Clinical Quiz

What is the Diagnosis?

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Our patient is a 14-year-old boy who referred to genetic clinic for assessment because of intellectual disability and facial dysmorphism. He was born at 36 weeks gestation with birth weight of 2.75 kg. The perinatal and family history was unremarkable. He was noted to have glandular hypospadias at birth and abdominal distension at 36 hours of life. Abdominal X-ray showed dilated bowel loops with absence of rectal gas. Rectal biopsy showed absence of ganglion cells at the rectosigmoid colon. The bowel problem was treated surgically without complication. He also had congenital heart disease namely patent ductus arteriosus and aortic stenosis. Because of multiple congenital malformations, karyotype and fluorescence in situ hybridization for Di George syndrome was performed during neonatal period which were normal. He was followed up regularly by paediatricians and surgeons afterward. He subsequently developed microcephaly and epilepsy that were well controlled by anticonvulsant. Developmental assessment at 5 years old showed he had moderate to severe grade mental retardation. Clinical photographs taken during consultation at 14 years of age can be seen in Figure 1.

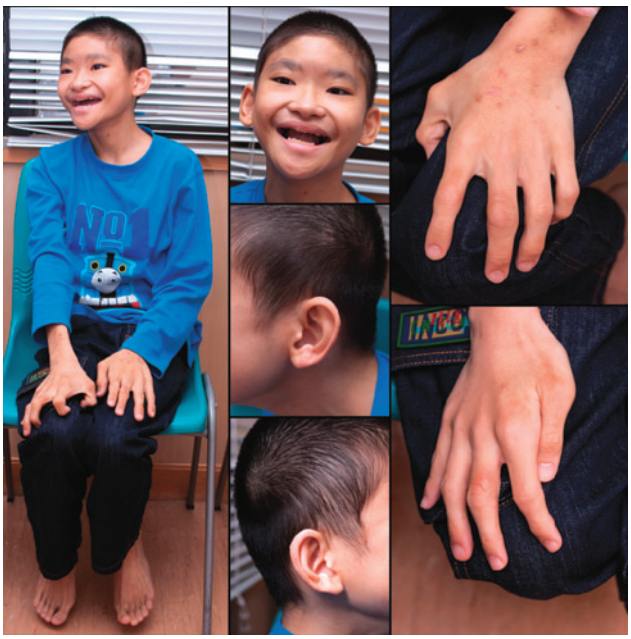

Figure 1 Facial profile of patient taken during consultation at 14 years of age. (Parental consent obtained for the clinical photo)

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