Congenital Absence of the Nasal Columella

MICHAEL L. LEWIN, M.D.

Three cases of congenital absence of the nasal columella are presented: one male and twin females. The male also had bilateral congenital cataracts. The nose and the septum were otherwise well developed and appeared normal. The missing segment involved the medial crura of the alar cartilages with their tegumental mantle. There was no deformity of the upper lip or alveolus. Two procedures for reconstruction of the columella that were used in these cases are described in detail. The embryonic pathogenesis of this deformity is suggested.

KEY WORDS: columella, nose congenital anomaly of the columella, nose reconstruction of the columella

Congenital absence of the nasal columella occurs in arhinencephaly, a severe midfacial defect associated with an anomaly of the brain (De Meyer, 1967; Mazzola, 1976). A more common congenital defect is the bifid columella. This occurs (with varying degrees of severity) together with other midfacial anomalies (bifid nose, median cleft lip, hypertelorism) in a syndrome known under different designations, such as frontonasal dysraphia (Francescon and Fortunato, 1969), frontonasal dysplasia (Sedano et al, 1970), median cleft lip syndrome (De Meyer 1967), or facial cleft #0 (Tessier classification cited in Kawamoto, 1976).

No reference could be found in the literature to isolated absence of the columella with a normally developed nose and upper lip (Gorlin, 1986). Possibly, these cases have not found their way into the literature because the deformity is not conspicuous and the individuals do not seek medical attention until late childhood or adolescence.

Three cases of isolated absence of the columella, including a set of twins, are reported here.

Case Reports

Case #1 and Case #2. Dizygotic twin sisters (J.M. and G.M., Figs. 1A and 2A), aged 16, were brought in by their mother in 1954 because of an identical defect of the nose. Viewed in front, the nose appeared normal and well-shaped. The lip was normal with no scars and with a small philtral dimple. It was only when the head was tilted backward that the absence of the fleshy columella was clearly visible. The septum was straight, supporting the distal nose and ending in the vestibulum, slightly cephalad to the plane of the nostril. The defect involved the medial crura of the alar cartilages, the enveloping skin, and the membranous septum. The lateral crura of alar cartilages could be palpated easily, and there was no separation between them. The alveolus had a full complement of normal-looking teeth.

The twins were born at term after an uneventful pregnancy. The deformity was noted shortly after birth, but since it was not conspicuous, nothing was done until the girls reached adolescence and became self-conscious about their abnormalities.

The family came from Newfoundland, Canada, and knew little about their antecedents. There was no consanguinity between the parents who had 13 living children, including another pair of fraternal twins. Three siblings died in infancy. There was no history of similar or other congenital defects in the family.

Both sisters had the columella reconstructed (Figs. 1B, C; 2B, C. See, Reconstruction of Columella). The sisters were seen again 30 years later. Both were married, and both seemed of average intelligence. J.M.D. was childless; G.M.H. had one son, aged 25, and had had two miscarriages. Their medical histories offered no further information about any other congenital abnormalities. They could account for 25 children among their own siblings, with no anomalies.

Except for minor, inconspicuous irregularities and asymmetry of the nostrils, the reconstructed columellae of both siblings looked normal (Figs. 1D, 2D).

Case #3. J.R., a 9-year-old black male child, was referred in 1974 by another surgeon for consultation because of absence of the columella since birth. The external nose was well-formed and the upper lip was normal, without scars, and with a well-developed...
FIGURE 1  A, J.M.: Preoperative absence of columella.  B, and C, Front and lateral views of reconstructed columella (see Fig. 4).  It is too square at its base.  D, Thirty years later.
The sharp edge of the cartilagenous septum ended within the vestibulum, and the structure distal to it was missing. Dental development was normal. The child was one of nine siblings. The mother had been declared incompetent (nothing was known about the father), and the children were placed in foster homes. Allegedly, the children were normal. The patient had bilateral congenital cataracts. After removal of the cataracts, he developed hyphema of the left eye and was also treated for glaucoma. It was suggested to the referring surgeon that the reconstruction of the columella be carried out with a composite graft from the ear or, as a second choice, by the Smith procedure. The surgeon chose the latter.

The patient was seen 10 years later (Fig. 3 A, B). There was some minor asymmetry of the nostrils. The reconstructed columella was bulky, somewhat short, and hyperpigmented. In a black individual, such hyperpigmentation is unavoidable after any reconstructive procedure. Because of the patient’s severely limited vision and learning disabilities, he attended a special school for handicapped children. He was still with his foster family and had no contact with his biological siblings.

Regretfully, the genetic medical history of these three cases was not very reliable. They all came from large, widely dispersed families and were poor historians.

**RECONSTRUCTION OF THE COLUMELLA**

Converse (1976) considered the columella as one of the most difficult structures to reconstruct. Paletta and Van Norman (1962) reviewed the plethora of methods used for columella reconstruction. They include forehead flaps, cheek or lip flaps, cervical skin tubes, and flaps from the dorsum of the hand. Most of the columella defects were the result of trauma, infection, or excision of a tumor. The composite free graft from the ear was considered applicable only in cases of partial loss of the columella.

