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Antenatal ultrasound findings in choanal atresia: A case report and review of the literature

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ABSTRACT

Choanal atresia occurs in about 1 in 5000 births and is associated with other structural and genetic abnormalities. Choanal atresia is usually diagnosed postnatally due to respiratory distress, and rarely diagnosed antenatally. Here, a woman with severe polyhydramnios is described, whose fetus was diagnosed antenatally with isolated bilateral choanal atresia, as evident by persistent absence of flow through the nostrils on ultrasound. A literature review is presented of the antenatal findings of choanal atresia, using ultrasound and other imaging modalities. An association of choanal atresia with polyhydramnios should be considered. Examining flow through the fetal nose, using color Doppler, might aid in diagnosing choanal atresia. If this condition is suspected, a detailed ultrasound scan should be done to rule out other anomalies. Fetal magnetic resonance imaging has been suggested as an additional imaging tool in selected patients. Genetic counselling and invasive prenatal testing should be offered.

1. Introduction

Choanal atresia (CA) is an uncommon craniofacial malformation that occurs in about 1 in 5000 live births. It leads to blockage in the connection between the nasal airway and the nasopharynx, most often due to combined membranous and bony atresia [1]. CA can be unilateral or bilateral; it can be isolated or associated with other conditions, such as CHARGE syndrome (Coloboma, Heart defect, Atresia choana, Retarded growth and development, Genital hypoplasia, Ear anomalies/deafness), Treacher Collins syndrome or abnormal karyotype. The pathogenesis of CA is poorly understood and may involve several genetic pathways [1].

Antenatally diagnosed CA has been reported sparsely in the literature. Bilateral atresia is usually diagnosed postnatally, after the presentation of symptoms of respiratory distress or feeding difficulties. Unilateral CA typically presents later, with unilateral nasal discharge or stuffiness; the diagnosis is usually delayed or even missed [2].

This report describes an antenatal diagnosis of isolated bilateral CA, which was confirmed postnatally. A review was performed of reports on antenatally suspected CA that were confirmed by postnatal diagnosis. To this end, a PubMed computer search using the terms "choanal atresia" and "antenatal diagnosis" was done. This was completed by manual searching for references of reports and review articles for relevant case reports.

2. Case Presentation

A 22-year-old healthy woman presented at 30 + 2 weeks of gestation with severe polyhydramnios. Her medical history revealed no abnormal findings. She had had one previous uncomplicated gestation and a normal vaginal delivery of a healthy baby at term. Ultrasound (US) scan showed an amniotic fluid index of 42 cm. The estimated fetal weight was equivalent to 31 weeks of gestation. There were no placental abnormalities. Neither structural abnormalities nor hydrops of the fetus were confirmed. A color Doppler (CD) study revealed normal swallowing movements and persistent absence of flow through both fetal nostrils during a prolonged scan (Fig. 1A and B). A pattern of shallow breathing movements was detected. Other parameters of the biophysical profile were normal. Fetal echocardiography revealed no arrythmia or structural abnormalities. A 100-g oral glucose tolerance test was normal. Serological tests for TORCH (toxoplasma, rubella, cytomegalovirus, herpes virus), parvovirus and syphilis revealed no recent infections.

Abbreviations: CA, choanal atresia; CD, color Doppler; CT, computed tomography; MRI, magnetic resonance imaging; US, ultrasound.

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Fig. 1C and D show the pattern of normal CD flow through fetal nostrils in another pregnant woman with isolated polyhydramnios.

In follow-up visits, US scans showed moderate to severe polyhydramnios and the same above-mentioned US findings. As the patient did not exhibit any symptoms, such as dyspnea or preterm contractions, amnioreduction was not performed. Despite the recommendation of the medical team, the patient refused genetic counselling and invasive prenatal testing. She eventually presented to the delivery room at 34 + 3 weeks with preterm labor, and had an uncomplicated vaginal delivery. A male baby weighing 2462 g was delivered with an Apgar score of 8/7. The newborn had intermittent attacks of cyanosis and respiratory distress soon after birth, and the small feeding tube failed to pass through the posterior choana. A computed tomography (CT) scan revealed bilateral CA dominated by membranous obstruction (Fig. 2). The absence of other abnormalities excluded CHARGE syndrome.

At age two days, the infant underwent transnasal endoscopic repair of the obstructed choana under synchronized intermittent mandatory ventilation. There were no intraoperative or postoperative complications and he was discharged in good condition.

3. Discussion

Definitive CA diagnosis is established postnatally by the inability to pass a nasal catheter to the pharynx. Antenatally, CA can be suspected in the presence of nose anomalies, mainly nasal septal deviation or the presence of a single nostril [1]. Bronshtein et al. [2] reported four cases of CA suspected antenatally by US in the early second trimester, which were confirmed postnatally. Three were unilateral CA and one was bilateral CA. A transient large nasal cavity was detected by routine US anatomy scan at 14–17 weeks in all four fetuses. Accumulated fluid in the obstructed cavity had apparently led to deviation of the nasal septum. Thus, the authors concluded that transient fluid collection appearing within the obstructed nasal cavity in early US is a sign of CA.

