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Hans汉斯

扩张型心肌病的研究进展

谭瑶瑶[1,2]，蔡国强[3*]，夏　丽[4*]

[1]重庆医科大学第一临床学院，重庆
[2]重庆市垫江县人民医院全科医学科，重庆
[3]重庆市垫江县人民医院重症医学科，重庆
[4]重庆医科大学附属第一医院全科医学科，重庆

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摘　要

扩张型心肌病是一种以心室腔扩大和收缩功能逐渐下降为特征的心肌疾病，常导致充血性心力衰竭。扩张型心肌病具有明显的家族性特征，相关致病基因的突变约占扩张型心肌病患者的50%。随着对相关致病基因的研究日益深入，它无疑将为扩张型心肌病的治疗提供更多潜在靶点和治疗途径。扩张型心肌病易引发心力衰竭、心律失常甚至猝死，国内≥35岁成人患病人数约177万，病死率高。因此，准确且及时的临床诊断及治疗至关重要。目前，扩张型心肌病的诊断主要依赖于患者的实验室和影像学检测，基因检测的重要性日益受到重视。扩张型心肌病的治疗方法是药物治疗、心脏再同步治疗、心脏移植等等。本文通过参考现有的临床研究和实践，总结扩张型心肌病的病因、病理生理机制、诊断和治疗方法，为临床医生提供参考。

关键词

扩张型心肌病，病因，病理生理机制，诊断，治疗

Advances in Research on Dilated Cardiomyopathy

Yaoyao Tan[1,2], Guoqiang Cai[3*], Li Xia[4*]

[1]The First Clinical College, Chongqing Medical University, Chongqing
[2]Department of General Practice, Dianjiang General Hospital, Chongqing
[3]Intensive Care Unit, Dianjiang General Hospital, Chongqing
[4]Department of General Practice, The First Affiliated Hospital of Chongqing Medical University, Chongqing

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*通讯作者。

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Abstract

Dilated cardiomyopathy is a myocardial disease characterized by the gradual enlargement of the ventricular cavity and the decline of systolic function, often leading to congestive heart failure. Dilated cardiomyopathy has a distinct familial trait, with mutations in related pathogenic genes accounting for approximately 50% of patients with dilated cardiomyopathy. As research on related pathogenic genes deepens, it will undoubtedly provide more potential targets and treatment approaches for the treatment of dilated cardiomyopathy. Dilated cardiomyopathy is prone to cause heart failure, arrhythmia, and even sudden death. In China, the number of adults aged 35 and above suffering from this disease is about 1.77 million, with a high mortality rate. Therefore, accurate and timely clinical diagnosis and treatment are of vital importance. Currently, the diagnosis of dilated cardiomyopathy mainly relies on laboratory and imaging tests of patients, and the importance of genetic testing is increasingly recognized. The treatment methods for dilated cardiomyopathy include drug therapy, cardiac resynchronization therapy, and heart transplantation, etc. This article summarizes the etiology, pathophysiological mechanism, diagnosis, and treatment methods of dilated cardiomyopathy by referring to existing clinical research and practice, providing a reference for clinicians.

Keywords

Dilated Cardiomyopathy, Etiology, Pathophysiological Mechanism, Diagnosis, Treatment

1. 引言

扩张型心肌病(Dilated cardiomyopathy, DCM)的定义是左室扩张和收缩功能障碍的存在，而不能仅由异常负荷条件或冠心病来解释[1]。扩张型心肌病每年的发病率估计为每 10 万人 5~8 例[2]，但由于不完全确定，这被认为是低估的。近年来，随着我国医疗技术的发展、提升，越来越多的扩张型心肌病患者得以明确诊断，扩张型心肌病的患病率呈持续升高趋势。根据近年来的研究估计，DCM 的发病率约为 1/250，即使在接受相关治疗后，其 3 年死亡率约为 20% [3] [4]。尽管存在流行病学上的限制，但扩张型心肌病在参与射血分数降低、因心力衰竭需要住院治疗或接受心脏移植的心力衰竭临床试验的患者中占比高达 40% [5]。

通常扩张型心肌病的患者临床预后较差，5 年生存率约为 50%，10 年生存率为 25% [6] [7]。其死亡率也较高，根据国外既往的研究提示扩张型心肌病患者 1 年死亡率为 25%~30%，随着医疗水平的提升和目前对于疾病治疗手段的增加，使得其死亡率有所下降，但其 5 年死亡率仍为 45% [8]。扩张型心肌病引起死亡的原因多数是心肌扩张导致的泵衰竭，约占 70%，心律失常导致的心源性猝死(SCD)占剩余的 30% [9]，男性患者多于女性患者[10]。国外一篇文献报道每 250 人中就有 1 人受到扩张型心肌病的影响，其病因可以是遗传的、环境的、免疫的或特发性的[11] [12]。它是卫生保健系统日益增长的医疗和经济负担。尽管扩张型心肌病的治疗取得了进展，但结果很差[13]。因此，临床医师迫切需要了解扩张型心肌病这类疾病的致病因素及治疗方法，以改善预后。

2. 扩张型心肌病的病因及病理生理学机制

2.1. 病因

2.1.1. 遗传性因素

扩张型心肌病的遗传原因一直在研究，因为这种疾病具有高度遗传性，在30%~40%的病例中被认为是家族性的[14]。家族性扩张型心肌病的遗传模式通常为常染色体显性。人们越来越认识到扩张型心肌病遗传结构的多样性：单基因、多基因或包括环境暴露在内的多因素[15]。其中主要的致病基因包括肌联蛋白(TTN)基因、核纤层蛋白(LMNA)基因、肌球蛋白重链7 (MYH7)基因、肌钙蛋白T-2 (TNNT2)基因、肌动蛋白(ACTC)基因、钠离子通道蛋白(SCN5A)基因、桥粒斑蛋白(DSP)基因、肌球蛋白结合蛋白C (MYBPC)基因[16]-[20]，其中 TTN 基因约占家族性扩张型心肌病的25%~30% [21]。随着基因学研究的发展和基因检测技术的提升，越来越多的国内外研究正在不断地发现扩张型心肌病致病的基因。引起扩张型心肌病患者的部分基因可见表1。

