Cureus
Part of SPRINGER NATURE

Open Access Case Report

Palpitations Unmasking Inherited Dilated Cardiomyopathy in a 21-Year-Old Woman: A Case Report

Review began 10/07/2026
Review ended 11/22/2026
Published 11/25/2026

© **Copyright** 2026
Panja et al. This is an open access article distributed under the terms of the Creative Commons Attribution License CC-BY 4.0., which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

DOI: 10.7759/cureus.120233

Kushal Panja [1], Muskan Khatoon [1], Raana Alamgir [2]

1. General Medicine, North Manchester General Hospital, Manchester University NHS Foundation Trust, Manchester, GBR 2. Cardiology, North Manchester General Hospital, Manchester University NHS Foundation Trust, Manchester, GBR

Corresponding author: Muskan Khatoon, muskankhatoon88@hotmail.co.uk

Abstract

Palpitations in young adults are often attributed to benign arrhythmia or anxiety, but they may occasionally be the first clue to inherited structural heart disease.

We report the case of a 21-year-old woman who presented with recurrent palpitations and breathlessness and was found to have severe left ventricular systolic dysfunction. Further evaluation with echocardiography and cardiac magnetic resonance (CMR) imaging supported a diagnosis of dilated cardiomyopathy (DCM). In view of her striking family history of DCM and cardiac arrest, genetic testing was performed and identified a likely pathogenic TTN variant consistent with autosomal dominant inherited DCM. She was started on medical therapy and subsequently underwent primary prevention subcutaneous implantable cardioverter-defibrillator (ICD) implantation.

This case highlights the importance of recognising recurrent palpitations as a possible early clue to inherited DCM in young patients, particularly when accompanied by a significant family history.

Categories: Cardiology, Genetics, Internal Medicine
Keywords: dilated cardiomyopathy (dcm), genetic cardiomyopathy, implantable cardioverter-defibrillator (icd), supraventricular tachycardia (svt), ttn gene

Introduction

Dilated cardiomyopathy (DCM) is defined by left ventricular or biventricular dilatation with systolic dysfunction that is not explained solely by abnormal loading conditions or coronary artery disease [1]. Genetic disease accounts for a substantial proportion of apparently idiopathic DCM, and identifying an inherited cause can influence diagnosis, family screening, risk stratification, and management [1,2].

The TTN gene encodes titin, a large sarcomeric protein that plays an important structural and mechanical role in cardiac muscle. Truncating TTN variants introduce premature stop signals that can impair normal titin production and are among the most common genetic causes of DCM [3,4].

Inherited cardiomyopathies do not always present with overt heart failure and may first come to clinical attention through arrhythmic symptoms [1,5]. Arrhythmias, including supraventricular and ventricular arrhythmias, are well recognised in DCM and may be part of the initial clinical presentation [5,6].

Accordingly, current guidance recommends careful family history assessment, phenotypic evaluation, and consideration of genetic testing in patients with young-onset disease, unexplained ventricular dysfunction, or a family history of cardiomyopathy or sudden cardiac death [1,2].

Cardiac magnetic resonance (CMR) plays an important role in DCM by confirming phenotype, aiding tissue characterisation, and helping exclude alternative diagnoses [1]. In nonischaemic DCM, mid-wall fibrosis or late gadolinium enhancement on CMR has been associated with adverse cardiovascular outcomes, including ventricular arrhythmias, sudden cardiac death, and mortality [7-9].

Case Presentation

A 21-year-old woman presented to the Emergency Department with a sudden-onset episode of palpitations and shortness of breath lasting approximately five minutes while on her way to the gym. She denied chest pain or syncope. Over the preceding two weeks, she had experienced intermittent light-headedness and recurrent episodes of palpitations.

She had no significant past medical history. She lived with her family, worked as a support worker, and

reported drinking alcohol socially. She denied any recreational drug use. A strong family history of cardiac disease was present, predominantly on the paternal side. Her father had DCM, suffered an out-of-hospital cardiac arrest at the age of 52 years, and had a secondary prevention implantable cardioverter-defibrillator (ICD) with subsequent shocks. Her paternal grandmother had DCM. Her paternal aunt and paternal uncle were also affected by DCM. There was also a history of cardiac arrest in a paternal uncle at 55 years of age (Figure *1*).

