SPINAL MUSCULAR ATROPHY TYPE 4: A CASE REPORT

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Background

Spinal muscular atrophies (SMA) are a group of inherited neuromuscular disorders caused by degeneration of alpha motor neurons in the spinal cord, resulting in progressive muscle weakness and atrophy. Most forms of SMA are autosomal recessive and associated with biallelic deletions or mutations in the SMN1 gene on chromosome 5q13. Clinically, SMA is classified based on age at onset and maximal motor function: type 1 (non-sitters), type 2 (sitters), type 3 (walkers with childhood onset), and type 4 (adult onset). All types present with symmetrical proximal muscle weakness, more pronounced in the lower limbs, and reduced or absent deep tendon reflexes. Diagnosis relies on clinical evaluation, EMG/EMNG findings, and confirmatory genetic testing.

Case report

We report a case of a 67-year-old woman evaluated in June 2016 by a physiatrist after surgical treatment of a left femur fracture. She had a previous forearm fracture in March 2016. Rehabilitation was slow, with persistent gait impairment and frequent falls. Clinical examination revealed proximal lower limb weakness and impaired ambulation using forearm crutches. EMNG findings showed generalized lower motor neuron involvement. Genetic testing confirmed SMA type 4. Risdiplam therapy (0.75 mg/mL once daily) was initiated in December 2023. Following an influenza infection in January 2024, the patient required mechanical ventilation. Post-recovery, she experienced worsening gait difficulties, painful knee recurvatum, and transitioned to a walker. In February 2024, motor assessments showed: MMT, RHS 32/69, RULM 43/43. Barthel Index was 68; 6-minute walk test was not feasible due to knee pain. She was prescribed KAFO orthoses and enrolled in a personalized rehabilitation program focusing on individual kinesitherapy.

Conclusion

This case underscores the importance of recognizing adult-onset SMA, which may present subtly and be misdiagnosed. Timely diagnosis, initiation of disease-modifying treatment, and ongoing rehabilitation are crucial for maintaining function, reducing disability, and improving quality of life in adult SMA patients.

Keywords: SMA, weakness, neurorehabilitation