We present a 30 years-old patient, G3P1A1, referred to our high-risk center for anomalies on the 20 week ultrasound. She was previously referred to our genetic department because of a fetal nuchal translucency of 6.1mm. Level II anatomy ultrasound demonstrated a nuchal translucency of 8.8mm with associated edema. The nasal bone was present. Transcervical chorionic venous sampling was normal for QF-PCR and CGH and Noonan panel was negative. At 16 weeks, nuchal fold was thickened and there were minimal pleural effusions. The ribs were noted to be short, the long bones were in the 30th percentile. Fetal echocardiography and TORCH investigations were normal. At the 20 weeks scan, pleural effusions were resolved but edema of the head, lower limbs and dorsal portion of the hands was present which remained on subsequent ultrasounds.

She delivered via Cesarean at 37 weeks because of spontaneous membrane rupture and breech presentation. Postnatal physical exam showed swelling of the neck, head, arms, legs and feet. All genetic evaluations were negative and a diagnosis of congenital lymphoedema was posed.

Congenital lymphoedema is a condition present at birth in which lymph collects in tissues and causes swelling. Only a few cases of congenital lymphoedema have been reported.