A 16-year-old male patient with known Becker’s muscular dystrophy (BMD), with deletion of exon 48–49, was noted to have recent deterioration in already limited functional capacity. The transthoracic echocardiogram revealed moderately impaired left ventricular systolic function, with ejection fraction of 44% (Figure 1A), and preserved relaxation (Figure 1B). A cardiovascular magnetic resonance (CMR) study was performed and confirmed high-normal indexed left ventricular volumes with moderately reduced global ejection fraction (48%). No active inflammation was seen. Late gadolinium-enhanced images showed typical subepicardial fibrosis of the lateral and inferior wall from base to mid-ventricular level (Figure 1C and D).

BMD is an X-linked inherited progressive myopathic disorder caused by mutations in the dystrophin gene; the dystrophin protein plays an essential role in signal pathways and stabilization of the cardiomyocyte cell membrane. It is characterized by proximal skeletal muscle weakness and wasting. Cardiac muscle can be affected, with degeneration of fibers, replacement fibrosis and fatty infiltration. The pathophysiology is not entirely understood; it is postulated that diffuse myocardial metabolic abnormalities lead to fibrosis, starting, as in this patient, in the subepicardium of the inferolateral wall, due to greater mechanical stress in this region. Progressive cardiomyopathy, often subclinical in the early stages, is a major cause of morbidity and mortality. Advanced echocardiographic assessment of diastolic function and tissue characterization by CMR enable early detection of cardiac involvement and timely initiation of heart failure treatment to prevent ventricular remodeling.
Figure 1  (A) Transthoracic echocardiogram in parasternal view depicting a dilated left ventricle with moderately reduced ejection fraction. (B) Tissue Doppler imaging of lateral mitral inflow velocities, with an E/e’ ratio of 5.3, suggestive of normal filling pressures. (C and D) Cardiovascular magnetic resonance images with late gadolinium enhancement showing typical subepicardial inferolateral fibrosis (white arrows), also involving the mid-septum (dashed arrow), consistent with Becker’s cardiomyopathy.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this investigation.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data and that all the patients included in the study received sufficient information and gave their written informed consent to participate in the study.

Right to privacy and informed consent. The authors have obtained the informed consent of the patients and/or subjects mentioned in the article. The corresponding author is in possession of this document.

Conflict of interest

The authors have no conflicts of interest to declare.