Clinical Quiz

What is the Diagnosis?

HM ***Tam***, JY ***Leung***, IFM ***Lo***, HM ***Luk***

Our patient is currently a 12-year-old boy. He was born at full term to a non-consanguineous Chinese couple with birth weight of 3.0 kg. His perinatal history was unremarkable. He was noted to have subtle facial dysmorphic features and mild developmental delay at early infancy. He was then referred to clinical genetic service by his general paediatrician at the age of 2 for assessment.

Physical examination at the genetic clinic showed he had long palpebral fissures, everted lateral 1/3 of lower eyelids, arched eyebrows, dense eyelashes, hypertelorism, short columella, high arched palate, depressed bulbous nasal tip and large prominent cupped ears (Figure 1). There were persistent fetal fingertip pads, brachydactyly, 5th clindactyly, short 4th and 5th metatarsals and joint laxity. The past medical history was significant that he had right dysplastic kidney with grade 4 right vesico-ureteric reflux. The brain MRI and echocardiogram were normal. Based on the clinical feature, a syndromic diagnosis was made and subsequently confirmed by molecular testing.

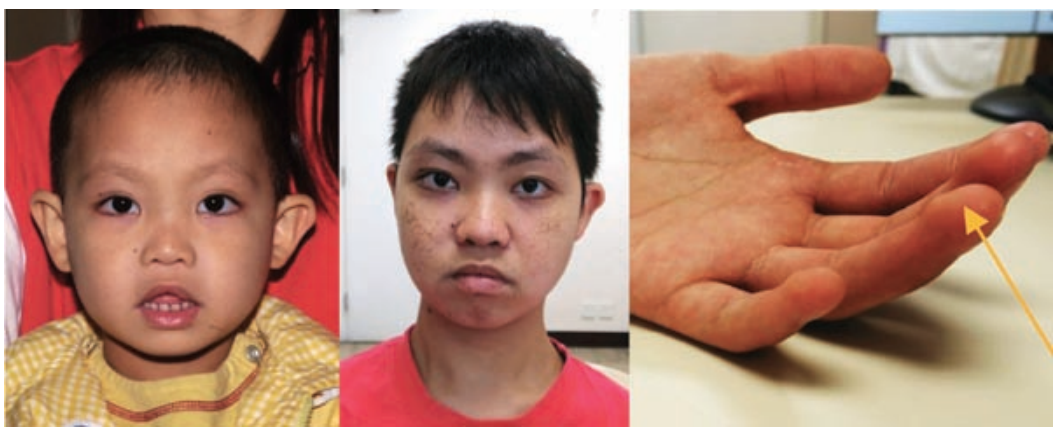

Figure 1 The clinical photos of our patient at 3 and 11 years of age which showed the specific facial gestalt and persistent finger pads (with consents for publication by parents).

The clinical quiz was prepared by:

HM **Tam**
Year 3 medical student, The University of Hong Kong, Hong Kong, China

JY **Leung**
Year 2 medical student, The University of Edingburgh, United Kingdom

IFM **Lo**
HM **Luk**
Clinical Genetic Service, Department of Health, Hong Kong, China

Answer to "Clinical Quiz" on Pages 65-67

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