

Prenatal diagnosis of total arhinia

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ABSTRACT

Complete or total arhinia, in which there is absence of the soft tissue of the nose, is extremely rare. The embryological origin of the defect is thought to be maldevelopment of the paired nasal placodes. Available neonatal case descriptions have reported the frequent coexistence of other facial anomalies. This report is the first to describe the antenatal diagnosis of total arhinia confirmed after delivery. Sonographic views of the fetal profile, showing an absence of the nose led to consideration of the diagnosis.

INTRODUCTION

Total arhinia, defined as the complete absence of the soft tissue of the nose with/without associated abnormalities is an exceedingly rare birth defect. In a recent report, Meyer identified fewer than 20 cases of surviving infants with arhinia in the medical literature¹. In all of the prior reports, the diagnosis of arhinia was made in the neonatal period^{1–4}. This report is the first to describe the prenatal diagnosis of total arhinia. A review of the embryological origins of this rare birth defect and of the sonographic features is provided.

CASE REPORT

JP was a 28-year-old G4 P2 Hispanic female initially referred for ultrasound dating and anatomical survey at 18 weeks' gestation. Initial targeted ultrasound revealed fetal size consistent with dates and normal anatomical survey although the fetal face/profile could not be visualized due to fetal position. Gestational diabetes was diagnosed in the late second trimester, and subsequently a follow-up ultrasound for growth at 29 weeks was performed. Examination of the fetal face was markedly abnormal. On a midline sagittal section of the fetal face, the upper lip appeared prominent and the profile was flattened (Figure 1). No soft tissue structures of the fetal nose could be visualized between the upper lip and forehead. Coronal sections of the fetal face confirmed

absence of the soft tissue of the fetal nose (Figure 2). The fetal lips appeared intact without cleft, and although the fetal orbits appeared subjectively small, the inner orbital distance was normal for gestational age. On deep coronal sections of the face, it appeared that the bones of the nasal cavity were present. The fetal intracranial anatomy revealed normal ventricles and midline structures (Figure 3). There were no other fetal abnormalities noted. Although no other fetal markers of aneuploidy were identified, she was offered, but declined amniocentesis for fetal karyotyping. The patient was followed with serial third trimester examinations which revealed an estimated fetal weight above the 90th percentile together with increased amniotic fluid volume (AFI: 22 cm). Referral to a regional hospital with a pediatric craniofacial team for consultation and delivery was arranged.

Vaginal delivery occurred without complications at 38 weeks gestation following the spontaneous onset of labour. The male infant was vigorous at delivery, APGAR scores were recorded of 8 and 9. Birthweight was 8 pounds 15 ounces. Clinical evaluation revealed arhinia, requiring placement of an oral airway. On physical examination, the cranial vault appeared normal. The ears were well formed and in normal position. Eye examination revealed microphthalmia and microcornea. The entire nasal bridge and external nares were absent (Figure 4). No apparent piriform recess was seen. The lips were intact without cleft; the soft and hard palates were normal. The remainder of the neonatal exam was unremarkable. A CT scan on day 1 showed absence of the cribriform plate, intact bone below the nasal area and no nares. A MRI scan performed on day 8 showed a grossly normal brain. On Day 3, intubation was required because of worsening oxygen saturations and hypotension. A diagnosis of probable sepsis was made and the infant responded well to IV antibiotics. The infant was extubated on Day 5 and maintained on an oral airway. The infant was able to tolerate short periods without the oral airway.

Following discharge, the baby exhibited episodes of cyanosis requiring readmission and subsequent tracheotomy placement at 11 weeks of age. Following the

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Figure 1 Sagittal section of fetal head showing the facial profile. Note the prominent upper lip and the total absence of the nose.

establishment of the airway, the infant has been stable. The baby is being fed via a G tube until the time when the oral airway can be removed. In an effort to avoid complications due to mid-face development, surgical nasal reconstructive surgery will be delayed until 5–6 years of age.

