Polymyoclonus aggravated by neck flexion as the isolated presenting symptom of Hirayama disease: case report

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thyroid function, and autoimmune-related tests, were all within normal limits. Electroencephalography and nerve conduction findings were unremarkable. However, needle electromyography revealed unit potentials in muscles (abductor pollicis brevis, first dorsal interosseous and triceps muscles) innervated by the C7 to T1 roots that were of significantly higher amplitude than other muscles. With a working diagnosis of HD, we ordered cervical magnetic resonance imaging (MRI) in both neutral and flexed positions, which revealed a clear anterior displacement of the posterior dural sac compressing the C7–8 roots (Fig. 1), which is an imaging hallmark of HD. With the above clinical, electrophysiological, and imaging findings, the patient was diagnosed with HD.

Discussion and conclusions
The pathogenic mechanism of HD is yet to be elucidated, but recent imaging studies have strongly suggested microcirculatory ischemia in the anterior horn cells caused by direct cord compression resulting from forward effacement of the posterior dural sac [6]. This can be easily observed using cervical MRI in the flexed posture, which shows direct compression of the C7-T1 levels, corresponding with the muscle weakness. Another hypothesis postulates that the pathogenesis of HD can be explained by an imbalance between growth of the vertebral column and the spinal canal [1]. It is noteworthy that a large cohort study revealed a 90% male preponderance, which indirectly reflects the important role of accelerated vertical growth during puberty. Besides weakness, cold paresis is known to be commonly observed among HD patients, as is tremor, which has a reported prevalence of approximately 70–80% among HD patients in contemporary studies [1, 2].

The underlying mechanism of hand tremor has not yet been elucidated, but a recent report of multiparametric brain MRI on an HD patient revealed brain hyperactivation that may explain the hand tremor associated with HD. A case report described coexisting juvenile myoclonic epilepsy in a young male HD patient [5]. Another hypothesis is the muscle myoclonus that originate from the motor nerve injury cause by neuroexcitatory mechanism [7]. Whatever is the cause, it is important to recognize HD-associated tremors, and narrowing down the differential diagnosis is pivotal for arriving at the appropriate treatment. Most publications that describe the clinical features of HD describe a high prevalence of hand tremor or polymyoclonus among the comorbid symptoms, but tremor as a predominant clinical symptom of HD is not well described.

Although HD is a self-limiting disease, it is important to recognize the disease that should lead to the avoidance of neck flexion because it usually aggravates the symptoms. Few reports have described positive results after cervical surgery or cervical collar therapy that reduced functional disability among HD patients [8, 9].

Only recent publications have begun to illustrate and emphasize polymyoclonus as a clinical feature of HD. A recent case report described a 17-year-old boy who presented with polymyoclonus along with decreased muscle bulk associated with weakness. Of note, the myoclonus was aggravated by outstretching the arms [3]. Another report described a 20-year-old male with asymmetric muscle weakness and myoclonic tremors in resting and arm-outstretched postures [5]. These reports emphasized the importance of suspecting HD when a patient presents with predominant polymyoclonus along with mild muscle weakness.

Our report is of clinical significance as it describes the first reported patient who presented with isolated polymyoclonus without muscle weakness or muscle atrophy. The patient also had no complaints related to muscle weakness or muscle atrophy. Furthermore, the myoclonus was aggravated by neck flexion, corresponding well with the imaging findings of the posterior dural sac compression in HD. This suggests that neck flexion should be avoided as it may exacerbate the symptoms.
with the hypothesis of direct cord compression, which is generally accepted as the main pathophysiologic mechanism of HD. Clinicians should be aware of a self-limiting disease, such as HD, when evaluating young, most frequently male patients who present with predominant functional polymyoclonus that is worse with neck flexion and perform an electromyography study for the earlier diagnosis that may prevent further clinical deterioration.

Supplementary information
Supplementary information accompanies this paper at https://doi.org/10.1186/s12883-020-01904-z.

Additional file 1: Video 1. The video shows polymyoclonus of the right hand which is aggravated by neck flexion.

Abbreviation
HD: Hirayama disease

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Consent for publication
The informed and written consent was obtained from the patient for publication of this case report.

Competing interests
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