

Deletion in the 15q11.2 region in a fetus with pulmonary agenesis: a case report

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OBJECTIVE

Pulmonary agenesis (PA) is a rare condition (24–34 per 1,000,000 live births) and the exact cause remains unknown. The clinical presentation of the disease is quite variable and most reported cases describe right agenesis. The objective is to report a case of left pulmonary agenesis diagnosed antenatally and its association with an abnormal microarray and the importance of a multidisciplinary follow up.

METHODS

The case reports a fetus with left lung agenesis associated to left multicystic dysplastic kidney, complex cardiac abnormality (heart axis to the left with backward-facing apex, parallel to the spine; double outlet right ventricle with tetralogy of Fallot; total anomalous drainage of the pulmonary veins and subaortic interventricular communication), vicariant right lung, right single umbilical artery and growth restriction diagnosed in the anomaly scan, cardiac scan, fetal MRI and postnatal exams.

RESULTS

After birth, prenatal findings were confirmed and other anomalies were also diagnosed. Vertebrae processes agenesis, left ear malformation, left facial muscles paralysis and vocal fold paresis were also reported and microarray test confirmed partial deletion of 476kb in the long arm of chromosome 15 (15q11.2) that carries gene NIPA1; and duplication of 393 kb in the long arm of chromosome 5 (5q14.3), that carries parts of gene ADGRV1.

