

Clinical Quiz

What is the diagnosis?

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A baby boy was born at 40 weeks with birth weight of 2.99 kg by normal spontaneous delivery. Parents were non-consanguineous healthy Chinese couple. Antenatal and postnatal history were unremarkable.

Upon physical examination shortly after birth, he was noted to have multiple upper limb abnormalities including bilateral hypoplastic thumb, syndactyly between thumbs and index fingers, and radial club hand (Figure 1). X-ray of upper limbs showed radial hypoplasia and thumb hypoplasia (Figure 2). Echocardiogram found a large secundum atrial septal defect with subsequent repair surgery done at 14 months old. He was otherwise well with

normal development and no craniofacial dysmorphism. There was no family history of similar presentation.



Figure 1 (a) Clinical photo of our proband taken at 5 months showing bilateral hypoplastic thumb, syndactyly of thumb and index fingers and radial hypoplasia. (b, c) Post-pollicisation at 20 years old.

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Figure 2 X-ray images demonstrating bilateral thumb and radial hypoplasia

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