Table 1. Possible causes of dilated cardiomyopathy
表 1. 扩张型心肌病可能的病因

Genetic	Evidence	Comments
Titin (*TTN*)	Definitive	20%~25% in familial DCM, 8%~15% in acquired DCM; higher rate of left ventricular reverse remodelling (in up to 70%) in the first 2 years before declining, and a higher risk of atrial tachyarrhythmias
LaminA/C (LMNA)	Definitive	Higher risk of sudden cardiac death: with early indication for primary prevention, ICD implantation should be considered (guided by risk factors as detailed); near 100% risk of atrial fibrillation with associated high risk of stroke; frequent progression to end-stage heart failure
Myosin heavy chain (*MYH7*)	Definitive	Early age of onset; high phenotypic expression; low left ventricular reverse remodelling; frequent progression to end-stage heart failure
Filamin C (*FLNC*)	Definitive	Mainly truncating variants with subsequent haploinsufficiency are associated with DCM and cardiac arrhythmias
RNA-binding motif-20 (RBM20)	Definitive	Highly penetrant; high rates of heart failure, arrhythmias, and sudden cardiac death
Troponin-T (TNNT2)	Definitive	Presentation as HCM or DCM; mild dysfunction
Troponin-C1 (TNNC1)	Definitive	Presentation as HCM or DCM; mild dysfunction but disproportionally prone to arrhythmias
Phospholamban (PLN)	Definitive	Specific PLN-R14del-associated DCM is prone to treatment-resistant heart failure and arrhythmias
Desmoplakin (DSP)	Definitive	Gene with overlapping DCM and AC presentation; prone to develop and present as acute myocarditis of unknown pathomechanism, even without typical imaging criteria
BLC2-associated athanogene 3 (BAG3)	Definitive	High disease penetrance in patients older than 40 years
Cardiac alpha-actin (ACTC1)	Moderate definitive	Can present as HCM or DCM; specific variants also lead to atrial-septal defect
Sodium channel alpha unit 5 (SCN5A)	Moderate definitive	Initially described as a gene of long QT syndrome; can present as AC or DCM
Tropomyosin-1 (TPM1)	Moderate definitive	Can present as HCM, DCM, or restrictive CMP
Vinculin (VCL)	Moderate	Lower penetrance; often paediatric onset of disease
Nexilin (NEXN)	Moderate	Can present as HCM or DCM
Myosin-binding protein C (MYBPC3)	Low	Mainly presents as HCM but can progress into HCM with reduced LVEF, which can be misdiagnosed as DCM

续表

Acquired		
Infection (viruses, bacteria, or parasites)	..	Virus presence in the heart only relevant if higher viral load or systemic infection and detection of cardiac inflammation
Immune-mediated diseases (e.g., rheumatoid arthritis, systemic lupus erythematosus, or dermatomyositis)	..	Immunosuppressive therapy in giant cell or eosinophilic myocarditis, sarcoidosis, or vasculitis, and in selected patients with increased cardiac inflammation of unknown origin based upon multidisciplinary counselling (cardiology and immunology)
Toxic (e.g., alcohol, amphetamines, or cocaine)	..	Except for alcohol, will cause irreversible injury or fibrosis to the heart, with frequent myocarditis and vascular involvement
Cancer treatment (anthracyclines, trastuzumab, and immune checkpoint inhibitors)	..	Anthracyclines can cause DCM long term, whereas immune checkpoint inhibitors can cause fatal acute myocarditis in 1%~2% of patients
Peripartum (pregnancy)	..	Pregnancy or delivery can cause haemodynamic and hormonal alterations, and can trigger DCM in (genetically) susceptible women

注：此表引用于 Linde, C. (2024). CRT for Dilated Cardiomyopathy. JACC: Clinical Electrophysiology, 10(7), 1465~1467.

2.1.2. 获得性因素

可以导致扩张型心肌病的非遗传性因素包括以下几类[15] [16]：1) 药物[22]-[24]，包括抗肿瘤药物(如蒽环类化疗药物、单克隆抗体、酪氨酸激酶抑制剂等)、精神科药物(如氯氮平、奥氮平、锂剂等)、氯喹等；2) 酒精[25]，可导致酒精性心肌病；3) 免疫性或炎症性因素[17]，包括自身免疫性疾病、感染、炎症、接触毒素等；4) 内分泌代谢、营养性疾病及神经肌肉原因；5) 其他，如心动过速所诱导的心肌病、围生期心肌病等等[26]。其中，扩张型心肌病最相关的获得性病因是心肌炎。据估计，20%的心肌炎患者在 1 年后可发展为扩张型心肌病[27]。然而，进展速度仍然高度不确定，这主要是由于心肌炎及其潜在病因的诊断困难。

2.2. 病理生理机制

扩张型心肌病的发生和发展过程是由基因突变和(或)环境因素的共同作用导致心肌的收缩和(或)舒张功能异常，伴有心脏的前后负荷异常，早期机体可以通过多种心脏本身的储备机制和心脏外的代偿机制进行代偿，心脏出现适应性重构；后期出现失代偿时，机体出现神经内分泌系统的异常激活，包括交感神经系统(sympathetic nervous system, SNS)、肾素血管紧张素-醛固酮系统(renin angiotensin ald osterone system, RAAS)、利钠肽、内皮素、血管加压素等，共同促进和维持病理性心脏重构的发生和发展。此外，细胞死亡(包括坏死、凋亡、自噬等形式)、心肌纤维化、慢性炎症及免疫反应、心肌能量代谢异常等也参与了扩张型心肌病的发生和发展过程[28]。最终导致心脏扩大伴收缩功能降低，并出现相应的临床表现[29]。

3. 扩张型心肌病的诊断

所有已知或疑似扩张型心肌病患者的诊断检查包括临床病史、实验室检查、心电图和超声影像学检查，见图 1。利钠肽是被广泛应用于心衰诊断和预后的生物标志物。肌酸激酶升高可提示明显或亚临床骨骼肌参与肌营养不良症、线粒体心肌病、糖原蓄积病和肌性心肌病[30]。高敏感性肌钙蛋白是扩张型心肌病急性心肌炎或心肌应激引起的心肌损伤的代表性生物标志物，与不良临床结局和心脏重构相关。反复出现高敏感性肌钙蛋白水平升高且排除冠状动脉疾病的患者，应检查是否有心肌炎和心脏炎症性疾病[31]。

尽管传统观点认为扩张型心肌病的心电图异常是非特异性的，但与致心律失常或肥厚性心肌病相反，扩张型心肌病有特定的 ECG 模式，提示特定的遗传或获得性形式的扩张型心肌病。例如，窦性心动过缓、

