Case report:

Patient is a 29 yr-old Malay, G2 P1 with gestational diabetes on diet control. First Trimester Screening noted a nuchal translucency of 2.3 mm, visible nasal bone, and a low risk of trisomy 21, 18 and 13. The screening scan at 19-1 weeks showed short limbs and a 2-vessel cord. At 20-5 weeks a repeat scan confirmed 2-vessel cord, normal head (HC) and abdominal circumferences (AC), and short femur length (FL) and other long bones just above the 5th centile. There was no frontal bossing or uterine artery notching. A provisional diagnosis of SGA due to fetal growth restriction and genetic syndromes was made. Growth scans at 24+5 and 28+5 weeks noted FL on 5th centile and other long bones just below 5th centile, with these parameters falling below the 5th centile at 32+6 weeks. Doppler analysis showed normal uterine artery and MCA PI, and the cerebro-placental ratio CPR<5th centile. The estimated fetal weight was at the 2.4th centile at 35+6 weeks scan, with a CPR <5th centile.

She delivered at 37+4 weeks via NVD to a female baby 2210 g; Apgar scores 8 and 9. Neonatal investigations revealed short limbs, anteriorly placed anus, single umbilical artery, normal renal ultrasound and incidental finding of coarctation of aorta on 2D echocardiography.

**Chromosome Analysis** revealed Karyotype 45, X[42]/ 46,X, r(X) [8]. Ish r(X) (DXZ1+, XIST+). The result is consistent with a **Variant Turner Syndrome phenotype**.