

Beyond the second trimester: early ultrasound findings in Pallister-Killian syndrome

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Introduction

Pallister-Killian syndrome (PKS) is a rare genetic disorder caused by mosaic tetrasomy of chromosome 12p with craniofacial dysmorphism, intellectual disability, hypotonia, and multiple congenital malformations. First-trimester prenatal diagnosis remains difficult, as ultrasound findings are often normal or non-specific.

Case description

A 33 year-old woman, gravida 6, para 3 presented for a first consultation. Her obstetric history included a first child affected by polycystic kidney disease who died at nine months old, two healthy children and one spontaneous first-trimester miscarriage. Her most recent pregnancy ended in intrauterine fetal demise at 21 weeks of gestation (W), with postmortem findings of growth restriction and bilateral clubfoot.

A first-trimester ultrasound at 13+1W demonstrated increased nuchal translucency (2.9 mm; >95th percentile), mediastinal shift with rightward deviation of the cardiac axis and posterior displacement of the stomach, raising suspicion of congenital diaphragmatic hernia. Additional findings included cardiac chamber disproportion, an enlarged cisterna magna, a small 4th ventricle and elongation of the distal sacrum. The calculated sonographic risk for trisomy 21 was intermediate (1:182).

Fetal echocardiography at 16+1W confirmed mediastinal shift with dextrocardia, right-sided cardiac predominance and tricuspid regurgitation. An intrathoracic stomach was demonstrated, confirming the diagnosis of congenital diaphragmatic hernia. Chorionic villus sampling with microarray analysis revealed a mosaic duplication involving the 12p region, consistent with PKS. The couple received genetic counseling and remains undecided regarding continuation of pregnancy.

Conclusion

Prenatal diagnosis of PKS is challenging due to variable phenotype and mosaicism, with many cases undetected until birth or childhood. The recognition of cardiac, central nervous system and musculoskeletal anomalies in the first-trimester scan should raise suspicion and prompt further evaluation.



Figure 1. First-trimester ultrasound showing increased nuchal translucency and an enlarged cisterna magna