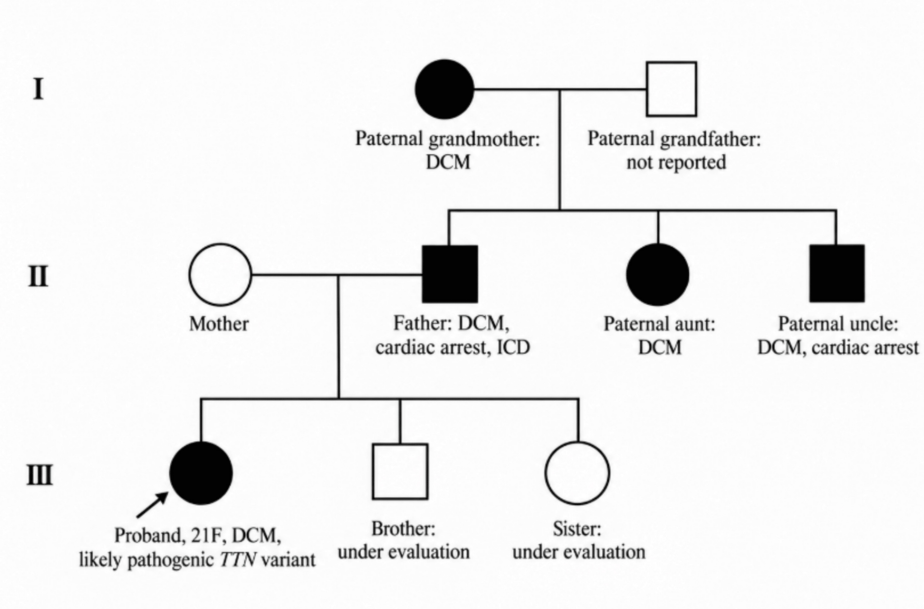


FIGURE 1: Family pedigree chart demonstrating familial DCM

Filled symbols indicate individuals affected by DCM, while open symbols indicate individuals without reported DCM or those undergoing evaluation. Squares represent males and circles represent females. The arrow identifies the proband, a 21-year-old woman with DCM and a likely pathogenic TTN variant.

DCM, Dilated Cardiomyopathy; ICD, Implantable Cardioverter-Defibrillator

A 72-hour Holter monitor performed approximately nine months before admission for recurrent shortness of breath and light-headedness had demonstrated sinus rhythm with a low supraventricular ectopic burden (2%) and 89 episodes of supraventricular tachycardia, with the longest episode lasting 3 minutes 38 seconds at a rate of up to 201 beats per minute. It also showed a low ventricular ectopic burden (2%), including couplets, triplets, and short runs of bigeminy and trigeminy.

On presentation, she was haemodynamically stable, with a blood pressure of approximately 120/78 mmHg, heart rate of 92 beats per minute, oxygen saturation of 99% on room air, and no fever. Physical examination was unremarkable. Initial blood investigations, including serial troponins, were within normal limits, with troponin values remaining at 4 ng/L. N-terminal pro-B-type natriuretic peptide during admission was 761 ng/L.

Twenty-four hours after admission, she experienced a further self-terminating episode of supraventricular tachycardia on standing. Cardiology review was sought, and an informal echocardiogram raised concern for severe left ventricular systolic impairment. The Cardiology team initiated bisoprolol 2.5 mg once daily and dapagliflozin 10 mg once daily. Bisoprolol was increased to 2.5 mg twice daily the following day. Ivabradine 2.5 mg twice daily was subsequently introduced because the resting heart rate remained approximately 85-92 beats per minute and increased to over 100 beats per minute on mobilisation. Further beta-blocker uptitration was limited by symptomatic low blood pressure, typically around 90-100/60 mmHg. Ivabradine was then increased to 5 mg twice daily, and sacubitril/valsartan 24/26 mg twice daily was commenced.

Formal transthoracic echocardiography subsequently demonstrated a severely dilated left ventricle with global hypokinesia and severely impaired systolic function, with a visual left ventricular ejection fraction (LVEF) of approximately 30%. Right ventricular systolic function was also impaired. No significant valvular abnormality was identified (Figure *2*).