DISCUSSION

The prenatal detection of abnormalities of the fetal nose have been described previously, most frequently in the setting of fetal holoprosencephaly with or without an underlying chromosomal abnormality (trisomy 13)^{5,6}. In such cases a rudimentary nose or proboscis can be seen. Likewise in Binder Syndrome (maxillonasal dysplasia), the nose is normally positioned although hypoplastic⁷. In contrast, the total lack of any nasal tissue combined with normal intracranial anatomy led us to consider total arhinia. Although neonatal case descriptions have been reported, no prior documentation exists describing the prenatal diagnosis of this condition.

In his review of arhinia, Cohen *et al.* provided an extended classification system for arhinia⁴. In total arhinia, the entire nose and olfactory bulbs are lacking whereas in



Figure 2 Coronal section of the fetal face showing a small dimple (arrow) in the region normally occupied by the nose.



Figure 3 Neonatal profile revealing total absence of the soft tissue of the nose.

partial arhinia, at least one nostril and olfactory nerve is present. Cases without other structural malformations are considered pure. In the previously reported cases of arhinia other abnormalities of the face, eyes, ears or central nervous system have been seen⁴. Although chromosomal analysis was unavailable in many of the previously reported cases, abnormalities of chromosome 9 (mosaic trisomy, inversion) were present in two cases. Neonatal survival is strongly dependent on the presence and severity of associated anomalies⁴.

An understanding of the origins of arhinia can be gained from a study of the normal embryology⁸. At the end of the fourth week, the paired nasal placodes develop from ectodermal thickenings on the frontonasal prominence. During the fifth week, mesenchymal proliferation at the placode margins lead to the formation of horseshoe shaped elevations called the medial and lateral nasal prominences, the primordia of the external nose. The nasal placodes, now lying within the created depression, are referred to as nasal pits. With progressive in-growth of the nasal pits, nasal sacs are formed which extend ventro-dorsally to form the primordia of the nasal cavity. Some cells within the nasal placodes differentiate into the primary neurosensory cells of the future olfactory bulb. By the end of the fifth



Figure 4 Neonatal image showing intact lips, absent nose and a small dimple corresponding to that seen prenatally.

week, these cells develop axons that extend to connect with the most cranial portion of the telencephalon. In the sixth week, the nasal prominences merge towards the midline with the medial prominences fusing to form the bridge and septum of the nose. The unfused lateral prominences form the alae of the nose. In the area where the axons of the primary neurosensory cells contact the cerebral hemispheres, outgrowths called the olfactory bulbs form that contain the secondary neurosensory cells. With differential growth of the fetal brain and face, the distance between the olfactory bulbs and cerebrum increases, thus forming the olfactory tracts. In arhinia, lack of development of one (partial) or both (total) nasal placodes results in failure of growth of the lateral and medial nasal prominence resulting in absence of the soft tissue of the nose on the affected side. Secondly, lack of development of the nasal sac leads to absence of the olfactory bulb and tract.

We believe this case report is the first to describe the prenatal diagnosis of total arhinia. The diagnosis is best established on a midline, sagittal section of the fetal face (fetal profile) which reveals a striking absence of the soft tissue of the fetal nose and a prominent upper lip. The finding can easily be missed on a coronal section of the fetal face. A thorough sonographic evaluation of the fetus is indicated, with close attention being paid to the intracranial anatomy to rule out holoprosencephaly. When absence of the fetal nose is seen in the presence of normal intracranial anatomy, a diagnosis of total arhinia should be entertained. Other fetal sonographic markers

for aneuploidy should be sought and amniocentesis for fetal karyotyping considered. Consultation with appropriate neonatal craniofacial personnel to discuss early neonatal as well as long-term medical and surgical management is indicated. Delivery should occur in a hospital with specialized neonatal services and all well-organized craniofacial team to ensure appropriate neonatal stabilization and long-term paediatric follow-up including nasal reconstruction.

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