Congenital cystic adenomatoid malformation

Mary Norris

Nepean Hospital, Penrith New South Wales, Australia
Correspondence to Mary Norris via ASUM. Email author@asum.com.au

Examples of Congenital Cystic Adenomatoid Malformation under ultrasound.

Introduction

Congenital cystic adenomatoid malformation (CCAM) is an uncommon fetal lung anomaly involving cystic changes to the terminal bronchioles. The condition requires close monitoring during the antenatal period with ultrasound in addition to input from the neonatal and paediatric surgical teams. This case study involves a 27-year-old female, G2P1, who was referred by her obstetrician to the Maternal Fetal Medicine Unit (MFM-U) with a morphology scan finding of a cystic mass in the left hemi thorax, most likely a CCAM. The report described a multicystic mass in the left chest occupying at least 50% of the left hemi-thorax. The right lung appeared normal as did the fetal heart and no other anomalies were detected at this initial scan.

The morphology scan was repeated at the MFM-U at 20 weeks gestation. This scan confirmed an isolated left-sided Type 1 (macro cystic) CCAM with evidence of mediastinal shift. In addition to these findings, an isolated choroid plexus cyst (CPC) and an echogenic focus in the left cardiac ventricle were also identified. Fetal anatomy was otherwise normal. At this stage the patient was counselled about the variable prognosis of CCAM, which primarily is dependent on whether fetal hydrops (heart failure) develops. At this pre-viable gestational age, the patient was counselled that the cysts could be drained in the event of further enlargement with the aim of avoiding cardiac compromise and subsequent development of hydrops fetalis. In addition, while the findings of CPC and the echogenic intracardiac focus did increase this patient's risk of a chromosomal problem slightly (risk trisomy 21 = 1:270), the patient was counselled that chromosomal problems are not reported in association with CCAM thus making aneuploidy unlikely in this case. The patient declined further testing.

Ultrasound evidence of a large CCAM lesion and early mediastinal shift placed this pregnancy at very high risk for significant complications including non-immune hydrops. Accordingly, a clear management plan with multidisciplinary counselling was instituted early for the patient and her family including close ultrasound surveillance, at least every two weeks, for the monitoring of fetal well-being and the early detection of fetal hydrops and polyhydramnios. Counselling by the neonatology and paediatric surgical teams was organised to provide important information regarding management issues in the neonatal period. A fetal echocardiogram was performed at 24 weeks showing a structurally normal fetal heart with deviation of the fetal heart to the right side of the chest. In addition, the large left-sided complex lung cysts seen on imaging were not causing any compression of the cardiac chambers or causing any cardiac inflow obstruction.

While regular ultrasound scanning of the fetus did not identify any signs of fetal hydrops, gradually enlarging fetal lung lesions, persisting right mediastinal shift and polyhydramnios complicated the pregnancy. Growth and Doppler studies remained normal. In an effort to lessen the effect of mediastinal shift on normal lung development, in-utero thoracocentesis was performed on three occasions. In addition,

Examples of Congenital Cystic Adenomatoid Malformation under Doppler (left) and ultrasound (right).

the patient underwent amnioreduction on two occasions to relieve the discomfort associated with the polyhydramnios. The patient received a course of Betamethasone prior to the first invasive procedure.

A plan was made in conjunction with the neonatology and paediatric surgical teams to induce labour at 36 weeks. The patient had an uncomplicated labour and normal delivery of a live female infant weighing 2770 g with apgars of 6 and 7 at 1 and 5 minutes respectively. The baby was subsequently admitted to a Level 3 Neonatal Intensive Care Unit (NICU). Unfortunately, the baby developed severe respiratory distress during her admission to the NICU. At two days of age, the baby was taken to theatre for a left thoracotomy and excision of the large CCAM but died the same day. The cause of death was cited as hypoxia secondary to the CCAM, which was not amenable to surgery and supportive therapy.

CCAM is a rare developmental anomaly of the lower respiratory tract histologically characterised by an overgrowth of terminal respiratory bronchioles that forms cysts of various sizes and by a lack of normal alveoli[1]. The malformation occurs sporadically, is generally unilateral and there is no sex predilection. The blood supply is usually derived from the pulmonary circulation. Associated congenital anomalies are uncommon[2,3].

The widespread use of antenatal ultrasound has resulted in an increase in the prenatal diagnosis of CCAM with the majority of cases being detected by the 18–20 week fetal morphologic examination. While the reported incidence of CCAM varies in the literature, data obtained from the Western Australia Birth Defects Registry places the incidence of CCAM at approximately 1.2 per 10,000 births[4,5].

Sonographically, CCAM lesions are categorised according to appearance/cyst size (Stocker Classification) as outlined by Goldstein[2] and Sanders[6] into three types: Type 1 or macro cystic (2–10 cm cysts); Type 2 or mixed (cysts <2 cm); and Type 3 or micro cystic (not cystic, but echogenic areas in the fetal lungs). In all forms, colour flow Doppler sonography will show a normal blood supply from the pulmonary arteries. The differential diagnosis for CCAM includes pulmonary sequestration, bronchogenic cyst, diaphragmatic hernia, mediastinal cyst teratoma and congenital pulmonary emphysema[4,5,6].

It is well recognised in the literature that the majority of patients with CCAM detected antenatally have a good outcome, however, the course of the malformation in pregnancy can be variable[6,7]. While it is reported that up to 15–20% of CCAM lesions decrease in size during the gestation, there is consensus in the literature that prognosis depends on the size of the lung mass and the secondary pathophysiologic effects. The larger the thoracic mass the worse the prognosis tends to be[3,4,7,8,9,10]. Large CCAM lesions can compress the normal developing lung leading to pulmonary hypoplasia. The mass can also obstruct the oesophagus causing impaired fetal swallowing, polyhydramnios and preterm labour. Of more concern is the potential for the development of non-immune fetal hydrops and fetal demise secondary to vena caval obstruction and cardiac compression from large lesions causing extreme mediastinal shift[9,10]. In addition, careful maternal assessment for the development of Mirror Syndrome (preeclampsia) is essential in those cases with fetal hydrops[1,6,9].

Ultrasound evidence of a large CCAM lesion and early mediastinal shift, such as in this case, places the pregnancy at very high risk for significant complications including non-immune hydrops. Therefore, a clear management plan should be instituted for the patient and her family including close ultrasound surveillance to monitor fetal well being and the early detection of fetal hydrops and polyhydramnios. Counselling by the neonatology and paediatric surgical teams provides important information regarding management issues in the postnatal period including the eventual need for surgical excision of the CCAM lesion to avoid the long term risks of morbidity from infection, pneumothorax or, more rarely, malignancy[1,5,6,8,9,10,11].

In the event that fetal hydrops develops in the pregnancy, families need to clearly understand that fetal gestational age is an important factor dictating management[4,6,9]. After 32 weeks gestation, any evidence of fetal compromise should prompt steroid administration to enhance fetal lung maturity and early delivery[2,6,9]. Alternatively, pregnancies complicated by fetal hydrops and polyhydramnios prior to 32 weeks could consider in-utero thoracocentesis or a thoracoamniotic shunt if the mass is predominantly a single cyst. In-utero fetal surgery is restricted to specialised fetal therapy centres only[2,9,11].

In conclusion, CCAM detected antenatally requires a clear management plan with multidisciplinary input from maternal fetal medicine, neonatology and paediatric surgery. Prognosis ultimately depends on the size of the lung mass. The larger the lung lesion, the greater is the risk for pulmonary hypoplasia in addition to severe mediastinal shift and the development of non-immune hydrops. Close monitoring with ultrasound is a key component of care.

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