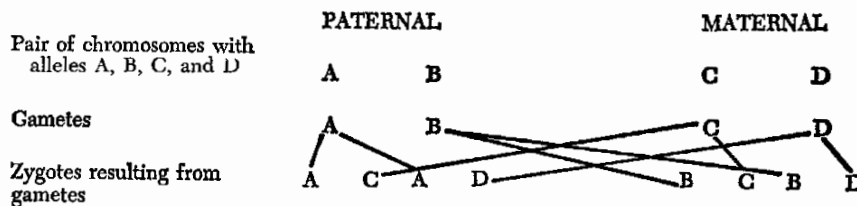


FIGURE 1. Segregation of allelic genes on a pair of chromosomes.

Genotype and Phenotype

The term *genotype* is used to indicate the genetic makeup of an individual. It is used in a wider sense to indicate the overall genetic constitution of the individual. The term *phenotype* is used to describe the recognizable effects of the genetic makeup either in the limited or more general sense.

Homozygosity and Heterozygosity

When an individual has identical alleles at a given locus, he is termed homozygous, or the homozygote, or as having a homozygote genotype. Per contra, if the two alleles in an individual are not identical, the individual is described as heterozygous or the heterozygote.

Dominance and Recessiveness

In the strict sense, the terms *dominant* and *recessive* apply to traits determined by genes. Thus, traits determined in heterozygotes for a mutated gene and the normal allele, where the individual need only receive a gene from one parent, are termed *dominant*. It follows that the trait is transmitted from generation to generation and, except for those individuals who are the first recipients of mutations arising in parental germ cells, if the transmission is regular and every heterozygote shows its effects, each affected person will show a detectably affected parent. If only one parent is affected, on the average, half of the offspring will be affected.

The original definition of a dominant trait was one where the characteristics of the heterozygote and the homozygote for the abnormal or mutated gene were identical. In man, homozygotes for mutations with harmful manifestations in the heterozygote state are extremely rare. In the few cases that have been reported, the homozygote is much more severe and probably dies in utero.

Recessive genetic traits are those expressed only in homozygotes and there should be no manifestation of the heterozygote genotype. In some situations (for example, most cases of inborn errors of metabolism), the heterozygote is frequently recognizable with the sophisticated biochemical and other techniques.

As a matter of convenience in medical genetics, it is customary to describe those traits usually noticed in heterozygotes as dominants, and those commonly seen in homozygotes are recessive.

The terms *dominance* and *recessiveness* apply to traits and not to genes or mutations. This is the convention of tradition and is the logical usage. However, it is often convenient and acceptable to apply the terms to the genes that cause these traits. Figures 2 and 3 demonstrate idealized pedigrees for autosomal dominant and autosomal recessive traits.

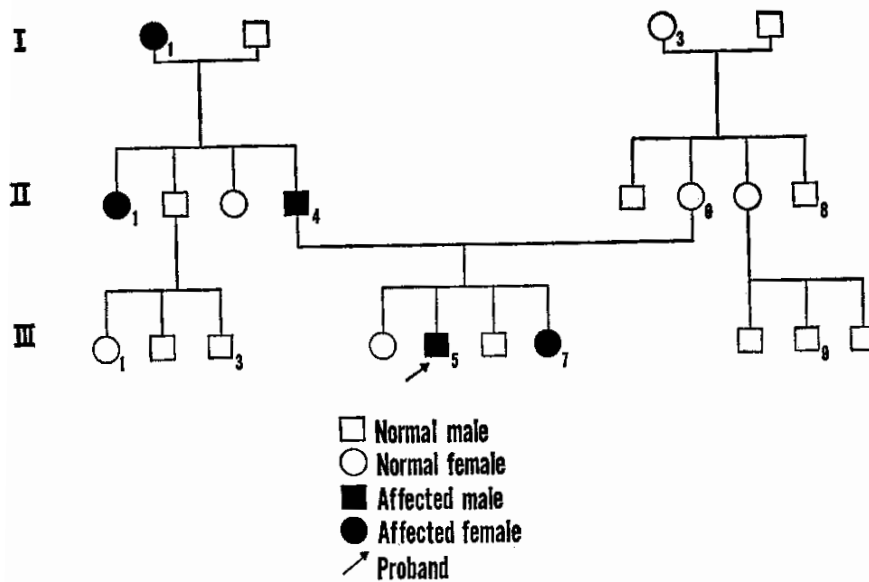


FIGURE 2. An idealized pedigree for an autosomal dominant trait.

