Clinical Quiz

What is the Diagnosis?

The propositus was born to a non-consanguineous healthy Chinese couple at 40 weeks of gestation by Ventouse extraction. Apgar scores were 8 at 1 minute and 9 at 5. No resuscitation was required. Antenatal scan at 36 weeks gestation showed a small-for-date fetus with rhizomelia. The femoral length, tibial length and fibular length corresponded to 27-28 weeks of gestation, while radial and ulnar length, together with thoracic circumference corresponded to 34-35 weeks of gestation. There was macrocephaly. Antenatal history was otherwise uneventful. Mother was 34 years old while father was 32 years old and both had normal height. Family history was insignificant.

Examination of the newborn revealed a birth weight of 2800 gram (10th percentile), birth length of 46 cm (3rd percentile) and head circumference of 36 cm (75th percentile). Systematic examination was only significant for a small nose with flat nasal bridge. There was no sign of respiratory distress. In view of the antenatal findings, a babygram was ordered.

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Answer to “Clinical Quiz” on Pages 241-242

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