
Clinical Quiz

What is the Diagnosis?

WY Yu, HM Luk

The Big Toe Points to the Diagnosis

A 3-year-old male child is referred to genetic clinic for suspected syndromal disease. He was born in full term with birth weight of 3.2 Kg by non-consanguineous Chinese couple. The perinatal history was unremarkable. He was noted to have bilateral deformity of big toes at birth (Figure 1). X-ray showed bilateral hallux valgus without bone destruction. Systemic review was normal. He had received up-to-date immunisation without adverse side effects. The family history was non-contributory. Based on the toe deformity, a rare genetic disease was suspected that confirmed by molecular testing.



Figure 1 The proband has bilateral deformity of big toes at birth (with consents for publication by parents).

The clinical quiz was prepared by:

WY Yu

HM Luk

Clinical Genetic Service, Department of Health, Hong Kong

Answer to "Clinical Quiz" on Pages 114-116

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