Prenatal diagnosis of Pleuropulmonary Blastoma as a differential diagnosis of congenital pulmonary airway malformation

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INTRODUCTION

Congenital Pulmonary Airway Malformations (CPAM) are a combination of conditions involving lungs, pleura and airway. Given the advance in prenatal diagnosis, we have been more exposed to this congenital malformation. We present a case of prenatal diagnosis, neonatal outcome and genetic study of a pleuropulmonary blastoma as a differential diagnosis of CPAM.

CASE REPORT

35 years old female, with a previous full-term pregnancy and past medical history of hypothyroidism after a total thyroidectomy due to nodular goiter. Current pregnancy with normal ultrasounds until the 32 weeks scan, findings: left pelvic kidney and dextrocardia due a thoracic mass with high vascularity at doppler (CVR 0.03cm2) (Figure 1). Fetal MRI showed ovoid unilocular cystic lesion with higher signaling in T2 than T1 surrounded by a capsule measuring 2,9x2,5x2cm (7,5cc volume). As possible diagnosis a bronchogenic cyst and CPAM were considered (Figure 2).

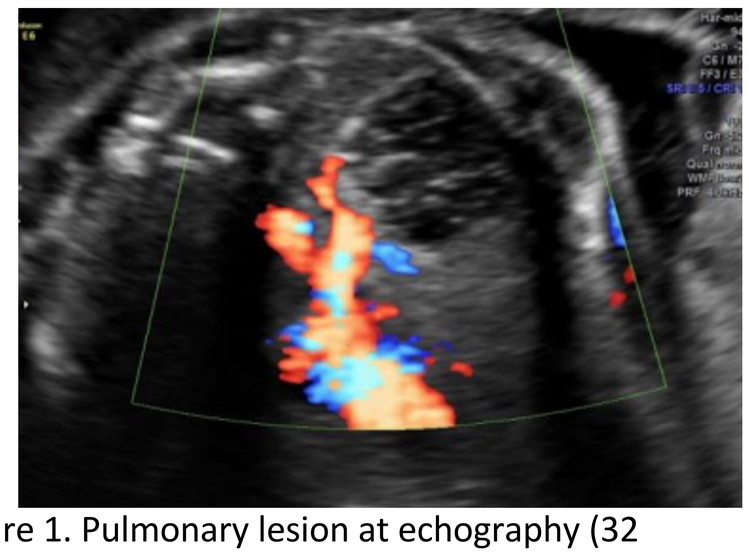

Figure 1. Pulmonary lesion at echography (32 weeks)

On ultrasound follow up, the mass increased its volume up to 6cm (58cc volume, CVR 1.6cm2). A new MRI was performed at 37 weeks. As new findings, presence of multiple cysts in the lesion and deviation of mediastinum were described. The MRI suggested a CPAM and less likely a pleuropulmonary blastoma as possibles diagnoses (Figure 3). The newborn was delivered at 38 weeks (by vaginal delivery), birthweight 3915g, 54cm, Apgar score 8-9, and was admitted to the neonatal care unit for study.

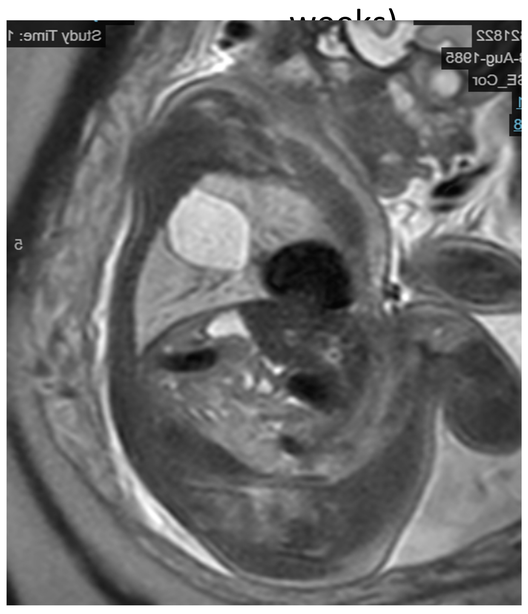

Figure 2. Fetal MRI, 32 weeks

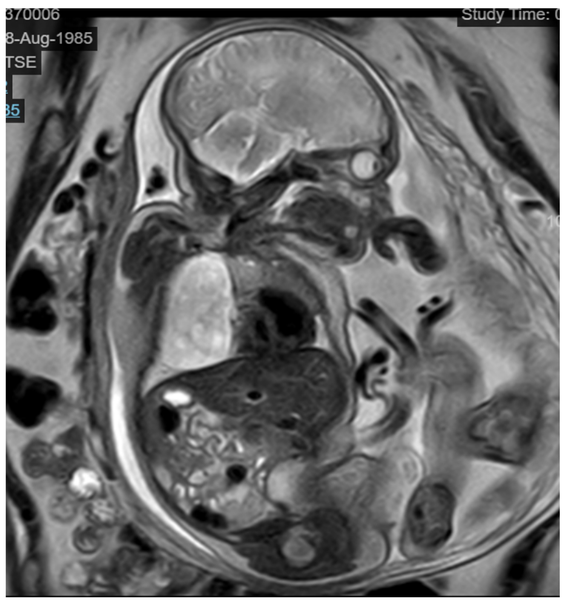

Figure 3. Fetal MRI, 37 weeks.

The newborn developed respiratory distress requiring intubation. An Angio CT was performed on day 7, reporting similar findings to the prenatal MRI (Figure 4). On day 9 a left superior lobectomy was performed. The evolution was satisfactory; on day 12th was successfully extubated, and on day 22 he was discharged home. Biopsy result of the surgical specimen report a type 1 pleuropulmonary blastoma. Due its association with specific mutations, genetic studies were performed: Normal karyotype (46, XY) and a pathogenic variant of the DICER1 gene was found on sequencing. Genetic testing was performed to the mother due to the association to multinodular goiter, and she was also found to have a heterozygous pathogenic variant of the DICER 1 gene (information of importance for subsequent pregnancies)

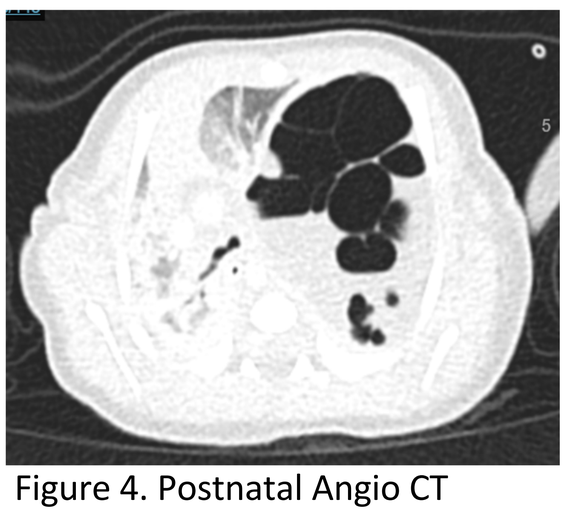

Figure 4. Postnatal Angio CT

At 3 month of age after follow-up, the patient required a second resection of pulmonary mass and chemotherapy treatment. He's been followed up for 2 years without recurrence nor other tumor according to DICER1 syndrome screening.

CONCLUSION

CPAM are malformations that involve lung, pleura and airway. Prenatal ultrasound findings have been described to predict its evolution (CVR). In our case, the patient presented with an unusual evolution, with an increase in the volume of the mass. The prenatal follow up is important, associated with the multidisciplinary approach with different specialists to analyze and find rare presentations and differential diagnosis.