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CLINICAL IMAGE

Recurrent pulmonary embolism complicated with myotonic dystrophy type 1

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hatchet face, myotonic dystrophy type 1, pulmonary embolism

A 40-year-old man fell down the stairs from the second to the first floor and was transported to the hospital with the suspect of high-energy trauma. He had a history of deep vein thrombosis and pulmonary embolism (DVT/PE) at the age of 35. At that time, ACA, PCA, PSA, ANA, aCLA, β2-GPIA, and coagulation function tests were performed to confirm the absence of congenital coagulation abnormalities, all of which were within the reference values. An ECG examination was also performed at that time, but no abnormal waves were identified. Therefore, both DVT/PE events were judged to be idiopathic. Two previous transports in which he fell down the stairs because of fainting, both determined to be because of DVT/PE. When he came to our hospital, we confirmed from the patient that there was no confusion after the fall. His general condition was so-so, with blood pressure of 120/84 mmHg, respiratory rate of 15, and heart rate of 75. General examination revealed minor injury; no other systemic findings of note including urinary incontinence and tongue bite. Laboratory tests including creatinine kinase and blood glucose level were within normal range other than a D-dimer of 880 ng/mL. There were no abnormal findings on the electrocardiogram or transthoracic echocardiography, and there was no brady- or tachyarrhythmia on the monitored electrocardiogram. Since the patient had a history of recurrent syncope and fell down derived from DVT/PE of unknown cause, contrast CT of the chest and thighs was performed to rule out the recurrence. The result shows, however, no pulmonary artery defects nor thrombi in either femoral vein. When we observed the patient's physical examination to confirm the cause of the fall, we found a frontal bald head and an ax-like facial expression (Figure 1), and the patient had poor articulation. Manual muscle test showed distal-predominant muscle weakness. Myotonia of the grasp was also observed (Figure 2). Myotonic dystrophy was

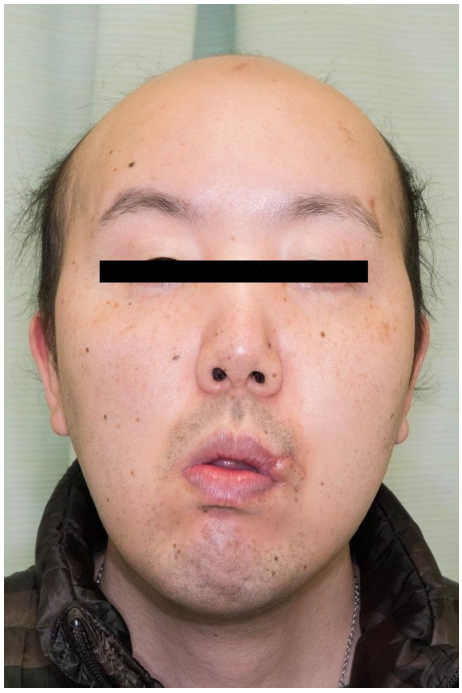

FIGURE 1 Frontal baldness and ax-like facial features are noted. Scar on the left upper lip was made at the time of the transfer.

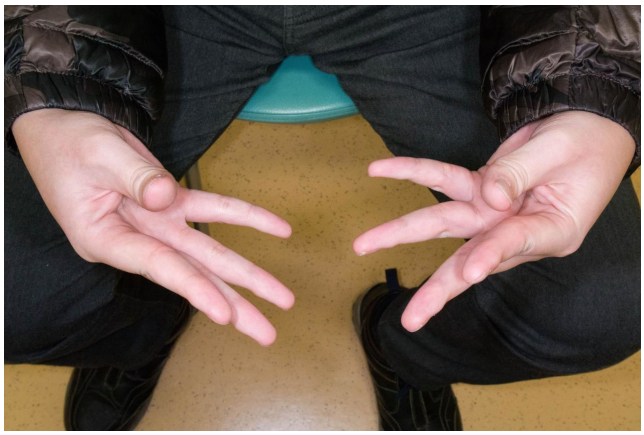

FIGURE 2 The patient instructed to hold his hand tightly and open it immediately after. It shows how the grasping muscles are difficult to relax.

strongly suspected, and leg tangle because of muscle weakness was thought to be the cause of his fall down. The patient was consulted by a neurologist who confirmed the CTG repeat extension of the myotonic dystrophy protein kinase gene, leading to a diagnosis of myotonic dystrophy type 1. The patient was followed by a neurologist while anticoagulants were continued.

Myotonic dystrophy is a hereditary neurological disease that causes multiple organ problems, including cardiovascular diseases.[1] In recent years, it has begun to be reported that myotonic dystrophy is more likely to cause DVT/PE than other dystrophies[2] or healthy individuals.[3] Referring to the results of a multivariate analysis presented in a previous report,[3] dyspraxia and a history of DVT were risk factors. RNA toxicity,[3] mis-splicing of factors related to the coagulation process,[3] activation of coagulation by muscle degeneration itself,[4] and factor-V(leiden) mutation[5] may be considered of the causative factor. The fact that D-dimer was elevated at the time of initial treatment in this case and there was no gross thrombosis suggests that a coagulation/fibrinolysis mismatch may have occurred in this case. Following the comment that DVT/PE has prognostic implications,[3] we initiated anticoagulation therapy; no conclusion has been reached regarding the statistical usefulness of introducing it to the best of our knowledge. In this case, the first two falls were considered to be the result of DVT/PE associated with myotonic dystrophy, and only the third fall was considered to be because of entanglement of the legs caused by muscle weakness. In conclusion, it will be useful to suspect myotonic dystrophy based on physical examination when repeated DVT/PE with an unknown predisposing factor is observed and to provide appropriate consultation.

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CONFLICT OF INTEREST STATEMENT

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REFERENCES

1. Gutiérrez Gutiérrez G, Díaz-Manera J, Almendrote M, Azriel S, Eulalio Bárcena J, Cabezudo García P, et al. Clinical guide for the diagnosis and follow-up of myotonic dystrophy type 1, MD1 or Steinert's disease. Neurologia (Engl ed). 2020;35(3):185–206.
2. Wahbi K, Sochala M, Porcher R, Stojkovic T, Behin A, Louis SL, et al. Venous thromboembolism in adult patients with inherited myopathies: a high-risk in myotonic dystrophy. Neuromuscul Disord. 2017;27:S179.
3. Sochala M, Porcher R, Stojkovic T, Bécane HM, Béhin A, Laforêt P, et al. High risk of fatal and nonfatal venous thromboembolism in myotonic dystrophy. Circulation. 2018;138:1169–71.
4. Gallinoro E, Papa AA, Rago A, Sperlongano S, Cassese A, Cioppa ND, et al. Myotonic dystrophy type 1 and pulmonary embolism; successful thrombus resolution with dabigatran etexilate therapy. Acta Myologica. 2018;37:227–31.

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