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Letter to the Editor

Myosin Storage Myopathy Due to Slow/?-Cardiac Myosin Heavy Chain Gene Mutation without Cardiac Involvement

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In their paper Tajsharghi *et al.*, report four patients from two unrelated families carrying the missense-mutation Arg1845Trp in the MYH7-gene, which encodes for the slow/β-cardiac myosin-heavy-chain (Tajsharghi, H. *et al.*, 2003). Though MYH7 is expressed in slow type-I-muscle-fibers and cardiomyocytes, and although MYH7 mutations were reported to exclusively cause familial hypertrophic cardiomyopathy, the described patients only presented with myopathy without overt cardiac involvement. The 71-year-old index-patient had atrial fibrillation (AF) with enlarged atrium and in his mother a pacemaker was implanted because of AF at age 80y. No information on cardiac investigations and abnormalities of the other patients or the clinically unaffected relatives is given,

The described findings raise the following concerns:

1. The etiology of AF remains uncertain. No results of coronary angiography, stress-test, or thyroid function are reported. Though the two patients with AF were of increased age, which is a risk factor of AF, it cannot be excluded that AF was due to the underlying mutation and manifested not before late adulthood.

2. Rhythm-abnormalities are frequent and sometimes the exclusive manifestation of cardiac involvement in primary myopathies, even before skeletal muscle manifestations (Finsterer, J., & Stöllberger, C. 2000). It is not reported if any of the patients or relatives complained about palpitations or syncope and if 24h-ECG-recordings were done.

3. Absence of myocardial thickening on echocardiography does not exclude cardiomyopathy (Elliott, P. M. *et al.*, 2001). Previous reports showed that cardiomyopathy might exclusively manifest as myocardial disarray or fibrosis on histological examinations, without echocardiographic abnormalities (Varnava, A. M. *et al.*, 2000). In such cases cardiac-MRI or endomyocardial biopsy are useful to assess abnormal myocardial texture (Moon, J. C. *et al.*, 2003).

4. Cardiac involvement in myopathies may also manifest as left-ventricular hypertrabeculation (LVHT), frequently overlooked or misinterpreted on echocardiography. Was LVHT present on echocardiography or cardiac-MRI in any of the cardiologically investigated patients?

5. Despite normal echocardiographic findings, it cannot be excluded that the MYH7 mutation also manifested in the myocardium or cardiac conduction-system. Thus, it is of interest if endomyocardial biopsy was abnormal and if the MYH7-mutation was also detected in cardiomyocytes. Given that the MYH7-mutation indeed manifested exclusively in the skeletal muscle, an explanation is required why the skeletal muscle but not the myocardial isoform of the slow/β-cardiac myosin-heavy-chain was mutant.

6. Since cardiac involvement in myopathies frequently manifests as discrete abnormalities and increases with age, regular follow-ups and autopsy studies are warranted.

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Overall, cardiac involvement in a myopathy due to a mutation in a gene expressed in the skeletal muscle and heart can be excluded only if thorough, comprehensive, non-invasive and invasive cardiologic examinations, including regular follow-ups are carried out.

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