Clinical Quiz

What is the diagnosis?

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Clinical History

A female baby was delivered at 40 weeks of gestation by spontaneous vaginal delivery with birth weight of 2.955 kg. She was the third child to non-consanguineous Chinese parents who were both alpha thalassaemia carriers, but amniocentesis was negative for alpha thalassaemia major. Antenatal and family history were otherwise unremarkable.

She was noted to have microcephaly and poor feeding soon after birth, and required hospital admission at two months of age for failure to thrive and feeding difficulties with choking and respiratory distress requiring tube feeding. She had generalised hypotonia and significant craniofacial dysmorphism featured by hypertelorism, low-set ears, a smaller lower jaw and alternating divergent squint. She cried with a monotonous, high-pitched voice.

She suffered from oromotor dysfunction, gastro-oesophageal reflux and recurrent aspiration pneumonia. She also had obstructive sleep apnoea and laryngomalacia with episodic stridor and desaturation during crying. Flexible laryngoscopy detected insucking of arytenoids and short aryepiglottic folds during inspiration. She was put on percutaneous gastrostomy feeding at 1-year-old.

She had severe developmental delay. By 18 months old, she had significant microcephaly and short stature. Her head circumference was 40 cm (significantly below 3rd-percentile), body height 76 cm (3-10th percentile) and body weight was 9.5 kg (25th percentile) on gastrostomy feeding. She cannot stand unsupported and was unable to perform pincer grasp. Echocardiogram revealed a small muscular ventricular septal defect, which was spontaneously closed at 1-year-old. Extensive workup including metabolic and neuroimaging were unremarkable.

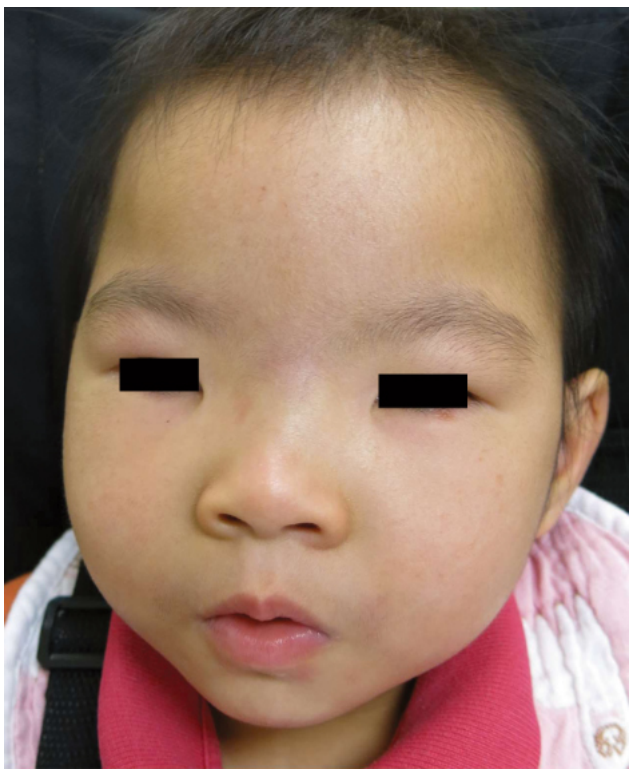

Figure 1 Clinical photo of the patient at 3 years old showing significant microcephaly and craniofacial dysmorphism – hypertelorism, epicanthic folds, low-set ears, wide and flat nasal bridge, downturned corners of mouth, a small lower jaw.

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