All flaps from the face share the serious disadvantage of leaving a visible scar. The distant flaps are cumbersome, time-consuming, inconvenient to the patient, and require multiple stages.

The two procedures used in the twin sisters avoided a visible scar. In patient J.M., the reconstruction of the columella was done with a skin-lined mucosal flap from the undersurface of the upper lip, as described by Smith (1959) (Fig. 4). Sagittal incisions were made 1.5 cm apart on the undersurface of the upper lip, extending approximately 1 cm from the vermilion border to the

---

**FIGURE 3** J.R. Original (preoperative) picture not available. A, B, Front and oblique views after reconstruction, as in Figure 4. The reconstructed columella is too short and bulky; it is clearly delimited by its hyperpigmentation.
A bipedicle flap was created, consisting of mucosa and some submucosal tissues. A full-thickness postauricular skin graft was placed underneath the flap with their raw surfaces in apposition. A mild pressure dressing was applied to the upper lip for a few days.

Three weeks later, the attachment of the bipedicle flap close to the gingiva was severed, the lip was everted, and the freed end of the flap was sutured to a denuded area on the undersurface of the nasal tip. The mucosal defect in the lip was closed by approximation. The lip was kept in an everted position by use of sutures passed through a small lead plate. These sutures were attached to the cheek with adhesive. Two weeks later, the remaining attachment to the lip was severed, the mucosa was incised superficially in the midline, trimmed, and sutured to the edge of the septum and along on the floor of the vestibulum. The skin-grafted surface faced outward, and the narrow strips of mucosa were well concealed within the vestibulum. A depressed scar at the attachment to the tip of the nose was revised a few months later (Fig. 1B, C).

This method produced an acceptable columella. However, it required multiple stages—albeit all minor—that could be performed without hospitalization.

In G.M., a composite graft from an earlobe was used. There was some concern about whether the meager area of contact with the recipient tissue would be sufficient to revascularize a composite graft of the required size. For a total reconstruction of a columella, a graft or flap with a skin surface of 2 × 2 cm is needed. Instead of the conventional pie wedge (Dupertuis, 1946) the composite graft was taken from the lower periphery of the earlobe above its attachment to the cheek. The segment of full-thickness earlobe, 2.5 × 1 cm, was tapered at its superior end so that, after closure of the defect, a smooth outline of the earlobe was restored. In patients with reasonably well-developed earlobes, reduction of 1 cm does not attract attention. The skin surface of the graft is doubled, since it includes the medial and lateral aspects of the earlobe.

The distal end of the septum was incised, freeing the mucoperichondrial flaps for a few millimeters. Small areas were denuded on the undersurface of the tip and on the floor of the nose at the junction with the upper lip. The earlobe graft was trimmed and sutured in place. The entire graft survived. A few months later an adjustment was performed because the reconstructed columella was too flabby and drooping. The tip was well-positioned; there was
no need for a supporting columella strut (Fig. 2, B and C).

EMBRYOLOGY

During the early period of organogenesis, the paired medial nasal processes join together to form the philtral portion of the upper lip, columella, nasal tip, and septum (Mazzola, 1976). Incomplete merger of these processes may affect all these structures, but in varying degree. Partial expression of this malformation may be limited to a bifid columella although, in most cases, there is a noticeable separation of the alar cartilages.

The explanation of a complete agenesis of a small morphologic unit consisting of medial crura of the alar cartilages with their tegumental mantle can be only speculative (Burdi, 1986). The assumption is that the organogenesis of the frontonasal area proceeds normally. At the end of this period, about the sixth week of gestation, a recognizable nose and lip are formed. However, the nose continues to grow and take shape during subsequent months. Its morphogenesis and growth are accomplished by differentiation and proliferation of mesenchymal elements that differentiate into bone, cartilage, and subcutaneous tissue.

Patterns of cellular differentiation and proliferation are selective in different parts of the nose and their timing varies (Newman and Burdi, 1981; Wilson, 1973). During the critical time of its peak development, a morphologic unit is most susceptible to a teratogen.

The columella develops in front of the growing septum. Assuming that the critical time for development of the columella is later than that of the adjacent parts of the nose, a teratogen introduced at a later date may selectively arrest the cellular penetration and chondrification of this structure alone. The surrounding structures of the nose and the lip will develop normally, except for the columella.

A better and more detailed understanding of the later stages of human embryonic development of the nose is needed to confirm this interpretation.

Acknowledgement. The author thanks Dr. Burdi for his helpful suggestions and Dr. R.D. Landres for reviewing the manuscript.

REFERENCES

Burdi HR. (1986). Personal communication to the author.
Gorlin RJ. (1986). Personal communication to the author.