

## Abnormal sympathetic innervation of the heart in a patient with Emery-Dreifuss muscular dystrophy

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A 33-year-old man was admitted for general malaise and vomiting. An electrocardiogram showed a complete atrioventricular block and an echocardiogram showed right atrial dilatation and normal wall motion of left ventricle (LV). Gene analysis showed nonsense mutation in the *STA* gene, which codes for emerlin, and Emery-Dreifuss muscular dystrophy was diagnosed. An endomyocardial biopsy of right ventricle showed mild hypertrophy of myocytes. Myocardial scintigraphic studies with Tc-99m methoxyisobutylisonitrile (MIBI) and I-123-betamethyl-*p*-iodophenylpentadecanoic acid (BMIPP) scintigrams showed no abnormalities. In contrast, I-123 metaiodobenzylguanidine (MIBG) scintigrams showed a diffuse and severe decrease in accumulation of MIBG in the heart. Six months later, his LV wall motion on echocardiograms developed diffuse hypokinesis. These results suggest that the abnormality on I-123 MIBG myocardial scintigrams may predict LV dysfunction in Emery-Dreifuss muscular dystrophy.

**Key words:** Emery-Dreifuss muscular dystrophy, metaiodobenzylguanidine (MIBG), scintigraphy, *STA* gene