

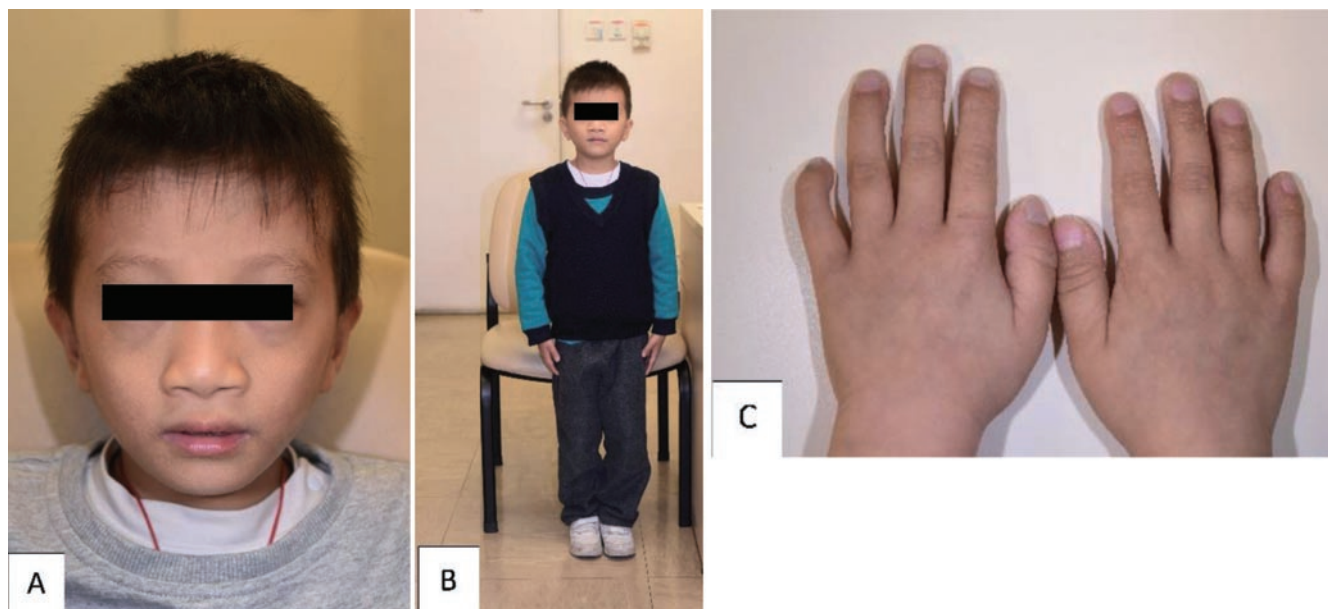
## Clinical Quiz

### What is the Diagnosis?

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The proband is a 7-year-old boy. He was born preterm (33 weeks) to a non-consanguineous Chinese couple due to antepartum haemorrhage. Antenatal check-up all along showed intrauterine growth restriction. His birth weight was 1.42 kg. He was referred to clinical genetic service at the age of 3 due to failure to thrive and global developmental delay. Physical examination at that time revealed microcephaly (47 cm, <3 rd centile), short stature (78.7 cm, SD -4.3), low body weight (9.258 kg, SD -3.0). He had triangular face, bitemporal narrowing, midface hypoplasia, prominent eyes, epicanthic fold, long nose with

broad tip and short philtrum and thin lips. Skeletal features included short neck, brachydactyly, fifth finger clinodactyly and hyperextended proximal interphalangeal joints (Figure 1). He had micropenis with descended testes. He also had mild bilateral grade hearing loss, myopia, and astigmatism. Cardiovascular and abdominal examinations were unremarkable. X-ray of hands showed delayed bone age. He had mild to moderate grade intellectual disability and studied in a special school. Hypernasality and dysarthria were noted.



**Figure 1** (A) Facial profile at 3 years of age. (B) Proportional short stature. (C) Brachydactyly, 5th clinodactyly (with consent for publication by parents).

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Answer to "Clinical Quiz" on Pages 281-282

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