Disabling Head Tremor in a Patient with DYT1 Mutation

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Dystonic head tremor is known to be a feature in some patients with DYT1 mutation. However, isolated tremor of the head without relevant cervical dystonia has rarely been described. We report here a patient with the three-bp GAG deletion in the DYT1 gene (904_906delGAG) who had severe head tremor in the frame of a generalized limb dystonia.

Key Words: DYT1 gene, Dystonia, Tremor.

Although the spectrum of dystonia produced by the DYT1 GAG deletion is broad, in-
including both childhood and adult onset and variable progression of signs from focal to widespread involvement, the typical phenotype usually is early-onset dystonia developing before the age of 26 years in a limb, particularly the leg, and rapidly generalizes within 5 years from onset. Selective or pronounced cranio-cervical involvement is said to be atypical in DYT1 dystonia. Onset of dystonia in the facial or neck muscles appears to be a much rarer feature. In the vast majority of cranio-cervical involvement, dystonia begins in a limb, and subsequently spread to the cranial-cervical region in the frame of a generalized phenotype.

Tremor in DYT1 dystonia is usually dystonic tremor or rarely isolated postural hand tremor. Head tremor accompanying cervical dystonia is known to be a feature in some patients. However, disabling head tremor in the absence of relevant cervical dystonia has rarely been described in DYT1 dystonia. Although co-occurrence of essential tremor was a diagnostic consideration, some of the clinical features shown by this patient would be quite unusual for such a diagnosis. He used maneuvers to attenuate the head tremor, which were similar to “sensory tricks” observed in patients with cervical dystonia. There was also striking change in his tremor on his head position, which is typical of dystonia. Isolated head tremor associated with these features may be initial manifestation of cervical dystonia. In these cases, neck deviation did not become apparent for many years, and eventually developed obvious cervical dystonia. Therefore, long-term clinical follow-up will confirm the nature of his head tremor.

We describe here atypical clinical features in a patient with DYT1 mutation who had isolated disabling head tremor in the frame of a generalized limb dystonia.

REFERENCES