Biard et al. reported using CD US to examine inferior nasal patency during fetal breathing movements in suspected CA [3]. In one fetus, CA was suspected due to the absence of CD flow through the choana, using coronal and parasagittal planes of the face, but the diagnosis was not confirmed at autopsy. Nevertheless, CD is used in other etiologies of fetal upper airway obstruction. Using CD in the transverse plane, Bardin et al.

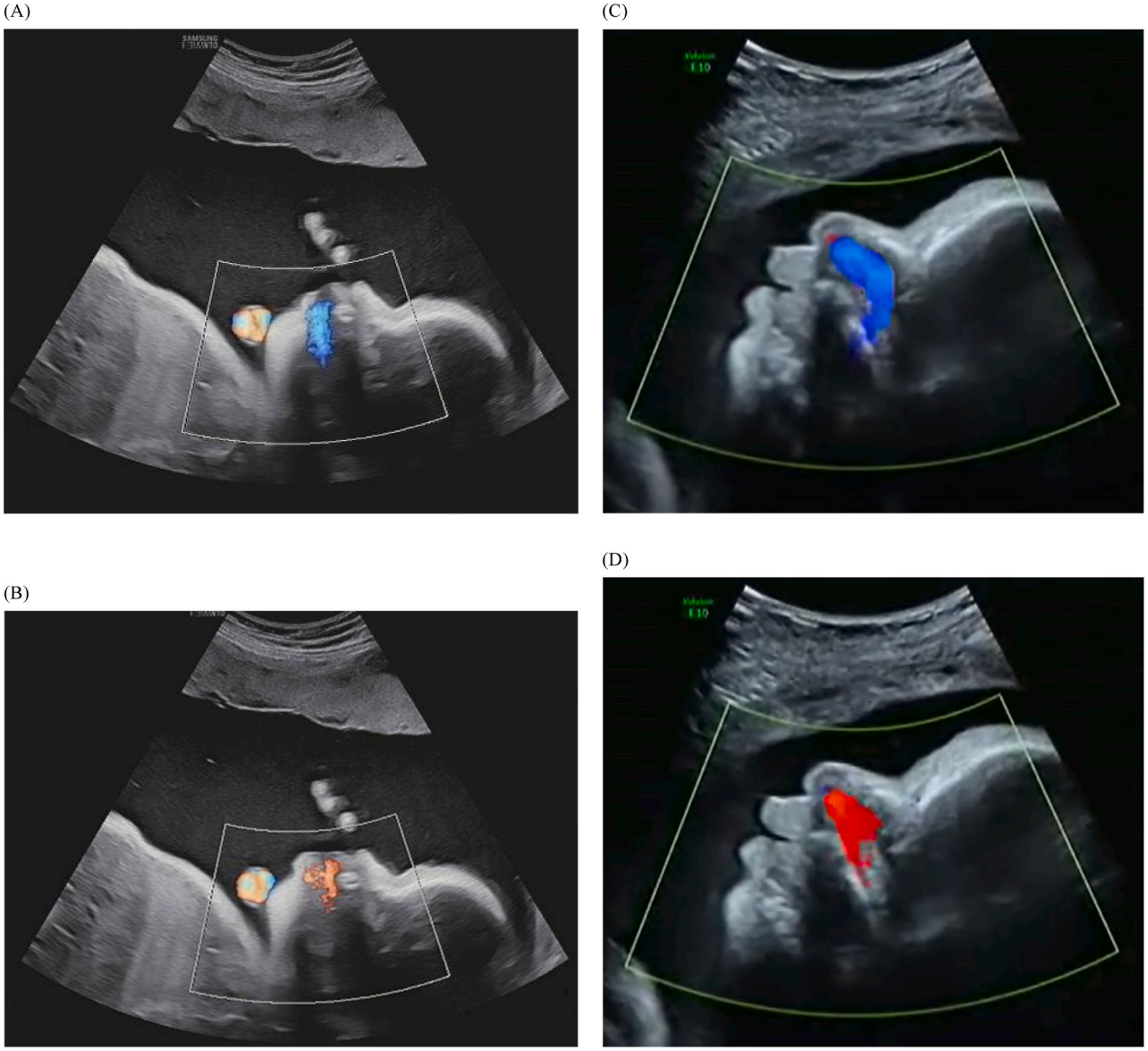

Fig. 1. Color Doppler ultrasound illustrating persistent absence of flow through the nostrils. Note the normal color Doppler flow through the mouth (A) during inspiration and (B) expiration. Another fetus in which color Doppler ultrasound illustrated normal flow through the nostrils during (C) inspiration and (D) expiration in a woman at 36 weeks+6 days of gestation with isolated polyhydramnios.

