A Proposed New Classification of Craniofacial Anomalies

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for
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Committee on Nomenclature and Classification of Craniofacial Anomalies

Introduction
In the fall of 1976, Kenneth R. Bzoch, President of the American Cleft Palate Association, requested the formation of an Ad Hoc Committee for Reclassification of Craniofacial Anomalies. The impetus for this was the rapid growth of new knowledge and understanding about the malformations that had occurred as a result of the development of craniofacial surgery during the previous few years. The original committee consisted of the following people: Linton A. Whitaker, M.D., Chairman; Howard Aduss, D.D.S.; Asa Berlin, Ph.D.; Mutaz Habal, M.D.; and Joseph Reichman, M.D. Each of these people helped in the early formulation of ideas about the new classification. Subsequently, Malcolm Johnson, D.D.S., and Hermine Pashayan, M.D. were added to the committee. The paper, as finally developed and edited, was written by Drs. Whitaker, Reichman, and Pashayan and was completed in the spring of 1980. Since it was thought to be desirable to present the information at the International Cleft Palate Meeting in Acapulco in May, 1981, publication has been withheld until following that meeting.

KEY WORDS: craniofacial anomalies, classification of anomalies, malformations

Malformations of the craniofacial complex abound with great variation. Previously, facial malformations were usually categorized by two means: 1) pathogenic mechanisms and 2) anatomic conditions. These two areas were often mutually exclusive. The confusion in nomenclature arises because the names of syndromes (i.e., Crouzon’s, Treacher-Collins, etc.) do not fulfill the criteria necessary for either the basic scientists or the clinician. They arose serendipitously or by isolated observations with no effort to fit them into a system as a whole. Because of such occurrences, there is now the need for a comprehensive system that will allow the classification of new syndrome entities without the use of eponyms. In formulating a new system, it is desirable to satisfy both the basic scientist and the clinician.

The basic scientist desires an embryologic or biochemical classification, while the clinician requires simplicity with clinical applicability. Such a system should be broadly based both etiologically and anatomically and should, ideally, combine treatment goals and principles.

A review of some of the more widely used classification systems is given below. Anomalies of the face have most often been classified according to a major structure involved, i.e. lip and palate, eye, nose, mandibular, and maxillary abnormalities.

Pathogenetic Classification

I. Lip and Palate

Of all facial deformities, classification of cleft lip and palate has received the most attention. Superb reviews of classification systems have been done by Millard (1976) and Berlin (1971). While the earlier classification systems of Davis and Ritchie (1922) and Veau
(1931) were based upon "operative" (anatomic) findings, recent advances in understanding the embryology of cleft lip and palate laid the foundations of a more complete classification. These advances evolved into the present systems:

A. **International Classification**: Classification of the lip, alveolus, and palate (based on embryological principles) (Berlin, 1971):

1. **Group 1**: Clefts of the anterior (primary) palate
   - a. Lip: right and/or left
   - b. Alveolus: right and/or left
2. **Group 2**: Clefts of the anterior and posterior (primary and secondary) palates:
   - a. Lip: right and/or left
   - b. Alveolus: right and/or left
   - c. Hard palate: right and/or left
3. **Group 3**: Clefts of the posterior (secondary) palate
   - a. Hard palate: right and/or left
   - b. Soft palate: median

(For further subdivisions, the terms "total" and "partial" should be used.)

**Rare Facial Clefts Based Upon Topographical Findings**

- a. Median clefts of the upper lip with or without hypoplasia or aplasia of premaxilla
- b. Oblique clefts (oro-orbital)
- c. Transverse clefts (oro-auricular)
- d. Clefts of the lower lip, nose, and other very rare clefts.

B. **Kernahan and Stark (1955)**: Classification using the incisive foramen as a boundary marker:

1. Clefts of the primary palate: lip and premaxilla
2. Clefts of the secondary palate: hard and soft palate posterior to incisive foramen.

Further description as to left, right, complete, and incomplete are added.

