

Clinical Quiz

What is the Diagnosis?

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Our proband is a 4 years old boy. He was born full term with body weight of 3.8 kg to a non-consanguineous Chinese couple conceived by *in vitro* fertilisation.

Apart from neonatal jaundice requiring phototherapy, there was no medical history of note. He presented to the Duchess of Kent Children's Hospital for speech delay at the age of 38 months, with developmental assessment showing global developmental delay and autism spectrum disorder.

Physical examination showed unremarkable growth parameters with body height of 75th percentile, body weight of 97th percentiles, and head circumference of 50th percentile. He had decreased eye contact with minimal

vocalisation, high pitched voice, repetitive self-stimulatory behavior such as hand flapping, intermittent smelling of objects, as well as self-injurious behavior such as intermittent head banging. He was also noted to have dysmorphic features with tall forehead, small ears, small teeth, lax joint, buried penis, small toes and overweight. Clinical photographs of our proband can be seen in Figure 1. Examination of his respiratory and cardiovascular systems was unremarkable. His abdomen was soft, there was no organomegaly and his testes were not enlarged. There was no neurocutaneous marker and no focal neurological sign.



Figure 1 Clinical photographs of our patient at 3 years old (with consent for publication by parents).

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