

Clinical Quiz

What is the Diagnosis?

KPT Yu, IFM Lo, HM Luk

A 3-year-old girl was referred to genetic clinic for developmental regression. She was born at full term with birth weight of 2.91 kg, from a non-consanguineous Chinese couple. She had normal development till 1.5 years old. She was initially able to speak a few meaningful words but with loss of speech since 1.5 years old. She was also reported to have on and off hyperventilating episodes and falling off head circumference from 50th to 25th percentile. She had no clinical seizure. Family history was not contributory. Physical examination at 3 years old showed growth parameters with head circumference at 25th percentile, body weight at 25th-50th percentile and body height at 3rd-10th percentile. There was no craniofacial dysmorphism, but frequent stereotypic handwashing movement and no purposeful hand use were noted during consultation. Hyperventilation and teeth grinding were also being observed. She got no single meaningful words during assessment. Examination of other systems was essential normal. Brain imaging and baseline routine workup included metabolic investigation in regional paediatric unit was negative.



Figure 1 Clinical photographs of our patient at age of 4 (consent has been taken for publication).

The clinical quiz was prepared by:

KPT Yu

IFM Lo

HM Luk

Clinical Genetic Service, Department of Health, Hong Kong, China

Answer to "Clinical Quiz" on Pages 192-194

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