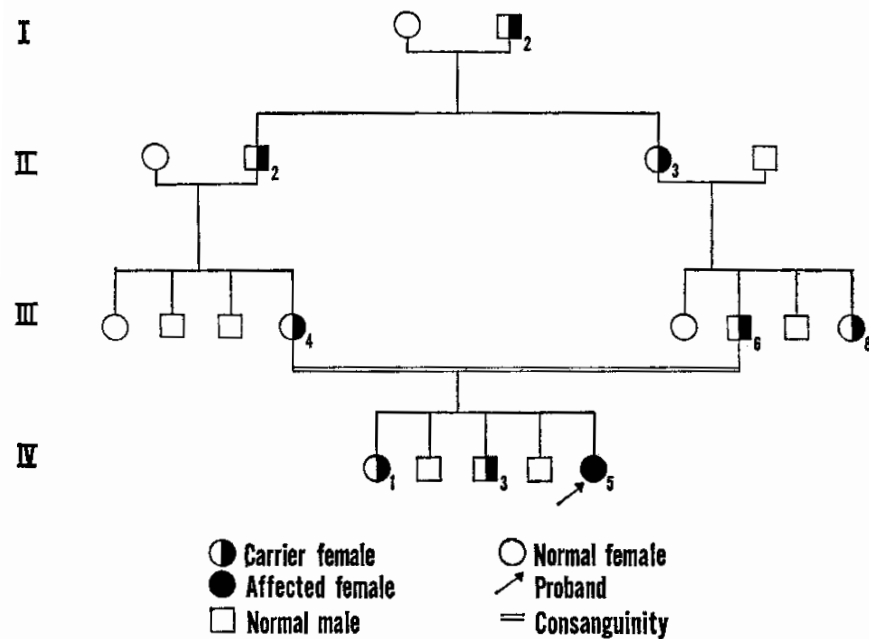


FIGURE 3. An idealized pedigree for an autosomal recessive trait.

The criteria for autosomal dominant inheritance are these:

1. Males and females are affected in approximately equal proportion.
2. Approximately 50% of the offspring of an affected parent are affected.
3. Male to male transmission is the criterion for diagnosis of this type of inheritance.
4. Every affected person has one affected parent, except for fresh cases in which none of the parents or the sibs is affected.
5. For an affected parent, the risk of having an affected offspring is 50% in every pregnancy.

The criteria for autosomal recessive inheritance are these:

1. Males and females are affected in approximately equal proportion.
2. Approximately 25% of the offspring are affected.
3. The parents are phenotypically normal.
4. A history of consanguinity is frequent.
5. For parents who have had one affected offspring, the risk of recurrence is 25% in every pregnancy.

X-Linked Inheritance

Like autosomal traits, those determined by genes on the X chromosome may be either dominant or recessive. The female with two X chromosomes may be either heterozygous or homozygous for a given mutant gene. The trait in the female can demonstrate either recessive or dominant behavior. But the male with one X chromosome can have only one genetic constitution, namely hemizygous; and regardless of the behavior of the gene in the female, whether recessive or dominant, it is always expressed in the male.

The critical characteristic of X-linked traits, both dominant and recessive, is the absence of male-to-male (father-to-son) transmission. This is an important point, as the X chromosome in the male is transmitted to none of his sons and to all of his daughters. An idealized X-linked recessive pedigree is shown in Figure 4.

