

Clinical Quizzes

What is the Diagnosis?

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This girl is the second child of a non-consanguineous Chinese couple. She was born at 38 weeks of gestation by Caesarean section with a birth weight of 2.55 kg. She had mild torticollis and mild neonatal jaundice. She was noted to have global delay with very slow progress since early infancy and developed into severe grade intellectual disability, with central hypotonia and dysmorphisms. She also has mild eczema, moderate conductive hearing impairment in the right ear, bilateral myopia and mild cerebral atrophy. She has obstructive sleep apnoea syndrome requiring continuous positive airway pressure and adenoidectomy at 2 years old. She takes anti-epileptics for seizure. She requires anti-psychotic for her behavioural problems and autistic features.

Physical examination showed all growth parameters were below the 3rd percentile. She had brachycephaly, midface retrusion, micrognathia, anterior facing ear lobes, upturned upper lip, short neck, low-set posterior hairline, widely spaced nipples, self-inflicted skin wounds, scoliosis, bilateral second and third toes syndactyly. Facial features and toes can be seen in Figure 1. There were self-inflicted superficial wounds on both of her upper limbs (Figure 2). She had fleeting eye contact with lots of hand stereotypies. Neurological examination showed no focal neurological signs. Her karyotype is 46,XX. Magnetic resonance imaging of the brain showed dysmorphic looking brain with ventriculomegaly and small skull base.

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Figure 1 Clinical photographs of the patient (with consent for publication by parents).



Figure 2 Self-inflicted superficial wounds on the patient's upper limbs.

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