Clinical Quiz

What is the Diagnosis?

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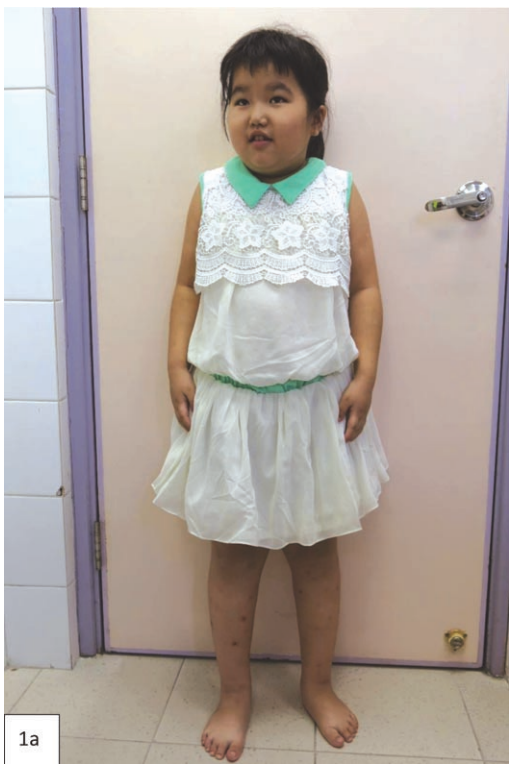
Our patient was referred to the Clinical Genetic Service at 8 years old for retinitis pigmentosa. She was born to a non-consanguineous couple via normal spontaneous vaginal delivery at 35 weeks of gestation with a birth weight of 2.5 kg after an uncomplicated pregnancy. She complained of visual impairment since the age of 5 and was subsequently diagnosed to have retinitis pigmentosa by ophthalmologist. She had developmental delay and was educated in a school for the visually impaired. She had a history of an atrial septal defect with spontaneous closure and vestibular anus with operation done. She also had postaxial polydactyly over the left hand and the right foot with operation done. Physical examination revealed central obesity and brachydactyly.

Clinical photos of the proband are shown in Figure 1.



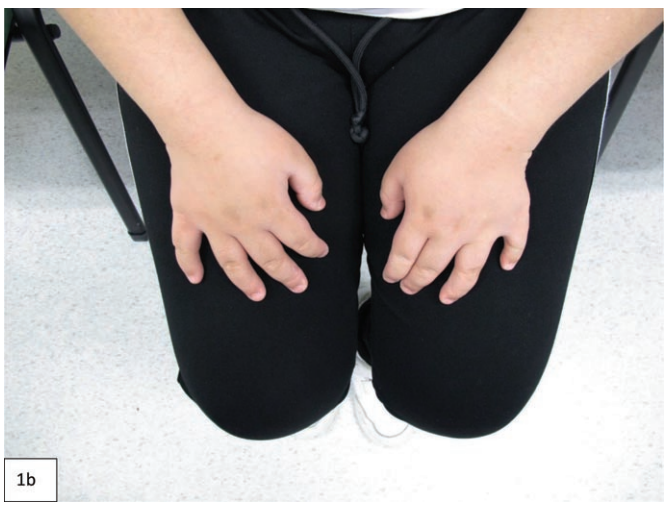


Figure 1 (a) Clinical photos of our proband at the age of 8 showing central obesity and (b) brachydactyly (with consent for publication by parents).

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Answer to "Clinical Quiz" on Pages 156-157

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