There are two criteria for sex-linked inheritance:

1. In sex-linked recessive traits, the carrier female does not show any obvious manifestations of the trait. On the average, 50% of her male offspring are affected and 50% of her female offspring are carriers.
2. In sex-linked dominant traits, the carrier female shows evidence of the trait, and the condition may be lethal in the male. The risk of recurrence is the same as for sex-linked recessive traits. The carrier females show obvious manifestations of the trait.

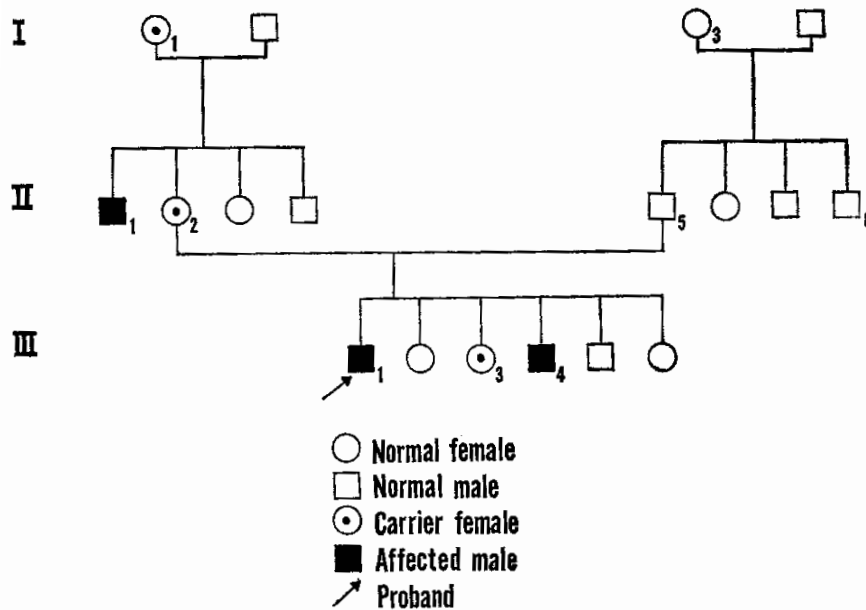


FIGURE 4. An idealized pedigree for an X-linked recessive trait.

Multifactorial

These traits are determined by many individually small effects of genes and environmental influences. Criteria for a multifactorial/threshold model are:

1. The genes contributing to multifactorial predisposition are assumed to be additive. A threshold point exists in the developmental process, and beyond the threshold, normal development is impossible.
2. An affected offspring identifies the parents as jointly contributing sufficient predisposition to their offspring for one to be affected.
3. The frequency in subsequent children appears to rise after recessive affected children have been born. Similarly, if a second child has been affected, the matings have a higher risk than in the general population.
4. Prevalence in close relatives of the index case is higher than in the general population (that is, in first-degree relatives of an index case, the frequency may be between three and 15 times that in the general population).
5. In relatives of lesser degree, the frequencies fall off rapidly.

Figure 5 shows an idealized pedigree of a case of cleft palate as an example of a multifactorial trait.

With this background in elementary genetics, let us now consider the genetics of hearing disorders.

