Pediatrics-1

Spinal Muscular Atrophy Type I: Cases of Normal Cognitive Function Despite having Limited Motor Function and Physical-Environmental Interaction

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Objective: To examine cognitive development in children with Spinal Muscular Atrophy Type 1 (SMA 1), a population with extremely limited motor function. SMA is an autosomal recessive neurodegenerative disease of spinal cord motor neurons. The most severe form, Type 1, presents prior to 6 months with extreme weakness, inability to sit independently, and early death (often by age 2 years). Recent medical advances have increased longevity in these children. It is unknown whether their limited ability to interact physically with their environment may negatively impact intellectual development. We hypothesized that children with SMA 1 have below average cognitive performance due to limited physical-environmental interaction. Method: Receptive vocabulary and nonverbal reasoning skills were examined in 4 children with SMA 1 (3 males and 1 female, ages: 4, 8, 12, 18 years). Measures included: Peabody Picture Vocabulary Test – 4 (PPVT), Raven’s Coloured Progressive Matrices (RCPM), and Comprehensive Test of Nonverbal Intelligence (CTONI). Information on birth and medical history was collected and a physical therapist performed examination of motor function. Highest motor function ever achieved was sitting with support. Results: Receptive vocabulary and nonverbal reasoning were at or above average in the three younger children (PPVT range 55–97%ile; RCPM range: 86–100%ile). The older participant scored slightly lower, yet still within normal limits (PPVT = 23%ile; CTONI = 8%ile). Conclusion: Children with SMA 1 have normal cognitive function despite having limited motor function and physical interaction with their environment. Better understanding of their cognitive skills will contribute to increased quality of life for the children and their families.