房室传导阻滞和心房颤动的扩张型心肌病可能具有致病性或致病性 LMNA 变异[32]。侧 t 波倒置、低 QRS 电压和频繁的室性早搏可能提示 DSP、FLNC 和 PLN 可能是致病或致病变异。虽然这些发现可能在某些形式的扩张型心肌病中更常见，但 ECG 没有足够的诊断准确性来取代心脏超声在诊断中的核心作用。

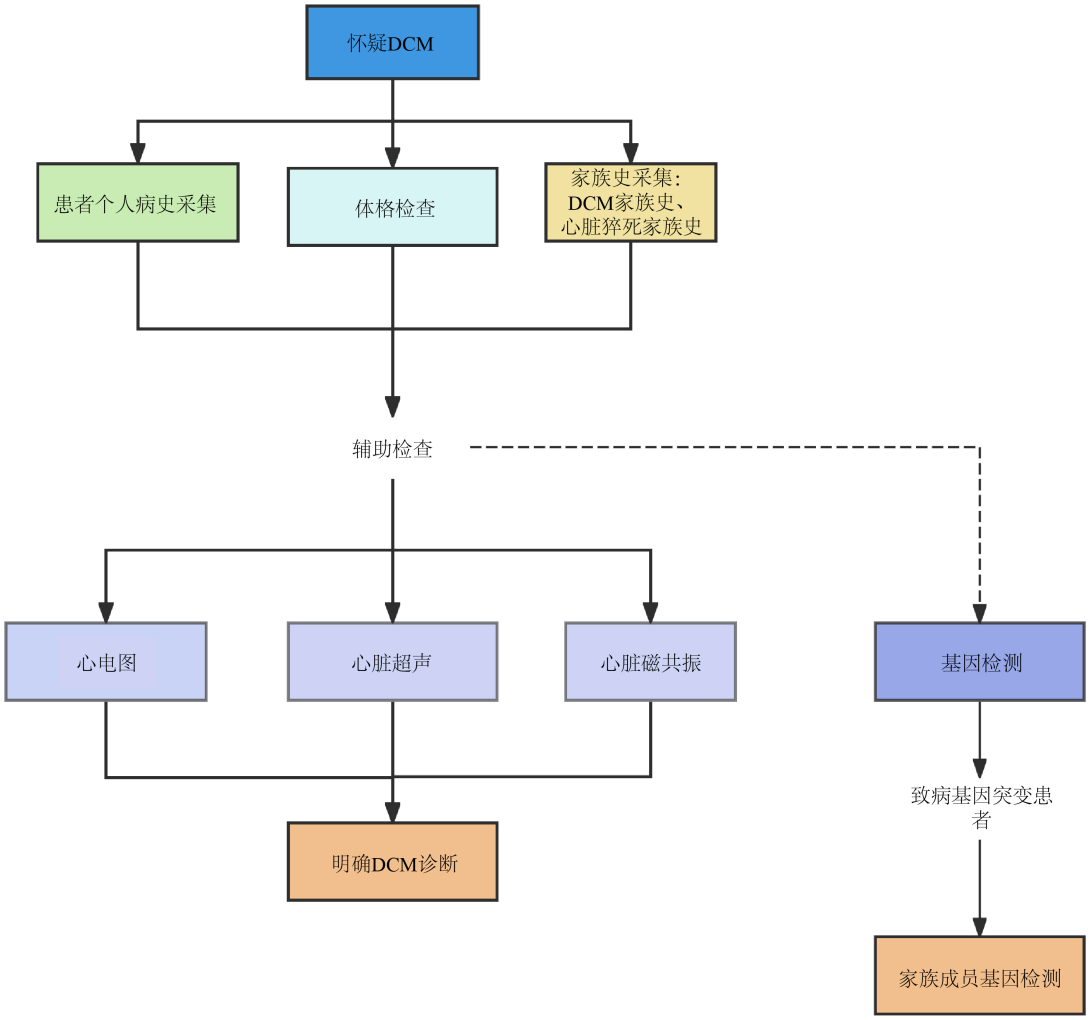

Figure 1. Diagnostic process of dilated cardiomyopathy
图 1. 扩张型心肌病的诊断流程

超声心动图是心脏收缩功能诊断和监测的核心，左心室射血分数(LVEF)仍然是扩张型心肌病事件的最强预测因子[33]。而心脏磁共振(CMR)成像是评估双心室容积、收缩功能和组织的金标准技术特征分析[34]。心脏纤维化和应变参数，特别是左心室整体纵向应变，与心血管事件相关，与 LVEF 的预后价值增加有关[35]。与扩张型心肌病相关的肌节变异个体即使没有异常的 LVEF，也表现出整体纵向应变降低[36]。左房导管应变已成为扩张型心肌病的独立预后预测指标，优于 LVEF 和左房容积，并逐渐增加到晚期钆增强[37] [38]。

传统上，扩张型心肌病的诊断需要同时识别心室扩张和收缩功能障碍；然而，如前所述，将诊断范围扩大到无左室扩张性收缩功能障碍的患者已形成共识。此外，在某些病因的情况下，最初的表型表现可能仅限于传导疾病或心律失常。虽然这些不能构成无收缩功能障碍的扩张型心肌病的诊断，但它们可

能代表疾病的早期阶段。对于全身性疾病、有毒物质(化疗、酒精和药物)、心脏或神经肌肉疾病或年轻时(<50 岁)心源性猝死的家族史，应询问详细的问题[39]。如果两个或两个以上一级或二级亲属有该病史，或一个一级亲属尸检证实有扩张型心肌病和年龄小于 50 岁的猝死，则可考虑为家族性心肌病。相关研究表明对于有扩张型心肌病家族史、心电图 QRS 波幅低、骨骼肌病、无高血压或左束支传导阻滞的患者[40]-[42]，其基因检测结果阳性。考虑到扩张型心肌病对患者及其家庭成员的高患病率和临床相关性，在有条件的情况下，扩张型心肌病的患者都应考虑进行基因检测和咨询。

4. 扩张型心肌病的治疗

4.1. 药物治疗

扩张型心肌病以左或双室扩张和运动障碍为特征[43]，其治疗以改善心功能和心肌代谢、避免感染、预防栓塞等并发症为中心，目前缺乏特异性治疗。这种疾病被认为是心力衰竭最常见的原因之一，在心力衰竭患者中占 1:250~400，在普通人群中占 1:2500 [44]。其治疗的终点是避免心力衰竭和心律失常[45]。

