

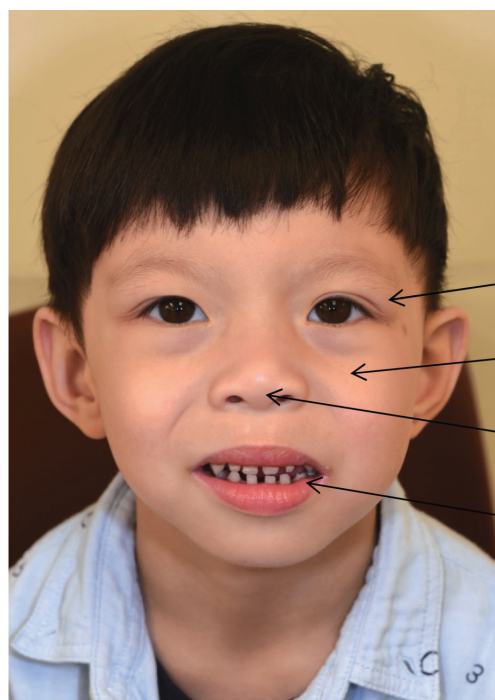
Clinical Quiz

What is the Diagnosis?

SSW CHENG, IFM LO, HM LUK

The proband is a 4-year-old boy. He was born at full term with body weight of 2.27 kg to a non-consanguineous Chinese couple. Antenatal course was uneventful except he had mild IUGR since 30 weeks' gestation. Postnatally he was noted to have cardiac murmur with echocardiogram showed he had mild supra-ventricular aortic stenosis. He also had mild to moderate developmental delay that studied in special school. He was then referred to genetic clinic for assessment. Physical examination showed he had unremarkable growth parameters with body height of

50th centile, body weight of 25th-50th centile and head circumference of 10th centile. However, facial dysmorphism (Figure 1) was noted that including periorbital puffiness, flat nasal bridge, malar hypoplasia, full nasal tip, long philtrum, prominent lips, small and widely spaced teeth and wide mouth. Cardiovascular examination revealed Grade 3 over 6 ejection systolic murmur at left upper sternal border that radiated to neck. The rest of examinations were normal. He is very talkative and friendly during consultation.



- Periorbital puffiness
- Malar hypoplasia
- Bulbous nasal tip
- Small and widely spaced teeth,
Prominent lips

Figure 1 Clinical photo of our patient (consent has been taken for publication).

The clinical quiz was prepared by:

SSW CHENG
IFM LO
HM LUK

Clinical Genetic Service, Department of Health, Hong Kong, China

Answer to "Clinical Quiz" on Pages 239-242

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