Congenital malformations of the nasal ala may occur as isolated unilateral nasal alar defects or as part of more severe facial clefing syndromes. The embryologic significance of the isolated alar deformity seems separate from other craniofacial deformities and justified comment. Clinical examples of isolated nasal alar malformations are correlated with a study of 60 human embryos ranging from seven to 10 weeks gestation. These studies indicate that the nasal and alar developmental fields each have a separate critical time during which cells are most susceptible to altered development.

KEY WORDS: Nasal ala, congenital defects, embryology, craniofacial deformities

Introduction

Congenital notching or clefting of the nasal ala is a rare anomaly which is most often associated with other craniofacial deformities but may occur as an isolated paramedian nasal defect. Nasal alar clefting has been characterized as part of a spectrum of craniofacial anomalies. Our emphasis is both on the patterns of presentation and on the developmental significance of isolated alar clefts. We have observed isolated notching and clefts of the ala nasi involving only that portion of the lateral nasal wall inferior and lateral to the nasal bone. The embryologic significance of these malformations appears to be separate from other craniofacial deformities and justifies comment.

The reported spectrum of anomalies associated with alar clefting has been expanded by numerous observers. The malformations most commonly associated with alar clefting are those of 1) orbital hypertelorism, 2) broadening of the nasal root, and 3) median facial clefting, anterior cranium bifidum, or both. De Myer (1967) called the condition "median cleft face syndrome with hypertelorism." Sedano et al. (1970) grouped the anomalies under the term "frontal nasal dysplasia" subdividing them into type B, C, and D, which may be associated with unilateral or bilateral alar notching, but no examples of isolated alar notching were reported. Facies B consists of orbital hypertelorism, broadening of the nasal root, and a deep median facial groove or unilateral cleft of the nose, the upper lip, or both. In facies C, unilateral or bilateral notching of the nasal ala may occur together with orbital hypertelorism and broad nasal root. The combination of facies B and C constitutes facies D.

In his analysis of 14 personal cases and a literature review, Mazzola (1976) included notching of the nasal ala as part of the spectrum of "paramedian frontal nasal anomalies" which, according to Pfeifer (1974), occur along the border of a line extending from that of the unilateral cleft lip passing through the nasal ala and maxillary bone and extending upwards across the eyebrow and forehead. Mazzola concluded that a notch of the nasal ala is a rare anomaly and that, when it is seen, it is generally combined with other paramedian or median facial anomalies.

All of the authors who have most recently speculated on the etiology and pathogenesis of notching of the nasal ala and its associated anomalies have had different views. Cohen (1971) suggests that the group of anomalies classified as frontal nasal dysplasia relate to
failed development of the nasal capsule. Specifically, the sagittal portion of the nasal portion of the nasal capsule is described as relating developmentally to the tip of the nose and nares. Based on his analysis of the developing face, Schaeffer (1935) pointed out that the differentiation of the nasal capsule into the alar cartilage is incomplete until after the sixth fetal month, but he does not speculate on the impact of developmental insults occurring after the seventh fetal week.

Clinical Observations

Selected from our collection of nasal anomalies are four examples of isolated unilateral paramedian nasal clefts of varying severity. Of the cases illustrated (Figures 1 through 4), each was born after an uneventful term pregnancy; there was no family history of craniofacial anomaly; and each child developed normally except for the nasal defect. None of these patients had clefts of the lip or palate, and all had normal interorbital distances, normal orbits, and normal cranial development. With the exception of a valgus deformity of the right foot (Case 3), none had extracranial defects.

Each of the nasal defects illustrated include a unilateral full thickness deformity of the lateral nasal wall and all or part of the alar cartilage, but the nasal bones, septum, and contralateral nasal ala are spared. In view of the absence of other anomalies generally attributed to frontal nasal dysplasia or lip and palate clefts, the embryological significance of isolated alar clefts should be reviewed.