进入 21 世纪以来，随着医药卫生行业的不断发展，用于治疗心力衰竭的许多新型药物相继问世，如血管紧张素受体脑啡肽酶抑制剂(ARNI) [46]、钠葡萄糖协同转运蛋白 2 抑制剂(SGLT2i) [47] [48]等。血管紧张素 II 受体抑制剂能够拮抗肾素－血管紧张素－醛固酮系统(RAAS)的作用延缓病情的发展[49]。PARAGON-HF [50]研究证实了沙库巴曲缬沙坦能给 HFpEF 患者带来获益，减少 HFpEF 患者主要终点事件。EMPA-REG [51]、CANVAS-PROGRAM [52]和 DKCLARE [53]、DAPA-HF [54] [55]研究揭示出，SGLT2 抑制剂同样可以改善心血管死亡风险和心衰住院时间。2021 年欧洲心脏病学会(ESC)心力衰竭指南将钠葡萄糖协同转运蛋白 2 抑制剂(SGLT2i)、ARNI 作为 I，A 类推荐[56]。标志着射血分数减少型心力衰竭的药物治疗模式由“金三角”模式晋级为“新四联”(ARNI/ACEI/ARB、MRA、BB、SGLT2i)模式。除此之外，VICTROIA [57]研究显示，对于有症状、近期发生过心力衰竭加重事件、LVEF < 45 的心力衰竭患者，推荐在标准治疗基础上尽早加用维立西呱(IIa，B 类推荐)，以降低心血管死亡和再住院率。至此，心力衰竭的药物模式进入“五朵金花”模式(即“新四联”+ 可溶性鸟苷酸环化酶刺激剂)。

随着中国中医药行业的发展，中医药可应用于 DCM 各阶段，通过辨证论治，提高心功能、逆转心室重构、改善长期预后[58]。芪苈强心可改善葡萄糖和脂肪酸氧化，减轻氧化应激，缓解促炎和促纤维化信号传导，增强线粒体健康和线粒体自噬[59]。有相关指南推荐 HFrEF 患者，联合应用芪苈强心(B 级证据，强推荐)，改善心功能、降低 NT-pro BNP、提高生活质量[60]。除此之外，黄芪保心汤(C 级证据，弱推荐)、抗纤益心方(B 级证据，强推荐)、芪参益气滴丸(B 级证据，强推荐)、生脉散(B 级证据，强推荐)、益心舒胶囊(C 级证据，弱推荐)等等中药制剂和方剂都对扩张型心肌病患者心功能的改善有一定作用[61]。尽管中医药在治疗方面具有其独特价值，但其仍存在标准化不足、疗效证据不充分、急症与结构病受限、质量控制复杂、不能被不同地区患者接受等等局限；与此同时其并非完全无毒副作用，如不当使用可造成肝肾损伤、过敏、心血管风险。

4.2. 器械治疗

扩张型心肌病是引起心力衰竭、心律失常甚至猝死的常见心肌疾病。研究证实，超过 1/3 的慢性心力衰竭患者存在心室收缩不同步，心电图 QRS 波增宽。CRT 已被大量证据证实是心脏收缩不同步患者的有效治疗手段[62] I 类推荐，CRT 使左心室体积和尺寸减少、射血分数增加和二尖瓣反流减少[63]。植入式心律转复除颤器(ICD)是一种植入式电子设备，能够监测心脏节律，并在检测到致命性心律失常时自动释放电击进行除颤，控制可能导致猝死的室性心律失常[64]。植入式心脏收缩调节器(CCM)是通过微创手术

植入患者体内的一种小型心脏起搏器，其于心室绝对不应期发放双向强刺激，进而调节钙离子浓度。相关临床预测模型表明，LVEF 25%~45%的患者若经其治疗，再住院率可降低 75% [65]。

4.3. 外科治疗

对于扩张型心肌病所致的终末期心脏病患者的外科治疗主要有心脏移植或左心室辅助装置(LVAD)植入术。心脏移植目前仍是晚期心力衰竭治疗的金标准，但是由于供体数量的匮乏，许多患者无法进行心脏移植，处于等待供体状态。而 LVAD 的出现很好地解决了这一问题，其为终末期患者提供支持治疗。Theochari [66]等 Meta 分析的 MOMENTUM 3 试验均说明 LVAD 与心脏移植患者群体的 2 年生存率相当[67]。

4.4. 新型治疗

4.4.1. 干细胞治疗

间充质干细胞(Mesenchymal stem cells, MSC)被认为通过免疫调节和细胞保护因子等机制，在促进血管生成、减少炎症和凋亡、减轻疤痕重塑和心肌纤维化等方面发挥着至关重要的作用[68]。内皮功能障碍是 DCM 的病理生理机制之一。Premer 报道同种异体而非自体间充质干细胞可以通过释放基质衍生因子-1a 改善 DCM 患者的内皮功能[69]。Sun 的研究证明，骨髓间充质干细胞衍生的外泌体可以通过改善心肌炎症微环境，特别是通过调节巨噬细胞的活性来缓解炎症性心肌病，这有望为 DCM 的临床治疗带来希望[70]。虽然干细胞治疗在 DCM 患者中的应用似乎是合理的，但这一结论仅限于临床研究环境，正在进行的研究结果可能需要对这些结论进行修订[71]。干细胞治疗在后续的治疗方法中具有巨大的潜力，但仍存在来源标准化不足、归巢存活效率低、分化调控困难、长期疗效与安全性不明确等局限；同时也存在免疫排斥、致瘤致癌、栓塞、感染、异常分化等潜在风险。因此，在作为治疗选择时，可能存在一定的风险。

