Clinical Quiz

What is the Diagnosis?

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The proband is a 5 years old boy. He was born at full term in China to a non-consanguineous Chinese couple. Antenatal checkup was normal. Postnatally, he had long segment Hirschsprung's disease with colectomy and ileocolic anastomosis performed at 1 year of age. He was referred to genetic clinic for multiple congenital anomalies. Physical examination at 19 month old of age showed microcephaly (44.7 cm, <3rd centile), bilateral low set and posteriorly rotated ears, hypertelorism, upturned nostrils, bilateral postaxial polydactyly, bilateral 2/3 syndactyly of toes (Figure 1). He had normal male genitalia. His body weight and body height were 9.2 kg (3-10th centile) and 77.5 cm (3-10th centile) respectively. He had borderline developmental delay. He had failure to thrive with body weight and body height centiles at 3rd centile in subsequent follow up.

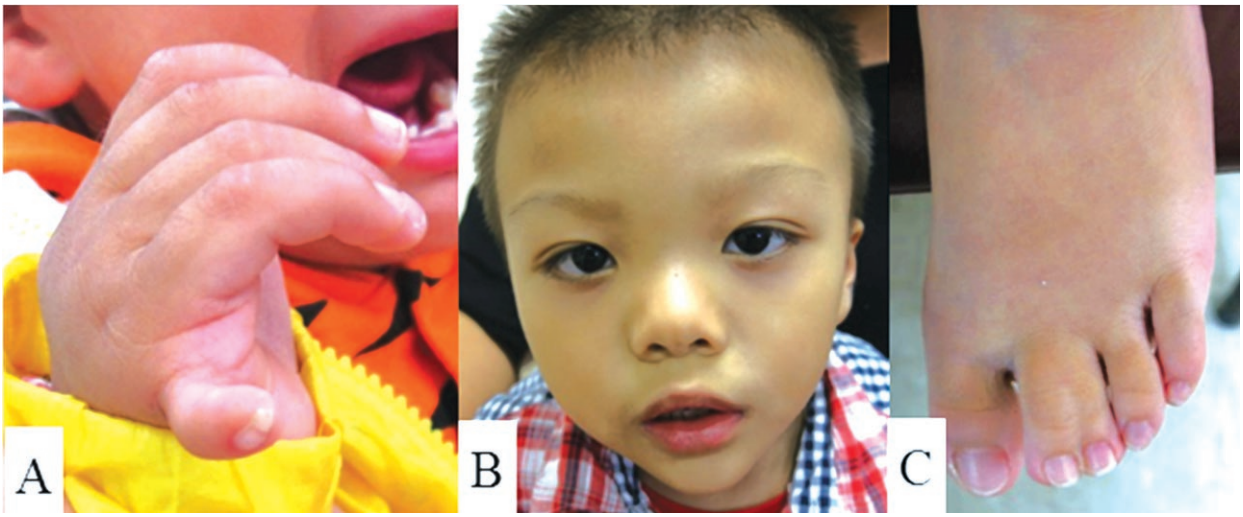

Figure 1 (A) Post-axial polydactyly of right hand; (B) Facial profile; (C) 2/3 syndactyly of left toes. (Consent for publication has been obtained).

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Answer to "Clinical Quiz" on Pages 266-267
N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.