C. **American Cleft Palate Association** (Haskins et al., 1962):

1. Prepalate: unilateral, bilateral, and median lip clefts; also alveolar clefts
2. Palate: all palatal clefts to incisive foramen
3. Prepalate and palate: unilateral and bilateral complete clefts; incomplete clefts of lip and palate
4. Rare clefts other than prepalatal or palatal types.

In the International Classification, several problems are apparent:

1. Numbering of groups may cause confusion with the classifications of Veau and Davis and Ritchie. (1972)
2. The term "anterior palate" is used rather than "primary palate" (Kernahan and Stark, 1955) or prepalate (ACPA) (1968)
3. Median clefts are listed under facial clefts rather than clefts of the lip.
4. The lip is not truly an embryologic part of the "primary" palate.

It seems that the classifications of Kernahan and Stark (1955) and of the American Cleft Palate Association (Haskins et al., 1962) are the best. Both are relatively simple and incorporate three major areas:

1. Clefts anterior to the incisive foramen
2. Clefts posterior to the incisive foramen
3. Combinations of both types of clefts

While there is no agreement as to which system is best, a more visual mode of categorization was presented by Kernahan (1973) in the form of a "Y." This was modified by Elsahy (1973) and Millard (1976):

- a) Cleft: area stippled
- b) Submucosal muscle and bony cleft: horizontal lines
- c) Nasal deformity marked with horizontal lines proportionate to severity

**II. Face and Skull**

"The most extensive overall compilation of facial malformation with regard to pathophysiology is that proposed by Pruzansky (1975):

1. Sociogenic and iatrogenic factors
2. Intrauterine mechanical factors
3. Focal necrosis
4. Morphokinetic arrest
5. Primary malformations of the skull with secondary effects on the brain
6. Primary malformations of the CNS with secondary deformities of the skull
7. Chromosomal abnormalities
8. Environmental factors
9. Inborn errors of metabolism
10. Syndromes of unknown etiology

Sociogenic and iatrogenic factors are not relevant to the present classification. The ramifications of intrauterine mechanical factors are uncertain. There is experimental data
(Trasler, et al., 1956) demonstrating that amniotic sac puncture constricts the embryo and may cause cleft palate by mechanical interference. Furthermore, amniotic strands have been found to be attached to the head and mouth of human fetuses (Torpin, 1968). Amniotic strands have also accounted for amputations of digits.

The role of focal necrosis as a causal mechanism has been pursued in recent years. Poswillo (1973, 1975b) has demonstrated an experimental model for hemifacial microsomia by the induction of hemorrhage of the primitive stem of the stapedial artery. The extravascular blood clot results in the sequential processes of focal tissue death, repair, and redifferentiation. It is felt that the primitive pattern of morphodifferentiation observed in the affected neonate is a result of varying degrees of primary damage followed by a greater or lesser degree of “catch-up” remodeling. A similar model (Poswillo, 1975a) is postulated for mandibulofacial dysostosis but with a different pathogenic mechanism. There is early destruction of neural crest cells of the facial and auditory primordia which migrate to the first and second brachial arches.

An example of morphokinetic arrest is that of hypertelorism. It is conceivable that the angulation between the orbits might remain in the fetal state because of a midline aberration.

Craniostenosis is an example of a primary malformation of the skull which may affect the brain. Premature closure of sutures is associated with increased intracranial pressure or facial deformity.

DeMeyer, et al. (1964) believe that the “face predicts the brain”, i.e., some facial malformations predict brain malformations. They assessed facial malformations based on embryological types of classifications.

The face can be separated, embryologically, into horizontal and vertical planes. The vertical segments are:

1. Fronto-nasal segment (fronto-nasal process)

Median plane facial skeletal elements derived from the frontonasal prominence:

- a. frontal bones
- b. crista galli
- c. ethmoid bone
- d. nasal bones
- e. vomer and cartilaginous nasal septum
- f. premaxillary bone
- g. anterior primary palantine triangle

2. Lateral brachial arch segments (2)

Skeletal elements of the face derived from the brachial arches:

- a. temporal bone, in part, and ossicles
- b. zygomatic arch
- c. maxillary bone
- d. mandible
- e. hard palate

The face can then be divided into three horizontal planes. Malformations may affect one horizontal segment solely or predominantly. The upper (frontal) horizontal segment derives solely from the frontal (frontonasal) process. The middle (maxillary) segment is derived from the maxillary process of the first brachial arch and the prolabium (from the frontonasal process). The third horizontal segment, the lower (mandibular) segment is derived from the mandibular process of the first brachial arch.

