Congenital Midline Defects of the Midface

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The early, so-called classical description of the embryological development of the midface was first propounded by Dursy and His in the 1890's. Those concepts were based on the study of animal material with techniques for microscopic study and material preparation which, by present standards, are relatively crude. In that material, those writers offered the concept of peninsular masses of ectoderm-covered mesoderm which are surrounded by clefts, growing forward and downward, meeting, fusing, and reforming to produce the various components of the face. Thus, any interruption or delay or any abnormal fusion of these masses could produce the cleft deformities seen in infants. The descriptive terminology applied to the various prominences and areas are still considered applicable by many and are in common use (*8, 9, 12*).

Veau, in 1930, theorized a basic ectodermal wall into which mesenchyme grew, migrated, and infiltrated to form the processes which are not truly free peninsular masses, but rather connected tissues. The lack of mesodermal support subsequently caused a breakdown of the areas of poor support or absent mesenchyme with a resultant cleft formation. Veau's theory differs in essence from the classical interpretation in that no fusion of masses occurs except, of course, in the formation of the posterior or true palate.

Meurer and Hoepke (in Stark, *9*), in their study of a human embryo with a cleft lip and palate, felt that there was a mesodermal streaming and proliferation in an attempt to heal a cleft in utero.

In his extensive study of six human embryos with clefts, Stark (*9, 10, 11*) determined actual mesodermal volumes, and found a paucity of this tissue on the cleft side. He further noted no unusual mitotic or migratory activity to support the concept of embryonic attempts to heal a pre-existing cleft. In addition, he offered support of Veau's interpretation of midfacial development, stating that if mesoderm existed on either side, but not in the prolabium or premaxilla, a midline cleft would exist.

According to Patten (*8*), development of the midportion of the face is

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a result of a marked downward growth of paired nasomedial processes, with a merging of the unpaired frontal process. This merging implies both mesodermal infiltration and fusion of parts with the obliteration and eventual disappearance of ectodermal tissues included within the fusion. This can easily explain the midline clefts in this area, as well as dermoid cysts of the septum and midline cysts connected to the skin. Any point of fixation of developing integument in the area of merging of the nasomedial processes would cause a cyst. The depth of these cysts indicate the degree of merging of parts over the frontal process. In a series of 1,000 facial clefts collected by Davis (4), seven were associated with the midline.

We have collected 10 cases which we feel illustrate the development of the midface as put forth by the above reports.

Figure 1 illustrates a five-week embryo, showing the various processes labeled. The deformities described can be visualized simply by extrapolating a merger of the various portions of the nasomedial processes to the adjacent area at the midline. Minimal deviations from this embryo will demonstrate all of the deformities illustrated.

Following are 10 subjects who are presented for demonstration purposes.

SUBJECT 1. (Figure 2). This patient has a bilateral macrostomia, with a moderate notching of the upper and lower lips, which becomes more apparent on attempting complete apposition of the lips. This situation illustrates the most minimal loss of a downward growth potential, or an inadequate infiltration or volume of mesodermal elements.

SUBJECT 2. (Figure 3). This child has an incomplete cleft of the lip with a normal premaxilla and normal palate. There is a deep philtrum and the skin is adherent to the buccal mucosa above the lip. As described by Patten, the labial components of the nasomedial processes were reasonably well

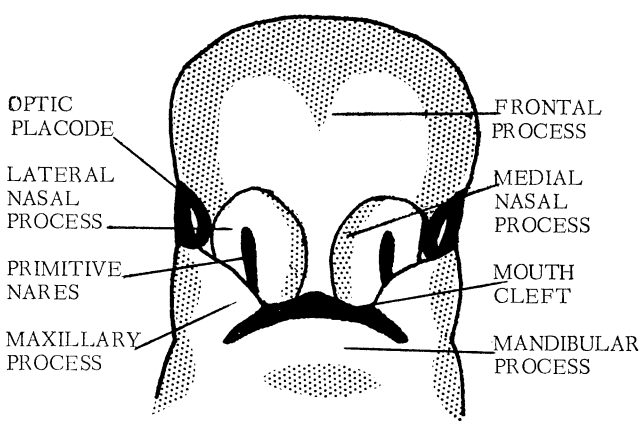

Fig. 1: 10 mm Embryo

FIGURE 1. Illustration of five-week embryo.

