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## Clinical Quiz

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### What is the Diagnosis?

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The patient was referred to us at 30 months old for developmental delay. She lived with her parents and her two siblings (elder sister 4 years old; younger brother 9 months old) in Fujian, China. Her parents were non-consanguineous. There was no family history of intellectual disability or neurological condition. Born at full term by normal vaginal delivery, she weighed 2.4 kg when she was delivered at a private hospital in Hong Kong. Antenatal and postnatal courses were smooth. She enjoyed good health. Parents noticed that child seemed to be slow in responding to stimulation when she was at 3-4 months old. Doctor in Fujian suspected that the child had hearing problem. The girl was diagnosed by doctors in Beijing to have developmental delay. MRI Brain in Heilongjiang showed suspected white matter disease when the child was 14 months old. She never had seizure before. She attended speech therapy and some training related to sensory integration in Fujian for about 1 year. She was also given some intravenous medication in Mainland with the aim to support and protect the brain.

She was first seen at the developmental clinic at the Duchess of Kent Child Assessment Centre at 30 months old. She had delayed motor milestones. She was able to walk without support at 18 months old. She could climb upstairs but not downstairs at 30 months old. She was right handed. She could build tower of 3 cubes. She was able to hold the pencil with palmar grasp and could mark on paper. She could hold the spoon but used it messily. She had no meaningful word and could understand simple verbal commands with gesture cues. She still wet and soiled. No behavioral issues were reported by her parents. At chronological age 30 months old, her mental age was at 21.5 months old (developmental quotient 72). She weighed 13 kg (75-90th percentile). Her height and her head circumference were 90 cm (90th percentile) and 45.5 cm (3rd percentile) respectively. Photographs of the child were shown on Figure 1. She was impressed to be hypotonic. The rest of the examination was unremarkable. Optometric and audiological assessments were normal. Blood tests including muscles enzymes and thyroid function test were normal.

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**Figure 1** Clinical photographs taken at 3 years old were as follow (with consent for publication by parents).

Answer to "Clinical Quiz" on Pages 212-214

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