Embryological Observations

The preceding clinical cases served to show that defects of the alar region need not involve the total nasal structures. In these cases, we see isolated defects of the nasal ala associated with normal anatomy of the nasal bones and nasal cavities. This selectivity can be explained in several ways.

Classical embryological information (Schaeffer, 1961; Schaeffer 1935) suggests that notching of the nostril borders may be due to

FIGURE 1. A Caucasian female infant with an isolated cleft notch of the left alar rim. The nasal bone, nasal cartilages, and soft tissues of the lateral nasal wall were otherwise normal. Note the mid-line sinus.
failure or alteration of early mesenchymal cells to migrate into the area and to divide into the number of cells to fill out the medial and lateral nasal processes which surround the developing nostril of the human embryo. This migration begins as early as five weeks after conception. Whatever the underlying mechanism which leads to altered cellular proliferation and diminished filling out of the subcutaneous tissue fields, the notching we do see in the nasal alae is comparable in appearance to clefts of the uvula, which have been explained as a failure of a merging of cells (Burdi and Faist, 1967).

In the context of this particular study, the localization of isolated clefting of the nasal alae was also addressed in terms of developmental relationships between the alae and nasal cavities. A sample of 60 human embryos ranging in fertilization age from approximately seven through 10 weeks had their external nasal and nasal capsular regions graphically reconstructed from serially sectioned histologic sections. As early as seven weeks, there were already clear signs of progressive chondrification of the anterior nasal capsule and its extensions into the basicranium (or chondrocranium). At this point, the cartilage of the nasal capsule did not extend anteriorly into the tissues of the external nose. Beginning at approximately the eighth week, the previously undifferentiated tissues of the external nose first began to show differentiation of mesenchymal cells into cell clusters taking on deeper staining properties with trichrome connective tissue staining used. As these cell clusters were followed into older embryos up to ten weeks, these clusters began to differentiate into cartilage cells in discrete areas that were identified as presumptive greater and lesser alar cartilages along with the separate lateral nasal and accessory cartilages of the external nose.

These observations in human embryos can be summarized by Figure 5, which shows structures comprising the nasal capsule and
FIGURE 4. A Caucasian male infant with an isolated lateral nasal cleft through the left lower and upper alar cartilages. The nasal bones, septum, and turbinates were normal.

FIGURE 5. Shown above are key stages in development of the nasal capsule and external nasal regions at seven and 10 prenatal weeks as compared to the postnatal morphology. Figure 5-A shows that the overall mesenchymal field in the nasal region can be divided into separate capsular and alar fields of cells which later differentiate into cartilage. At 10 weeks (Figure 5-B), the previously undifferentiated capsular field appears as a cartilaginous skeleton for the nasal cavities. Alar field mesenchyme differentiates into separate cartilage fields which correspond to the cartilages of the nasal alae seen postnatally (Figure 5-C).
framework of the external nose derived from two distinct (but contiguous) areas referred to as the capsular and alar fields respectively. The capsular field chondrifies first into the nasal capsule and its forward midline extension called the nasal septum. It is this capsule which provides the morphologic template around which bones of the upper facial skeleton develop. Developing somewhat later and in the alar mesenchymal field anterior to the capsular field are the alar cartilages. At ten weeks, the skeletal elements of the nasal capsule and the external nose are well defined.

These observations show that the alar cartilages do not develop from the forward-most regions of the cartilaginous nasal capsule per se. Instead, the alar cartilages chondrify a week or more later from a separate mesenchymal field. The implications of these findings are that the nasal and alar developmental fields each have separate critical time periods during which cells are most susceptible to altered development (Wilson, 1973). Thus, it is possible for alar field cells to be affected selectively. Compared to the capsular field, the alar field has a longer critical period during which its cellular proliferations and chondrifications can be altered to produce defects. Of course, if an environmental event, such as the introduction of a teratogen occurs early enough and is of sufficient magnitude, then both the capsular and alar mesenchymal fields can be affected. This series of events would lead to altered structure of the skeletal parts derived from each of the fields.

References