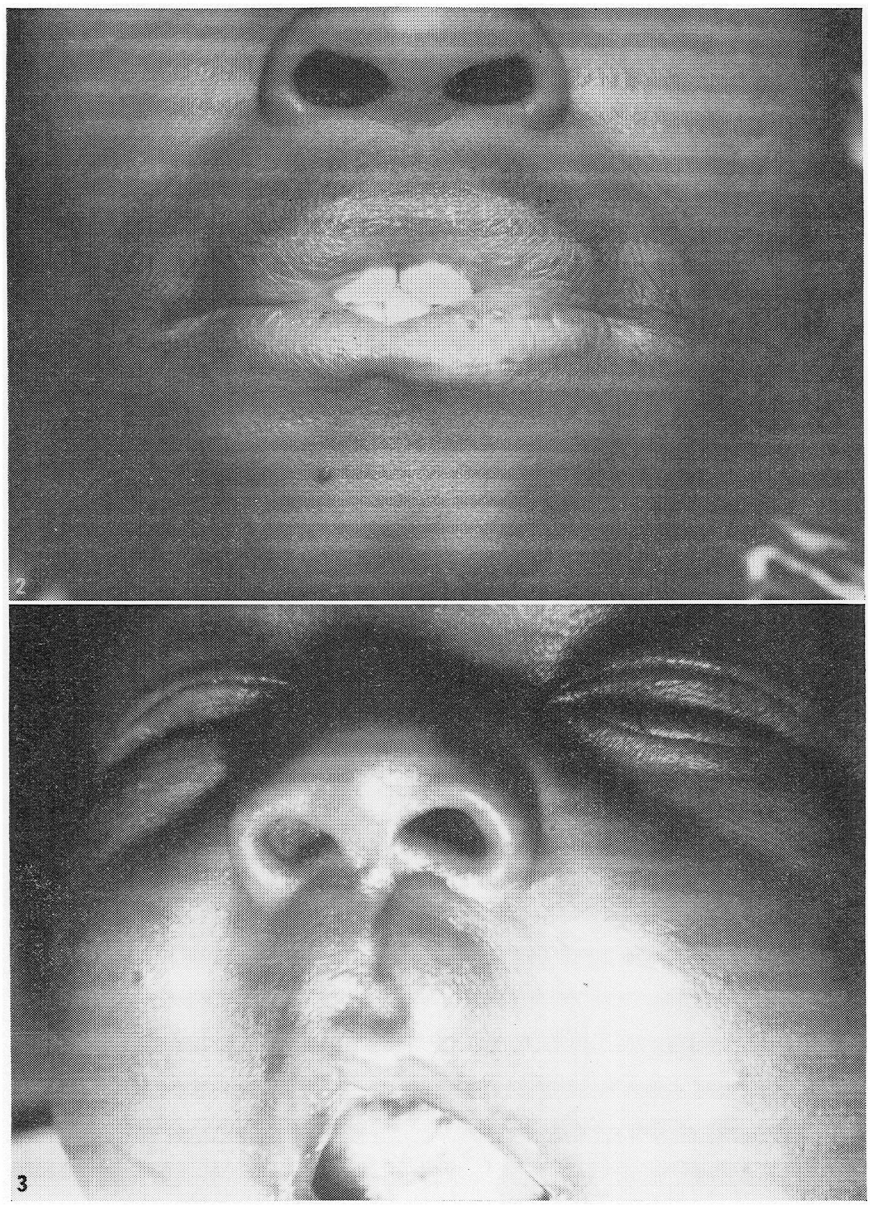

FIGURE 2. Macrostomia, associated with midline notching.
FIGURE 3. Subcutaneous midline cleft lip.

developed, and fused to the maxillary processes but only partially to each other at the midline.

