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Case Report

Late onset of large benign ductus arteriosus aneurysm presented with increased nuchal translucency and cystic hygroma at first trimester Down syndrome screening

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ABSTRACT

Objective: Fetal ductus arteriosus aneurysm (DAA) is a rare but potentially risky congenital heart disease. It is often not diagnosed until the third trimester because of its asymptomatic nature and late onset. In rare occasions, DAA may result in serious complications; therefore, prenatal diagnosis is helpful.
Case Report: Herein, we report the case of a foetus with cystic hygroma and increased nuchal translucency in the first trimester (but regressed at 20-week anomalous scan). Karyotyping indicated a 46 XY genotype. A large vascular mass was noted at the apex of the left lung by Doppler ultrasound at 38 weeks of gestation, with a diameter of 12.5 mm. After birth, echocardiography showed a patent ductus arteriosus with aneurysmal dilatation (17 mm as the largest diameter); thus, DAA was impressed. Chest computed tomography and three-dimensional angiography confirmed the large aneurysmal dilatation of the ductus arteriosus with a closed end at the pulmonary arterial side.
Conclusion: The male infant survived, but presented mild respiratory distress at birth. He was discharged at 24 days of age. At that time, DAA had regressed partially (diameter of 8.5 mm and much less blood flow), and it fully regressed at 40 days of age.
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Introduction

Fetal ductus arteriosus aneurysm (DAA) is a rare but potentially risky congenital heart disease [1]. An incidence of approximately 1.5–8.8% is reported [2]. Usually, DAAs are diagnosed in the third trimester, and a tortuous ductus arteriosus is excluded. This condition is not always diagnosed in utero because of its asymptomatic nature and late onset [3,4]. In rare occasions, DAA may result in serious complications, such as thrombus formation, infection, rupture, and even mortality [5–7]; hence, it cannot be overlooked and a prenatal diagnosis is beneficial.

Case Presentation

A 28-year-old female, gravida 3, para 1, spontaneous abortion 1, was transferred to our department for thickened nuchal fold at 13^{+0} weeks of gestational age. A cystic hygroma of ~7.6 mm in diameter was noted without skin oedema or fetal hydrops. The cystic hygroma did not progress. Therefore, we arranged karyotyping by chorionic villus sampling and the result indicated a 46 XY genotype. The patient underwent uneventful antenatal examinations until 38 weeks of gestation. A large vascular mass at the apex of the left lung field was incidentally found and the Doppler ultrasound showed an anechogenic cyst with turbulent flow at the apex of the lung with the largest diameter as 12.5 mm (Figure 1).

A 3562 g male baby was born by caesarean section at 40 weeks of gestation. The Apgar score was 7 (at 1 minute) and 9 (at 5

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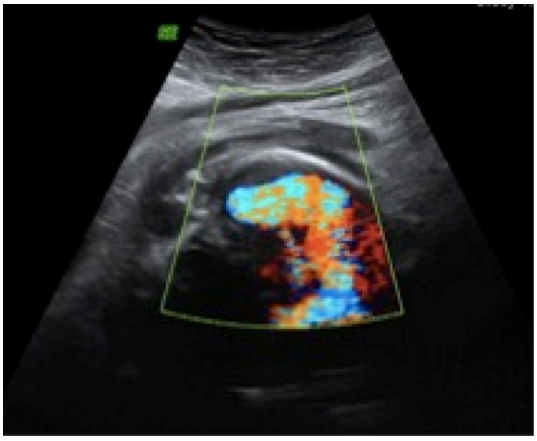

Figure 1. Vascular structure with a turbulent flow under color Doppler ultrasound.

minutes). Immediately after birth, tachypnea, mild subcostal retraction, and desaturated pulse oximetry were noted. Respiratory support with O_2 using a nasal cannula was provided and was well tolerated by the infant. Moreover, an echocardiography was performed on the 2nd day after birth. It showed a patent ductus arteriosus with the largest diameter as 17 mm and an aneurysm near the upper part of the descending aorta. Computed tomography was performed, and it showed a large ductus aneurysm with a diameter of 8.5 mm and length of 17 mm (Figures 2A and 2B). A three-dimensional angiography clearly revealed an aneurysmal dilatation without compression onto the adjacent structures (Figures 2C and 2D); thus, DAA was diagnosed. The male infant was discharged after the respiratory condition was stabilized, and the follow-up echocardiography showed spontaneous closure at 40 days of age.

Discussion

DAA develops during late gestation. The theories of pathogenesis that have been reported include delayed closure at the aortic end, congenital wall weakness, increased velocity through the ductus arteriosus and connective tissue disease [1]. None of these hypotheses can explain the aneurysm formation in all conditions; thus, the aetiology remains elusive. According to the latest studies [2,3], DAA is not as rare as previously thought. Jan et al [2] reported a DAA incidence of 8.8% by a prospective study in 2002, and Tseng et al [3] found a similar incidence (8.1%) in 2005, although neither of them were remarkably large incidences.