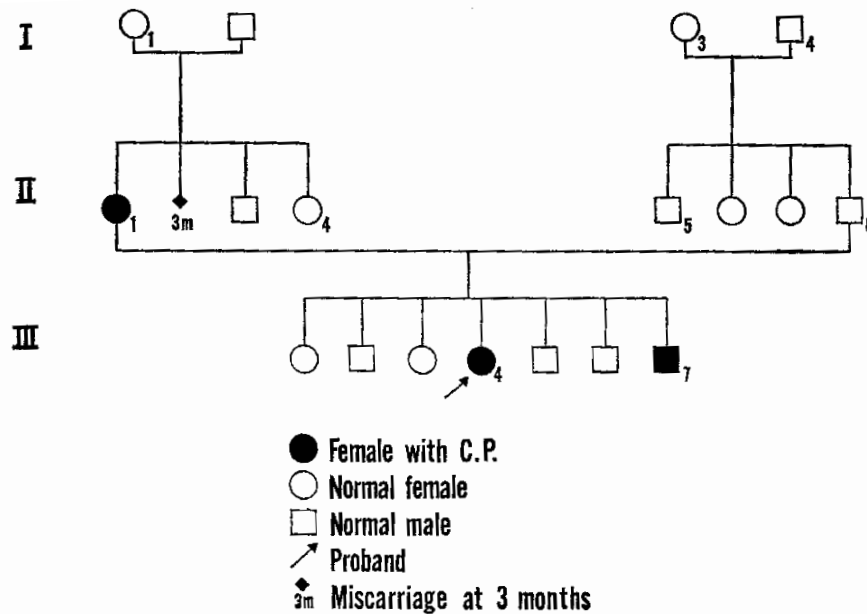


FIGURE 5. An idealized pedigree for a multifactorial trait.

HEREDITARY HEARING DISORDERS

Hereditary hearing disorders may appear in many forms. Being a geneticist, I have found it easier to classify the different forms according to the mode of inheritance. Transmission may be dominant, as in Waardenberg's disease; it may be recessive, as in Pendred's disease; or it may be sex-linked, as in congenital, sex-linked deafness of early onset. There are, however, other important factors of differentiation that have to be considered, such as whether the hearing loss is conductive, as in mandibulofacial dysostosis; neural, in Alport's disease; or mixed, as in Crouzon's disease. The time of onset may be congenital, as in Waardenberg's disease; in the teens, as in Alport's disease; or in adult life, as in otosclerosis. The frequency affected is another point of differentiation.

Although there are several kinds of hereditary deafness with no associated defects, in the majority the specific genetic defect results in abnormalities in other systems as well. The structures that are most frequently involved in the different types of familial hearing loss are the external ears, integumentary system, nervous system, skeletal system, urinary system, and endocrine system.

The known types of hereditary deafness in man are listed in the Appendix. As mentioned previously the different types are classified according to the mode of inheritance.

ACKNOWLEDGMENT

This work was supported in part by grant number PHS-DE 02872 from the National Institutes of Health and the Maternal and Child Health Services, Department of Health, Education, and Welfare.

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APPENDIXES

A. Hereditary Hearing Loss Transmitted by an Autosomal Dominant Gene

Type	Onset	Severity	Neural Loss	Conductive Loss	Vestibular Findings	Associated Features	Ref. No.*
Dominant Congenital Severe Deafness	Congenital	Severe	+	-	Normal	-	1
Dominant Progressive Nerve Deafness	Childhood	Mild to severe	High-frequency progressing to all frequencies	-	Normal	-	2
Dominant Unilateral Deafness	Congenital	Severe	+	-	Normal	-	3
Dominant Low-Frequency Hearing Loss	Childhood	Mild to severe	+	-	Normal	-	4
Dominant Midfrequency Hearing Loss	Childhood	Mild to severe	+	-	Normal	-	5
Otosclerosis	Adult	Mild to moderate	-	+	Normal	-	6

*Refer to numbered reference list preceding these appendices for literature relating to the type of hearing loss described.