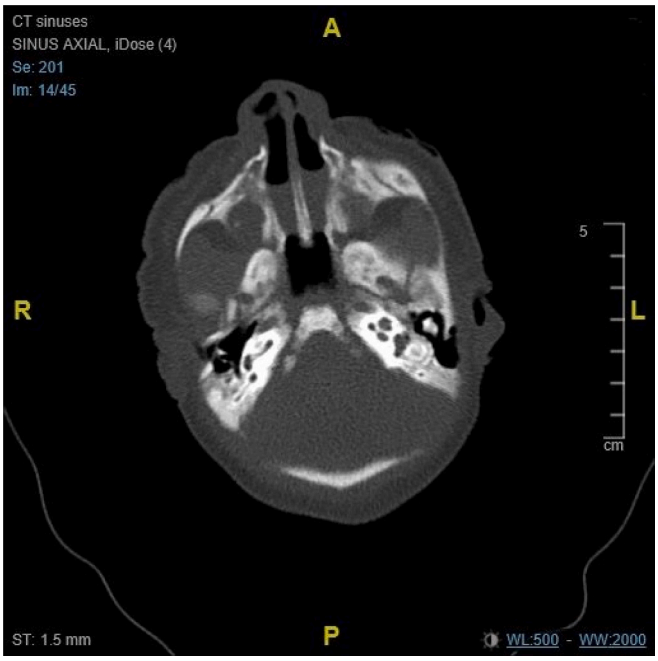

Fig. 2. Bilateral membranous choanal atresia.

[4] demonstrated a patent nasal airway on the left, and absence of flow in the right obstructed nasal airway, due to a dacryocystocele. They suggested that CD be implemented as a simple method also for prenatal diagnosis of CA.

A literature search did not find any reports of isolated CA associated with polyhydramnios or abnormal fetal breathing movements. However, nasal airway obstruction might lead to polyhydramnios, due to the interference of the amniotic fluid exchange. Salvetet et al. [5] reported bilateral dacryocystocele associated with polyhydramnios. The cyst intranasal extension might lead to an obstruction and disrupted amniotic fluid regulation. Polyhydramnios reported in CHARGE syndrome could be attributed primarily to the severe malformations accompanying this syndrome rather than CA. The obstructed nasal cavities might lead to abnormal shallow breathing movements, as were observed in the described fetus, as fetuses are obligate nose breathers.

Other imaging modalities have been suggested for suspected CA. Choanal evaluation by CT scan was described in one fetus with CHARGE syndrome [3]. CA was suspected in an antenatal CT scan and subsequently confirmed after birth. Typical changes in the nasal cavity on the atretic side on the CT scan include medialization of the lateral nasal wall on the level of the choana, and thickening of the posterior part of the vomer and the medial plate of the processus pterygoideus [6]. Yet, utilization of antenatal CT scans is highly restricted, and its contribution to antenatal diagnosis of CA is unclear. The presence of a septum in the hyposignal T2 in sagittal or axial planes on fetal magnetic resonance imaging (MRI) has been reported as a sign of CA [7]. Nevertheless, MRI lacks optimal resolution; optimizing technical parameters such as using 2-mm-thinner sections could improve its accuracy [7].

CA may present either as an isolated congenital anomaly or as a part of a craniofacial malformation such as CHARGE, Treacher Collins, Pfeiffer, Apert, mandibulofacial dysostosis, and Crouzon syndromes. CA associated with methimazole embryopathy was also reported [8]. Among 129 individuals with CA, CA was an isolated finding in 34 (26.4%), and associated with other anomalies in 95 (73.6%) [9]. CHARGE syndrome was the most common diagnosis (33 patients (25.6%)). CHARGE syndrome is a rare multiple anomaly syndrome caused by mutations in the CHD7 gene [3]. Antenatal diagnosis of CHARGE syndrome is uncommon. Busa et al. [10] reported prenatal US findings of 12 children with CHARGE syndrome, of whom three were diagnosed postnatally with CA. CA was suspected antenatally in only one, although US findings were not reported by the authors. Given that CA is frequently associated with other fetal anomalies and genetic disorders, especially CHARGE syndrome [9], antenatal suspicion of CA should prompt offering genetic testing.

4. Conclusion

A woman with severe polyhydramnios was described, whose fetus was suspected antenatally of having CA, due to the detection on US of persistent absence of flow through both nostrils. Although an uncommon condition, CA should be ruled out in women with polyhydramnios, as the latter may indicate upper airway obstruction. In this scenario, flow through the nostrils should be assessed using CD during fetal breathing movements. Additional indicators that suggest CA include nasal septal deviations, the presence of a single nostril and temporarily enlarged nasal cavities. Other potential causes of upper airway obstruction should be explored, such as obstructive masses and dacryocystocele. Once CA is suspected, a detailed US scan should be done to rule out other anomalies, especially those associated with CHARGE syndrome. MRI can serve as an additional imaging tool in selected circumstances. Genetic counselling and invasive prenatal testing should be offered.

Contributors

Inshirah Sgayer contributed to the conception of the case report and patient care, drafted the manuscript, undertook the literature review, and revised the article critically for important intellectual content.

Lior Lowenstein contributed to the literature review, and revised the article critically for important intellectual content.

Marwan Odeh contributed to patient care and the literature review, and revised the article critically for important intellectual content.

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Patient consent

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This article was not commissioned and was peer reviewed.

Conflict of interest statement

The authors declare that they have no conflict of interest regarding the publication of this case report.

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