Spinal Muscular Atrophy with Two SMN2 Copies and Preserved Deep Tendon Reflexes: An Extremely Rare Case

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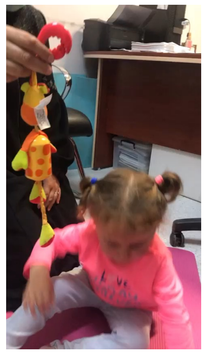
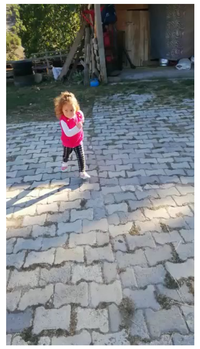
Spinal muscular atrophy (SMA) type 1 is the most severe form of SMA, in which deep tendon reflexes cannot be elicited. It accounts for 50-70% of cases of childhood onset SMA. Our search of the literature reveal no instances of SMA with preserved reflexes except for one case report[1].

The present report describes a child aged 35 months with SMA with two SMN2 copies with preserved deep tendon reflexes, an extremely atypical phenomenon.

We report this extremely rare case together with video images. The patient was diagnosed with SMA type 1 (two SMN2 copy numbers) as a result of SMN gene analysis. She had two siblings who died due to SMA at six months of age. No hypoxia was present in her prenatal, natal, or postnatal histories. Intrathecal nusinersen therapy was initiated at the age of 1.5 months. The case was evaluated together with history and neurological examination findings.





The patient was able to walk independently, run, and climb stairs at physical examinations performed at 35 months of age. Her HFMSE score was 57. She was capable of using a fork and spoon and of forming three or four-word sentences. Bilateral deep tendon reflexes were evaluated as normoactive since she was 1.5 months years old (++/++ from the patella) (video images are available).

When she was 27 month of age, her electromyelography was referred as normal action potentials and conduction velocities in motor and sensory nerves. Normal muscle unit action potentials were detected in the right biceps brachii/ right deltoid and right tibialis anterior and medial head of right gastrocnemius muscle. The denervation potentials was not detected.

A number of other genes associated with SMA have also been identified. NAIP is one of them. In a study, it was reported that the deterioration in respiratory system functions was more rapid in cases with a deletion in the NAIP gene.Some pathogenic variants have been reported to show milder clinical phenotype. Hyperactive deep tendon reflexes may indicate an underlying comorbidity such as hemiplegia or diplegia. However, from these perspectives, birth history and neurological examination findings did not indicate any of these factors in our patient.

This is an extremely interesting case in terms of the preserved deep tendon reflexes as well as a favorable course in the prognosis of motor functions.

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