

Clinical Quiz

What is the Diagnosis?

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Our proband is a 5-month-old female who was referred from Maternal & Child Health Centre for suspected hypotonia with decreased limb movement since birth. She was born to a non-consanguineous couple via vaginal delivery at 40 weeks of gestation after an uneventful pregnancy, weighing 3.27 kg at birth. The family history was unremarkable and there was no history of neurometabolic diseases. Mother also had a history of first trimester spontaneous miscarriage, the cause of which was unknown.

Parents reported paucity of limb movement since birth. There was not much anti-gravity movement observed and the child did not withdraw with painful stimulation. No seizure-like activity or cyanosis was noted. Growth was satisfactory despite some choking, with body weight at 25-50th centile, height at 90th centile and head circumference

at 3-10th centile at 5 months of age. Clinical photographs can be seen in Figure 1.

Physical exam at 5 months of age showed a responsive non-dysmorphic baby with generalized hypotonia with frog like posture, positive scar sign and heel ear test and complete head lag. She also had tongue fasciculation and generalized areflexia. Minimal horizontal movement over lower limbs and some passive grasping over fingers were noted. She was not distressed in room air but there was some paradoxical breathing. Examination of other systems was unremarkable.

Basic blood tests were unremarkable and chest X-ray showed signs of atelectasis/aspiration likely related to weak respiratory muscle. She was referred to clinical genetics for further evaluation.



Figure 1 Clinical photographs of our patient at 6 months old (Consent for publication has been obtained).

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