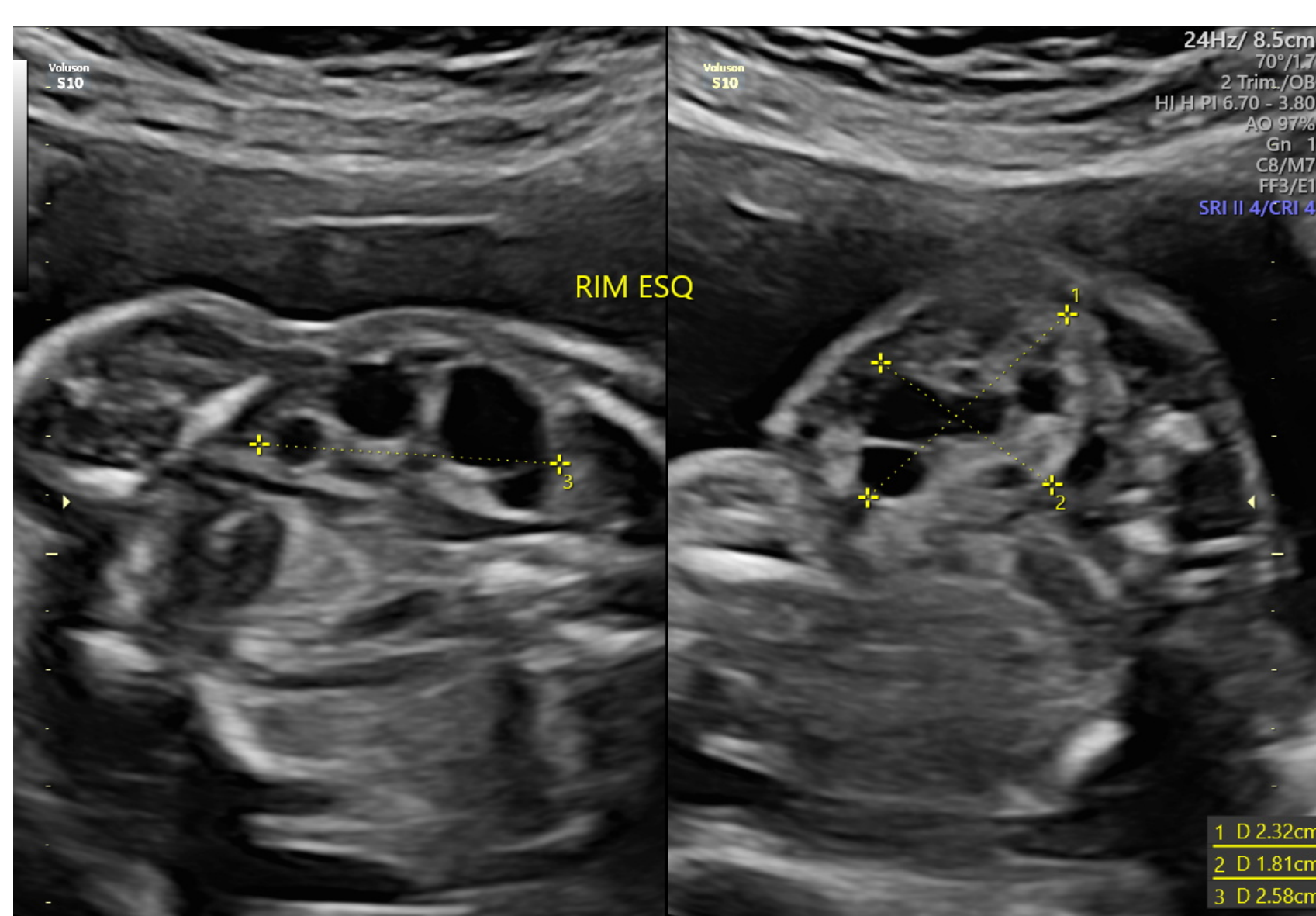


Figure 1. Left multicystic dysplastic kidney

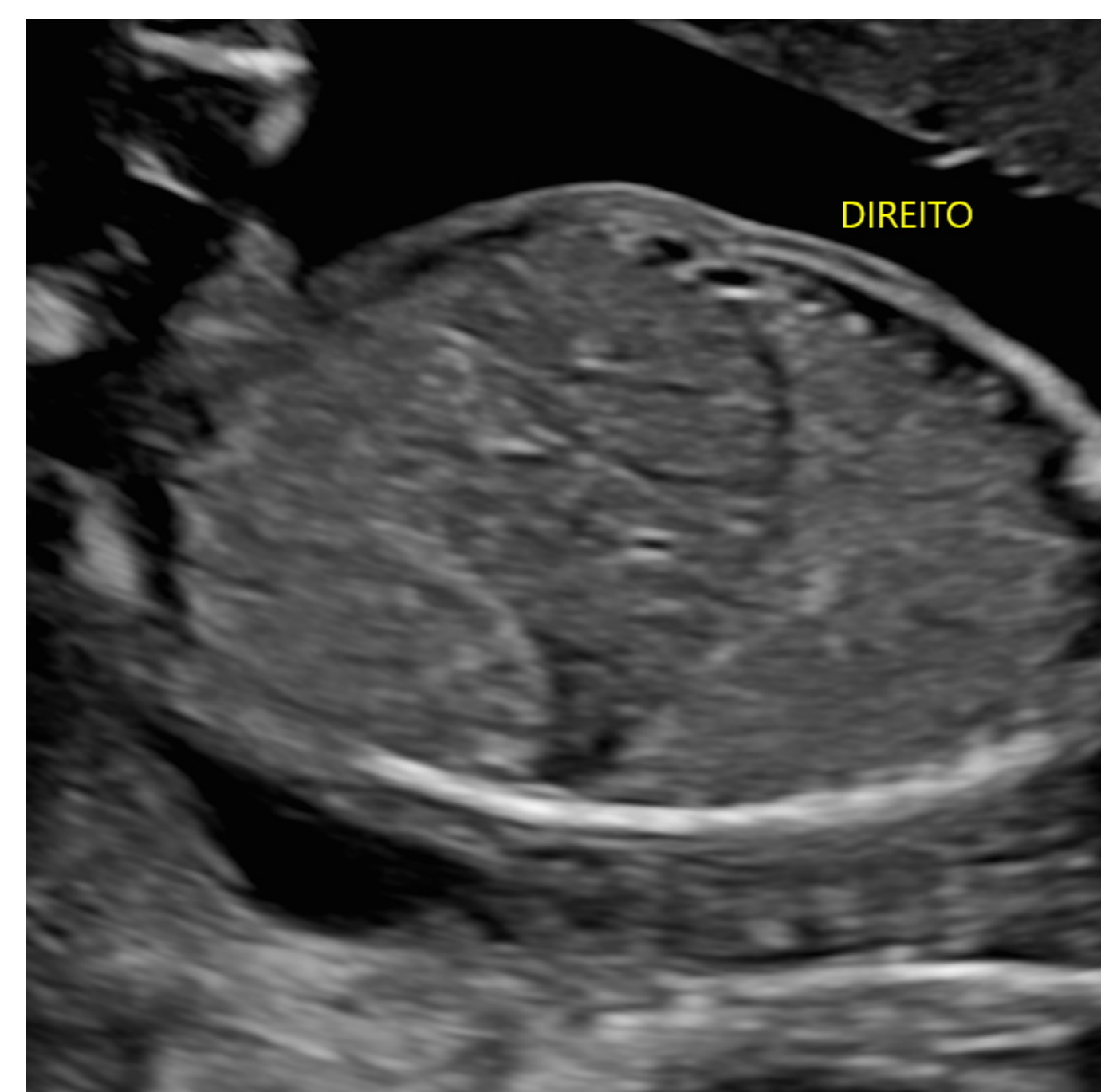


Figure 2. Vicariant right lung;