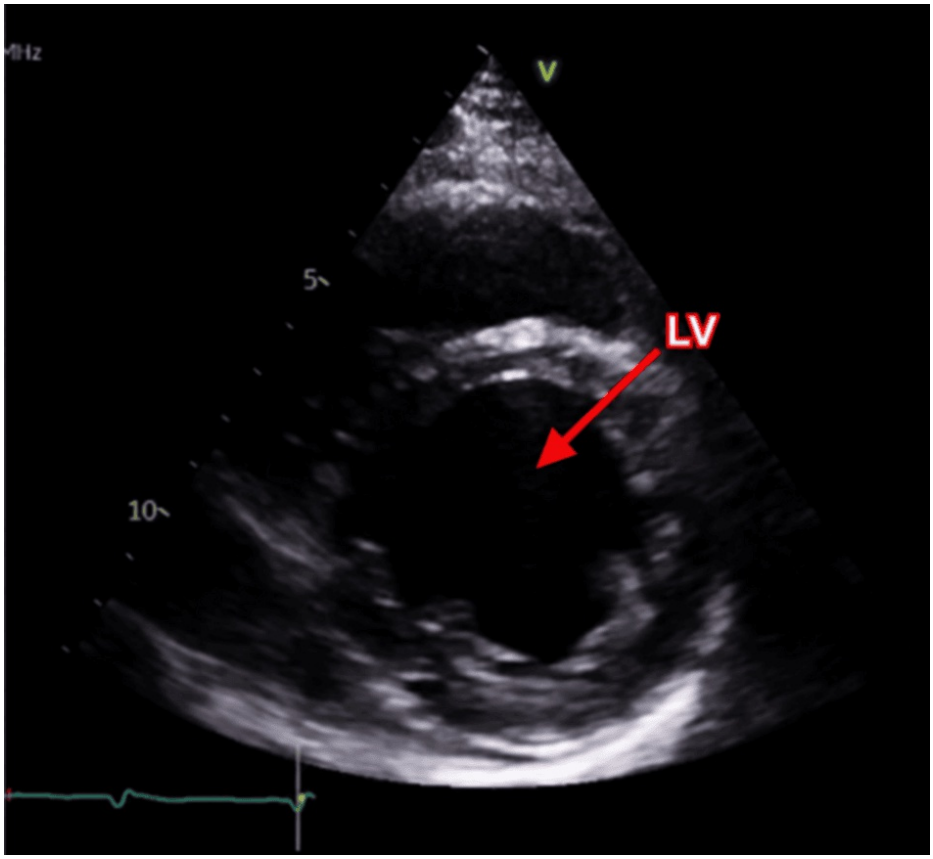


FIGURE 2: Transthoracic echocardiographic parasternal short-axis view showing a markedly dilated left ventricle

The red arrow indicates the left ventricular (LV) cavity.

CMR imaging performed three days later demonstrated a moderately dilated left ventricle with an end-diastolic volume of 201 mL (108 mL/m^2) and an end-systolic volume of 142 mL (76 mL/m^2), with a LVEF of 30%. The right ventricle was normal in size and function, with an ejection fraction of 51%, an end-diastolic volume of 139 mL (75 mL/m^2), an end-systolic volume of 67 mL (36 mL/m^2), and tricuspid annular plane systolic excursion of 20 mm. Mid-wall late gadolinium enhancement was seen in the basal-to-mid anteroseptum and inferoseptum, as well as in the basal inferolateral wall, with additional enhancement at the inferior right ventricular insertion point. T2 mapping values were normal, with no evidence of myocardial oedema. Mid-wall fibrosis was seen within the basal-to-mid septum and basal inferolateral segment (Figure *3*).