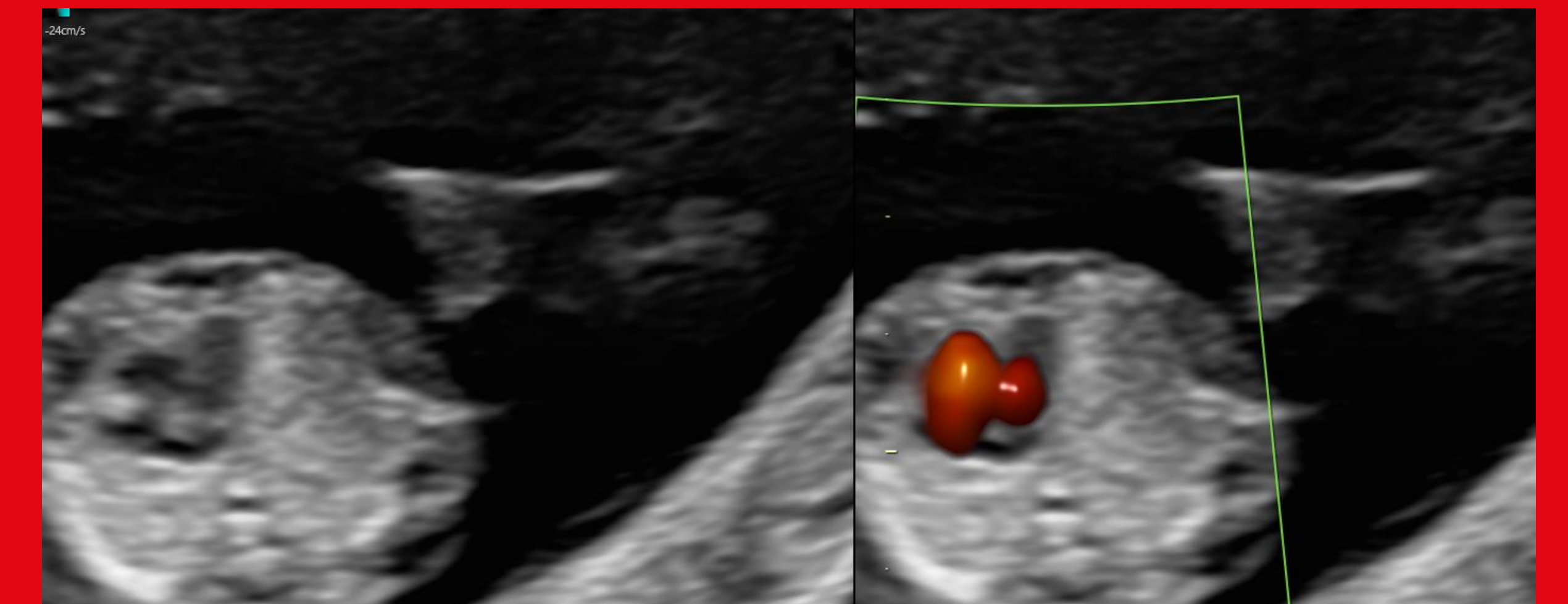


Figure 2. First-trimester ultrasound revealing cardiac chamber disproportion

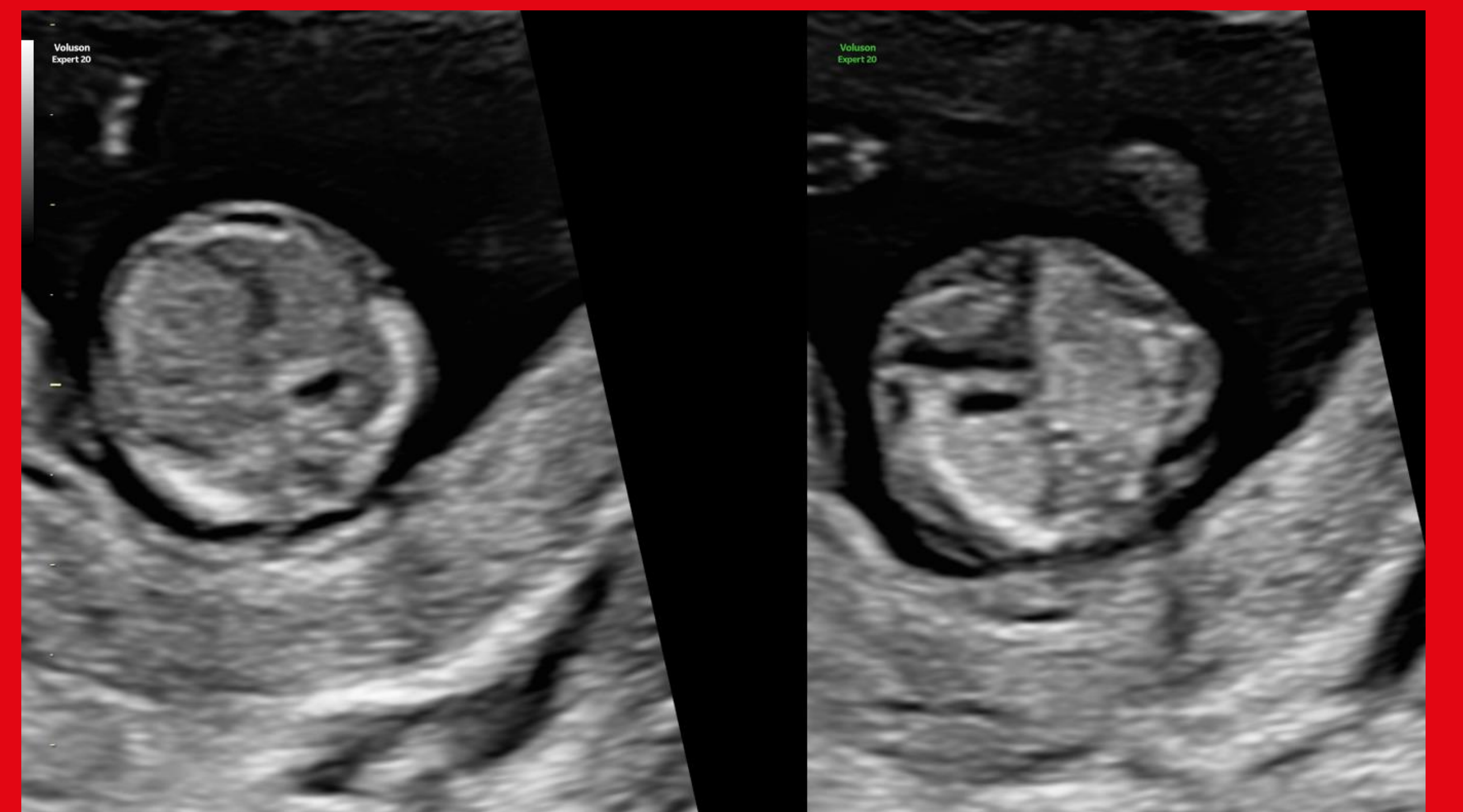


Figure 3. First-trimester ultrasound demonstrating mediastinal shift with rightward deviation of the cardiac axis and posterior displacement of the stomach