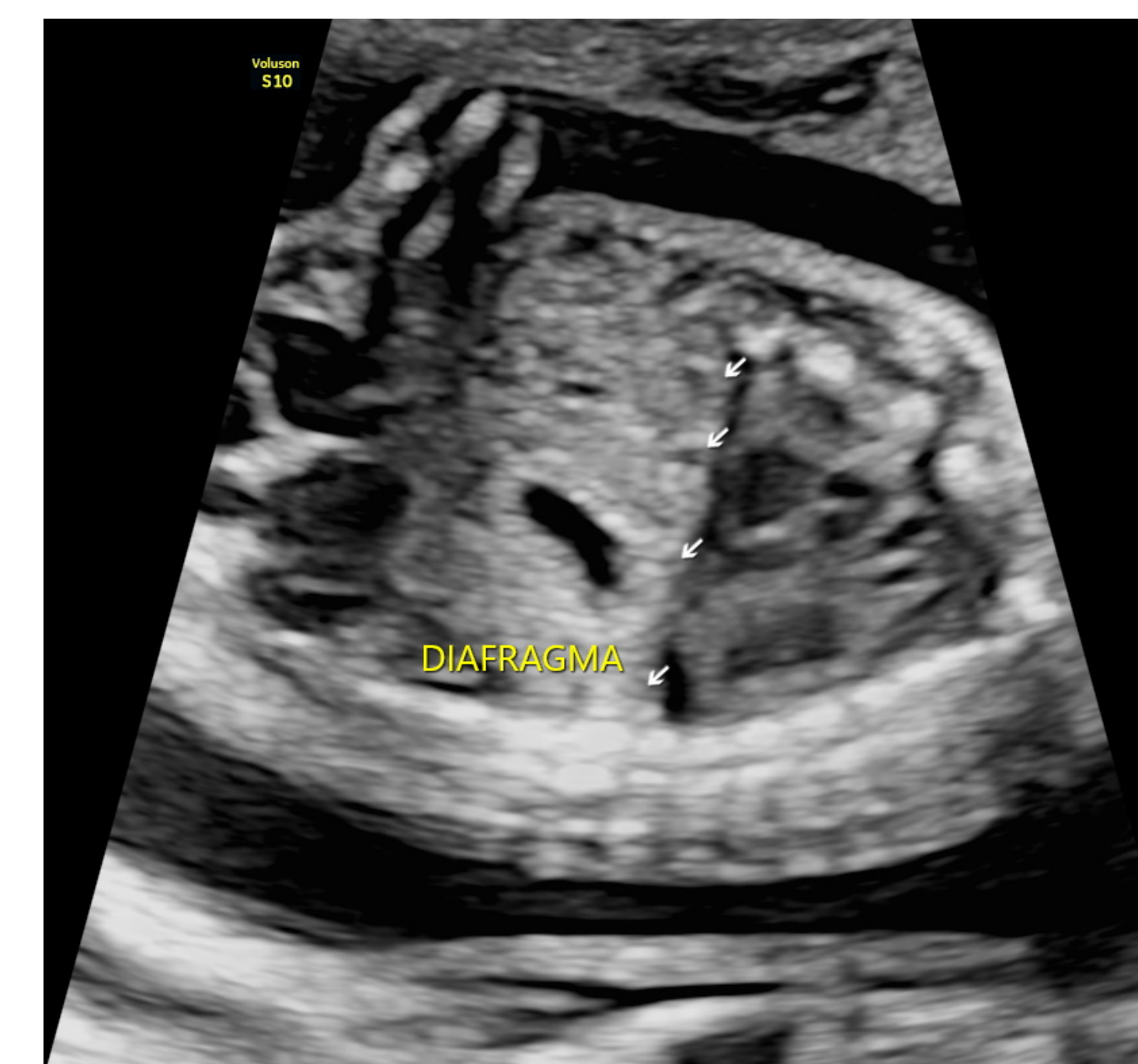


Figure 3. Arrows showing intact diaphragm and no left lung parenchyma

CONCLUSION

Although PA is a rare congenital anomaly, the search for an accurate diagnosis is critical for prenatal counseling and better postnatal management to reduce mortality and improve prognosis. Moreover, to our knowledge this is the first time that this microdeletion and duplication are associated with lung agenesis.

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2. Malcon MC, Malcon CM, Cavada MN, Caruso PEM, Real LF. Unilateral pulmonary agenesis . J Bras Pneumol. 2012;38(4):526-529