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Background

Binstein-Taybi syndrome is a rare autosomal dominant genetic disorder. It has multisystem clinical manifestations involving the face, limbs, and nervous system. Prenatal ultrasound (US) diagnosis lacks specificity due to the rarity of the disease, so there are few related reports globally. Diagnosis usually depends on clinical features and genetic testing.

Objectives

By analyzing prenatal and postpartum US results and follow-up findings, we explored the prenatal US sonogram features related to the syndrome, aiming to offer a basis for obstetric management.

Case Presentation

★**Prenatal Findings:** A 37-year-old pregnant woman (G3P2) underwent multiple prenatal US, which revealed fetal anomalies including persistent left superior vena cava, mild right heart enlargement, tricuspid regurgitation, restricted foramen ovale flow and left foot polydactyly (6 toes). ★**Genetic Testing:** Amniocentesis showed a normal fetal karyotype, but chromosomal microarray analysis identified a pathogenic variant at 16p13.3. Parental genetic testing was normal, confirming a de novo mutation. Following genetic counseling, the patient opted to continue the pregnancy due to its precious nature. ★**Postpartum Outcomes:** The patient delivered a 2510g male infant via full-term cesarean section. Postpartum US revealed atrial septal defect, mild tricuspid regurgitation, persistent left superior vena cava, subependymal cysts, a short corpus callosum, and cryptorchidism. Thick thumbs and halluces, with abducted halluces, a single palmar crease, six toes on the left foot, wide intercanthal distance, conjunctivitis, and abundant hair on the upper back and upper limbs. During the follow-up at 3 months postpartum, the infant had feeding difficulties, slow response, and was unable to lift his head.

Conclusion

Fetal RSTS presents relevant prenatal ultrasound features with multiple malformations, including limb abnormalities (polydactyly, Thick thumbs and toes), cardiovascular anomalies (persistent left superior vena cava, slightly enlarged right heart, restricted foramen ovale flow), facial anomalies (wide intercanthal distance), and neurological abnormalities (corpus callosum dysplasia). Since RSTS is mostly diagnosed postnatally and fetal cases are rare, prenatal diagnosis of RSTS holds important clinical guiding significance and provides a basis for obstetric management.

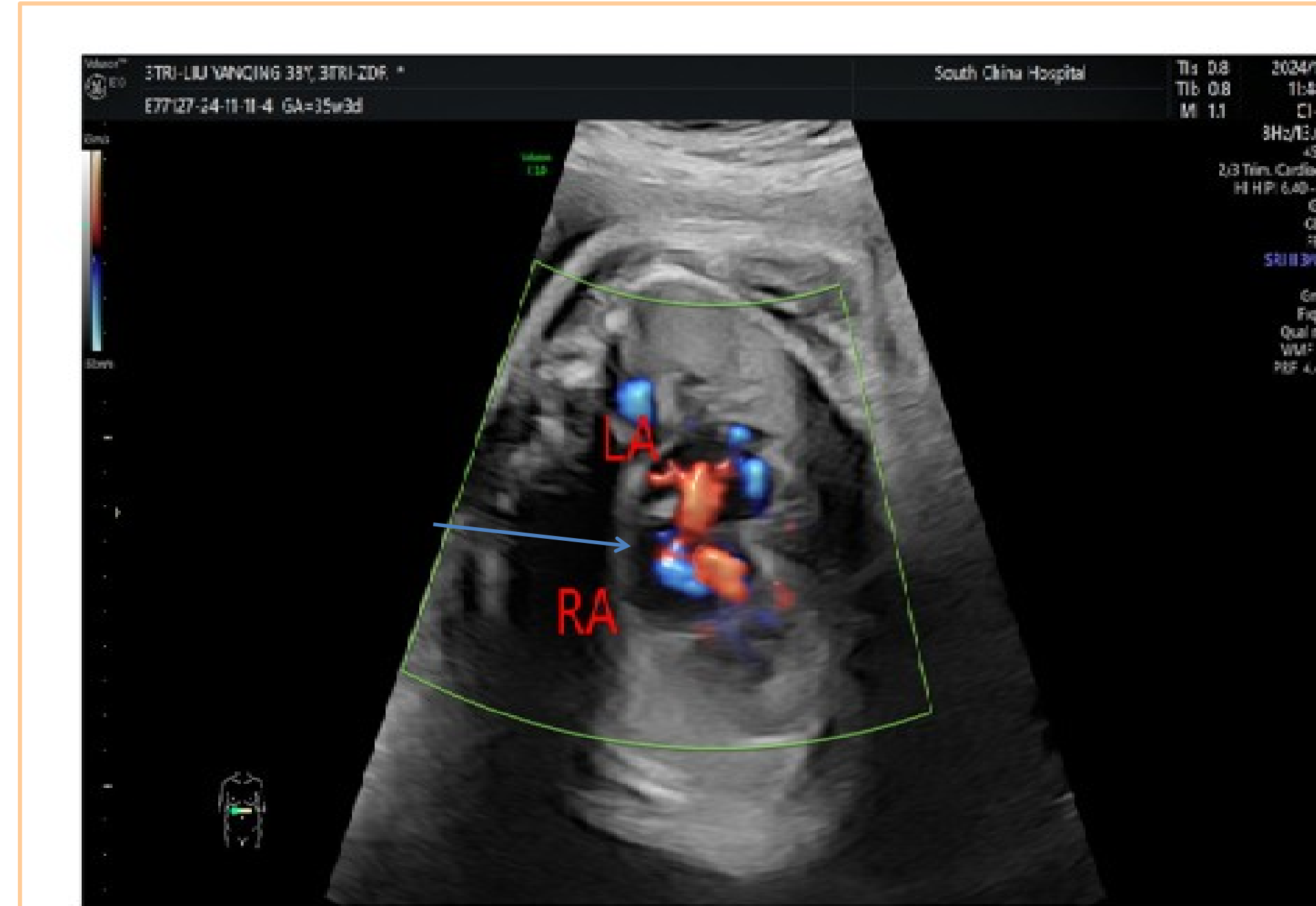


Figure 1. → prenatal ultrasound color Doppler shows thin foramen ovale flow

Figure 2: Photograph of postnatal fingers with a big broad hallux