4.4.2. 基因治疗

近年来，DCM 的基因治疗领域取得了重大进展。使用腺相关病毒(AAV)载体，传递 CRISPR 碱基编辑器，在小鼠模型中成功修复了 RBM20 中的 P635L 和 R636Q 突变。心脏细胞核定位的恢复和 TTN 的正常剪接导致心脏射血分数和心室扩张的显著改善。在小鼠模型中，AAV 介导的基因编辑已达到约 70%的心肌细胞修复率，且全基因组测序未发现脱靶效应[72]。目前，针对 LMNA 突变的 CRISPR-Cas9 治疗还处于探索阶段，需要解决核膜稳定性问题[73]。未来的优化方向包括调整 AAV 载体的剂量并增强其心肌靶向性(如 AAV9 变异)，监测基因编辑和免疫反应的持久性，以及设计针对多基因突变的联合疗法(如 TTN 复合杂合突变)。此外，有必要进一步开发针对其他高频突变基因(如 TTN 和 LMNA)的精确编辑工具，并探索基因治疗与现有治疗方法(如 β 受体阻滞剂和血管紧张素转换酶抑制剂)的协同作用。基因治疗为当前的治疗提供了新思路，但仍存在递送效率低、表达调控难、适用范围窄、长期安全性不确定等局限；同时存在免疫反应、插入突变、脱靶效应、致癌、肝肾损伤等潜在风险。目前仍需在载体优化、精准编辑、安全性评估、伦理规范上进一步突破。

5. 总结与展望

近年来，扩张型心肌病的研究取得了巨大进展，指南也不断更新和完善，这使得患者病情的控制、生活质量有了显著改善。但是，目前对于这种疾病仍没有特定的治愈方法。因此，扩张型心肌病仍是一个具有挑战性但有益的领域，对其治疗的探索仍是医学领域的重大难题。此外，对于扩张型心肌病患者，应“早发现、早诊断、早治疗”，只有从源头开始实施管理，才能为其治疗开启新篇章。

参考文献

[1] Gigli, M., Stolfo, D., Merlo, M., Sinagra, G., Taylor, M.R.G. and Mestroni, L. (2024) Pathophysiology of Dilated Cardiomyopathy: From Mechanisms to Precision Medicine. *Nature Reviews Cardiology*, **22**, 183-198. https://doi.org/10.1038/s41569-024-01074-2

[2] Hazebroek, M.R., Moors, S., Dennert, R., van den Wijngaard, A., Krapels, I., Hoos, M., *et al*. (2015) Prognostic Relevance of Gene-Environment Interactions in Patients with Dilated Cardiomyopathy: Applying the MOGE(S) Classification. *Journal of the American College of Cardiology*, **66**, 1313-1323. https://doi.org/10.1016/j.jacc.2015.07.023

[3] McNally, E.M. and Mestroni, L. (2017) Dilated Cardiomyopathy: Genetic Determinants and Mechanisms. *Circulation Research*, **121**, 731-748. https://doi.org/10.1161/circresaha.116.309396

[4] Halliday, B.P., Cleland, J.G.F., Goldberger, J.J. and Prasad, S.K. (2017) Personalizing Risk Stratification for Sudden Death in Dilated Cardiomyopathy: The Past, Present, and Future. *Circulation*, **136**, 215-231. https://doi.org/10.1161/circulationaha.116.027134

[5] Tsao, C.W., Aday, A.W., Almarzooq, Z.I., *et al*. (2022) Heart Disease and Stroke Statistics-2022 Update: A Report from the American Heart Association. *Circulation*, **145**, e153-e639.

[6] Tayal, U., Ware, J.S., Lakdawala, N.K., Heymans, S. and Prasad, S.K. (2021) Understanding the Genetics of Adult-Onset Dilated Cardiomyopathy: What a Clinician Needs to Know. *European Heart Journal*, **42**, 2384-2396. https://doi.org/10.1093/eurheartj/ehab286

[7] Verdonschot, J.A.J., Hazebroek, M.R., Krapels, I.P.C., Henkens, M.T.H.M., Raafs, A., Wang, P., *et al*. (2020) Implications of Genetic Testing in Dilated Cardiomyopathy. *Circulation*: *Genomic and Precision Medicine*, **13**, 476-487. https://doi.org/10.1161/circgen.120.003031

[8] (2016) Global, Regional, and National Incidence, Prevalence, and Years Lived with Disability for 310 Diseases and Injuries, 1990-2015: A Systematic Analysis for the Global Burden of Disease Study 2015. *The Lancet* (*London, England*), **388**, 1545-1602.

[9] Halliday, B.P., Gulati, A., Ali, A., Newsome, S., Lota, A., Tayal, U., *et al*. (2018) Sex- and Age-Based Differences in the Natural History and Outcome of Dilated Cardiomyopathy. *European Journal of Heart Failure*, **20**, 1392-1400. https://doi.org/10.1002/ejhf.1216

[10] Wilsbacher, L.D. (2024) Dilated Cardiomyopathy Presentation, Early Outcomes, and Female Sex: A Paradox Revealed. *JACC*: *Heart Failure*, **12**, 364-365. https://doi.org/10.1016/j.jchf.2023.11.013

[11] García-Hernandez, S. and Iglesias, L.M. (2022) Genetic Testing as a Guide for Treatment in Dilated Cardiomyopathies. *Current Cardiology Reports*, **24**, 1537-1546. https://doi.org/10.1007/s11886-022-01772-8

[12] Tschöpe, C., Ammirati, E., Bozkurt, B., Caforio, A.L.P., Cooper, L.T., Felix, S.B., *et al*. (2020) Myocarditis and Inflammatory Cardiomyopathy: Current Evidence and Future Directions. *Nature Reviews Cardiology*, **18**, 169-193. https://doi.org/10.1038/s41569-020-00435-x

[13] Tayal, U., Newsome, S., Buchan, R., Whiffin, N., Halliday, B., Lota, A., *et al*. (2017) Phenotype and Clinical Outcomes of Titin Cardiomyopathy. *Journal of the American College of Cardiology*, **70**, 2264-2274. https://doi.org/10.1016/j.jacc.2017.08.063

[14] Liu, C., Leon, S. and Tang, W.H.W. (2026) Epigenetic Regulation in Dilated Cardiomyopathy. *Heart Failure Clinics*, **22**, 29-43. https://doi.org/10.1016/j.hfc.2025.08.002

[15] Argirò, A., Ho, C., Day, S.M., van der Velden, J., Cerbai, E., Saberi, S., *et al*. (2022) Sex-Related Differences in Genetic Cardiomyopathies. *Journal of the American Heart Association*, **11**, e024947. https://doi.org/10.1161/jaha.121.024947

[16] Mazzarotto, F., Tayal, U., Buchan, R.J., Midwinter, W., Wilk, A., Whiffin, N., *et al*. (2020) Reevaluating the Genetic Contribution of Monogenic Dilated Cardiomyopathy. *Circulation*, **141**, 387-398. https://doi.org/10.1161/circulationaha.119.037661

