Case Report

Congenital Arhinia: Case Report of a Rare Congenital Anomaly

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Abstract

Congenital absence of the nose or arhinia is a rare defect of embryogenesis often associated with other anomalies. Arhinia is a life-threatening condition that requires a highly skilled neonatal resuscitation team in the delivery room. The associated anomalies often have a significant effect on the immediate as well as long-term outcome of the neonate. This report presents a case of congenital arhinia and reviews the management of such cases.

Keywords: arhinia, congenital, neonate

Introduction

Congenital absence of the nose (arhinia) is extremely rare. A review of the literature located only 29 cases of arhinia since 1931.[1–26] Arhinia causes severe airway obstruction and poor feeding in the affected neonate. There is an association with other facial anomalies, especially defects of the eyes, ears, palate and midline defects.

Case Report

A full-term boy was born via an uncomplicated vaginal delivery. The mother was 35 years old and she had a normal pregnancy. Prenatal care that included sonographic findings was normal. The mother had four uncomplicated pregnancies before this conception. There was no history of amniotic fluid imbalances or drug consumption during pregnancy.

Just after delivery, the resuscitation team encountered a baby without a nose and eyes. The neonate was breathing with his mouth by the oral airway. Physical examination of other organs was normal. Laboratory findings were within normal limits (Table 1).

Echocardiography of the neonate was normal as was a chest X-ray. Both abdominal and brain sonographies were normal. CT scan revealed the absence of a nose and eyes.

After delivery, due to ineffective breathing, the neonate was intubated and transferred to the NICU. The neonate needed a tracheostomy due to the continuous need for respiratory support. He was hospitalized for 30 days in NICU and then transferred to a nursery for observation. After 2.5 months, the baby died due to sepsis (Figure 1).

Figure 1. Arhinia.

Table 1. Laboratory findings.

CBC
WBC= 9300 (Poly: 70%, Lymph: 30%)
Hb=16.5
Hct=47.4%
MCV=112.9
MCH=39.3
MCHC=34.8
Platelets: 226000
Arterial blood gas
pH=7.44
PaO_2=61.1
$PaCO_2$=34.4
HCO_3=22.9
Biochemistry
BS=82
BUN=3
Cr=0.5
Ca=9.5
Na=130
K=4.1
CRP: Neg
B/C: Neg

Discussion

Embryological development of the nose occurs between the third and eighth week of life.[27] The formation of the face is preceded by wave-like migrations of cranial neural crest cells from the region of the trigeminal nerve to the face.[28] These cells create the mesoderm that develops into facial structures. At day 24, the face consists of a superior frontal process, paired bilateral maxillary process in the mid-face and paired bilateral mandible process, caudally.[29] The maxillary and mandible process are separated by the primitive mouth (stomodeum).[28] The nasal alae are formed by fusion of the nasal lateral and medial process.[29] The medial nasal process fuses at midline with the frontal prominence and results in the formation of the frontonasal process that gives origin to the columella, philtrum, upper lip, nasal bones, cartilaginous nasal capsule and superior alveolar ridge.[29] The nasal placodes, which are local thickening of surface ectoderm, develop laterally to form the frontal process between the lateral and medial nasal process during the fourth week of life.[28] The nasal placodes invaginate to form the nasal pits during the fifth week of life. The nasal pits form the nostrils. Deeper within the face, fusion of the maxillary and frontal process forms the rudimentary palatal shelves at six weeks of life. Cells within the nasal pits continue to migrate posteriorly to form the primitive nasal cavities, which are separated from the

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buccal cavity by the rudimentary palatal shelves.[30] By the ninth week of life, the cartilaginous nasal septum, which results from persistence of neural crest cells between nasal cavities, directly overlies the buccal cavity.[30] The palatal shelves of the maxillae migrate medially as the septum migrates inferiorly. By the tenth week of life, the palatal shelves and the inferior septum fuse to form the secondary palate.[28] At this point, the posterior nasal cavities are separated from the buccal cavity by the bucco-nasal membrane. This membrane ruptures, establishing communication between the nasal and buccal cavities. The primary posterior choanae formed as the nasal cavities canalize are promptly filled by epithelial plugs. These plugs eventually resort to form the secondary (permanent) posterior choanae and establish the potency of the nasal cavities.[30] It has been traditionally accepted that the epithelial plugs are present at the unit 24th week of life.[28] However, recent evidence suggests that these plugs may actually absorb as early as the 15th week of life.[30]

The pathogenesis of arhinia is poorly understood. It has been postulated that lack of development of the nose results from failure of the medial and lateral nasal process to grow, but it is also possible that overgrowth and premature fusion of the nasal medial process results in the formation of an artretic plate. Arhinia may also result from lack of resorption of the nasal epithelial plugs during the 13th to 15th weeks of gestation. Another explanation may be abnormal migration of neural crest cells to this region, resulting in aberrant flow of the multiple mesodermal structures required to establish a normal nose and its cavities.

Congenital arhinia is a rare defect of embryogenesis. It appears that only 29 cases have been reported in the literature to date, with no specific gender preponderance.[1–27] Most cases are sporadic; however, familial cases have been described. Olsen and associates reviewed the literature through 2001 and noted 22 additional cases.[3] McGlone and associates collected 27 cases until 2003 and investigated common abnormalities.[4] As was true in our case, most of the cases had an uneventful antenatal history.

The clinical consequences of congenital arhinia are severe airway obstruction and inability to feed. Placement of an oral airway should be performed in an acute setting. All cases required airway management in the neonatal period, most commonly a surgical tracheostomy, which allows the infant to feed orally and precludes the complications associated with orogastric tubes. Most authors agree that surgical reconstruction of the external nose and inner cavities should be delayed at least until preschool years, when facial development is nearly complete.[15,17] One case has been reported in which simultaneous reconstruction of both the internal and external nose was undertaken in the newborn period.[18]

The degree of nasal absence varies from case to case. In our case, a thick bony atretic plate was present anteriorly in place of the nostrils and ophthalmic structures were absent.

In summary, congenital arhinia is a rare defect of embryogenesis often associated with other anomalies that significantly influence the immediate as well as long-term outcome of the neonate. It is a potentially life-threatening condition and requires the presence of a highly skilled neonatal resuscitation team at the time of delivery. Parental counseling is vital and a multidisciplinary team approach is required to optimize neonatal outcome.

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