Blueprint Genetics

Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel

Test code: CA1801

Is a 33 gene panel that includes assessment of non-coding variants.

Is ideal for patients with a clinical suspicion of left ventricular non-compaction cardiomyopathy.

About Left Ventricular Non-Compaction Cardiomyopathy (LVNC)

Left ventricular noncompaction (LVNC) is a cardiomyopathy characterized by a spongy appearance of the left ventricle and prominent trabeculations. Individuals may be asymptomatic, but common symptoms include arrhythmias, heart failure, and thromboembolism. LVNC can be seen in the context of congenital structural heart disease in children. LVNC is considered to have a significant amount of overlap with hypertrophic (HCM) and dilated cardiomyopathies (DCM). Many patients with clinical diagnosis of LVNC carry established mutations normally found in patients with clinical diagnosis of HCM or DCM. As the understanding of LVNC phenotype is still limited, we recommend genetic testing to follow HCM scheme if patient's myocardial thickness exceeds 15 mm at any segment or DCM scheme when the LV size and function fulfill DCM criteria even when significant hypertrabeculation co-exists. Our Cardiomyopathy Panel provides highest diagnostic utility in cases with complex phenotype.

Availability

4 weeks

Gene Set Description

Genes in the Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel and their clinical significance

Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
ABCC9	Atrial fibrillation, Cantu syndrome, Dilated cardiomyopathy (DCM)	AD	27	46
BAG3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar	AD	39	62
CTNNA3	Arrhythmogenic right ventricular dysplasia	AD	7	46
DES	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Scapuloperoneal syndrome, neurogenic, Kaeser type	AD/AR	64	124
DMD	Becker muscular dystrophy, Duchenne muscular dystrophy, Dilated cardiomyopathy (DCM)	XL	832	3915
DSC2	Arrhythmogenic right ventricular dysplasia with palmoplantar keratoderma and woolly hair, Arrhythmogenic right ventricular dysplasia	AD/AR	32	87
DSG2	Arrhythmogenic right ventricular dysplasia, Dilated cardiomyopathy (DCM)	AD	44	129
DSP	Cardiomyopathy, dilated, with wooly hair, keratoderma, and tooth agenesis, Arrhythmogenic right ventricular dysplasia, familial, Cardiomyopathy, dilated, with wooly hair and keratoderma, Keratosis palmoplantaris striata II, Epidermolysis bullosa, lethal acantholytic	AD/AR	177	296

DTNA	Left ventricular noncompaction 1	AD	3	7
EMD	Emery-Dreifuss muscular dystrophy	XL	48	113
FBXO32	Dilated cardiomyopathy (DCM)	AD/AR		2
FLNC*	Myopathy	AD	54	109
HCN4	Sick sinus syndrome, Brugada syndrome, Left ventricular non-compaction cardiomyopathy (LVNC)	AD	8	34
JPH2	Hypertrophic cardiomyopathy (HCM)	AD	3	13
JUP	Arrhythmogenic right ventricular dysplasia, Naxos disease	AD/AR	8	46
LAMP2	Danon disease	XL	62	101
LMNA	Heart-hand syndrome, Slovenian, Limb-girdle muscular dystrophy, Muscular dystrophy, congenital, LMNA-related, Lipodystrophy (Dunnigan), Emery-Dreiffus muscular dystrophy, Malouf syndrome, Dilated cardiomyopathy (DCM), Mandibuloacral dysplasia type A, Progeria Hutchinson-Gilford type	AD/AR	250	564
MIPEP	Combined oxidative phosphorylation deficiency 31	AR	5	8
MYBPC3	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	482	1048
MYH6	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM), Atrial septal defect 3	AD	14	123
MYH7	Hypertrophic cardiomyopathy (HCM), Myopathy, myosin storage, Myopathy, distal, Dilated cardiomyopathy (DCM)	AD	305	986
PKP2*	Arrhythmogenic right ventricular dysplasia	AD	150	289
PLEKHM2	Dilated cardiomyopathy (DCM), left ventricular noncompaction	AR	1	1
PLN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD/AR	8	30
RAF1	LEOPARD syndrome, Noonan syndrome, Dilated cardiomyopathy (DCM)	AD	45	53
RBM20	Dilated cardiomyopathy (DCM)	AD	19	47
RYR2	Ventricular tachycardia, catecholaminergic polymorphic, Arrhythmogenic right ventricular dysplasia	AD	124	372
SCN5A	Heart block, nonprogressive, Heart block, progressive, Long QT syndrome, Ventricular fibrillation, Atrial fibrillation, Sick sinus syndrome, Brugada syndrome, Dilated cardiomyopathy (DCM)	AD/AR/Digenic	234	899
TCAP	Muscular dystrophy, limb-girdle, Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD/AR	12	28
TNNT2	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM)	AD	61	148
TPM1	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	34	98

TTN*	Dilated cardiomyopathy (DCM), Tibial muscular dystrophy, Limb-girdle muscular dystrophy, Hereditary myopathy with early respiratory failure, Myopathy, early-onset, with fatal cardiomyopathy (Salih myopathy), Muscular dystrophy, limb-girdle, type 2J	AD	818	327
VCL	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	8	30

*

Some, or all, of the gene is duplicated in the genome. Read more.

