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Complex cerebral malformation in prenatal diagnosis: consider Joubert Syndrome

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Introduction

Congenital pulmonary airway malformation (CPAM) is a rare developmental anomaly of the fetal lung characterized by cystic or solid pulmonary lesions resulting from abnormal airway development. Prenatal ultrasound allows early detection and assessment of lesion size and potential complications.

Case

We report a case diagnosed at 15 weeks of gestation during routine prenatal ultrasound examination. Ultrasound revealed a large echogenic mass within the fetal thorax, consistent with congenital pulmonary airway malformation. The lesion occupied a significant portion of the fetal lung and was associated with mediastinal shift, raising concern for progressive thoracic compression (*Figure 1*).

Given the early gestational age at diagnosis and the severity of the lesion, detailed counseling was provided regarding prognosis and potential complications, including pulmonary hypoplasia and fetal hydrops. After multidisciplinary discussion and parental counseling, the patient opted for termination of pregnancy.

Fetopathological examination confirmed the presence of a congenital pulmonary airway malformation(*Figure 2*).

Conclusion

This case highlights the value of early prenatal ultrasound in detecting fetal thoracic malformations. Early diagnosis allows timely counseling regarding prognosis and management options, including close prenatal surveillance or termination of pregnancy in severe cases.

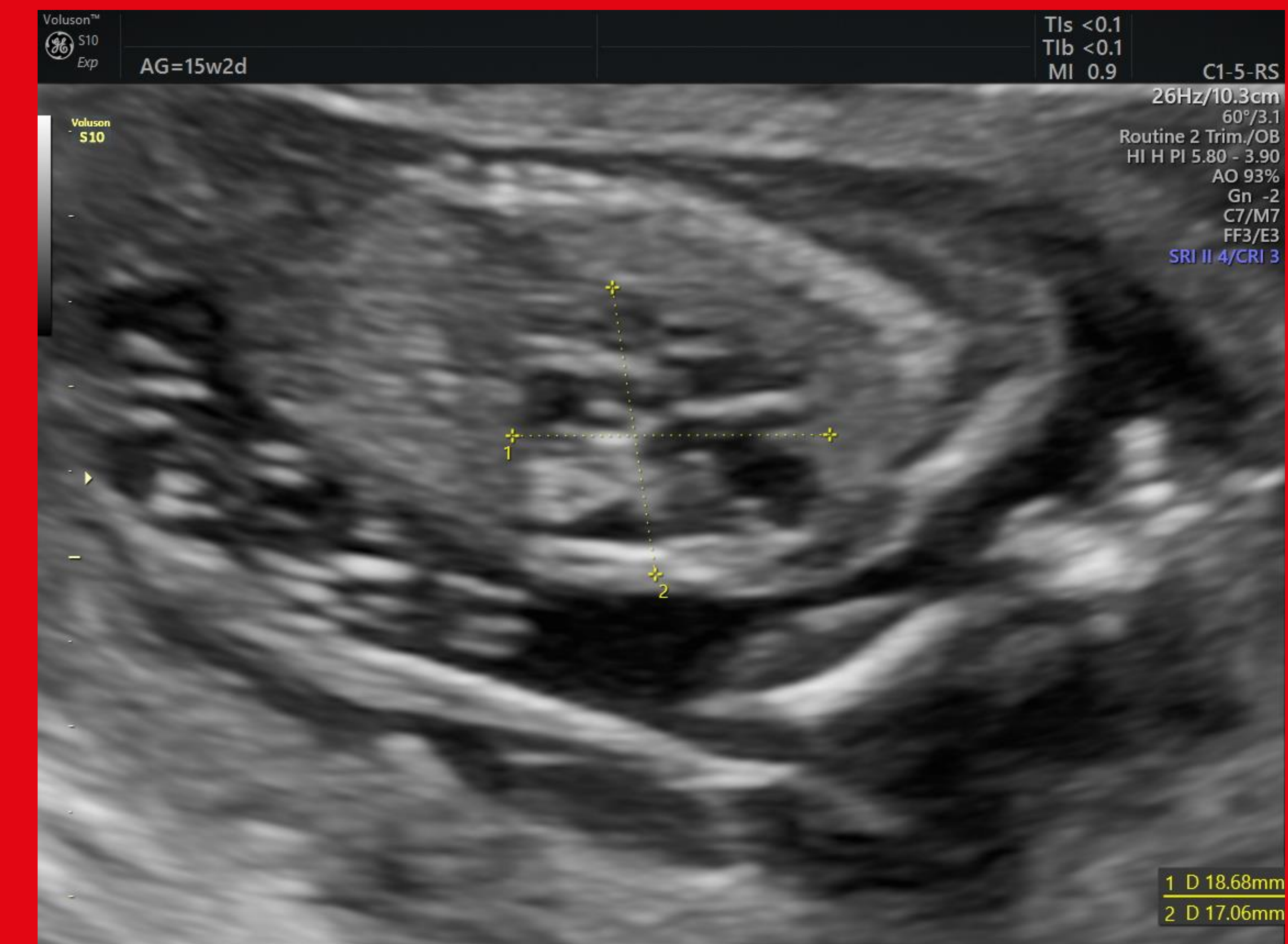


Figure 1: prenatal ultrasound examination

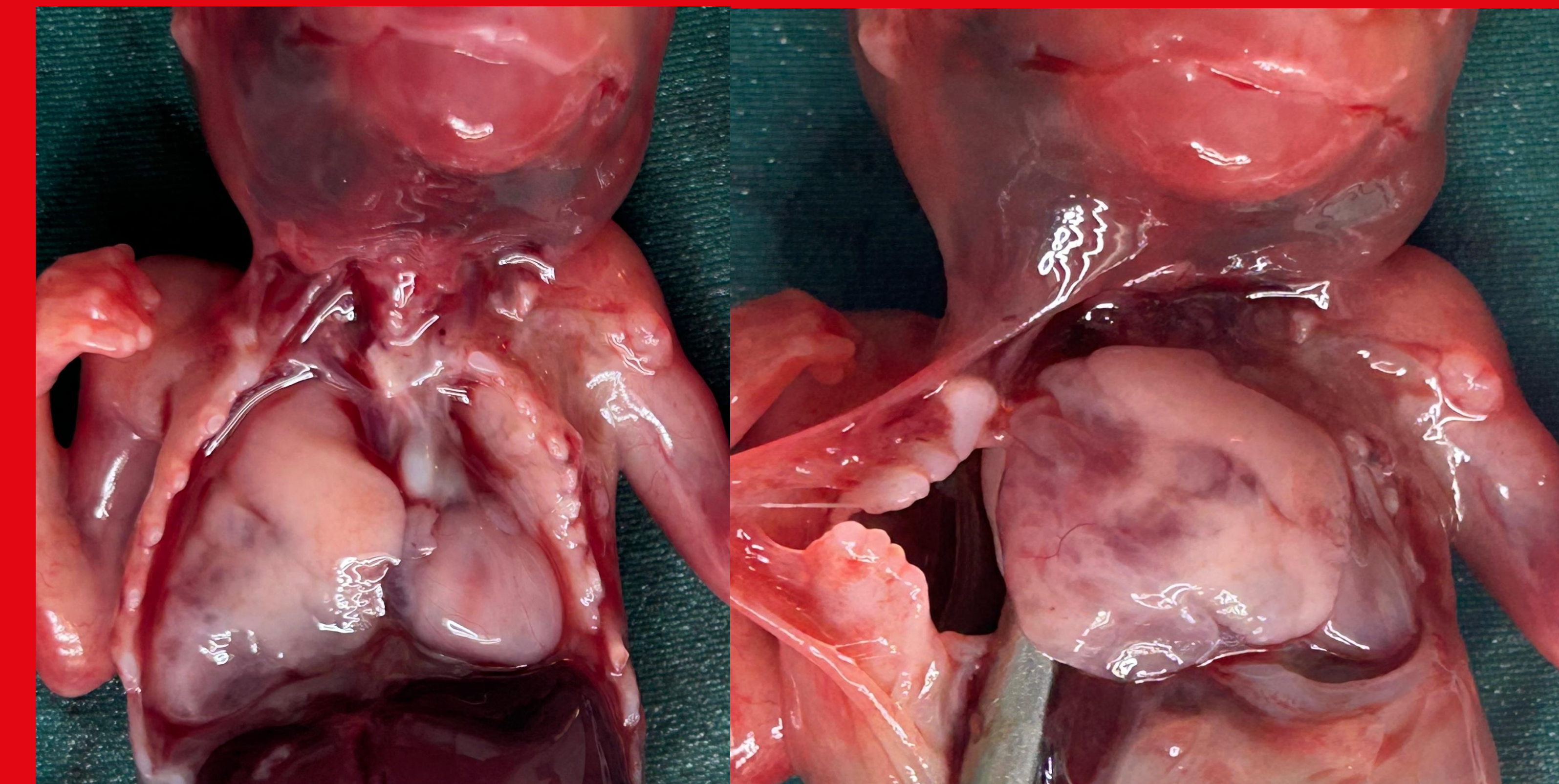


Figure 2 Fetopathological examination