[17] Jordan, E., Peterson, L., Ai, T., Asatryan, B., Bronicki, L., Brown, E., *et al*. (2021) Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. *Circulation*, **144**, 7-19. https://doi.org/10.1161/circulationaha.120.053033

[18] Stroeks, S.L.V.M., Hellebrekers, D.M.E.I., Claes, G.R.F., Tayal, U., Krapels, I.P.C., Vanhoutte, E.K., *et al*. (2021) Clinical Impact of Re-Evaluating Genes and Variants Implicated in Dilated Cardiomyopathy. *Genetics in Medicine*, **23**, 2186-2193. https://doi.org/10.1038/s41436-021-01255-1

[19] Romano, R., Ghahremani, S., Zimmerman, T., Legere, N., Thakar, K., Ladha, F.A., *et al*. (2022) Reading Frame Repair of *ttn* Truncation Variants Restores Titin Quantity and Functions. *Circulation*, **145**, 194-205. https://doi.org/10.1161/circulationaha.120.049997

[20] Kamel, S.M., van Opbergen, C.J.M., Koopman, C.D., Verkerk, A.O., Boukens, B.J.D., de Jonge, B., *et al*. (2021) Istaroxime Treatment Ameliorates Calcium Dysregulation in a Zebrafish Model of Phospholamban R14del Cardiomyopathy. *Nature Communications*, **12**, Article No. 7151. https://doi.org/10.1038/s41467-021-27461-8

[21] Reichart, D., Lindberg, E.L., Maatz, H., *et al*. (2022) Pathogenic Variants Damage Cell Composition and Single Cell Transcription in Cardiomyopathies. *Science*, **377**, eabo1984.

[22] Wallace, K.B., Sardão, V.A. and Oliveira, P.J. (2020) Mitochondrial Determinants of Doxorubicin-Induced Cardiomyopathy. *Circulation Research*, **126**, 926-941. https://doi.org/10.1161/circresaha.119.314681

[23] Bhagat, A., Shrestha, P. and Kleinerman, E.S. (2022) The Innate Immune System in Cardiovascular Diseases and Its Role in Doxorubicin-Induced Cardiotoxicity. *International Journal of Molecular Sciences*, **23**, Article No. 14649. https://doi.org/10.3390/ijms232314649

[24] Yousif, L.I., Tanja, A.A., de Boer, R.A., Teske, A.J. and Meijers, W.C. (2022) The Role of Immune Checkpoints in Cardiovascular Disease. *Frontiers in Pharmacology*, **13**, Article ID: 989431. https://doi.org/10.3389/fphar.2022.989431

[25] Kundnani, N.R., Di Luca, F., Meche, V., Sharma, A., Popa, M., Nicula-Neagu, M., *et al*. (2025) Revisiting Secondary Dilative Cardiomyopathy. *International Journal of Molecular Sciences*, **26**, Article No. 4181. https://doi.org/10.3390/ijms26094181

[26] Orphanou, N., Papatheodorou, E. and Anastasakis, A. (2021) Dilated Cardiomyopathy in the Era of Precision Medicine: Latest Concepts and Developments. *Heart Failure Reviews*, **27**, 1173-1191. https://doi.org/10.1007/s10741-021-10139-0

[27] Mueller, K.A.L., Mueller, I.I., Eppler, D., Zuern, C.S., Seizer, P., Kramer, U., *et al*. (2015) Clinical and Histopathological Features of Patients with Systemic Sclerosis Undergoing Endomyocardial Biopsy. *PLOS ONE*, **10**, e0126707. https://doi.org/10.1371/journal.pone.0126707

[28] Monda, E., Palmiero, G., Rubino, M., Verrillo, F., Amodio, F., Di Fraia, F., *et al*. (2020) Molecular Basis of Inflammation in the Pathogenesis of Cardiomyopathies. *International Journal of Molecular Sciences*, **21**, Article No. 6462. https://doi.org/10.3390/ijms21186462

[29] Ampong, I. (2022) Metabolic and Metabolomics Insights into Dilated Cardiomyopathy. *Annals of Nutrition and Metabolism*, **78**, 147-155. https://doi.org/10.1159/000524722

[30] Arbustini, E., Di Toro, A., Giuliani, L., Favalli, V., Narula, N. and Grasso, M. (2018) Cardiac Phenotypes in Hereditary Muscle Disorders: JACC State-of-the-Art Review. *Journal of the American College of Cardiology*, **72**, 2485-2506. https://doi.org/10.1016/j.jacc.2018.08.2182

[31] Aimo, A., Januzzi, J.L., Vergaro, G., Ripoli, A., Latini, R., Masson, S., *et al*. (2018) Prognostic Value of High-Sensitivity Troponin T in Chronic Heart Failure: An Individual Patient Data Meta-Analysis. *Circulation*, **137**, 286-297. https://doi.org/10.1161/circulationaha.117.031560

[32] Finocchiaro, G., Merlo, M., Sheikh, N., De Angelis, G., Papadakis, M., Olivotto, I., *et al*. (2020) The Electrocardiogram in the Diagnosis and Management of Patients with Dilated Cardiomyopathy. *European Journal of Heart Failure*, **22**, 1097-1107. https://doi.org/10.1002/ejhf.1815

[33] Kim, I.C. and Yoo, B.S. (2022) Multidimensional Approach of Heart Failure Diagnosis and Prognostication Utilizing Cardiac Imaging with Biomarkers. *Diagnostics*, **12**, Article No. 1366. https://doi.org/10.3390/diagnostics12061366

[34] Smith, E.D., Lakdawala, N.K., Papoutsidakis, N., Aubert, G., Mazzanti, A., McCanta, A.C., *et al*. (2020) Desmoplakin Cardiomyopathy, a Fibrotic and Inflammatory Form of Cardiomyopathy Distinct from Typical Dilated or Arrhythmogenic Right Ventricular Cardiomyopathy. *Circulation*, **141**, 1872-1884. https://doi.org/10.1161/circulationaha.119.044934

[35] Raafs, A.G., Boscutti, A., Henkens, M.T.H.M., van den Broek, W.W.A., Verdonschot, J.A.J., Weerts, J., *et al*. (2022) Global Longitudinal Strain Is Incremental to Left Ventricular Ejection Fraction for the Prediction of Outcome in Optimally Treated Dilated Cardiomyopathy Patients. *Journal of the American Heart Association*, **11**, e024505. https://doi.org/10.1161/jaha.121.024505