In studying median facial anomalies, the author classifies five types of tissue deficiency disorders which he terms holoprosencephaly (DeMeyer, 1964):

- 1. Cyclopia
- 2. Ethmocephaly
- 3. Cebocephaly
- 4. With median cleft lip (premaxillary agenesis)
- 5. With median philtrum-premaxilla anlage

When there is a normal or an excess amount of tissue in the midline structure, DeMeyer (1967) favors the term median cleft face syndrome and describes seven features of this entity.

- 1. Orbital hypertelorism
- 2. U-shaped frontal hairline
- 3. Cranium bifidum occultum
- 4. Median cleft of upper lip
- 5. Median cleft of premaxilla
- 6. Median cleft of palate
- 7. Primary telecanthus

In conclusion, DeMeyer states that, when hypertelorism is present, there is almost certainly brain damage. When hypertelorism is associated with one of the other malformations, the probability of mental retardation is low but increases when the hypertelorism is extreme and is the sole facial anomaly.
When the clinician looks at these systems, there are many problems. While he is capable of assessing etiology of these disorders, he has virtually no information with regard to clinical classification. The first classification of rare craniofacial clefts was by Morian (1887). He described three types of clefts. Recently, there has been renewed interest in the rare clefts. The American Cleft Palate Association classification (Harkins et al., 1962) was mentioned earlier. This was then modified by Boo-Chai (1970) who subdivided oro-ocular clefts into Type I ( coursing from lateral to cupids bow to medial canthus) and Type II (from lateral to cupids bow to mid-lower lid or lateral canthus).

In 1966, Karfik proposed a detailed classification of rare clefts. Most recently, Tessier (1976) formulated a classification system based upon his extensive personal experience. This system uses the orbit as the frame of reference and the clefts are based around this axis.

A Proposed New Classification System

Based on extensive recent experience in the treatment of craniofacial anomalies, a new, practical, and simple classification system is suggested here.

All craniofacial anomalies may be classified into five categories based on etiology, anatomy, and current treatment principles:

I. Clefts
   Centric
   Acentric

II. Synostoses
   Symmetric
   Asymmetric

III. Atrophy—hypoplasia

IV. Neoplasia—hyperplasia

V. Unclassified

All may vary in their manifestation from subtle to extreme.

Clefts

Clefts may be subclassified in many ways. They all result in regions of tissue deficiency which require shifts of tissue for correction. Usually, both bone and soft tissue are involved. All gradations of hypoplasia may occur from the most minimal to wide defects of complete absence of tissue. A broad classification is one proposed by Tessier (1976) utilizing a clockface analogy from 0 to 14 (see Table 1). The point of reference for these clefts is the orbit with the clefts found in two different hemispheres. Those of the lower lid region are facial, while those of the upper lid are cranial. Clefts 0 through 4 have extensions downward to involve the maxilla and fit into the usual cleft lip and palate classifications. Their superior extensions are the more severe major craniofacial anomalies. Cleft 0 (Figures 1 and 2), with its cranial extension number 14, is a true midline abnormality frequently resulting in unilobed brain or holoprosencephaly, incompatible with life. Hypotelorism is usual with underdevelopment of tissue in the midline though, conversely, hypertelorism may occur. This cleft and 1 through 3, along with their cranial extensions 13 through 11, are centric (Figures 3, 4, and 5). The impor-

<table>
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<th>TABLE 1. A Modification of Tessier’s Cleft Classification System</th>
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<td><strong>A. Basic Considerations</strong></td>
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<tr>
<td>1. The point of reference is the orbit with the clefts found in two different hemispheres.</td>
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<tr>
<td>a. Those of the lower lid are classified as facial clefts</td>
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<tr>
<td>b. Those of the upper lid are classified as cranial clefts</td>
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<td>c. Combined or craniofacial clefts may occur</td>
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<td>2. This system describes both the surface and underlying bony anatomy.</td>
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<td>3. The extent of involvement of soft and bony tissue is variable.</td>
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<td><strong>B. The Classification</strong></td>
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**CENTRIC**