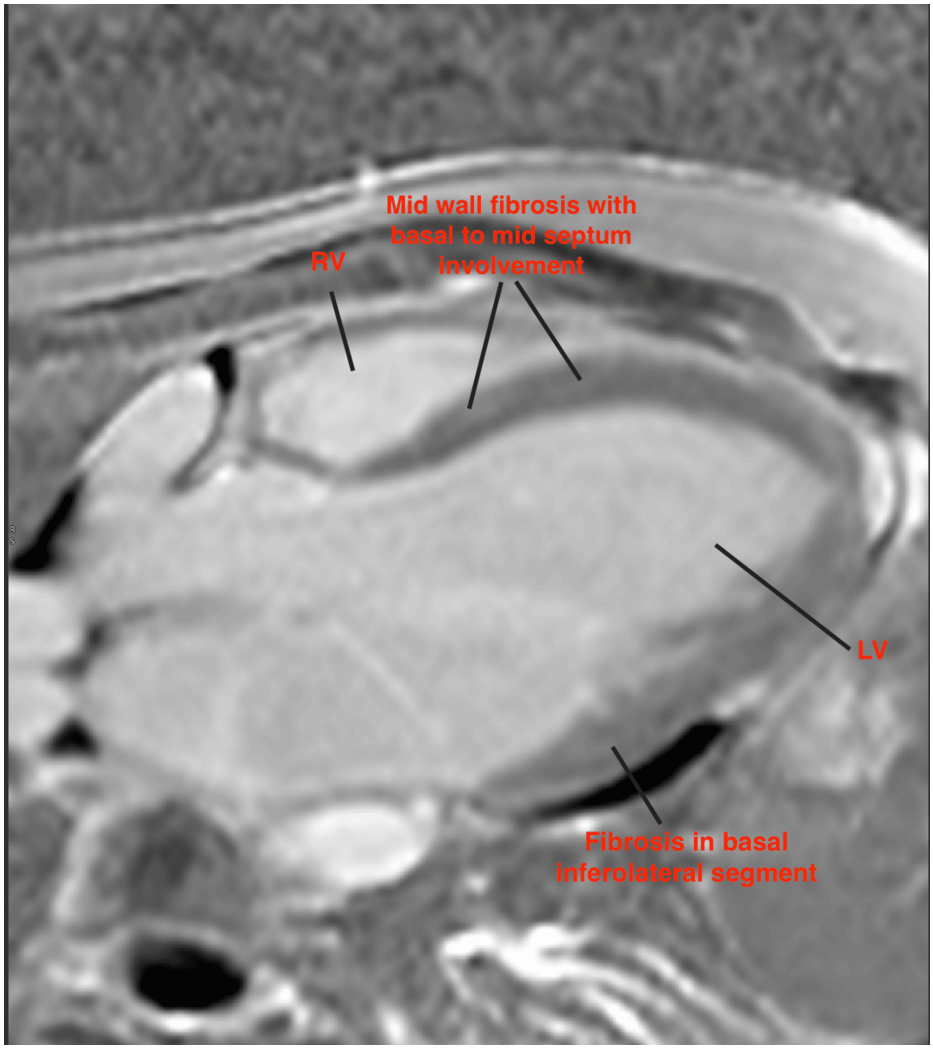


FIGURE 3: Cardiac magnetic resonance late gadolinium enhancement image showing mid-wall fibrosis within the basal-to-mid septum and basal inferolateral segment

RV, Right Ventricle; LV, Left Ventricle

There was no evidence of myocardial infarction. These findings supported the phenotype of DCM.

The apparent differences between the echocardiographic and CMR descriptions likely reflect modality-related differences in ventricular assessment. Both demonstrated significant left ventricular systolic dysfunction, with an LVEF of approximately 30%, while CMR provided quantitative ventricular volumes and demonstrated preserved right ventricular systolic function.

Thyroid function testing showed a mildly elevated free T4 with normal thyroid-stimulating hormone. Endocrinology review considered this most likely to represent a transient abnormality or sick euthyroid state rather than thyrotoxicosis, and outpatient repeat thyroid function testing was advised.

She was discharged on the maximum tolerated heart failure regimen at that stage: bisoprolol 2.5 mg twice daily, dapagliflozin 10 mg once daily, ivabradine 5 mg twice daily, and sacubitril/valsartan 24/26 mg twice daily, with plans for ambulatory rhythm monitoring, outpatient cardiology follow-up, and genetic testing for inherited cardiomyopathy.

A seven-day Holter monitor following discharge demonstrated sinus rhythm with low-burden supraventricular ectopy and a 3% burden of multimorphic ventricular ectopic beats, seen in isolation, couplets, and triplets. One ventricular run lasting seven beats at 128 beats per minute was recorded.

Given her young age, markedly abnormal cardiac imaging, and striking family history, genetic testing was

undertaken. This identified a heterozygous likely pathogenic TTN variant, c.65333G>A, p.(Trp21778Ter), supporting a likely diagnosis of autosomal dominant TTN-related DCM.

Genetic counselling was discussed following identification of the likely pathogenic TTN variant. First-degree relatives were advised to undergo clinical cardiac assessment and targeted genetic testing, where appropriate. At the time of writing, the patient's siblings were undergoing cardiac evaluation, but no confirmed genetic results were available.

During outpatient follow-up, she remained symptomatic despite continued heart failure therapy, reporting breathlessness, dizziness, headaches, and episodes of sudden sweating occurring both at rest and during activity. Her exercise tolerance remained markedly reduced compared with baseline. Further beta-blocker uptitration was limited by symptomatic hypotension, and she remained on the maximally tolerated regimen of bisoprolol 2.5 mg twice daily, dapagliflozin 10 mg once daily, ivabradine 5 mg twice daily, and sacubitril/valsartan 24/26 mg twice daily. Her most recent outpatient echocardiogram before device implantation showed a modest improvement in LVEF to 35%, compared with 30% during admission. Her most recently documented functional status before implantation was New York Heart Association (NYHA) class IV.

