

## Clinical Quiz

### What is the Diagnosis?

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A baby boy was born at 38 weeks to a non-consanguineous Chinese couple by elective Caesarean Section for history of previous Caesarean Section. The baby's birth weight was 2.425 kg (3rd centile), head circumference 34 cm (50th centile), body length 46 cm (10th centile). Apgar scores were 9 at 1 minute and 10 at 5. Antenatal ultrasonography was remarkable for dilated bowel loops, and liquor volume was normal.



Figure 1.

Multiple joint abnormalities were noted after birth, with adducted shoulders, flexed wrists, flexed and ulnar deviated fingers. The elbows showed very limited flexion. His left hip was held in an abducted position and flexion was limited, while the left knee was kept in an extended position (Figure 1). Muscle bulk was poor over the deltoids and biceps. There was generalised paucity of movement. Additionally, he had trismus, posteriorly rotated ears and a naevus flammeus over the forehead. Ultrasonography of hips and spine, however, were normal. Karyotype was 46,XY. Family history was negative for neuromuscular diseases.

In view of the antenatal ultrasonography result, water soluble contrast study was performed (Figure 2).

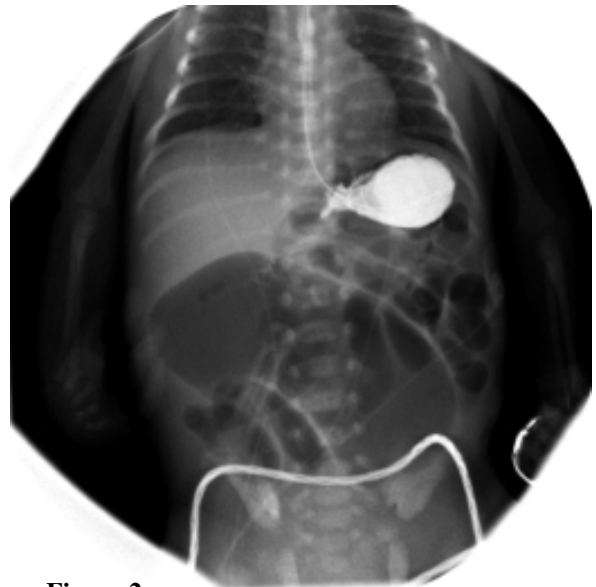


Figure 2.

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N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.