DAA usually has no symptoms, except occasionally it may present with respiratory stress and a heart murmur may occur. The complications include infection, rupture, thromboembolism, and

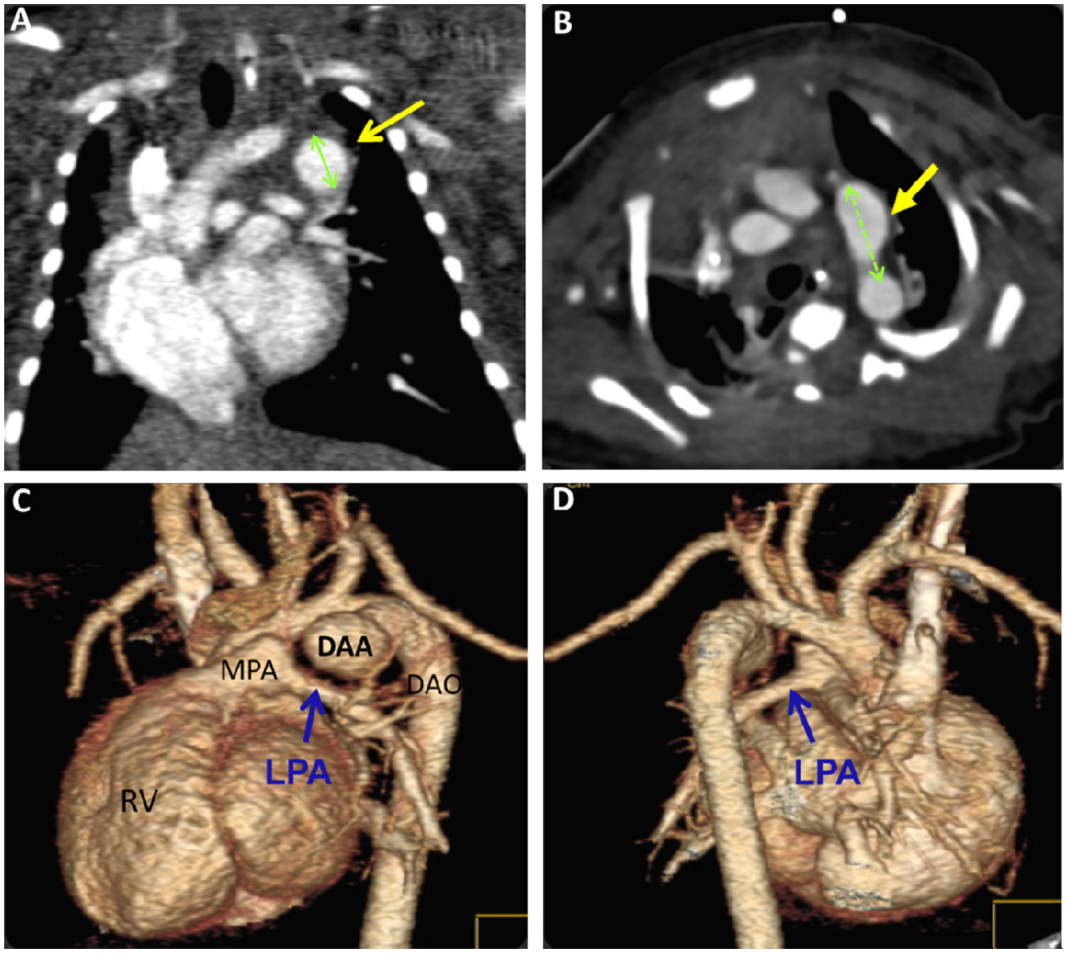

Figure 2. (A) Computed tomography shows a large DAA with a diameter of 8.48 mm; (B) DAA with a length of 17.02 mm; (C, D) three-dimensional computed tomography showing DAA arising from the descending aorta with a closed end at the pulmonary artery side. No compression of the adjacent structure. DAA = ductus arteriosus aneurysm; DAO = descending aorta; LPA = left pulmonary artery; MPA = main pulmonary artery; RV = right ventricle.

symptoms/signs related to compression onto the adjacent structures. However, a prospective study depicted that most DAAs are asymptomatic, and complications are very rare [2]. Tseng et al [3] reported that the foetuses with DAA had lower Apgar scores, but all cases encountered spontaneous regression within 2 months postdelivery with only expectant management. Tortuous ductus arteriosus should be excluded before diagnosis [8,9]. Herein, we report the case of an infant who had a huge aneurysm (diameter of 12.5 mm) and mild acute respiratory insufficiency but no other complications, and the outcome was favorable as those reported in previous literature [10].

Notably, it is well recognized that the incidence of congenital heart disease increases with increased nuchal translucency [11]. A large cohort study showed that nuchal translucency measurement of 2.5 times the median values or more (99^{th} percentile) in a foetus without aneuploidy is a marker for congenital heart disease and needs referral for fetal echocardiography [12]. We report this late onset case of DAA with cystic hygroma that was noted in the first trimester of pregnancy, which regressed spontaneously, but the thickened nuchal fold remained. Karyotyping was normal, and the targeted fetal echocardiography performed in the second trimester showed normal structures.

Although DAA may be a normal variant of patent ductus arteriosus [3] with favorable outcomes, rare complications may sometimes occur, and it is better to diagnose this condition before delivery so that neonatologists can be well prepared. Sporadic reports in the literature had described that DAA was not diagnosed until very late gestation [3,13]. In this case study the targeted 20-week fetal anomalous echocardiography scan revealed no structural anomalies. Therefore, clinicians should consider the possibility of late-onset DAA by examining the three-vessel view in the fetal upper chest during late gestation (i.e., in the 3^{rd} trimester), especially when thickened nuchal translucency has been previously noted (i.e., in the 1^{st} trimester) [4].

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

Acknowledgments

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