

## Letters to the Editor

# Left ventricular hypertrabeculation/noncompaction in juvenile neuronal ceroid lipofuscinosis

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(Rinsho Shinkeigaku (Clin Neurol) 2015;55:185)

With interest we read the article by Murata et al. about a 17yo female with a 10y history of severely disabling juvenile neuronal ceroid lipofuscinosis (JNCL) in whom left ventricular hypertrabeculation/noncompaction (LVHT) was diagnosed during hospitalisation for aspiration pneumonia and consecutive heart failure<sup>1)</sup>. We have the following comments and concerns.

The patient is interesting for the association between JNCL and LVHT, which has not been reported previously. Since LVHT is most frequently associated with neuromuscular disorders (NMDs) or chromosomal defects, it would be interesting to know how neuronal ceroid lipofuscinosis (NCL) was diagnosed. NCL are rare genetically inherited neurodegenerative disorders, most frequently in children, and classified according to age of onset as congenital, infantile, late-infantile, juvenile, or adult-onset or by mutated genes as CLN10/CTSD, CLN1/PPT1, CLN2/TPP1, CLN3, CLN5, CLN6, CLN7/MFSD8, or CLN8<sup>2)</sup>. Was the diagnosis ever established upon a biochemical or genetic basis or only based on lipofuscin-like material in peripheral lymphocytes? Enzyme activity assays are helpful in identifying several of these disorders; however confirmation of a mutation in one of the NCL genes is vital for the definitive diagnosis<sup>2)</sup>. A further argument challenging the diagnosis of a JNCL is that myocardial thickening is not a typical feature of JNCL, as mentioned in the discussion<sup>1)</sup>. Did the described posterior wall thickness of 11–13 mm include the non-compacted layer?

LVHT can occur familiarly<sup>3)</sup>. Irrespective of the causal background, it is thus essential that first-degree relatives of the patient are also investigated for both LVHT and JNCL. If both disorders do not co-segregate between the generations, a causal relation is rather unlikely. If other family members also have LVHT without JNCL, search for other LVHT-associated

disorders is strongly recommended. A further reason for considering other conditions more frequently associated with LVHT than JNCL is the absence of a causal relation between LVHT and JNCL. In particular, search for NMDs by determining muscle enzymes, nerve conduction studies, needle electromyography, and by investigating other family members, or for chromosomal defects has to be carried out.

Since LVHT may be also acquired we suggest to reviewing echocardiographic investigations prior to the one at age 17yo including intrauterine ultrasound. Overall, the presented case is interesting but the diagnosis needs to be genetically confirmed, other family members investigated for LVHT and JNCL, a NMD and chromosomal defect excluded, and previous echocardiographies reviewed. Not only regular echocardiographic investigations are required in LVHT but also long-term ECG recording, for example by implantation of a reveal, in order to detect arrhythmias, which are frequently associated with LVHT.

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

## References

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(Received: 16 February 2014)