

CC: ,Delayed motor development.,HX:, This 21 month old male presented for delayed motor development, ""jaw quivering"" and ""lazy eye."" He was an 8 pound 10 ounce product of a full term, uncomplicated pregnancy-labor-spontaneous vaginal delivery to a G3P3 married white female mother. There had been no known toxic intrauterine exposures. He had no serious illnesses or hospitalizations since birth. He sat independently at 7 months, stood at 11 months, crawled at 16 months, but did not cruise until 18 months.,He currently cannot walk and easily falls. His gait is reportedly marked by left ""intoeing."" His upper extremity strength and coordination reportedly appear quite normal and he is able to feed himself, throw and transfer objects easily. He knows greater than 20 words and speaks two-word phrases.,No seizures or unusual behavior were reported except for ""quivering"" movement of his jaw. This has occurred since birth. In addition the parents have noted transient left exotropia.,PMH: ,As above.,FHx:, Many family members with ""lazy eye."" No other neurologic diseases declared.,9 and 5 year old sisters who are healthy.,SHx:, lives with parents and sisters.,EXAM:, BP83/67 HR122 36.4C Head circumference 48.0cm Weight 12.68kg (70%) Height 86.0cm (70%),MS: fairly cooperative.,CN: Minimal transient esotropia OS. Tremulous quivering of jaw--increased with crying. No obvious papilledema, though difficult to evaluate due to patient movement.,Motor: sat independently with normal posture and no truncal ataxia. symmetric and normal strength and muscle bulk throughout.,Sensory: withdrew to

vibration.,Coordination: unremarkable in BUE.,Station: no truncal ataxia.,Gait: On attempting to walk, his right foot rotated laterally at almost 70degrees. Both lower extremities could rotate outward to 90degrees. There was marked passive eversion at the ankles as well.,Reflexes: 2+/2+ throughout.,Musculoskeletal: pes planovalgus bilaterally.,COURSE: ,CK normal. The parents decided to forego an MRI in 8/90. The patient returned 12/11/92 at age 4 years. He was ambulatory and able to run awkwardly. His general health had been good, but he showed signs developmental delay. Formal evaluation had tested his IQ at 87 at age 3.5 years. He was weakest on tasks requiring visual/motor integration and fine motor and visual discrimination skills. He was 6 months delayed in cognitive development at that time. On exam, age 4 years, he displayed mild right ankle laxity on eversion and inversion, but normal gait. The rest of the neurological exam was normal. Head circumference was 49.5cm (50%) and height and weight were in the 90th percentile. Fragile X analysis and karyotyping were unremarkable.