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References
The list of references is available in the online version of this paper.

CARDIOVASCULAR FLASHLIGHT

An unusual cause of dyspnoea in a young man
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A 20-year-old man presented with progressive exertional dyspnoea. His cardiovascular examination was unremarkable. The electrocardiogram showed sinus rhythm with a dominant R-wave in the early precordial leads and lateral Q-waves (Panel A). Laboratory investigation demonstrated elevated creatine kinase with normal Troponin-T levels. Echocardiography was non-diagnostic due to poor echo-windows. Cardiac magnetic resonance imaging showed mild hypokinesis of the basal infero-lateral wall with an ejection fraction of 50% (Panels B and C, Supplementary material online, Videos 1 and 2). Late-gadolinium enhancement (LGE) imaging demonstrated epicardial hyperenhancement in the basal-inferior and lateral walls of the left ventricle (Panels D and E, arrows) consistent with muscular dystrophy or viral-myocarditis. Genetic testing demonstrated a deletion mutation in the dystrophin gene encompassing exons 52–54 and subsequent muscle biopsy confirmed the diagnosis of Becker muscular dystrophy.

Becker muscular dystrophy is an X-linked disorder affecting the synthesis of dystrophin—a large, sarcolemmal protein which contributes to maintenance of cellular architecture and permits signal transduction between the cytoskeleton and the extracellular matrix. Progressive symptomatic cardiac involvement eventually occurs in 70–75% of cases by the age of 40. Therefore, these patients require long-term cardiac-follow-up looking for development of heart failure, cardiomyopathy, and arrhythmias. There is no correlation between severity of cardiac and skeletal muscle involvement and cardiac symptoms may present before skeletal muscle involvement is clinically apparent.

Electrocardiography may show a dominant R-wave in lead-V1 in approximately half of patients. Cardiac magnetic resonance imaging can demonstrate LGE—classically in the epicardial inferolateral wall—even in the absence of other abnormalities. There is emerging evidence that early initiation of heart failure therapy may delay the onset and progression of left ventricular dysfunction.

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