Increased ventilatory response to exercise in symptomatic and asymptomatic LMNA mutation carriers: a follow-up study.

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Abstract

BACKGROUND: LMNA mutations are an important cause of cardiomyopathy often leading to cardiac arrhythmias, heart failure and even heart transplantation. An increasing number of asymptomatic mutation carriers are identified, as family members of the index patients are screened. Our aim was to study the disease progression in asymptomatic LMNA mutation carriers and in patients with symptomatic cardiolaminopathy by repeated spiroergometric testing in a prospective clinical follow-up study.

METHODS AND RESULTS: We studied 26 LMNA mutation carriers once a year during 5 years up to 6 times by spiroergometry, clinical assessment, laboratory tests and echocardiography. The 23 control subjects underwent clinical assessment and spiroergometry once. Twelve of the mutation carriers were asymptomatic, and 14 had some clinical manifestations of the mutation ranging from clinically relevant rhythm disturbances to DCM and heart failure. Compared to controls, the symptomatic carriers showed a higher slope of the ventilatory equivalent for CO₂ (V˙E/V˙CO₂ slope) and a lower fraction of end-tidal CO₂ (FetCO₂). The asymptomatic mutation carriers also showed an increased ventilatory response to exercise during the follow-up as indicated by increased V˙E/V˙CO₂ slope and decreased FetCO₂.

CONCLUSIONS: The study suggests that an increased ventilatory response during exercise might reveal a preclinical manifestation of DCM in LMNA mutation carriers.

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KEYWORDS: FetCO₂; V˙E/V˙CO₂ slope; cardiomyopathy; clinical exercise testing; spiroergometry

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