[36] Lakdawala, N.K., Thune, J.J., Colan, S.D., Cirino, A.L., Farrohi, F., Rivero, J., *et al*. (2012) Subtle Abnormalities in Contractile Function Are an Early Manifestation of Sarcomere Mutations in Dilated Cardiomyopathy. *Circulation*: *Cardiovascular Genetics*, **5**, 503-510. https://doi.org/10.1161/circgenetics.112.962761

[37] Becker, M.A.J., Cornel, J.H., van de Ven, P.M., van Rossum, A.C., Allaart, C.P. and Germans, T. (2018) The Prognostic Value of Late Gadolinium-Enhanced Cardiac Magnetic Resonance Imaging in Nonischemic Dilated Cardiomyopathy: A Review and Meta-Analysis. *JACC*: *Cardiovascular Imaging*, **11**, 1274-1284. https://doi.org/10.1016/j.jcmg.2018.03.006

[38] Fong, L.C.W., Lee, N.H.C., Poon, J.W.L., Chin, C.W.L., He, B., Luo, L., *et al*. (2022) Prognostic Value of Cardiac Magnetic Resonance Derived Global Longitudinal Strain Analysis in Patients with Ischaemic and Non-Ischaemic Dilated Cardiomyopathy: A Systematic Review and Meta-Analysis. *The International Journal of Cardiovascular Imaging*, **38**, 2707-2721. https://doi.org/10.1007/s10554-022-02679-9

[39] Pinto, Y.M., Elliott, P.M., Arbustini, E., Adler, Y., Anastasakis, A., Böhm, M., *et al*. (2016) Proposal for a Revised Definition of Dilated Cardiomyopathy, Hypokinetic Non-Dilated Cardiomyopathy, and Its Implications for Clinical

Practice: A Position Statement of the ESC Working Group on Myocardial and Pericardial Diseases. *European Heart Journal*, **37**, 1850-1858. https://doi.org/10.1093/eurheartj/ehv727

[40] Heidenreich, P.A., Bozkurt, B., Aguilar, D., Allen, L.A., Byun, J.J., Colvin, M.M., *et al*. (2022) 2022 AHA/ACC/HFSA Guideline for the Management of Heart Failure: Executive Summary: A Report of the American College of Cardiology/American Heart Association Joint Committee on Clinical Practice Guidelines. *Journal of the American College of Cardiology*, **79**, 1757-1780. https://doi.org/10.1016/j.jacc.2021.12.011

[41] Wilde, A.A.M., Semsarian, C., Márquez, M.F., Shamloo, A.S., Ackerman, M.J., Ashley, E.A., *et al*. (2022) European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the State of Genetic Testing for Cardiac Diseases. *Europace*, **24**, 1307-1367. https://doi.org/10.1093/europace/euac030

[42] Escobar-Lopez, L., Ochoa, J.P., Royuela, A., Verdonschot, J.A.J., Dal Ferro, M., Espinosa, M.A., *et al*. (2022) Clinical Risk Score to Predict Pathogenic Genotypes in Patients with Dilated Cardiomyopathy. *Journal of the American College of Cardiology*, **80**, 1115-1126. https://doi.org/10.1016/j.jacc.2022.06.040

[43] Pelliccia, A., Solberg, E.E., Papadakis, M., Adami, P.E., Biffi, A., Caselli, S., *et al*. (2018) Recommendations for Participation in Competitive and Leisure Time Sport in Athletes with Cardiomyopathies, Myocarditis, and Pericarditis: Position Statement of the Sport Cardiology Section of the European Association of Preventive Cardiology (EAPC). *European Heart Journal*, **40**, 19-33. https://doi.org/10.1093/eurheartj/ehy730

[44] Ciarambino, T., Menna, G., Sansone, G. and Giordano, M. (2021) Cardiomyopathies: An Overview. *International Journal of Molecular Sciences*, **22**, 7722. https://doi.org/10.3390/ijms22147722

[45] Weintraub, R.G., Semsarian, C. and Macdonald, P. (2017) Dilated Cardiomyopathy. *The Lancet*, **390**, 400-414. https://doi.org/10.1016/s0140-6736(16)31713-5

[46] McMurray, J.J.V., Packer, M., Desai, A.S., Gong, J., Lefkowitz, M.P., Rizkala, A.R., *et al*. (2014) Angiotensin-Neprilysin Inhibition versus Enalapril in Heart Failure. *New England Journal of Medicine*, **371**, 993-1004. https://doi.org/10.1056/nejmoa1409077

[47] Wiviott, S.D., Raz, I., Bonaca, M.P., Mosenzon, O., Kato, E.T., Cahn, A., *et al*. (2019) Dapagliflozin and Cardiovascular Outcomes in Type 2 Diabetes. *New England Journal of Medicine*, **380**, 347-357. https://doi.org/10.1056/nejmoa1812389

[48] Bhatt, D.L., Szarek, M., Steg, P.G., Cannon, C.P., Leiter, L.A., McGuire, D.K., *et al*. (2021) Sotagliflozin in Patients with Diabetes and Recent Worsening Heart Failure. *New England Journal of Medicine*, **384**, 117-128. https://doi.org/10.1056/nejmoa2030183

[49] 孙海涛. 市级三甲医院慢性心力衰竭患者用药现状及影响因素分析[D]: [硕士学位论文]. 沈阳: 沈阳医学院, 2023.

[50] Vaduganathan, M., Mentz, R.J., Claggett, B.L., Miao, Z.M., Kulac, I.J., Ward, J.H., *et al*. (2023) Sacubitril/Valsartan in Heart Failure with Mildly Reduced or Preserved Ejection Fraction: A Pre-Specified Participant-Level Pooled Analysis of PARAGLIDE-HF and PARAGON-HF. *European Heart Journal*, 44, 2982-2993. https://doi.org/10.1093/eurheartj/ehad344

[51] Zinman, B., Wanner, C., Lachin, J.M., Fitchett, D., Bluhmki, E., Hantel, S., *et al*. (2015) Empagliflozin, Cardiovascular Outcomes, and Mortality in Type 2 Diabetes. *New England Journal of Medicine*, **373**, 2117-2128. https://doi.org/10.1056/nejmoa1504720

[52] Baglioni, P. (2017) Canagliflozin and Cardiovascular and Renal Events in Type 2 Diabetes. *The New England Journal of Medicine*, **377**, 2097-2098.