Type	Onset	Severity	Neural Loss	Conductive Loss	Vestibular Findings	Associated Features	Ref. No.
Dominant Preauricular Pits and Neural Hearing Loss	1st-2nd decade	Mild to severe	+	-	?	Preauricular pits, uni- or bilateral; branchial fistula, uni- or bilateral	7
Dominant Thick Ears and Incudostapedial Joint Abnormality	Congenital	Moderate	-	+	?	-	8
Ear Malformation and Conductive Hearing Loss	Congenital	Mild to moderate	-	+	?	Preauricular pits, uni- or bilateral; deformed auricles, uni- or bilateral	9
Familial Hearing Loss with Branchial Fistulas	Congenital	Mild to moderate	+	+	?	Cervical and preauricular branchial fistulae; malformed auricles and ossicles; hypoplastic mandible	10
Dominant Otofacio-cervical Abnormalities	Congenital	Moderate	-	+	?	Midface hypoplasia; preauricular pits and fistulae; hypoplasia of cervical muscles	11
Dominant Albinism and Congenital Deafness	Congenital	Severe	+	-	?	Scanty eyebrows; optic fundi and irides normal	12
Leopard Syndrome	Congenital	Moderate to severe	+	-	Normal	Pulmonary stenosis; growth and mental retardation; lentiginosis; ocular hypertelorism; abnormal genitalia	13

Dominant Anhidrosis and Progressive Hearing Loss	Adult	Mild to moderate	+	-	?	Anhidrosis	14
Dominant Keratopathy-derma, Digital Constrictions, and Deafness	Congenital	Severe	+	-	?	Hyperkeratosis of palms, soles, and pressure areas; constrictions of fingers and toes	15
Dominant Knuckle Pads, Leukonychia, and Hearing Loss	Congenital	Moderate to severe	+	+	+	Disorganized ear bones	16
Dominant Onychodysplasia, Coniform Teeth, and Hearing Loss	Congenital	Moderate to severe	+	-	?	Partial anodontia; elevated sweat electrolyte concentration	17
Dominant Saddle Nose, Myopia, Cataract, and Hearing Loss	Childhood	Moderate	+	-	?	Congenital or juvenile cataracts	18
Dominant Myopia, Hearing Loss, Peripheral Neuropathy, and Skeletal Abnormalities	1st or 2nd decade	Mild to moderate	+	-	?	Myopia, cataracts, retinitis pigmentosa, peripheral neuropathy with shooting pains; kyphoscoliosis; osteoporosis	19
Dominant Acoustic Neuromas	2nd or 3rd decade	Moderate to severe	+	-	Hypo-active	Bilateral acoustic neuromas	20
Dominant Sensory Radicular Neuropathy	2nd or 3rd decade	Moderate to severe	+	-	?	Dorsal root ganglion with shooting pains	21

Type	Onset	Severity	Neural Loss	Conduc-tive Loss	Vestibu-lar Findings	Associated Features	Ref. No.
Dominant Photomyoclonus, Hearing Loss, Diabetes, and Nephropathy	3rd decade	Moderate	+	-	?	Neuronal loss and gliosis of cerebral and cerebellar cortex	22
Dominant Proximal Symphalangism and Hearing Loss	Congenital	Mild to moderate	-	+	?	Lateral digits more affected	23
Crouzon's Disease	Congenital	Mild to moderate	+	+	?	Craniosenosis; exophthalmos; hypoplastic mandible; beaked nose; external canal atresia	24
Mandibulofacial Dysostosis	Congenital	Moderate to severe	-	+	?	Antimongoloid slant of palpebral fissures; midface hypoplasia; malformed ossicles; lower lid colobomas	25
Osteitis Deformans (Paget's Disease)	Adult	Mild to moderate	+	-	?	Narrowing of cranial nerve foramina and inner ear involvement	26
Cranio-metaphyseal Dysplasia (Pyle's Disease)	Childhood	Mild to moderate	+	+	?	Metaphyseal dysplasia of long bones and skull; other cranial nerves may be involved	27
Osteogenesis Imperfecta	3rd decade	Mild to moderate	-	+	?	Blue sclerae; frequent fractures	28

