

## Letter to the Editor

## Left Ventricular Noncompaction in Infantile Mitochondrial Disorder

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With interest we read the article by Scaglia *et al.*, on the frequency of clinical manifestations in 113 children with mitochondriopathy (MCP) (Scaglia, F. *et al.*, 2004). Forty-five of the 113 patients (45%) showed cardiac involvement (CI) on echocardiography. Among those with CI, 58% had hypertrophic and 29% dilative cardiomyopathy (CMP) and 13% left-ventricular hypertrabeculation (LVHT)/noncompaction. Disease onset was earlier in patients with CI than without. Survival of patients with CI was significantly reduced compared to those without CI. Sixty percent of those with CI had neuromuscular involvement. We have the following concerns:

How was CI defined? How many patients underwent ECG and how many echocardiography? How many had CI at follow-up who were normal at baseline? Did hypertrophic CMP progress into dilative CMP in any patient?

What is the explanation for the rapid deterioration of cardiac function? In our experience deterioration of CI in adult MCP is slow (Finsterer, J. *et al.*, 2001). Which was the cause of death in those who deceased?

Is there an explanation for the earlier onset in patients with CI compared to those without CI. Does CI in this respect indicate multi-organ involvement?

Frequency of CI was 100% in the group with CMP and myopathy. Which was the frequency of CI in the other 5 groups?

How to explain the low rate of rhythm abnormalities (11%) compared to the high rate of CMP (100%) in patients with CI? Is the cardiac conduction system less frequently involved than the myocardium? Is the cardiac conduction system less vulnerable to respiratory chain defects? In our experience with adult MCP patients the frequency of rhythm abnormalities is roughly the same as the frequency of CMP.

Which of the three possible definitions of LVHT was applied in the study (Chin, T. K. *et al.*, 1990; Oechslin, E. N. *et al.*, 2000; Stollberger, C., & Finsterer, J. 2004)?

Did LVHT also occur in combination with other cardiac abnormalities in any patient?

LVHT has been already previously described in patients with MCP (Finsterer, J. *et al.*, 2004a; Finsterer, J. *et al.*, 2004b; Finsterer, J., & Stollberger, C. 1998; Finsterer, J. *et al.*, 2000; Finsterer, J. *et al.*, 2003).

LVHT is not only a congenital abnormality but may be acquired in single cases (Finsterer, J. *et al.*, 2004b).

In addition to MCP and Barth syndrome, LVHT has been described in patients with Duchenne muscular dystrophy, [personal communication] Becker muscular dystrophy, myotonic dystrophy type 1, myoadenylate-deaminase deficiency (Finsterer, J. *et al.*, 2004c). Dystrobrevinopathy (Ichida, F. *et al.*, 2001), cypher gene mutations (Vatta, M. *et al.*, 2003), Pompe disease (Pipo, J. R. *et al.*, 2003), Friedreich ataxia (Alper, G., & Narayanan, V. 2003), and a number of

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rare non-neuromuscular genetic syndromes (Oechslin, E. N. *et al.*, 2000).

Overall, it is important to be aware of CI in MCP and to recognise it already in the subclinical stage. Adequate treatment of CI may improve quality of life and prolong survival and improve prognosis in infantile and adult cases.

#### REFERENCES

1. Scaglia, F., Towbin, J. A., Craigen, W. J., Belmont, J. W., Smith, E. B., Neish, S. R., ... & Wong, L. J. C. (2004). Clinical spectrum, morbidity, and mortality in 113 pediatric patients with mitochondrial disease. *Pediatrics*, 114(4), 925-931.
2. Finsterer, J., Stollberger, C., Blazek, G., & Spahits, E. (2001). Cardiac involvement in myotonic dystrophy, Becker muscular dystrophy and mitochondrial myopathy: a five-year follow-up. *Can J Cardiol*, 17, 1061-1069.
3. Chin, T. K., Perloff, J. K., Williams, R. G., Jue, K., & Mohrmann, R. (1990). Isolated noncompaction of left ventricular myocardium. A study of eight cases. *Circulation*, 82(2), 507-513.
4. Oechslin, E. N., Jost, C. H. A., Rojas, J. R., Kaufmann, P. A., & Jenni, R. (2000). Long-term follow-up of 34 adults with isolated left ventricular noncompaction: a distinct cardiomyopathy with poor prognosis. *Journal of the American College of Cardiology*, 36(2), 493-500.
5. Stollberger, C., & Finsterer, J. (2004). Left ventricular hypertrabeculation/noncompaction. *J Am Soc Echocardiogr*, 17, 91-100.
6. Finsterer, J., Stollberger, C., & Blazek, G. (2004). Left ventricular noncompaction suggests myopathy. *Circulation*, 109:e201-e202
7. Finsterer, J., Stollberger, C., & Schubert, B. (2004). Acquired left ventricular hypertrabeculation/noncompaction in mitochondriopathy. *Cardiology*, 102, 228-230.
8. Finsterer, J., & Stollberger, C. (1998). Hypertrabeculated left ventricle in mitochondriopathy. *Heart*, 80, 632.
9. Finsterer, J., Bittner, R., Bodingbauer, M., Eichberger, H., Stollberger, C., & Blazek, G. (2000). Complex mitochondriopathy associated with 4 mtDNA transitions. *Eur Neurol*, 44, 37-41.
10. Finsterer, J., Stollberger, C., & Kopsa, W. (2003). Noncompaction on cardiac MRI in a patient with nail-patella syndrome and mitochondriopathy. *Cardiology*, 100, 48-49.
11. Finsterer, J., Schoser, B., & Stollberger, C. (2004). Myoadenylate-deaminase gene mutation associated with left ventricular hypertrabeculation/noncompaction. *Acta Cardiol*, 59, 453-456.
12. Ichida, F., Tsubata, S., Bowles, K. R., Haneda, N., Uese, K., Miyawaki, T., ... & Towbin, J. A. (2001). Novel gene mutations in patients with left ventricular noncompaction or Barth syndrome. *Circulation*, 103(9), 1256-1263.
13. Vatta, M., Mohapatra, B., Jimenez, S., Sanchez, X., Faulkner, G., Perles, Z., ... & Bowles, K. R. (2003). Mutations in Cypher/ZASPin patients with dilated cardiomyopathy and left ventricular noncompaction. *Journal of the American College of Cardiology*, 42(11), 2014-2027.
14. Pipo, J. R., Feng, J. H., Yamamoto, T., Ohsaki, Y., Nanba, E., Tsujino, S., ... & Ohno, K. (2003). New GAA mutations in Japanese patients with GSDII (Pompe disease). *Pediatric neurology*, 29(4), 284-287.
15. Alper, G., & Narayanan, V. (2003). Friedreich's ataxia. *Pediatr Neurol*, 28, 335-341.