Clinical Quiz

What is the Diagnosis?

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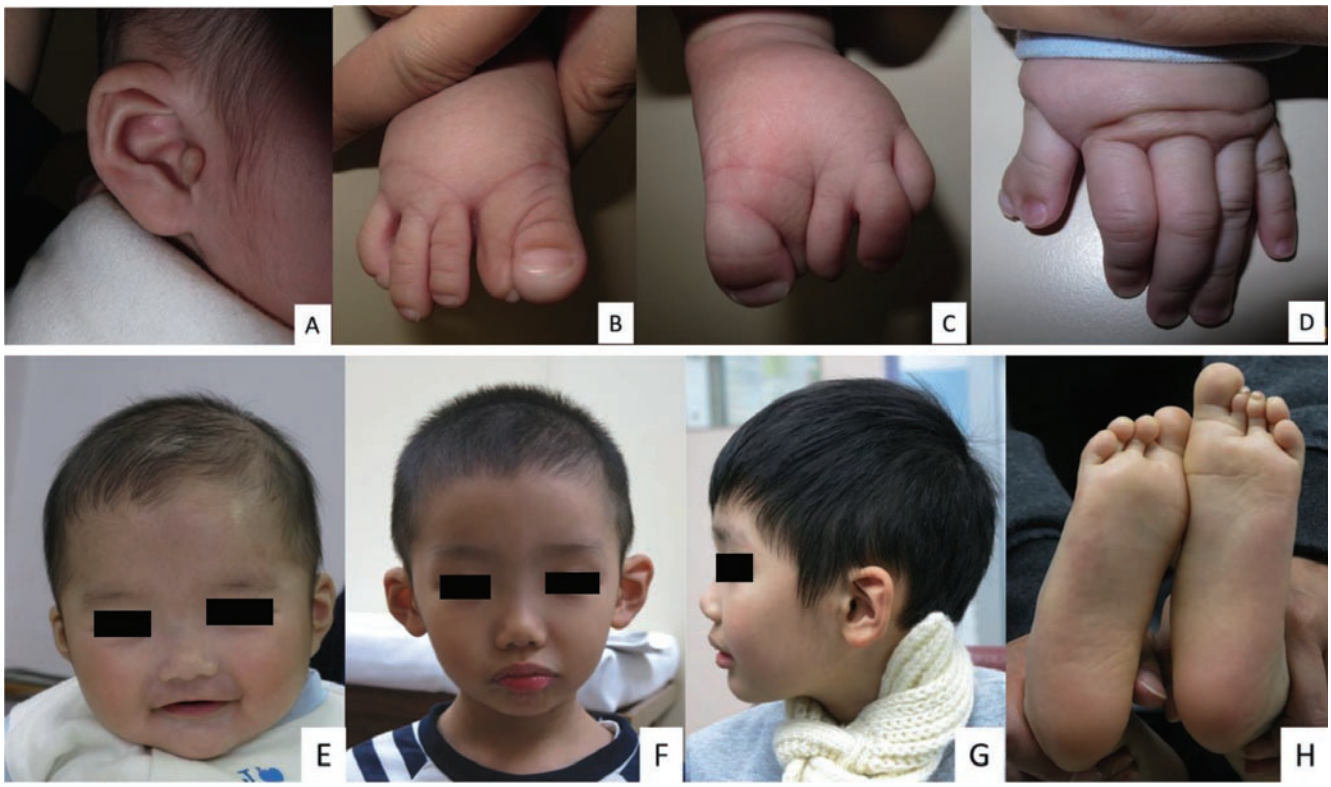
The proband is a 5-year-old boy. He was born full term in Hong Kong to a non-consanguineous Chinese couple. Antenatal checkup was normal. He was referred to genetic clinic for multiple congenital anomalies at 4 months of age. Physical examination at that time revealed normal growth parameters. He had bilateral mild hearing deficit, bilateral pre-auricular skin tags, left preaxial polydactyly, toes syndactyly and hypoplastic toes (see Figures A-D). He was hypeterloic without cleft lips and palate. He had normal male genitalia. His cardiovascular and abdominal examination was unremarkable. His renal ultrasound, echocardiogram and MRI brain were unremarkable. On further follow up, he had borderline developmental delay and chronic constipation since infant period. He had corrective surgery for pre-axial polydactyly and preauricular skin tag during infant period.



(A) Pre-auricular skin tag of right ear; (B) Right foot: variable 1st/2nd toes syndactyly, hypoplastic second toe and mild splitting between 2nd/3rd toes; (C) Left foot: variable 1st/2nd toes syndactyly, hypoplastic second toe and mild splitting between 2nd/3rd toes and 3rd/4th toes syndactyly; (D) Left preaxial polydactyly. (E) Facial profile at 4 months old; (F) Facial profile at 3 years old; (G) Lateral facial profile at 5 years old with mildly malformed ear; (H) Bilateral 1st/2nd toes syndactyly and displaced nails (with consent for publication by parents).

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Answer to "Clinical Quiz" on Pages 219-221

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