Alport's Disease	2nd decade	Mild to moderate	+	-	Normal	Nephritis, more severe in males; Ocular abnormalities	29 30
	2nd decade	Mild to moderate	+	-	?	Recurrent urticaria, amyloidosis with nephropathy and uremia	31
Dominant Mitral Insufficiency, Joint Fusion, and Hearing Loss	Congenital	Moderate to severe	-	+	?	Fusion of cervical vertebrae, carpal and tarsal bones	32
	Congenital	Mild to severe	+	-	Abnormal organ of corti; absent coils	Lateral displacement of medial canthi; prominent root of nose; Hyperplasia of medial third of brows; heterochromia iridium; white forelock; skin pigmentary changes	33
Stickler's Syndrome	Childhood	Mild to moderate	+	-	?	Myopia, retinal degeneration; arthritis, muscle pain; Marfanoid habitus; cleft palate	34 35
	Adult	Moderate	?	?	?	Abnormal calcium metabolism; corneal degeneration	36
Dominant Nephritis, Ichthyosis, Hearing Loss, and Prolinuria	Childhood	Mild to moderate	+	-	?	Renal cyst; variable degree of nephritis	37

Type	Onset	Severity	Neural Loss	Conduc- tive Loss	Vestibu- lar Findings	Associated Features	Ref. No.
Turner Phenotype Syndrome	Congenital	Moderate to severe	+	-	?	Short stature; mental retardation; low-set ears; webbed neck; low hairline; vertebral anomalies; congenital heart disease; abnormal genitalia	38
Albers-Schonberg Disease (Benign Osteopetrosis)	Adult	Mild to moderate	+	-	?	Narrowing of cranial nerve foramina	39

B. Hereditary Hearing Loss Transmitted by an Autosomal Recessive Gene

Type	Onset	Severity	Neural Loss	Conduc- tive Loss	Vestibu- lar Findings	Associated Features	Ref. No.
Recessive Congenital Severe Deafness	Congenital	Severe	+	-	Normal	More than one genotype	40
Recessive Early-Onset Neural Deafness	Infancy to childhood	Severe	+	-	Normal	-	41
Recessive Congenital Moderate Hearing Loss	Congenital	Moderate	+	-	Normal	-	42
Recessive Malformed Low-Set Ears and Conductive Hearing Loss	Congenital	Mild to severe	-	+	Normal	Unilateral or bilateral	43

Oculoauriculovertebral Dysplasia	Congenital	Moderate to severe	-	+	Normal	44	Epibulbar dermoid; coloboma of upper lids; preauricular skin tags; macrostomia; vertebral anomalies
Hereditary Piebaldness and Congenital Deafness (? Sex-Linked)	Congenital	Severe	+	-	Normal	45	Depigmentation of head and arms; hyperpigmented spots
Recessive Atopic Dermatitis and Neural Hearing Loss	Congenital	Moderate	+	-	Normal	46	Atypical atopic dermatitis, onset at age 10
Congenital Pili Torti and Hearing Loss	Congenital	Moderate to severe	+	-	?	47	Brittle and fragile hair
Recessive Onychodys- trophy and Deafness	Congenital	Severe	+	-	Hypoac- tive	48	Strabismus
Recessive Myopia and Hearing Loss	Congenital	Moderate to severe	+	-	Normal	49	Low intelligence
Recessive Retinitis Pigmentosa and Congenital Deafness	Congenital	Moderate to severe	+	-	Normal	50	Progressive visual loss
Refsum's Disease	1st or 2nd decade	Mild to severe	+	-	Normal	51	Progressive retinitis pigmentosa; hyper- trophic peripheral neuropathy; cerebellar ataxia; nystagmus; plasma phytanic acid

