

Reply from the Author

Juvenile neuronal ceroid-lipofuscinosis with hypertrophic cardiomyopathy and left ventricular noncompaction: a case report

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Thank you for your valuable comments and suggestions¹⁾ regarding our case report "Juvenile neuronal ceroid-lipofuscinosis with hypertrophic cardiomyopathy and left ventricular non-compaction"²⁾. Below, we provide our responses to your questions concerning: (1) our rationale for diagnosing the patient with juvenile neuronal ceroid-lipofuscinosis (JNCL); (2) the details of our echocardiographic findings in the patient, particularly at the site of left ventricular (LV) noncompaction; and (3) the results of our search for cardiac complications among the patient's family members.

In terms of your suggestion to perform DNA diagnostic testing for JNCL in this patient, we were also interested in the DNA diagnostic testing, but were unable to do it because the patient's family did not provide consent. Electron microscopy revealed lipofuscin granule-like material with high electron density in the cytoplasm of the patient's lymphocytes. On electrophysiological testing, the patient had attenuated responses on the electroretinogram. We therefore made a diagnosis of JNCL on the basis of these test results and the patient's clinical course. No diagnostic criteria have been established for JNCL, and "diagnosis is primarily made on clinical grounds, documented by appropriate neuroradiologic and electrophysiologic studies, and confirmed by the appropriate enzymatic or DNA-based laboratory tests"³⁾. Thus, although DNA diagnostic testing reinforces a diagnosis of JNCL, it is not necessarily essential.

On echocardiography, this patient showed asymmetrical LV wall thickening from the LV posterior wall to the LV apex. The patient also had normal LV contractility but showed LV diastolic dysfunction, leading us to a diagnosis of hypertrophic cardiomyopathy (HCM). At the same time, a reticulated pattern of trabeculation and deep interstice was observed medial to the

apex, and the ratio of the non-compacted layer (NC) to the outer compacted layer (C), or NC/C ratio, was ≥ 2 , indicating LV noncompaction⁴⁾. This noncompaction was limited to the apex, whereas the posterior wall only showed myocardial thickening.

We have regularly assessed the patient's HCM and LV noncompaction using echocardiography and long-term electrocardiography (ECG), but we have not to date discovered any thrombosis, embolism, or significant arrhythmia. However, we had not conducted regular testing of family members for cardiac complications until we received your suggestions. We performed chest radiography, ECG, and echocardiography on the patient's parents and siblings, but no particular abnormalities were identified. Nevertheless, since, as you pointed out, LV noncompaction is a form of primary cardiomyopathy in which genetic factors play an important role⁴⁾, we will continue to regularly assess the patient's family for cardiac complications.

Finally, we would like to thank you once again for your valuable comments and suggestions.

※ The authors declare there is no conflict of interest relevant to this article.

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