At electrophysiology follow-up, implantable defibrillator therapy was discussed. Although no sustained arrhythmia had been documented, the combination of persistent severe left ventricular systolic dysfunction, TTN-associated DCM, significant ongoing symptoms, and a strong family history of cardiac arrest prompted primary prevention device planning. After discussion, the patient opted for a subcutaneous ICD in view of the lower risk of lead-related complications. The device was implanted approximately 2.5 months after the diagnosis and initiation of heart failure therapy, during which she had remained on her maximum tolerated medical regimen.

Follow-up continued on an outpatient basis after subcutaneous ICD implantation. At approximately three weeks after implantation, she reported improved energy and exercise tolerance, with functional status improving to New York Heart Association (NYHA) class II. Approximately six weeks after implantation, she experienced two ICD shocks during exertion while hiking. Device interrogation demonstrated a sudden tachycardia that was terminated by shock. Eplerenone 12.5 mg once daily was introduced.

Over the following two to three months, she experienced intermittent palpitations, postural dizziness, presyncopal symptoms, blurred vision, and shortness of breath, with difficulty tolerating eplerenone. This led to temporary discontinuation and later attempts at reintroduction. She also had further emergency department attendances for palpitations and chest discomfort, although serial troponins, electrolytes, and inflammatory markers remained unremarkable, and no heart failure admission occurred.

Approximately three months after ICD implantation, electrophysiology review suggested that the tachycardia triggering the previous shocks was likely supraventricular, and the device therapy threshold was increased from 200 to 220 beats per minute to reduce the likelihood of inappropriate therapy during exertion. By approximately six months after implantation, she had experienced no further ICD shocks. She continued to report intermittent palpitations and postural symptoms, and her most recent functional status was documented as NYHA class II at the time of writing the report. No post-implantation echocardiogram or CMR imaging was available at the time of writing; therefore, no repeat LVEF measurement was available for comparison.

Discussion

This case highlights that recurrent palpitations in a young adult should not automatically be attributed to benign arrhythmia or anxiety, particularly when symptoms recur and there is a strong family history of cardiomyopathy [1,2,5].

In our patient, intermittent palpitations and documented supraventricular tachycardia preceded the recognition of severe left ventricular systolic dysfunction and a familial DCM phenotype. This pattern is clinically important because arrhythmias may be the presenting manifestation of genetic cardiomyopathy, and DCM is frequently accompanied by both supraventricular and ventricular rhythm disturbances [5,6].

Current cardiomyopathy guidance emphasises that a family history of cardiomyopathy, sudden cardiac death, or unexplained cardiac arrest should prompt consideration of an inherited aetiology and should lead to targeted family evaluation and genetic testing when appropriate [1,2]. The identification of a likely pathogenic TTN variant in our patient supported a diagnosis of autosomal dominant inherited DCM and provided a rationale for cascade screening of relatives. This is particularly relevant because TTN truncating variants are a major cause of heritable DCM and may show variable expressivity and age-dependent phenotypic manifestations [3,4,10].

An important alternative explanation in this case was whether the tachyarrhythmia burden itself might have caused or contributed to the cardiomyopathy.

Arrhythmia-induced cardiomyopathy is a recognised and potentially reversible cause of left ventricular dysfunction that may occur with sustained or recurrent tachyarrhythmias [6]. That possibility should be considered whenever tachyarrhythmia and ventricular impairment coexist [6]. However, several features in this case favoured a primary inherited cardiomyopathic process rather than a purely tachycardia-mediated cardiomyopathy, including the striking familial clustering of DCM, the likely pathogenic TTN variant, marked ventricular dilatation at diagnosis, and the presence of mid-wall fibrosis on CMR.

The CMR findings in this patient were also clinically meaningful. Mid-wall fibrosis in nonischaemic DCM has been associated with worse prognosis in cohort studies, and subsequent systematic reviews and meta-analyses have consistently linked late gadolinium enhancement with ventricular arrhythmias, sudden cardiac death, and adverse cardiovascular outcomes [7-9]. In our patient, CMR showed mid-wall fibrosis in the basal-to-mid septum and basal inferolateral segment without oedema or infarction, supporting a nonischaemic DCM phenotype and strengthening the case for careful arrhythmic risk assessment.

Our patient later underwent primary prevention subcutaneous ICD implantation after multidisciplinary assessment.

