An Unusual Presentation of Turner’s Syndrome
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Background
• This lady was a 29 year old para 1 with a history of Obstetric Cholestasis (OC), 3B tear and BMI>40. This pregnancy was complicated by OC and gestational diabetes (which was well controlled on diet).
• An increased nuchal thickness of 3.9mm was noted on dating scan. Non-invasive prenatal testing was low risk for trisomies 21, 18 and 13 and the parents declined invasive testing.
• The anomaly scan detected significant pedal oedema affecting both the dorsum and plantar aspects of the feet. This oedema extended up to the knees bilaterally, giving the appearance of rocker bottom feet.
• Adjunctive fetal echocardiography was normal, as was the rest of the anatomy. Her four weekly growth scans were normal but continued to demonstrate oedema affecting the lower legs and feet, although there was a subjective reduction in the swelling over the shin bones.
• The increased nuchal translucency noted at first trimester became less prominent on the subsequent scans.
• The fetus was phenotypically female and the parents were counselled that there was a strong possibility of Turner’s syndrome.

Discussion
Turner’s syndrome is described as a partial or total deficiency of one of the X chromosomes resulting in ovarian dysgenesis and is thought to affect 1-2% of all pregnancies. However, there is a high rate of pregnancy loss in Turner’s syndrome, especially as miscarriage and occasionally as still birth.1
The characteristic prenatal ultrasound findings of Turner syndrome are of cystic hygroma, increased nuchal thickness and coarctation of aorta. Cystic hygroma in Turner’s is a result of a deficiency in the lymphatic system of the fetus, and indeed congenital lymphoedema of the hands, feet and neck are common presentations at birth and in infancy.3
Notwithstanding this, reports of oedema of hands and feet on prenatal ultrasound are uncommon. Lymphedema in Turner syndrome is related to lymphatic hypoplasia or aplasia of the lymphatic tracts, which results in stasis of lymph fluid and swelling.
In this case, it was the lymphoedema of the legs and feet that were the main presenting feature on prenatal ultrasound.

Outcome
• The pregnancy was induced at 36 weeks for obstetric cholestasis with vaginal delivery of a phenotypic female infant with good Apgars. There were initially issues establishing feeding but she made good progress. Significant lower limb and pedal oedema was evident in the neonate. The parents agreed to karyotyping, which confirmed 45XO, Turner syndrome.

References