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Letter to the Editor

Non-Compaction in a Homozygous *NPHP1* Deletion Carrier

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In a recent article, Brndiarova *et al*., reported about a 17yo female with nephronophtisis type-1 due to a homozygous deletion in *NPHP1(*Brndiarova, M., *et al*.,2019). Her parents were third degree cousins and each of them carried the culprit mutation as well (Brndiarova, M.,*et al*.,2019). Interestingly, echocardiography at age 17y revealed left ventricular hypertrabeculation (LVHT), also known as non-compaction, which was not present on echocardiography at age 3y (Brndiarova, M.,*et al*.,2019). LVHT thus has to be classified as acquired (Finsterer, J., & Stollberger, C. 2017). We have the following comments and concerns.

LVHT is frequently associated with neuromuscular disorders (NMDs) (Finsterer, J.,*et al*.,2010). Were there any indications of neuropathy or myopathy in the index case? Normal neurological examination, as described in the article, in the light of chronic kidney disease is unusual since at least nephrogenic neuropathy is likely. We should know if creatine-kinase or serum lactate were elevated and if nerve conduction studies were indicative of subclinical neuropathy of the lower limbs.

Genetic work-up for LVHT is not reasonable as the number of mutated genes and chromosomal defects associated with LVHT is steadily increasing and as a causal relation between LVHT and any of these genetic defects has not been proven thus far. To find out if LVHT was associated with the *NPHP1* deletion it is crucial to screen the consanguineous parents for LVHT. If cardiac imaging in the parents also discloses LVHT, a causal relation is quite likely (familial LVHT) (Finsterer, J.,*et al*.,2013).

Since LVHT is frequently associated with chromosomal defects(Finsterer, J. 2009), we should know if the index patient carried a second trouble in addition to the *NPHP1* deletion in the form of a chromosomal deletion, duplication, translocation, or insertion. A chromosomal defect is likely in the light of the parents' consanguinity.

LVHT is frequently complicated by cardio-embolic events, systolic dysfunction or heart failure, ventricular arrhythmias, or sudden cardiac death (Stöllberger, C.,*et al*.,2015). We should know if the patient was regularly screened for complications of LVHT, and if her individual history was positive for any of these complications. Screening LVHT patients for potential complications is crucial as they may strongly determine the outcome of these patients.

Since the patient required kidney transplantation, and received immunosuppression, we should know if she tolerated the immunosuppression without side effects. Regularly screening transplanted patients is crucial as they may not only develop host-versus graft or graft-versus host reactions but also neuromuscular side effects due to the immunosuppressive medication. Particularly from steroids, cyclosporine, mycophenolate mofetil, sirolimus, and tacrolimus, it is well known that they are

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myotoxic and potentially trigger the development of a NMD.

The authors describe LVHT as congenital in the introduction but present a case with acquired LVHT. We propose revision of the echocardiography at age 3y to confirm that LVHT was truly absent and thus acquired.

Overall, this interesting case could be more meaningful if the index patient was screened for NMD, if the consanguineous parents were screened for LVHT, if a chromosomal defect was excluded, if surveillance for complications of LVHT was initiated, and if the index patient was prospectively screened for neuromuscular side effects of the immunosuppressive medication after kidney transplantation.

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