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<tr>
<th>Facial Clefts</th>
<th>Corresponding Cranial Extension of Facial Clefts</th>
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<td>No. 0</td>
<td>No. 14</td>
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**ACENTRIC**

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<th>Facial Clefts</th>
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<td>No. 4</td>
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tance of this distinction is that structures lat-
eral to this, including the cranium, orbits, and
zygoma-maxilla, are displaced laterally by the
pathologic process and must be repositioned
medially along with adding tissue to achieve
correction. The lower extensions of clefts 1, 2,
3, and 4 result in cleft lip and palate with
clefts of the maxilla and alveolus. Extension
onto the mandible occurs rarely with associ-
ated deformities such as notching of the lower
lip, bifid tongue, and deformities of the chin,
neck, and hyoid bone.

The other facial clefts, 4 through 8 and
their cranial counterparts 9 through 11, are
acentric and require addition of bone or soft
tissue. Four in its upward extension goes into
the orbit lateral to the nasolacrimal region
(Figure 6) but medial to the infraorbital
nerve. Five is a cleft of the maxilla and zyg-
oma extending from lateral to the infraorbital
nerve into the region of the canine teeth and
resulting in a true isolated facial cleft. This
cleft is extremely rare.

Clefts 6, 7, and 8 are associated with hem-
ifacial microsomnia if unilateral and with
Treacher-Collins Syndrome if bilateral. Six
extends through the zygoma at about the
zygomaticomaxillary suture, 7 through the
zygomaticotemporal suture region, and 8
through the zygomaticofrontal suture (Fig-
ures 7 and 8). All result in hypoplasia in these
regions, though, with hemifacial microsomnia,
the etiology is different from that of Treacher-
Collins Syndrome (Poswillo, 1975a). Clefts 9,
10, and 11 are associated with deformities of
the superior orbit-anterior cranial base and
are rare (Figures 9 and 10).

Treatment mechanisms involve reposition-
ing or reconstruction of structures that have
been displaced by the clefts. Number 0 and
14 is often incompatible with life and, there-
fore, not treated. Orbital hypotelorism may
be present as a component of these clefts.
When it is not associated with holoprosen-
cephaly and a true midline cleft, a similar de-
formity generally occurs with synostosis.
When the latter occurs, it is usually compat-
ible with life and may appear similar clini-
cally, but no cleft of the lip or nose is present.
It is, in fact, a masquerade of the true 0,14
cleft. It is important to distinguish between
the two as treatment and timing vary. The
synostoses are preferentially treated in the
first six months of life by removing or reposi-
tioning abnormal structures. Clefts may be
treated, if severe or extreme, in the first few
months of life by adding tissue or rearranging
it. The majority, however, are treated after
FIGURE 3. Centric cleft in #1 position with orbital hypertelorism, notched ala nasi, and laterally displaced left nasal bone. Cranial extension in #13 position.

FIGURE 4. Centric cleft in #2 position with orbital hypertelorism, bifid nasal bones, and nasal and lip deformities.

FIGURE 5. Centric cleft in #3 position with orbital hypertelorism, absent nasolacrimal drainage system, and lip and nose distortions. Cranial extension in positions 11 and 12 with forehead distortions.
FIGURE 6. Acentric cleft in #4 position with hypoplasia of left zygoma, extension downward to the lip with subcutaneous cleft and notching of the eyelid lateral to the lacrimal puncta.

the first year of life. Procedures involve major shifts of the orbit and are complex, prolonged operations. This is particularly true with upper extensions of clefts 1, 2, and 3 and their cranial components 13, 12, and 11. Clefts 1, 2, and 3 in their inferior extension are closed at the usual times for cleft lip and palate. Clefts 4 and 5 may be treated early, if extreme, and later if not so severe. Clefts 6, 7, and 8 are treated with established treatment principles and time considerations for hemifacial microsomia and Treacher-Collins Syndrome usually later in childhood. Clefts 9, 10, and 11 are also treated early if extreme, but later if they are not.