#

The gene has suboptimal coverage (means <90% of the gene's target nucleotides are covered at >20x with mapping quality score (MQ>20) reads), and/or the gene has exons listed under Test limitations section that are not included in the panel as they are not sufficiently covered with high quality sequence reads.

The sensitivity to detect variants may be limited in genes marked with an asterisk (*) or number sign (#). Due to possible limitations these genes may not be available as single gene tests.

Gene refers to the HGNC approved gene symbol; Inheritance refers to inheritance patterns such as autosomal dominant (AD), autosomal recessive (AR), mitochondrial (mi), X-linked (XL), X-linked dominant (XLD) and X-linked recessive (XLR); ClinVar refers to the number of variants in the gene classified as pathogenic or likely pathogenic in this database (ClinVar); HGMD refers to the number of variants with possible disease association in the gene listed in Human Gene Mutation Database (HGMD). The list of associated, gene specific phenotypes are generated from CGD or Mitomap databases.

Non-coding disease causing variants covered by the panel

Gene	Genomic location HG19	HGVS	RefSeq	RS-number
DMD	ChrX:31165653	c.10554-18C>G	NM_004006.2	
DMD	ChrX:31200680	c.9974+175T>A	NM_004006.2	
DMD	ChrX:31224814	c.9564-30A>T	NM_004006.2	
DMD	ChrX:31225211	c.9564-427T>G	NM_004006.2	
DMD	ChrX:31226400	c.9563+1215A>G	NM_004006.2	
DMD	ChrX:31229031	c.9362-1215A>G	NM_004006.2	
DMD	ChrX:31241047	c.9361+117A>G	NM_004006.2	
DMD	ChrX:31279293	c.9225-160A>G	NM_004006.2	
DMD	ChrX:31279418	c.9225-285A>G	NM_004006.2	
DMD	ChrX:31279420	c.9225-287C>A	NM_004006.2	
DMD	ChrX:31279780	c.9225-647A>G	NM_004006.2	rs398124091

DMD	ChrX:31279781	c.9225-648A>G	NM_004006.2	rs398124084
DMD	ChrX:31332523	c.9224+9192C>A	NM_004006.2	
DMD	ChrX:31382270	c.9085-15519G>T	NM_004006.2	
DMD	ChrX:31613687	c.8217+32103G>T	NM_004006.2	
DMD	ChrX:31627738	c.8217+18052A>G	NM_004006.2	
DMD	ChrX:31697714	c.7661-11T>C	NM_004006.2	
DMD	ChrX:31897527	c.6913-4037T>G	NM_004006.2	
DMD	ChrX:31983146	c.6614+3310G>T	NM_004006.2	rs797045526
DMD	ChrX:32274692	c.6290+30954C>T	NM_004006.2	
DMD	ChrX:32305833	c.6118-15A>G	NM_004006.2	
DMD	ChrX:32360414	c.5740-15G>T	NM_004006.2	
DMD	ChrX:32366860	c.5326-215T>G	NM_004006.2	
DMD	ChrX:32379144	c.5325+1743_5325+1760delTATTAAAAAATGGGTAGA	NM_004006.2	
DMD	ChrX:32398808	c.4675-11A>G	NM_004006.2	
DMD	ChrX:32460274	c.3787-843C>A	NM_004006.2	
DMD	ChrX:32470726	c.3603+2053G>C	NM_004006.2	
DMD	ChrX:32479316	c.3432+2240A>G	NM_004006.2	
DMD	ChrX:32479520	c.3432+2036A>G	NM_004006.2	
DMD	ChrX:32669100	c.961-5831C>T	NM_004006.2	rs398124099
DMD	ChrX:32669194	c.961-5925A>C	NM_004006.2	
DMD	ChrX:32716130	c.832-15A>G	NM_004006.2	rs72470513
DMD	ChrX:32756908	c.650-39498A>G	NM_004006.2	
DMD	ChrX:32827744	c.531-16T>A/G	NM_004006.2	
DMD	ChrX:32827744	c.531-16T>A	NM_004006.2	
DMD	ChrX:32827744	c.531-16T>G	NM_004006.2	
DMD	ChrX:32841967	c.265-463A>G	NM_004006.2	
DMD	ChrX:33032666	c.93+5590T>A	NM_004006.2	
DMD	ChrX:33192452	c.31+36947G>A	NM_004006.2	
DMD	ChrX:33229483	c.-54T>A	NM_004006.2	
DSC2	Chr18:28683379	c.-1445G>C	NM_024422.4	rs75494355
EMD	ChrX:153608559	c.266-27_266-10delTCTGCTACCGCTGCCCCC	NM_000117.2	

