Sarcouncil Journal of Medical Series

ISSN(Online): 2945-3550

Volume- 04| Issue- 06| 2025

Letter to the Editor **Received:** 02-05-2025 | **Accepted:** 25-05-2025 | **Published:** 12-06-2025

Cervical Myelopathy C5-6 in Myotonic Dystrophy-1 Due to Neck Muscle Weakness Should Not Be Confused with Hirayama Disease

Josef Finstere
MD, PhD Neurology Dpt., Neurology & Neurophysiology Center, Vienna, Austria

Keywords: myotonic dystrophy, Hirayama disease, muscle weakness, DMPK, electromyography.

LETTER TO THE EDITOR

We were interested to read the article by Dudhatra, *et al*. about a 31-year-old man with myotonic dystrophy type-1 (MD1) due to CTG repeat expansion >50 repeats in DMPK, who was also diagnosed with Hirayama disease (HD) [Dudhatra, A. V. *et al*., 2025]. Since the age of 26, the patient developed hand and forearm weakness, muscle wasting and bilateral myotonia with left-sided dominance without sensory disturbances and weakness of the neck extensor muscles [Dudhatra, A. V. *et al*., 2025]. The patient was treated with a neck brace and physiotherapy [Dudhatra, A. V. *et al*., 2025]. The study is noteworthy, but several points should be discussed.

The first point is that the diagnosis of HD is questionable. Several arguments can be made against the diagnosis. The patient is 31 years old, but HD usually begins in adolescence (15 to 25 years). The HD usually affects the segments C7 to Th1, but not C5-6, as in the index patient [Lay, S. *et al*., 2024]. Initially, the HD is unilateral and spreads to the other hand over time, a dynamic that was not described in the index patient. The brachioradialis muscle is spared in HD, but was described as "relatively spared" in the index patient [Boruah, D. K. *et al*., 2018]. A further argument against HD is that the hand and forearm muscle weakness cannot be explained by myelopathy C5-C6. As MD1 is often associated with a dropped head, we should know whether the index patient was able to keep his head upright without support.

The second point is that the exact number of expanded repeats has not been determined [Dudhatra, A. V. *et al*., 2025]. Since the number of CTG repeats determines the phenotype [Overend, G. *et al*., 2019], it is crucial to know the exact number of expanded CTG repeats to establish a genotype-phenotype correlation. As patients carrying 50-100 repeats may usually be mildly symptomatic or asymptomatic [Pratte, A. *et al*., 2015], the patient should be reassessed with a more specific test.

The third point is that the parents have not been tested for the mutation [Dudhatra, A. V. *et al*., 2025]. Since the mother or father could be carriers of a pre-mutation (38-50 repeats) that does not cause symptoms, and since CTG repeats may continue to increase in their offspring, a phenomenon known as anticipation, it would have made sense to test both parents for the mutation as well.

The fourth issue is that the index patient was not systematically screened for multisystem involvement [Dudhatra, A. V. *et al*., 2025]. MD1 is a multisystem disease that affects all organs and tissues, but especially the brain, nerves, muscles, eyes, heart, endocrine organs and the gastrointestinal tract. Although the patient denied involvement of body regions other than the upper limbs, involvement may be subclinical, which can be overlooked if not systematically searched for. Did the patient have a cataract or atrioventricular block I?

In summary, this interesting case report has limitations that affect the results and their interpretation. Addressing these limitations could strengthen the conclusions and support the message of the study. Cervical myelopathy C5-6 in myotonic dystrophy-1, which is thought to be due to neck muscle weakness, should not be confused with Hirayama disease.

AUTHOR CONTRIBUTION: JF was responsible for the design and conception, discussed available data with coauthors, wrote the first draft, and gave final approval. FS and CS: contributed to literature search, discussion, correction, and final approval

***Corresponding Author:** Josef Finsterer

DOI- https://doi.org/ 10.5281/zenodo.15659460

REFERENCES

1. Dudhatra, A. V., Arora, R. K. & Poncha, F. F. "Diagnostic crossroads: Coexistence of myotonic dystrophy type 1 and Hirayama disease in a case report." *Neurology India*, 73.2 (2025): 368–370.
2. Lay, S., Gudlavalleti, A., Munakomi, S, *et al.* "Monomelic amyotrophy (Hirayama disease) [Updated 2024 Nov 9]." *StatPearls [Internet]*. Treasure Island (FL): StatPearls Publishing; 2025 Jan–. Available from: https://www.ncbi.nlm.nih.gov/books/NBK499913/
3. Boruah, D. K., Prakash, A., Gogoi, B. B., Yadav, R. R., Dhingani, D. D. & Sarma, B. "The importance of flexion MRI in Hirayama disease with special reference to laminodural space measurements." *American Journal of Neuroradiology*, 39.5 (2018): 974–980.
4. Overend, G., Légaré, C., Mathieu, J., Bouchard, L., Gagnon, C. & Monckton, D. G. "Allele length of the DMPK CTG repeat is a predictor of progressive myotonic dystrophy type 1 phenotypes." *Human Molecular Genetics*, 28.13 (2019): 2245–2254.
5. Pratte, A., Prévost, C., Puymirat, J. & Mathieu, J. "Anticipation in myotonic dystrophy type 1 parents with small CTG expansions." *American Journal of Medical Genetics Part A*, 167A.4 (2015): 708–714.

Source of support: Nil;
Conflict of interest: Nil.

Cite this article as:
Finsterer, J. "Cervical Myelopathy C5-6 in Myotonic Dystrophy-1 Due to Neck Muscle Weakness Should Not Be Confused with Hirayama Disease." *Sarcouncil Journal of Medical Series* 4.6 (2025): pp 3-4.

Publisher: SARC Publisher