SUBJECT 3. (Figure 4). This infant has a midline cleft of the lip and palate associated with a meningocele in the region of the posterior septum at the base of the skull. It is suggested that there was such a lack of quality,

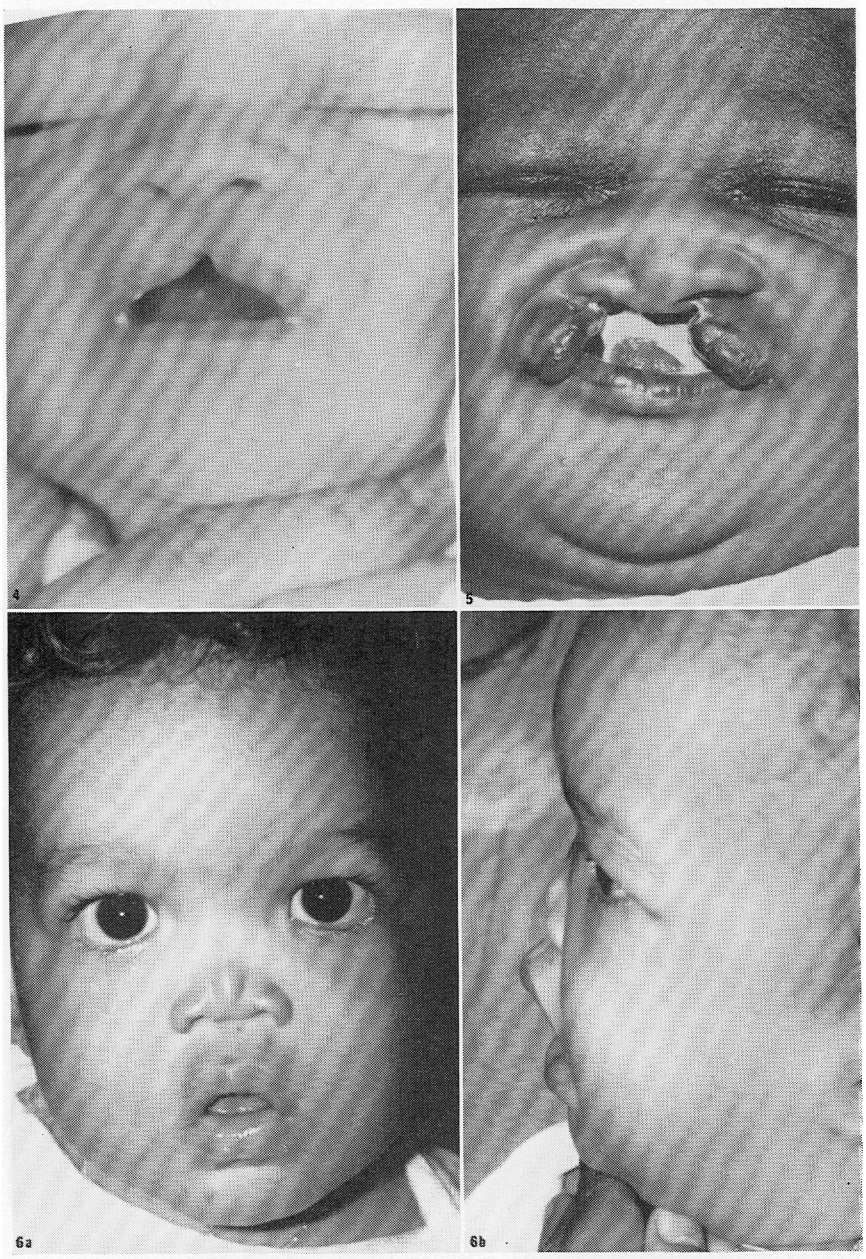

FIGURE 4. Midline cleft lip, cleft palate, and meningocelle of septum.
FIGURE 5. Midline cleft upper lip and absent premaxilla.
FIGURE 6A. Absent septum and bridge of nose.
FIGURE 6B. Profile view of subject shown in 6A, showing normal upper lip.