LMNA	Chr1:156100609	c.513+45T>G	NM_170707.3	
LMNA	Chr1:156105681	c.937-11C>G	NM_170707.3	rs267607645
LMNA	Chr1:156107037	c.1608+14G>A	NM_170707.3	
LMNA	Chr1:156107433	c.1609-12T>G	NM_170707.3	rs267607582
MYBPC3	Chr11:47353394	c.*26+2T>C	NM_000256.3	
MYBPC3	Chr11:47353821	c.3628-12C>G	NM_000256.3	rs371428751
MYBPC3	Chr11:47359371	c.2309-26A>G	NM_000256.3	
MYBPC3	Chr11:47360310	c.2149-80G>A	NM_000256.3	
MYBPC3	Chr11:47364709	c.1227-13G>A	NM_000256.3	rs397515893
MYBPC3	Chr11:47364832	c.1224-19G>A	NM_000256.3	rs587776699
MYBPC3	Chr11:47364865	c.1224-52G>A	NM_000256.3	rs786204336
MYBPC3	Chr11:47365750	c.1091-575A>C	NM_000256.3	
MYBPC3	Chr11:47367305	c.1090+453C>T	NM_000256.3	
MYBPC3	Chr11:47368602	c.906-22G>A	NM_000256.3	rs756267771
MYBPC3	Chr11:47368616	c.906-36G>A	NM_000256.3	rs864622197
PLN	Chr6:118869382	c.-271A>G	NM_002667.4	
PLN	Chr6:118869417	c.-236C>G	NM_002667.4	rs188578681
RYR2	Chr1:237730106	c.3423+32dupG	NM_001035.2	
SCN5A	Chr3:38639469	c.2024-11T>A	NM_198056.2	rs777987317
SCN5A	Chr3:38691021	c.-53+1G>A	NM_198056.2	
TPM1	Chr15:63349172	c.241-12_241-11delCTinsTG	NM_001018005.1	rs199476309

Test Strengths

The strengths of this test include:

- CAP accredited laboratory
- CLIA-certified personnel performing clinical testing in a CLIA-certified laboratory
- Powerful sequencing technologies, advanced target enrichment methods and precision bioinformatics pipelines ensure superior analytical performance
- Careful construction of clinically effective and scientifically justified gene panels
- Some of the panels include the whole mitochondrial genome (please see the Panel Content section)
- Our Nucleus online portal providing transparent and easy access to quality and performance data at the patient level
- ~2,000 non-coding disease causing variants in our clinical grade NGS assay for panels (please see 'Non-coding disease causing variants covered by this panel' in the Panel Content section)
- Our rigorous variant classification scheme
- Our systematic clinical interpretation workflow using proprietary software enabling accurate and traceable processing of NGS data
- Our comprehensive clinical statements

Test Limitations

The following exons are not included in the panel as they are not sufficiently covered with high quality sequence reads: *PKP2* (NM_001254727:6). Genes with suboptimal coverage in our assay are marked with number sign (#) and genes with partial, or whole gene, segmental duplications in the human genome are marked with an asterisk (*) if they overlap with the UCSC pseudogene regions. Gene is considered to have suboptimal coverage when >90% of the gene's target nucleotides are not covered at >20x with mapping quality score (MQ>20) reads. The technology may have limited sensitivity to detect variants in genes marked with these symbols (please see the Panel content table above).

This test does not detect the following:

- Complex inversions
- Gene conversions
- Balanced translocations
- Some of the panels include the whole mitochondrial genome but not all (please see the Panel Content section)
- Repeat expansion disorders unless specifically mentioned
- Non-coding variants deeper than ±20 base pairs from exon-intron boundary unless otherwise indicated (please see above Panel Content / non-coding variants covered by the panel).

