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Letter to the Editor

1q43 deletion and left ventricular hypertrabeculation / noncompaction

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Keywords: with left ventricular hypertrabeculation / noncompaction (LVHT), DTNA (dystrobrevin), or CSX, lamin A/C, cypher/ZASP, GAA, DMPK, AMPD1.

With interest we read the article by Kanemoto *et al.,* (2006) on a female infant patient with a de novo interstitial deletion of the band 1q43 associated with left ventricular hypertrabeculation / noncompaction (LVHT) (Kanemoto, N. *et al.,* 2006). The article raises some objections and concerns:

It is a widespread error to regard LVHT only as a congenital disorder. Though most likely congenital in the majority of the cases, LVHT has been also described to occur spontaneously after birth (Finsterer, J. *et al.,* 2004; Finsterer, J. *et al.,* 2006). Acquired LVHT has been described in association with mitochondrial disorder and Duchenne muscular dystrophy (Finsterer, J. *et al.,* 2004; Finsterer, J. *et al.,* 2006).

It is not clear according to which criteria LVHT was defined. At least 3 echocardiographic definitions are currently available (Chin, T.K. *et al.,* 1990; Oechslin, E.N. *et al.,* 2006; and Stöllberger, C. *et al.,* 2002). No echocardiographic figure of LVHT in the presented patient is provided. At which segments was LVHT located? Was LVHT confirmed by cardiac MRI? 'Were persisting sinusoids excluded by coronary angiography as a differential diagnosis?

LVHT has not only been described in association with mutations in the G4.5, DTNA (dystrobrevin), or CSX genes, but also in association with mutations in the lamin A/C, cypher/ZASP, GAA, DMPK, AMPD1, mitochondrial, frataxin, and PMP22 genes (Finsterer, J., *et al.,* 2005). Additionally, LVHT has been described in association with Turner syndrome, Ohtahara syndrome, Roifman syndrome, Noonan syndrome, patella syndrome, Melnick needles syndrome, MIDAS syndrome, DiGeorge syndrome, congenital adrenal hyperplasia, distal 4q trisomy and distal 1q monosomy, distal chromosome 5q deletion, trisomy 11, and trisomy 13 (Finsterer, J., *et al.,* 2005). Did the authors look for mutations in these genes and did they exclude the described disorders?

Why was the patient investigated for desmin mutations, which have been associated only with dilated cardiomyopathy but not with LVHT in humans? To our knowledge, only in a mouse model LVHT has been associated with desmin mutations so far.

LVHT is frequently associated with neuromuscular disorders and the presented patient showed hypotonia. Was hypotonia due to muscular weakness or of central origin? Was the girl investigated for neuromuscular disease? Did the patient undergo neurological investigations at all? Was the patient also investigated for dystrophinopathies, dystrobrevinopathies, myotonic dystrophy, zaspopathy, myoadenylate-deaminase deficiency, Charcot-Marie-Tooth disease, mitochondrial disorder, Barth syndrome, Friedreich ataxia, or Pompe's disease (Finsterer, J., et al., 2005)? Was a second trouble excluded to have caused LVHT?

How to explain that LVHT was found in the present case with the 1q32 deletion but not in other previously reported del1q syndromes?

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There is no information given on the further clinical course of the patient. Which was the long-term outcome of the patient? Did she develop stroke or embolism previously suspected to be caused by LVHT?

In the discussion the authors mention 24 cases with del1q syndrome previously described. In table 1, however, only 7 cases are mentioned. Did all these cases actually undergo cardiologic including echocardiographic examinations?

Overall, the cause of LVHT in the described patient remains speculative. It would be interesting to assess the 40 genes located in the deleted band 1q43 with regard to their regulatory function in the embryonic cardiac development.

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