

Isolated unilateral foetal limb lymphoedema in pregnancy: a rare presentation

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INTRODUCTION

Foetal lymphedema is a rare condition that can be associated with genetic syndromes, structural anomalies, or isolated developmental abnormalities. Milroy syndrome, an autosomal dominant disorder caused by *FLT4* gene mutations, is a recognized cause of congenital lymphedema, typically presenting with bilateral lower limb involvement. Prenatal detection of unilateral limb lymphedema is uncommon and may indicate localized lymphatic obstruction or an early manifestation of an underlying genetic condition. In suspected hereditary lymphedema syndromes, genetic testing via amniocentesis or chorionic villus sampling can help identify specific mutations, including those in *FLT4* or *FOXC2*.

THE CASE

We present a 28-year-old multigravida with no relevant medical or obstetric history, whose foetus was diagnosed with progressive unilateral left leg lymphedema during a routine 20-week anomaly scan. First-trimester combined screening indicated a low risk for Trisomy 21, 13, and 18.

Following the anomaly scan, a detailed foetal assessment in the foetal medicine unit revealed significant soft tissue thickening localized to the left leg (Figure 1), with no associated structural anomalies or hydrops. The oedema extended into the left inguinal region, scrotum, and gluteal area. foetal echocardiography and amniocentesis PCR were normal, and chromosomal microarray analysis did not reveal any genetic abnormalities. An MRI performed at 23+6 weeks confirmed extensive lymphatic malformation of the left lower limb and perineal region, causing structural displacement without evidence of invasion.

After extensive counselling regarding the foetal condition and prognosis, the patient opted for pregnancy termination at 24 weeks. No postmortem examination was performed.

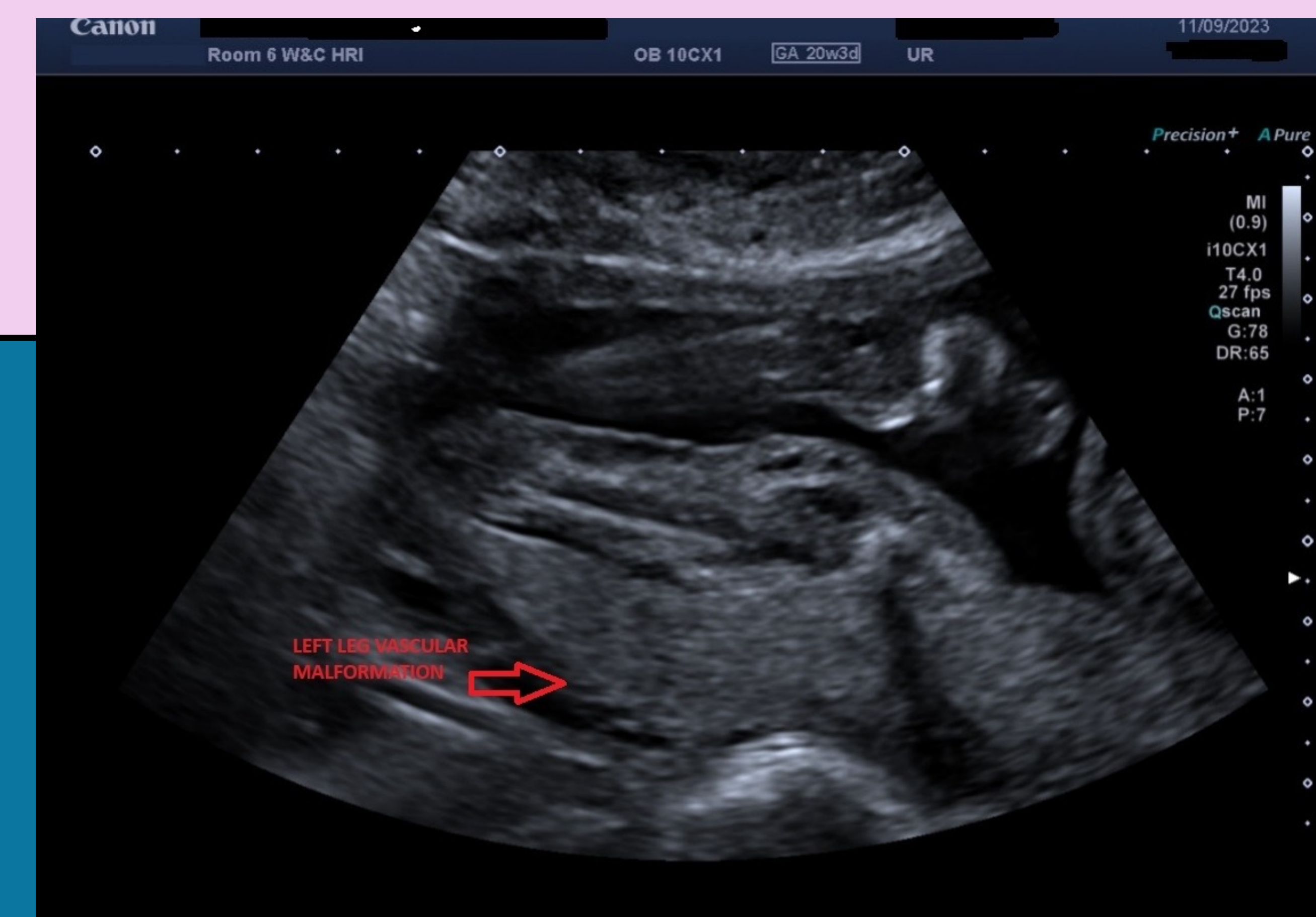


Figure 1. USS of Left lower limb lymphatic malformation in this foetus.

LEARNING POINTS:

Isolated unilateral foetal limb lymphedema is rare and presents diagnostic and prognostic challenges. While it may be an isolated finding, comprehensive antenatal imaging, genetic evaluation, and multidisciplinary counselling are crucial for accurate diagnosis and informed decision-making. This case highlights the importance of early recognition and thorough assessment of foetal lymphatic anomalies to guide clinical management.