[53] Špinar, J., Špinarová, L. and Vítovec, J. (2021) EMPEROR-Reduced-Empagliflozin in Patients with Heart Failure and Reduced Ejection Fraction. *Vnitřní Lékařství*, **67**, 43-47. https://doi.org/10.36290/vnl.2021.006

[54] Solomon, S.D., McMurray, J.J.V., Anand, I.S., Ge, J., Lam, C.S.P., Maggioni, A.P., *et al*. (2019) Angiotensin-Neprilysin Inhibition in Heart Failure with Preserved Ejection Fraction. *New England Journal of Medicine*, **381**, 1609-1620. https://doi.org/10.1056/nejmoa1908655

[55] Solomon, S.D., McMurray, J.J.V., Claggett, B., *et al*. (2022) Dapagliflozin in Heart Failure with Mildly Reduced or Preserved Ejection Fraction. *The New England Journal of Medicine*, **387**, 1089-1098.

[56] McDonagh, T.A., Metra, M., Adamo, M., *et al*. (2021) 2021 ESC Guidelines for the Diagnosis and Treatment of Acute and Chronic Heart Failure. *European Heart Journal*, **42**, 3599-3726.

[57] 王华, 李莹莹. 2022 年 AHA/ACC/HFSA 心力衰竭管理指南解读: 从新指南看心衰分类和诊断评估[J]. 中国心血管病研究, 2022, 20(6): 481-486.

[58] Packer, M. (2023) SGLT2 Inhibitors: Role in Protective Reprogramming of Cardiac Nutrient Transport and Metabolism. *Nature Reviews Cardiology*, **20**, 443-462. https://doi.org/10.1038/s41569-022-00824-4

[59] 孙卫卫, 王冬颖, 崔英凯, 等. 充血性心力衰竭气虚血瘀证治疗的研究进展[J]. 医学研究与教育, 2018, 35(4): 24-28.

[60] 田庄, 张抒扬. 《中国心力衰竭诊断和治疗指南 2024》要点解读[J]. 协和医学杂志, 2024, 15(4): 801-806.

[61] 陶诗怡, 于林童, 李军, 等. 扩张型心肌病中西医诊疗进展[J]. 世界中西医结合杂志, 2025, 20(11): 2326-2332.

[62] 黄晓华, 唐名扬, 欧登科, 等. 心脏再同步化治疗在心力衰竭中的研究进展[J]. 心血管病学进展, 2022, 43(7): 645-648, 656.

[63] Cazeau, S., Leclercq, C., Lavergne, T., Walker, S., Varma, C., Linde, C., *et al*. (2001) Effects of Multisite Biventricular Pacing in Patients with Heart Failure and Intraventricular Conduction Delay. *New England Journal of Medicine*, **344**, 873-880. https://doi.org/10.1056/nejm200103223441202

[64] Heymans, S., Lakdawala, N.K., Tschöpe, C. and Klingel, K. (2023) Dilated Cardiomyopathy: Causes, Mechanisms, and Current and Future Treatment Approaches. *The Lancet*, **402**, 998-1011. https://doi.org/10.1016/s0140-6736(23)01241-2

[65] Wiegn, P., Chan, R., Jost, C., Saville, B.R., Parise, H., Prutchi, D., *et al*. (2020) Safety, Performance, and Efficacy of Cardiac Contractility Modulation Delivered by the 2-Lead Optimizer Smart System: The FIX-HF-5C2 Study. *Circulation*: *Heart Failure*, **13**, e006512. https://doi.org/10.1161/circheartfailure.119.006512

[66] Theochari, C.A., Michalopoulos, G., Oikonomou, E.K., Giannopoulos, S., Doulamis, I.P., Villela, M.A., *et al*. (2018) Heart Transplantation versus Left Ventricular Assist Devices as Destination Therapy or Bridge to Transplantation for 1-Year Mortality: A Systematic Review and Meta-Analysis. *Annals of Cardiothoracic Surgery*, **7**, 3-11. https://doi.org/10.21037/acs.2017.09.18

[67] Mehra, M.R., Uriel, N., Naka, Y., Cleveland, J.C., Yuzefpolskaya, M., Salerno, C.T., *et al*. (2019) A Fully Magnetically Levitated Left Ventricular Assist Device—Final Report. *New England Journal of Medicine*, **380**, 1618-1627. https://doi.org/10.1056/nejmoa1900486

[68] Janssens, S.P. (2017) Mesenchymal Cell Therapy for Dilated Cardiomyopathy: Time to Test the Water. *Journal of the American College of Cardiology*, **69**, 538-540. https://doi.org/10.1016/j.jacc.2016.11.044

[69] Premer, C., Wanschel, A., Porras, V., Balkan, W., Legendre-Hyldig, T., Saltzman, R.G., *et al*. (2019) Mesenchymal Stem Cell Secretion of SDF-1α Modulates Endothelial Function in Dilated Cardiomyopathy. *Frontiers in Physiology*, **10**, Article No. 1182. https://doi.org/10.3389/fphys.2019.01182

[70] Sun, X., Shan, A., Wei, Z. and Xu, B. (2018) Intravenous Mesenchymal Stem Cell-Derived Exosomes Ameliorate Myocardial Inflammation in the Dilated Cardiomyopathy. *Biochemical and Biophysical Research Communications*, **503**, 2611-2618. https://doi.org/10.1016/j.bbrc.2018.08.012

[71] Diaz-Navarro, R., Urrútia, G., Cleland, J.G., Poloni, D., Villagran, F., Acosta-Dighero, R., *et al*. (2021) Stem Cell Therapy for Dilated Cardiomyopathy. *Cochrane Database of Systematic Reviews*, **2021**, CD013433. https://doi.org/10.1002/14651858.cd013433.pub2

[72] Grosch, M., Schraft, L., Chan, A., Küchenhoff, L., Rapti, K., Ferreira, A., *et al*. (2023) Striated Muscle-Specific Base Editing Enables Correction of Mutations Causing Dilated Cardiomyopathy. *Nature Communications*, **14**, Article No. 3714. https://doi.org/10.1038/s41467-023-39352-1

[73] Zhou, L., Liu, C., Zou, Y. and Chen, Z. (2022) Development and Verification of the Nomogram for Dilated Cardiomyopathy Gene Diagnosis. *Scientific Reports*, **12**, Article No. 8908. https://doi.org/10.1038/s41598-022-13135-y