Clinical Quiz

What is the Diagnosis?

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Our proband is currently a 2-year-old boy. He was born at full term with birth weight of 2.7 kg to a non-consanguineous Chinese couple. He presented with hypotonia and feeding difficulties during the neonatal period, otherwise the perinatal and family history was unremarkable. The patient was first referred to the Clinical Genetic Service at 5 months of age. Physical examination showed his head circumference was below 3rd centile with a body weight at 10th centile and a supine length between 3rd to 10th centile. He had subtle dysmorphic features including narrow bifrontal diameter, almond-shaped eyes, low-set ears, narrow nasal bridge, down-turned corners of the mouth and micrognathia. There was bilateral cryptorchidism and small hands and feet.

In the subsequent follow-up, he was diagnosed to have global developmental delay and received early education training. His feeding was improved but he developed obstructive sleep apnoea that required continuous positive airway pressure since 1 year old. Clinical photographs of the proband at the age of 21 months are shown in Figure 1.

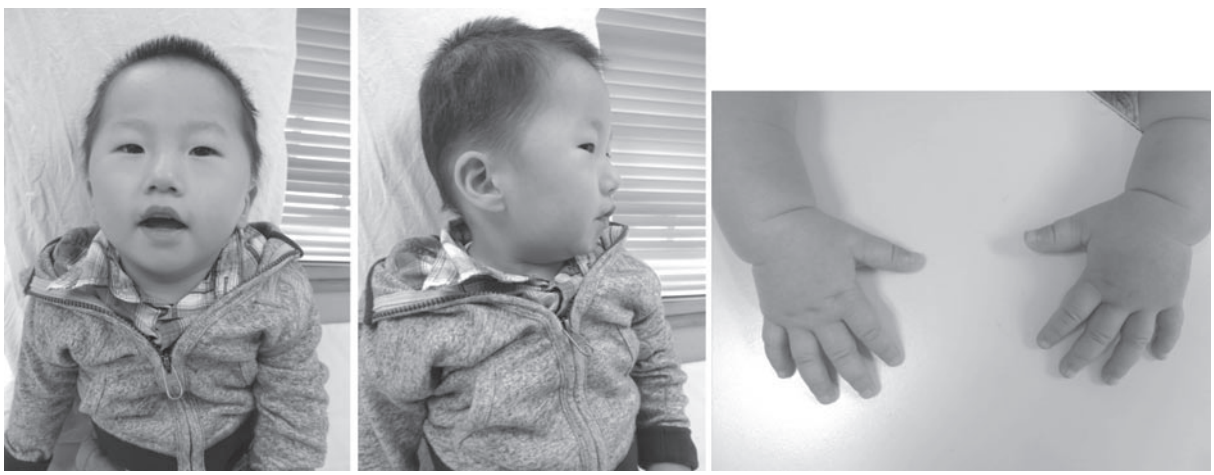

Figure 1 Clinical photographs of the proband at the age of 21 months. Facial dysmorphic features and small hands are shown (with consents for publication by parents).

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