Associated with clefts 10 through 14, encephalocele may occur. Clefts of the nostril may occur with 1, 2, or 3. Numbers 4 and 5 may result in a cleft of the lip as well as the zygoma-maxilla and a coloboma of the eyelid. Hemifacial microsomia corresponds to these clefts, but is etiologically based on Poswillo’s work (1973, 1974, 1975b), the result of a vascular accident in utero, resulting in under-development or hypoplasia of the mandible-zygoma-maxilla and orbit. This varies in severity from extremely subtle to severe. Ear problems are associated in their subtle forms with minor displacements and in the more severe forms with extreme displacement or absence of the ear. Temporomandibular joint problems sometimes occur, and lateral canthal displacement is common in association with the deformities in the lateral orbital region. In Treacher-Collins Syndrome, a tissue hypoplasia syndrome involving clefts 6, 7, and 8, lateral canthal displacement, and colobomas are frequent. In clefts 9, 10, and 11, globe abnormalities and cranial base deformities as well as eyebrow deformities may occur.

Synostoses

Symmetrical

Symmetrical synostoses result from closures of the metopic suture, the coronal suture, the sagittal suture, or one of the posterior sutures. Closure of the posterior sutures rarely requires surgery about the face. Bilateral coronal suture synostosis is associated with the craniofacial dysostosis syndromes, including Apert’s and Crouzon’s, with abnormalities of the forehead, the orbits, and the midface, including the maxilla (Figures 11 and 12). Nasal protrusion and high arched palatal deformities usually occur. Associated with these synostoses are abnormalities of the anterior cranial base, generally vertical positioning, and shallow orbits. Upper forehead protrusion, lower forehead retrusion, shallow orbits with protrusion of the globe or exorbitism, and zygomaticomaxillary hypoplasia with Class II malocclusion are found. Low set ears are frequent. Many variations in the synostoses occur. If there are associated hand abnormalities, Apert’s Syndrome is designated (Figure 13). Rieger’s Syndrome (Figure 14) is associated with acentric pupils. Telecanthus is frequent in all of the syndromes, and a minimal grade of orbital hypertelorism is often found. This occurs as a result of lack of forward growth of the skull with compensatory horizontal growth in coronal synostosis.

If the lower portion of the metopic or the sagittal suture closes prematurely, there is symmetrical lack of lateral rotation of the orbits and orbital hypotelorism. Often there is a triangulation defect of the skull anteriorly with scaphycephaly (Figure 15).

Etiologically, these structural changes are important in the light of current treatment methods. Cranietomies with release of the coronal suture and the metopic suture are performed in the first six months of life. At the same time, the orbits, forehead, and nose may be repositioned. The zygomaticomaxil-
lary region may be treated using the excess bone from the craniectomy.

The spectrum of anomalies in these syndromes varies, as it does in with others, from extremely subtle to severe.

Asymmetric

Unilateral closure of the coronal or lambdoid suture results in asymmetric deformities with orbital displacement, zygomaticomaxillary, and forehead or occipitoparietal problems. This is isolated craniofacial dysostosis or plagiocephaly. Commonly, the deformities result in forehead flattening on the involved side, verticalization and shortening of the anterior cranial base, and retrusion of the supraorbital and lateral orbital regions and of the zygoma on the involved side. In addition, there may be a slight shift of the nose to that side, downward displacement of the ear, and upward shift of the mouth. Variations include protrusion of the forehead on the opposite side and shifts of the opposite orbit. Commonly associated is a contralateral flattening of the occipitoparietal region (Figures 16 and 17).

Atrophy-Hypoplasia

A multitude of anomalies occur in this category. Romberg’s Syndrome or coup-de-sabre is an example of an important form of atrophy (Figure 18). Depending on the age of onset, there is skin, subcutaneous tissue, and underlying muscle and bony atrophy with inhibition of growth and deformity of the face. There may be displacement of the orbit, ear, nose, mouth, as well as of the forehead, zygomaticomaxillary region, and mandible. Its expression varies from subtle, if it occurs later in life when growth is nearly complete, to extremely severe if onset is early in life.