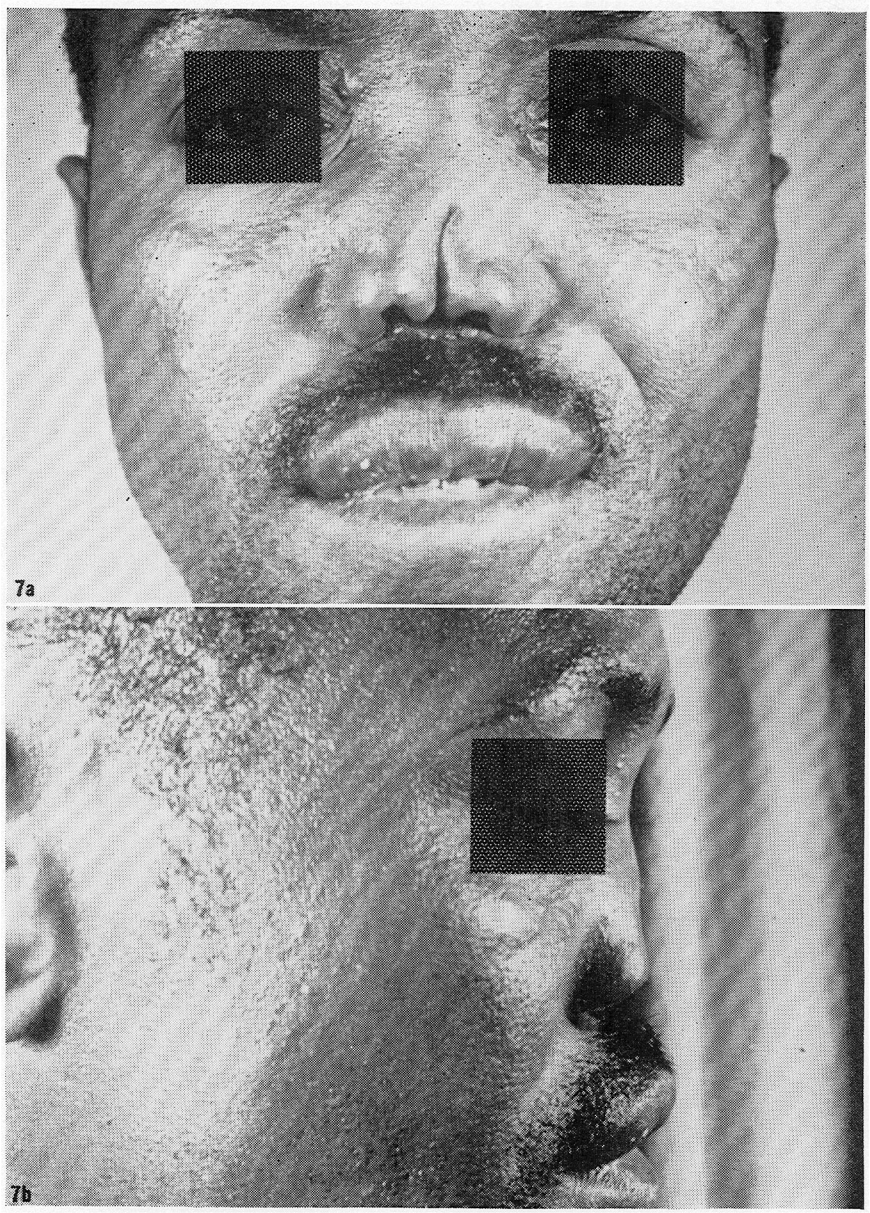

FIGURE 7A. Typical bifid nose.
FIGURE 7B. Profile, adult bifid nose.

vigor, or quantity of mesoderm that the mesenchyme at the midline failed to produce skeletal support to the forebrain as well as the soft tissue elements necessary to form over the frontal process, the midline labial component, and lower septal segment.

