

Clinical Quiz

What is the Diagnosis?

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Our patient was born at 38 weeks of gestation. Her antenatal history was uneventful. Her parents are non-consanguineous and are both originated from the Guangdong province in China. Her mother had 1 episode of spontaneous miscarriage at 1st trimester, otherwise her family history was unremarkable. Her birth weight was 2.81 kg. At 2 months old, she was found to have umbilical hernia. At 2 years of age, she was noted to have decreased range of movements in her major joints, especially in her upper limbs. Her weight and height were both at 50th percentile and her head circumference was between 50th and 75th percentile at 3 years of age. Her developmental assessment was normal. Ultrasound at 3 years of age showed splenomegaly and echocardiography showed mitral valve regurgitation. Her parents also noticed she had noisy breathing and snoring during sleep. Clinical photographs and X-rays taken at 3 years of age are shown in Figure 1 and Figure 2 respectively.



Figure 1 Clinical photographs of our proband at 3 years of age. (Parental consent obtained for the clinical photo)

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Figure 2 Selected X-rays of our proband at 3 years of age.

Answer to "Clinical Quiz" on Pages 52-55

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