Absence of the sphenoid wing or anterior cranial base unilaterally with exophthalmos in association with neurofibromatosis is an example of a mixed atrophy-hyperplasia syndrome. There is hyperplasia associated with
the tumor and aplasia of the orbit. With the brain against the ocular globe, there is resultant exorbitism (Figure 19).

Other atrophy-hypoplasia syndromes are: mandibular hypoplasia; isolated atrophy as a result of trauma, particularly if it occurred early in life; and scleroderma, which results in atrophy and hypoplasia. Hypoplasia of the maxilla occurs in association with clefts, mongolism, blepharonasal facial syndrome, and syndromes such as cutis laxa. There are other rare syndromes such as whistling face deformity with hypoplasia and globe deformities. Hallermann-Streiff Syndrome is a mixture of skin-subcutaneous tissue hypoplasia, orbit and globe hypoplasia, abnormalities of bone growth, and chin deformities. Aglossia-hypodactyly is a problem ranging from severe underdevelopment of the mandible to total mandibular absence (Figure 20). This has occurred in at least one case which included absence of the tongue and fusion of the mandibular ramus to the palate. The deformity was confined to the lower face. Clefts of the soft tissue of the nose, lips, ears, and lip, and pits, or grooves may all occur. The list is extensive.

FIGURE 9. Acentric cleft in positions 9 and 10. Lack of complete orbit rotation with orbital hypertelorism and coloboma of the right upper eyelid. Also, in this instance, fibro-fatty-glial tissue in upper eyelid.

FIGURE 10. Acentric cleft in positions 10 and 11 with absence of left ocular globe, excess interorbital distance, and absence of left nostril.

Neoplasia—Hyperplasia

Lymphangioma occurring early in life results in hyperplasia of all adjacent tissues, including skin, subcutaneous tissue, muscle, and underlying bone. Hemangiomas create similar problems, but there is less uniformity of tissue hyperplasia. Fibrous dysplasia (Figure 21) generally develops later in life and is treated at a different time. Mandibular prognathism develops prior to and during active adolescent growth unless there is associated lymphangioma-hemangioma or other tumors. Treatment is carried out at the end of this growth period in cases of isolated hyperplasia of the mandible. Lymphangioma, hemangioma, and fibrous dysplasia are treated in accordance with the severity of the problem, the age of the patient, and functional and psychological needs. Tongue, lip, ear, nose, and other single-organ enlargements may occur for all the reasons described along, with others. Multiple other tumors also occur. Treatment is planned according to the principles already outlined.

Unclassified

Other extremely rare problems occur, but they generally fit into the first four categories.

When these problems cannot be classified, they are best placed in this last category, which can best be subdivided into the following areas:

A. Multiple-Organ Involvement
B. Single-Organ Involvement
   a. Tongue
   b. Nose
   c. Eye and orbit
   d. Lips
   e. Ears
   f. Jaws

A. Multiple-Organ Involvement

FIGURE 12. Symmetric synostosis, craniofacial dysostosis or upper Crouzon’s variation. Has associated coronal synostosis, anterior cranial base deformity, lower forehead retraction, upper forehead protrusion, and slight midface hypoplasia.
These are numerous isolated occurrences which cannot be systematically classified except as described previously.

B. Single-Organ Involvement

a. Tongue

Macroglossia, or large tongue, may result from a hemangioma or lymphangioma. In muscular macroglossia, hypertrophy of individual muscle fibers has been observed. Treatment is surgical. Aglossia or hypoglossia is the absence of or a small tongue. The tongue may be cleft, lobed, or fissured. These conditions may not require treatment if tongue or oral function is not impaired.

b. Nose

The external nose and nasal septum are derived from a prolongation of the frontonasal process together with an infolding which produces the nasal septum. Unequal growth of the two sides of the nasal septum or excessive infolding of the septal portion without fusion of the two parts can account for the majority of the abnormalities of the nose and septum. The unusual presence of dermoids of dural origin can be accounted for by dural extensions which have become incorporated into the frontonasal process as it grows out from the anterior cranium. The specific defects involved are: absence of the nose, small nose, absence of the nasal septum, absence of half of the nose, persistent frontonasal process, half nose, and proboscis.