Type	Onset	Severity	Neural Loss	Conduc- tive Loss	Vestibu- lar Findings	Associated Features	Ref. No.
Alstrom's Disease	2nd decade	Moderate	+	-	?	Retinitis pigmentosa; early-onset diabetes mellitus; moderate obesity; cataract	52
Recessive Retinal Changes, Deafness, Muscular Wasting, and Mental Retardation	Childhood	Moderate to severe	?	?	?	Retinal detachment and telangiectasia	53
Recessive Optic Atrophy, Hearing Loss, and Juvenile Diabetes	Childhood	Moderate	+	-	Hypoaac- tive	Childhood onset of optic atrophy; early on- set of diabetes	54
Familial Hearing Loss, Polyneuropathy, and Optic Atrophy (? Sex- Linked Inheritance)	Infancy	Severe	+	-	?	Childhood onset of peripheral neuropathy; later optic atrophy and visual loss	55
Recessive Deafness, Mental Retardation, Ataxia, and Hypogonadism	Infancy	Moderate to severe	+	-	?	Truncal ataxia and muscle wasting starting in childhood; decreased estrogen and 17 ketosteroids	56
Oral-Facial-Digital Syndrome II	Congenital	Mild to moderate	-	+	?	Facial deformities; digital abnormalities; lobulated tongue	57

Otopalatodigital Syndrome	Congenital	Mild to moderate	-	+	?	Hypertelorism; frontal bossing; broad nasal root; dwarfism; wide-spaced and short 1st digits	58
Recessive Absence of Tibia and Deafness	Congenital	Severe	?	?	?	Absence of tibia, may be unilateral or bilateral; malformed fibulas	59
Recessive Split Hand and Foot Syndrome	Congenital	Moderate to severe	+	-	Hypoa-	Absent phalanges and syndactyly	60
Progressive Diaphyseal Dysplasia	2nd decade	Moderate	+	-	?	Thickened diaphyseal cortex of limb bones; skull thickening and cranial nerve palsies	61
Osteopetrosis	Childhood	Mild to moderate	+	+	?	Sclerotic and brittle bones; hepatosplenomegaly and anemia	62
Hyperostosis Corticalis Generalisata	Adult	Mild to moderate	+	-	?	Osteosclerotic overgrowth of skeleton	63 64
Recessive Renal, Genital, and Middle Ear Abnormalities	Congenital	Severe	+	-	?	Renal agenesis (unilateral or bilateral); vaginal atresia; malformed incus	65
Pendred's Disease	Congenital	Moderate to severe	+	-	De-	Goiter develops in adolescence	66
Recessive Goiter, Stippled Epiphyses, and Deafness	Congenital	Moderate to severe	+	-	?	Elevated PBI	67

Type	Onset	Severity	Neural Loss	Conduc- tive Loss	Vestibu- lar Findings	Associated Features	Ref. No.
Jervell and Lange- Nielsen Disease	Congenital	Severe	+	-	Atrophy of the organ of corti; atrophy and fibrosis of stria vascu- laris; loss of spiral ganglion cells	Prolonged QT intervals; recurrent Stokes-Adams attacks, with sudden death	68
							69
Cockayne's Syndrome	Infancy	Moderate	+	-	P	Severe mental retarda- tion; dwarfism; cataracts; unsteady gait	70
Laurence-Moon-Biedl Syndrome	Childhood	Moderate	+	-	P	Polydactyly-syndactyly; retinitis pigmentosa; mental retardation; obesity; genital hypoplasia	71
Fraser's Syndrome	Congenital	Moderate	-	P	+	Auricular defect; ocular abnormalities; abnormal genitalia	72
Recessive Congenital Deafness, Ataxia, and Retinitis Pigmentosa	Congenital	Severe	+	-	Abnor- mal response	Vestibular ataxia; mental deficiency and psychosis	73

