

Clinical Quiz

What is the Diagnosis?

SSW CHENG, IFM LO, HM LUK

The proband is a 8 years old boy. He was born full term to a non-consanguineous Chinese couple in Hong Kong. Antenatal checkup showed small head circumference at 3rd centile. His birth weight was 3.95 kg. Postnatally, he suffered from failure to thrive without developmental delay. He was referred to genetic clinic for dysmorphism at 1 year old. Physical examination at that time revealed microcephaly (41 cm, <3rd centile), short stature (70 cm, SD -2.2), low body weight (6.95 kg, SD-3.0), depressed nasal bridge, anteverted nostrils, bulbous nasal tip, long

philtrum, thin upper vermillion border and right hypoplastic ear. He had right hearing loss. His hands, feet and genitalia were normal. Cardiovascular examination and abdominal examination were unremarkable. Subsequently, he developed multiple exostoses at right scapula and left distal tibia at the age of 2. He studied in mainstream primary school with average performance. He had sparse hair and eyebrows and short hands. Orthopedic surgery was performed for exostoses on right knee at 7 years of age.

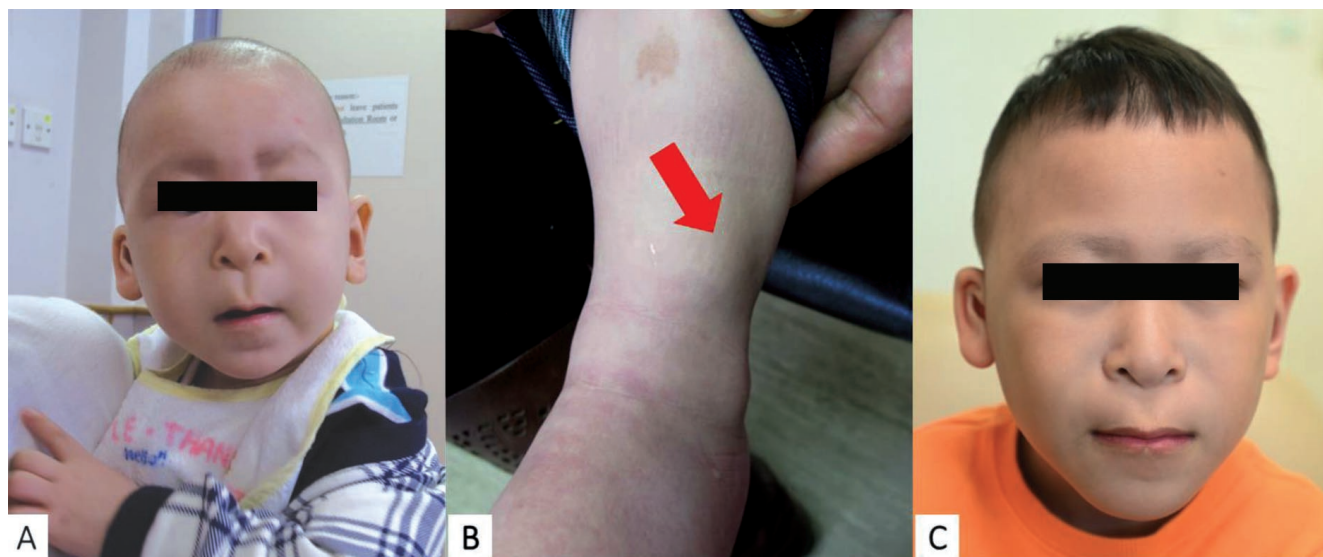


Figure (A) Facial profile at 1 years of age. (B) Left ankle region exostosis (arrow). (C) Facial profile at 8 years old (with consents for publication by parents).

The clinical quiz was prepared by:

SSW CHENG

IFM LO

HM LUK

Clinical Genetic Service, Department of Health, HKSAR

Answer to "Clinical Quiz" on Pages 62-64

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