SUBJECT 4. (Figure 5). This subject, from the records of Dr. Richard

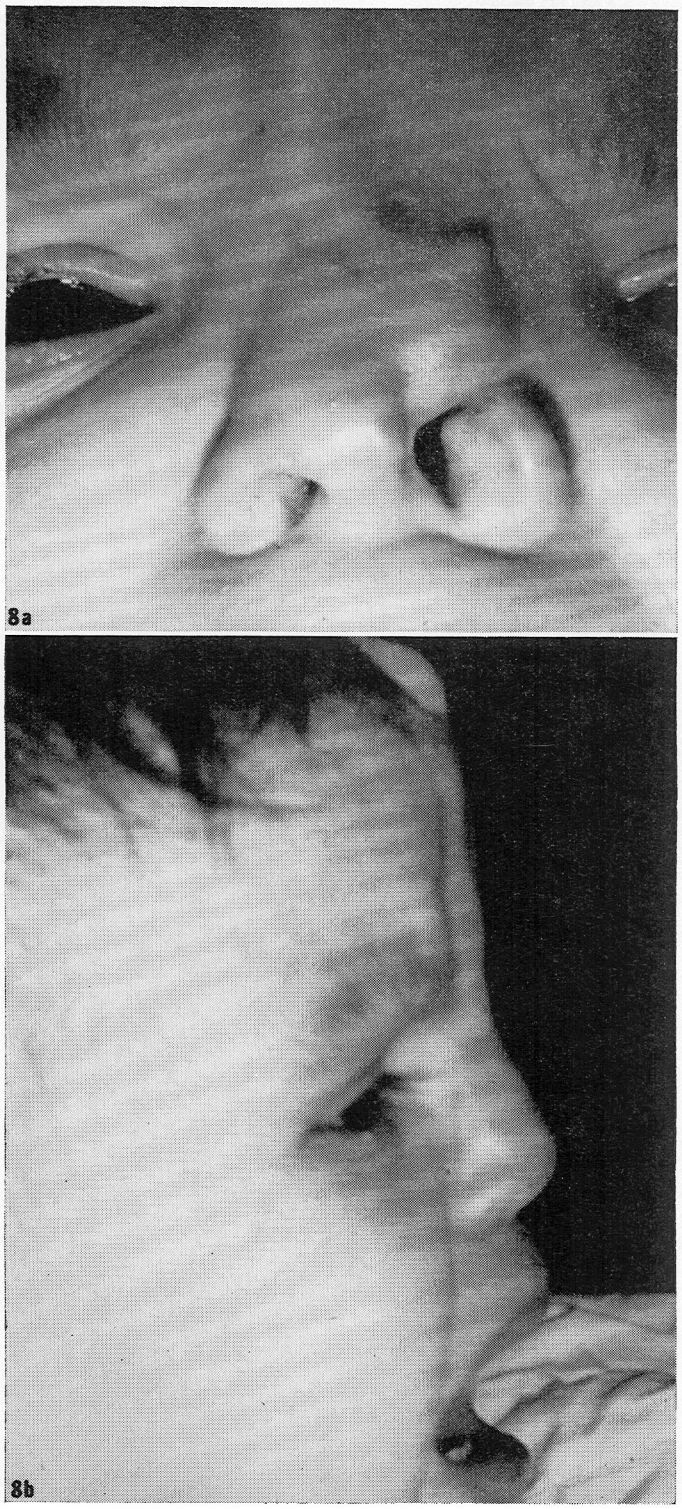

FIGURE 8A. Cleft nose, cyst of the septum, and a frontal midline encephalocele.
FIGURE 8B. Profile view of subject shown in 8A, showing a cleft nose, cyst of septum, and frontal midline encephalocele.

Stark, is similar to Subject 3 but exhibits more severe deformities of the lip and lower nasal structures.

SUBJECT 5. (Figures 6A and 6B). This child demonstrates an absence of cartilaginous septum and midnasal support, and, in profile, the midface is seen to be underdeveloped also. The normal lip structures would indicate that the lower nasomedial elements were normally formed, while the superior portion lacked the tissue to form the nasal bones and septal elements at the midline.

SUBJECT 6. (Figures 7A and 7B). This adult demonstrates a bifid nose, a condition which has been previously described by several authors (*1, 3, 6*). The profile again illustrates an underdeveloped midface.

SUBJECT 7. (Figures 8A and 8B). This child presents with a cleft in the nose (unilaterally), a midline cyst of the septum, and a frontal midline encephalocele. Air contrast studies of the ventricular system revealed an absence of the corpus callosum. It suggests that a deficiency or improperly timed development of the unpaired frontal process may well be reflected