Treatment: Operative repair of the defect or reconstruction is indicated if either functional or cosmetic problems are present.

The nose can have a variety of characteristics. It may be small, narrow, beaked, flat, or pear-shaped, and these may be syndrome-specific. More specific abnormalities include: transverse groove, where a horizontal depression or groove 1 to 3 mm wide and about 1 mm deep is located caudal to the ala nasi; and bifid nose, where a congenital median fissure is present. These may also be duplication of the bony structure of the nose with two additional nasal cavities that end blindly or connect to the surface medial to the normal ones, slit-like openings in the nasal columella, or nostril-like openings in the nasal columella.

Anterior atresia may occur when the epithelial plugs, normally present from the second to sixth month of intrauterine life fail to absorb. Clinically, it may represent a spectrum from narrow nostril(s) to bony or membranous stenosis of the nostril. The treatment of this condition is reconstructive surgery designed to provide an epithelial lined nasal...
FIGURE 14. Symmetric pseudosynostosis, Rieger’s syndrome. Midface hypoplasia without craniosenosis or cranial base deformity. Has acentric pupils and ear deformities as part of the syndrome.

FIGURE 15. Symmetric synostosis of metopic suture with anterior triangulation defect of skull (trigonencephaly), orbital hypotelorism, and decreased bitemporal distance. Coincidental ear deformities.

FIGURE 16. Asymmetric synostosis, plagiocephaly or isolated craniofacial dysostosis. Unilateral synostosis of coronal suture (left in this instance) and unilateral cranial base deformity. Manifestations include retrusion of left frontotemporal region, left orbit, and left zygoma. Left lateral canthus and ear frequently displaced inferiorly on involved side. Often there is contralateral occipitoparietal flattening.

FIGURE 17. Asymmetric synostosis. Isolated craniofacial dysostosis variant with unilateral coronal suture and cranial base abnormalities. In this instance, protrusion of the left forehead, inferior displacement of the left globe including both canthi, flattening with retrusion of left zygoma, and slight retrusion of right frontal area.

c. Eyes and Orbits

Epicanthus palpebris tarsalis inversus includes excessive skin at the root of the nose, poor development of the bones of the bridge of the
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The bands extend from the white line of one lid to that of the other lid, posteriorly to the cilia, and anteriorly to the meibomian orifice.

d. Lips

Paramedial lip pits or wounds originate as vestigial remnants of the lateral sulci appearing in the embryonic mandible at the stage when

nose, and retention of the fetal epicanthal fold.

Simple epicanthus is a variation of normal which disappears by puberty in Caucasians and should not be operated on prematurely for cosmetic reasons alone.

Filiform fusion of the eyelids may be an isolated feature or may be associated with cleft lip and
length is from 7.5 to 12.5 mm. Pits of the upper lip are the result of failure of complete fusion of the premaxilla and the maxillary processes. They may be associated with the popliteal pterygium syndrome or with cleft lip or palate.

e. Ear

The external ear (auricle) is developed from six swellings called auricular hillocks. These swellings, which develop around the margins of the first branchial arch, are produced by proliferation of mesenchyme from the first and second branchial arches. As the auricle grows, the contribution of the first branchial arch becomes relatively reduced. The lobule is the last part of the ear to develop. The auricles begin to develop in the upper part of the future neck region. As the mandible develops, the ears move to the side of the head and ascend to the level of the eyes.

Absence and hypoplasia of the auricles are rare. These problems are usually associated with other multiple congenital abnormalities, including deafness or malformations of other derivatives of the first arch. These conditions result from failure of the auricular hillocks to develop (anotia) or from abnormal or suppressed development of the auricular hillocks (microtia). Atresia of the external auditory meatus and middle ear abnormalities are usually also present.

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