Although TTN-related DCM does not carry the same classic high-arrhythmic-risk profile described for some other genotypes, TTN-associated disease is still clinically significant and may present with arrhythmia, systolic dysfunction, and adverse outcomes requiring standard heart failure therapy and device-based risk management where indicated [4,10]. Her ventricular ectopy, short ventricular run, severe left ventricular systolic dysfunction, and extensive family history of significant cardiac events all added weight to that decision.

This case reinforces several practical lessons. First, recurrent palpitations in a young patient may be the first clue to inherited structural heart disease rather than an isolated benign rhythm problem [1,5]. Second, family history remains one of the highest-yield components of the assessment in unexplained cardiomyopathy [1,2]. Third, echocardiography, CMR, and genetic testing are complementary investigations in suspected inherited DCM because they provide structural, tissue-level, and etiologic information that together refine diagnosis and management [1,2,7-9]. Finally, establishing a genetic diagnosis has implications beyond the index patient by enabling targeted screening and surveillance of relatives who may be at risk [1,2].

Conclusions

This case highlights that recurrent palpitations in a young adult should not always be assumed to be benign, particularly in the presence of a strong family history of cardiomyopathy or sudden cardiac death. In this patient, arrhythmic symptoms led to the recognition of severe DCM and subsequent identification of a likely pathogenic TTN variant. This case highlights the value of integrating family history, multimodality cardiac imaging, and genetic testing when evaluating young patients with recurrent palpitations and suspected inherited cardiomyopathy.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Kushal Panja, Muskan Khatoon

Acquisition, analysis, or interpretation of data: Kushal Panja, Raana Alamgir, Muskan Khatoon

Drafting of the manuscript: Kushal Panja

Critical review of the manuscript for important intellectual content: Kushal Panja, Raana Alamgir, Muskan Khatoon

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

References

1. Arbelo E, Protonotarios A, Gimeno JR, et al.: 2023 ESC guidelines for the management of cardiomyopathies. Eur Heart J. 2023, 44:3503-626. 10.1093/eurheartj/ehad194
2. Hershberger RE, Givertz MM, Ho CY, et al.: Genetic evaluation of cardiomyopathy - a Heart Failure Society of America practice guideline. J Card Fail. 2018, 24:281-302. 10.1016/j.cardfail.2018.03.004
3. Herman DS, Lam L, Taylor MR, et al.: Truncations of titin causing dilated cardiomyopathy. N Engl J Med. 2012, 366:619-28. 10.1056/NEJMoa1110186
4. Akhtar MM, Lorenzini M, Cicerchia M, et al.: Clinical phenotypes and prognosis of dilated cardiomyopathy caused by truncating variants in the TTN gene. Circ Heart Fail. 2020, 13:e006832. 10.1161/CIRCHEARTFAILURE.119.006832
5. Lukas Laws J, Lancaster MC, Ben Shoemaker M, et al.: Arrhythmias as presentation of genetic cardiomyopathy. Circ Res. 2022, 130:1698-722. 10.1161/CIRCRESAHA.122.319835
6. Huizar JF, Ellenbogen KA, Tan AY, Kaszala K: Arrhythmia-induced cardiomyopathy: JACC state-of-the-art review. J Am Coll Cardiol. 2019, 73:2328-44. 10.1016/j.jacc.2019.02.045
7. Assomull RG, Prasad SK, Lyne J, et al.: Cardiovascular magnetic resonance, fibrosis, and prognosis in dilated cardiomyopathy. J Am Coll Cardiol. 2006, 48:1977-85. 10.1016/j.jacc.2006.07.049
8. Di Marco A, Anguera I, Schmitt M, et al.: Late gadolinium enhancement and the risk for ventricular arrhythmias or sudden death in dilated cardiomyopathy: systematic review and meta-analysis. JACC Heart Fail. 2017, 5:316. 10.1016/j.jchf.2017.02.006
9. Kuruvilla S, Adenaw N, Katwal AB, Lipinski MJ, Kramer CM, Salerno M: Late gadolinium enhancement on cardiac magnetic resonance predicts adverse cardiovascular outcomes in nonischemic cardiomyopathy: a systematic review and meta-analysis. Circ Cardiovasc Imaging. 2014, 7:250-8. 10.1161/CIRCIMAGING.113.001144
10. Jansweijer JA, Nieuwhof K, Russo F, et al.: Truncating titin mutations are associated with a mild and treatable form of dilated cardiomyopathy. Eur J Heart Fail. 2017, 19:512-21. 10.1002/ejhf.673