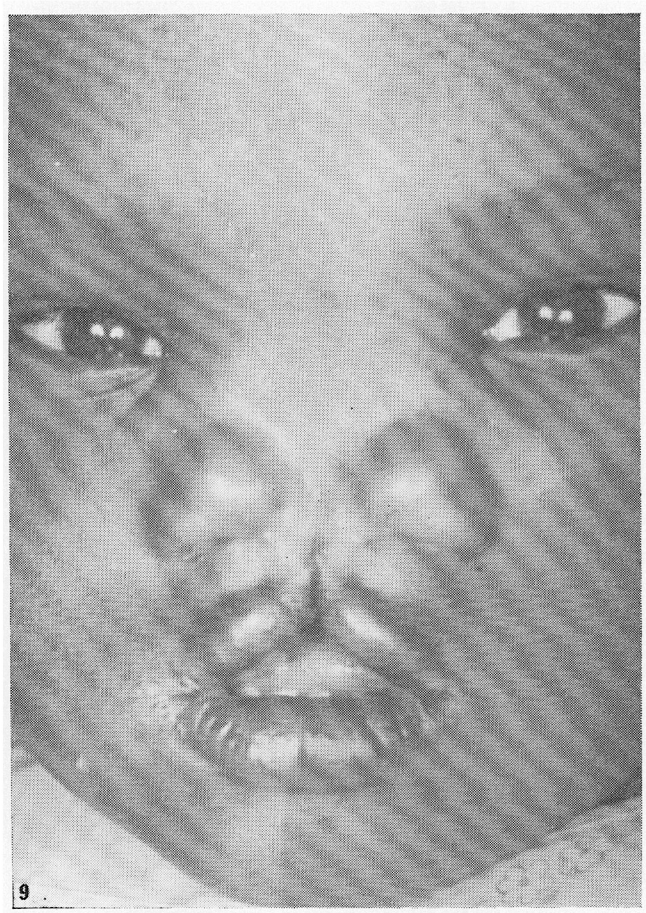

FIGURE 9. Severe bifid nose and cleft lip.

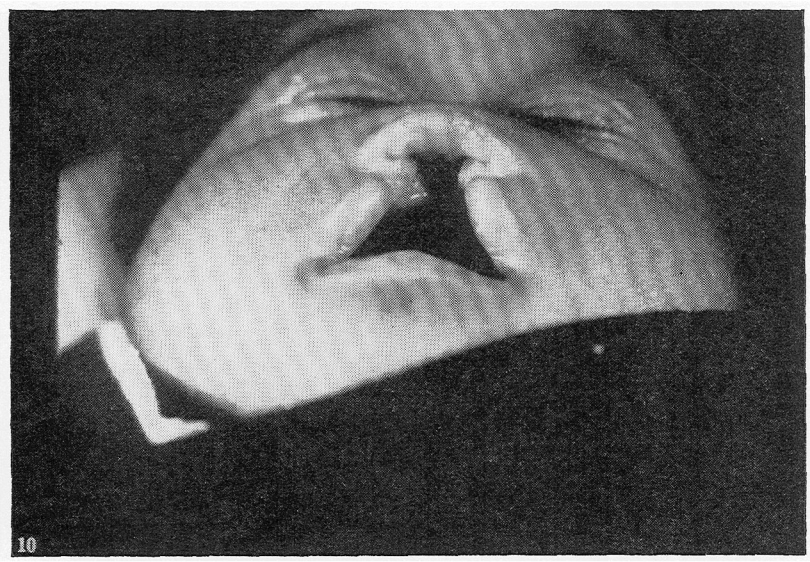

FIGURE 10. Arrhinencephaly, showing multiple midline deformities.

in the abnormal growth progress and subsequent merging of the paired nasomedial processes which fuse over this structure (*1, 3, 7*).

SUBJECT 8. (Figure 9). This child demonstrates a severe bifid nose with notching of the lip and a W-shaped nasal skeleton with the equivalent of two nasal bone arches. The similarity to the embryo is too obvious to merit a more detailed description. The multipotential tissues were, however, able to develop two sets of nasal bones in this case and the skin to cover them (*13*). (This material is from the records of Dr. J. C. Kelleher.)

SUBJECT 9. (Figure 10). This child is a case of arrhinencephaly, described previously by Brucker (*2*). This is a much more severe deformity of the frontal process with absence of the forebrain, septum, and cribiform plate as well as the obvious midface deficiencies in nose, columella, and upper lip. Ordinarily, there is an intact true plate which arises from the maxillary processes rather than the frontonasal processes.

