Clinical Quiz

What is the Diagnosis?

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Our proband is a 34-month-old boy who was referred from the private sector for global developmental delay, tremor, sleep disturbance and suspected neurometabolic disorder.

He was born via Caesarian section at 36 weeks of gestation for social reason after an uneventful pregnancy, weighing 2.9 kg at birth. Physical growth was apparently normal. Both his body weight and height were at the 50th-75th percentile, whilst his head circumference was at the 10th percentile**.** His development progressed slowly. At 21 months of age, he could sit without support but could not stand alone; had fine pincer grasp but could not release; responded to name calling but could not identify most common objects; could vocalise but did not yet have babbling; had strong stranger anxiety but could not wave bye-bye. Overall, his mental age was 9-10 months across all aspects of development except language expression, where he was discrepantly delayed with mental age of 3-4 months only.

He also had episodic tremor involving his hands and feet which progressively increased in frequency and was nearly continuous throughout daytime. His eyes were unaffected and there was no oculogyric crisis. Behaviour-wise, he had frequent bouts of laughter as well as sleep disturbances, and would wake up 2-3 times per night.

Physical examination was unremarkable except relative microcephaly and nearly continuous tremor during voluntary movement, which was superimposed with myoclonus. There was no dysmorphic feature, goiter or neurocutaneous marker. Clinical photo at 34 months of age can be seen in Figure 1.

Extensive investigations were performed in the mainland prior to his referral, including plasma amino acid, carnitine and acylcarnitine profile, thyroid function, copper and caeruloplasmin, electroencephalogram (EEG), MRI brain and nerve conduction studies. They were all unrevealing. Further neurometabolic examination including urine for organic acid and catecholamine, CSF for neurotransmitter profile and repeat thyroid function test were also normal. Multiple EEGs were performed, showing posterior dominant rhythm with generalised high amplitude 2-3 Hz activities admixed with or without spikes.

He was referred to clinical genetics for further evaluation.

Figure 1 Clinical photographs of our proband at 34 months 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.