Recessive Retinitis Pigmentosa, Spastic Diplegia, Skeletal Abnormalities, and Hearing Loss	Congenital	Moderate to severe	?	?	-	74	Microcephaly; spastic paraplegia; brachydactyly
Hurler's Disease (MPS I)	Infancy	Mild to moderate	+	-	?	75	Mental retardation; corneal opacity; skeletal deformities; increased dermatan and heparan sulfate in urine
Sanfilippo Disease (MPS III)	Childhood	Mild to moderate	+	-	?	76	Moderate mental retardation; skeletal deformities; increased heparan sulfate in urine
Scheie's Disease (MPS V)	Adult	Mild to moderate	+	-	?	77	Corneal opacities; mild bony changes; increased dermatan sulfate in urine
Maroteaux-Lamy Disease (MPS VI)	Childhood	Mild to moderate	+	-	Hypocative	78	Marked corneal clouding; moderate bone changes; increased dermatan sulfate in urine
Recessive Renal Acidosis and Progressive Hearing Loss	2nd decade	Moderate	+	-	Normal	79	Progressive renal disease with calculi and acidosis
Metaphyseal Dysostosis, Conductive Hearing Loss, and Mental Retardation	Childhood	Moderate to severe	-	+	?	80	Metaphyseal dysostosis; dwarfism (short-limbed); mild mental retardation; short, stubby digits
Opticocochleodentate Degeneration	Childhood	Moderate to severe	+	-	?	81	Progressive spastic quadriplegia; optic atrophy and dementia

C. Hereditary Hearing Loss Transmitted by a Sex-Linked Gene

<i>Types</i>	<i>Onset</i>	<i>Severity</i>	<i>Neural Loss</i>	<i>Conduc- tive Loss</i>	<i>Vestibu- lar Findings</i>	<i>Associated Features</i>	<i>Ref. No.</i>
Sex-Linked Congenital Neural Deafness	Congenital	Severe	+	-	-	-	82
Sex-Linked Early-Onset Neural Deafness	Congenital	Moderate to severe	+	-	-	-	83
Sex-Linked Moderate Hearing Loss	Childhood	Mild to moderate	+	-	-	-	84
Sex-Linked Pigmentary Abnormalities and Congenital Deafness	Congenital	Severe	+	-	De- pressed function	Skin normal at birth; spots of hypo- and hy- perpigmentation appear in childhood	85
Familial Hearing Loss, Polyneuropathy, and Optic Atrophy	Infancy	Severe	+	-	?	Visual problems appear during second decade ^c	55
Hunter's Disease (MPS II)	Childhood	Mild to Moderate	+	-	?	Mental retardation; dwarfism; hepatosplenomegaly; increased dermatan and heparan sulfate in urine	86
Hereditary Piebaldness and Congenital Deafness	Congenital	Severe	+	-	Normal	Depigmentation of head and arms and hyper- pigmented spots	45

X-linked Mixed Deafness with Congenital Fixation of the Stapedial Footplate and Perilymphatic Gusher

Congenital Moderate to severe + + P - 87

D. Hereditary Hearing Loss Syndromes Associated with Chromosomal Abnormality

Type	Onset	Severity	Neural Loss	Conduc- tive Loss	Vestibu- lar Findings	Associated Features	Ref. No.
Trisomy D Syndrome	Congenital	P Severe	+	-	Abnormal organ of corti	Multiple system involvement; trisomy of Group D (13-15) chromosomes; rarely, translocation of chromosome 13-15 group to another chromosome	88
XO Turner's Syndrome	Congenital	Moderate to severe	+ -	+ -	Normal	Short stature; short, webbed neck; low posterior hairline; shield chest; wide-set nipples; congenital heart disease; cubitus valgus	89

E. Hereditary Hearing Loss, Transmission Not Yet Defined

Type	Onset	Severity	Neural Loss	Conduc- tive Loss	Vestibu- lar Findings	Associated Features	Ref. No.
McCune-Albright Syndrome	Childhood	Moderate to severe	+	-	Normal	Fibrous dysplasia of long bones, pelvic bones, and facial bones; irregular skin pigmentation; sexual precocity; cranial nerve compression	90
Klippel-Feil Syndrome	Congenital	Moderate to severe	+	+	?	Short neck; fused cervical vertebrae; low posterior hairline; ossicle abnormalities	91
Congenital Aplasia of the Alae Nasi, Deafness, Hypothyroidism, Dwarfism, and Absent Teeth	Congenital	Moderate	+	-	?	Nasal abnormalities; anodontia; hypothyroidism; dwarfism; low birth weight; mental retardation; ectodermal scalp defect	92

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