SUBJECT 10. (Figures 11A and 11B). This child presents an extremely unusual deformity: An adhesion between the prolabium of the upper lip and the midportion of the lower lip, with the lateral labial segments similar to the cleft sides of a bilateral cleft lip. In addition, there is a cleft of the hard and soft palate, as seen in the usual bilateral lip. The premaxilla angulates abruptly behind the adhesion. The premise of mesoderm infiltration of a pre-existing ectodermal wall is difficult to theorize in this subject, since no ectodermal plate assumes this position in the developing embryo. The oropharyngeal plate lies over the embryological stomadeum which occupies an area approximately analogous to the tonsillar area in the adult, and no other structure is present in this location. The possibility of an exceedingly mild type of midline double teratology, as described by Conway and Goulian (*5*), can probably be discounted in the absence of the other stigmata (duplication of other oral parts and ball-like mass of tissue below the fusion). The most easily acceptable premise would be to revert to the

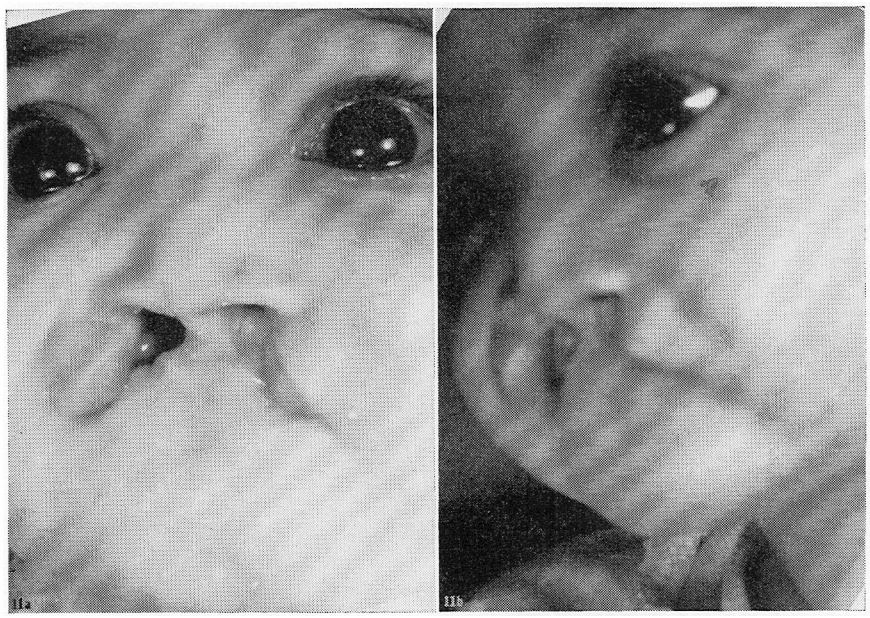

FIGURE 11A. A typical bilateral cleft; however, there is an adhesion between the prolabium and the lower lip.

FIGURE 11B. Profile view of subject shown in 11A.

classic concept of fusion of peninsular processes to obliterate free clefts. This would presuppose either an altered development of the frontal process which forces the lower nasomedial processes to fuse with the lower lip, an overgrowth of the mandibular process with a similar fusion, or an overabundance of material in the lower portion of the nasomedial process with projection to and merger with the lower lip with eventual obliteration of the ectodermal vestiges. This would also suggest the multipotential status of the early mesenchymal cell as it develops a part appropriate to its surrounding environment. Davis (*4*) illustrated one somewhat analogous case where there was a fusion of the mandibular to the maxillary gingival margins and an associated temporomandibular joint ankylosis.

Summary

A series of cases was presented with a variety of deformities of the midline structures of the face which appears to support the contention that there is a combination of factors to be considered in the etiology of the midline defect. There are indications that the midline facial structures develop from the unpaired frontal process which grows upward and away from the lower face. The paired nasomedial processes progress both toward the midline and downward, away from the frontal process and oropharynx by a process of mesodermal infiltration into the preexisting ectodermal plate. They grow over the frontal process and exhibit a marked growth and differentiation into the mesodermal structures of the midface, which merge and fuse as they grow forward, down-

ward, and mesialward obliterating the prominence of the frontal process. The merging of nasomedial masses and obliteration of ectodermal remnants is as important as the support of pre-existing walls of ectoderm by mesodermal infiltration which prevents dissolution as in the formation of clefts or rupture of the oropharyngeal plate. Additional material for study as well as the refinement of techniques may offer better explanations of the pathogenesis of these often severe anomolies, and may offer a better rationale of therapy or prevention rather than the present gross surgical corrections of anatomical deformities.

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