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CASE REPORT

Is mitochondrial disease the common cause of histiocytoid cardiomyopathy and non-compaction?

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Dear Sirs,

We appreciated reading the report by Edston and Perskvist [1] about the association of histiocytoid cardiomyopathy (hcCMP) and left ventricular hypertrabeculation (LVHT), also known as noncompaction, in a baby who was found dead at home at age three and a half months. The report evokes the following concerns: hcCMP and LVHT are occasionally associated with mitochondrial disease (MD). hcCMP was not only associated with the 8344A>G mtDNA mutation, but also with the 15498G>A transition. Furthermore, hcCMP was reported in a patient with lactacidosis and agenesis of the corpus callosum and also in a female infant with skeletal muscle myopathy and generally decreased cytochrome-c oxidase activity on muscle biopsy. Additionally, in cardiomyocytes of hcCMP patients the number of mitochondria is increased, their structure is abnormal, and the activity of succinate-cytochrome-c reductase and NADH-cytochrome-c reductase is decreased [2]. Why was neither the myocardial biopsy nor the skeletal muscle investigated for features of MD? Were any abnormalities found on immunohistochemistry, electron microscopy, or on biochemical investigations, which were indicative of mitochondrial disease?

Little information about the clinical findings of the baby are provided. Was the baby a floppy infant? Were tendon reflexes exaggerated or reduced? Was there a problem with sucking or swallowing, or were the postural reflexes absent? Did she develop seizures? Where there any dysmorphic features? Were creatine-kinase, lactate, or pyruvate ever elevated in the serum or organic acids reduced in the urine?

There is also little information concerning the family history of the baby. Was the family history positive for neuromuscular disease? LVHT is frequently associated with myopathy, with chromosomal abnormalities, and sometimes even with hereditary neuropathy. Most frequently LVHT is associated with Barth syndrome, mitochondrial disease, or the 1p36 deletion syndrome. Were the first degree relatives investigated by a neurologist? Were muscle enzymes or lactate determined in any of the relatives?

If the authors regard hcCMP as an X-linked disease, the infant's mother needs to be a carrier of the disease-linked mutation. She might have presented with rudimentary symptoms or signs of hcCMP. Did she ever report palpitations, exertional dyspnea, or edema of the legs? Was she ever investigated cardiologically? Did she ever undergo echocardiography?

Although it is a widely propagated opinion that LVHT is a congenital abnormality, there are isolated cases in which LVHT developed years after birth [3]. Acquired LVHT needs to be kept in mind when discussing the pathogenesis of LVHT. All cases with acquired LVHT were also suffering from muscle disease [4].

There are only few reports about the histology of the trabeculations in LVHT. Were abnormalities typical for

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hcCMP also found in the trabeculations? What was the ratio between the non-compacted layer and compacted layer? Where was LVHT located within the left ventricle? Were there sub-endocardial calcifications as has been previously reported [5]?

To further elucidate the pathogenesis of LVHT and hcCMP, the presented case and all first degree relatives require further diagnostic evaluation. There is also a need to review myocardial histology and, if available, muscle biopsy for neuromuscular disorders.

Conflict of interest statement There are no financial or other conflicts of interest.

References

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