This test may not reliably detect the following:

- Low level mosaicism in nuclear genes (variant with a minor allele fraction of 14.6% is detected with 90% probability)
- Stretches of mononucleotide repeats
- Low level heteroplasmy in mtDNA (>90% are detected at 5% level)
- Indels larger than 50bp
- Single exon deletions or duplications
- Variants within pseudogene regions/duplicated segments
- Some disease causing variants present in mtDNA are not detectable from blood, thus post-mitotic tissue such as skeletal muscle may be required for establishing molecular diagnosis.

The sensitivity of this test may be reduced if DNA is extracted by a laboratory other than Blueprint Genetics.

For additional information, please refer to the Test performance section.

Test Performance

The genes on the panel have been carefully selected based on scientific literature, mutation databases and our experience.

Our panels are sectioned from our high-quality, clinical grade NGS assay. Please see our sequencing and detection performance table for details regarding our ability to detect different types of alterations (Table).

Assays have been validated for various sample types including EDTA-blood, isolated DNA (excluding from formalin fixed paraffin embedded tissue), saliva and dry blood spots (filter cards). These sample types were selected in order to maximize the likelihood for high-quality DNA yield. The diagnostic yield varies depending on the assay used, referring healthcare professional, hospital and country. Plus analysis increases the likelihood of finding a genetic diagnosis for your patient, as large deletions and duplications cannot be detected using sequence analysis alone. Blueprint Genetics' Plus Analysis is a combination of both sequencing and deletion/duplication (copy number variant (CNV)) analysis.

The performance metrics listed below are from an initial validation performed at our main laboratory in Finland. The performance metrics of our laboratory in Seattle, WA, are equivalent.

Performance of Blueprint Genetics high-quality, clinical grade NGS sequencing assay for panels.

	Sensitivity % (TP/(TP+FN)	Specificity %

Single nucleotide variants	99.89% (99,153/99,266)	>99.9999%
Insertions, deletions and indels by sequence analysis		
1-10 bps	99.2% (7,745/7,806)	>99.9999%
11-50 bps	99.13% (2,524/2,546)	>99.9999%
Copy number variants (exon level dels/dups)		
1 exon level deletion (heterozygous)	100% (20/20)	NA
1 exon level deletion (homozygous)	100% (5/5)	NA
1 exon level deletion (het or homo)	100% (25/25)	NA
2-7 exon level deletion (het or homo)	100% (44/44)	NA
1-9 exon level duplication (het or homo)	75% (6/8)	NA
Simulated CNV detection		
5 exons level deletion/duplication	98.7%	100.00%
Microdeletion/-duplication sdrs (large CNVs, n=37))		
Size range (0.1-47 Mb)	100% (25/25)	

The performance presented above reached by Blueprint Genetics high-quality, clinical grade NGS sequencing assay with the following coverage metrics

Mean sequencing depth	143X
Nucleotides with >20x sequencing coverage (%)	99.86%

Performance of Blueprint Genetics Mitochondrial Sequencing Assay.

	Sensitivity %	**Specificity %**
ANALYTIC VALIDATION (NA samples; n=4)		
Single nucleotide variants		
Heteroplasmic (45-100%)	100.0% (50/50)	100.0%
Heteroplasmic (35-45%)	100.0% (87/87)	100.0%
Heteroplasmic (25-35%)	100.0% (73/73)	100.0%
Heteroplasmic (15-25%)	100.0% (77/77)	100.0%
Heteroplasmic (10-15%)	100.0% (74/74)	100.0%
Heteroplasmic (5-10%)	100.0% (3/3)	100.0%
Heteroplasmic (<5%)	50.0% (2/4)	100.0%

CLINICAL VALIDATION (n=76 samples)		
All types		
Single nucleotide variants n=2026 SNVs		
Heteroplasmic (45-100%)	100.0% (1940/1940)	100.0%
Heteroplasmic (35-45%)	100.0% (4/4)	100.0%
Heteroplasmic (25-35%)	100.0% (3/3)	100.0%
Heteroplasmic (15-25%)	100.0% (3/3)	100.0%
Heteroplasmic (10-15%)	100.0% (9/9)	100.0%
Heteroplasmic (5-10%)	92.3% (12/13)	99.98%
Heteroplasmic (<5%)	88.9% (48/54)	99.93%
Insertions and deletions by sequence analysis n=40 indels		
Heteroplasmic (45-100%) 1-10bp	100.0% (32/32)	100.0%
Heteroplasmic (5-45%) 1-10bp	100.0% (3/3)	100.0%
Heteroplasmic (<5%) 1-10bp	100.0% (5/5)	99,997%
SIMULATION DATA /(mitomap mutations)		
Insertions, and deletions 1-24 bps by sequence analysis; n=17		
Homoplasmic (100%) 1-24bp	100.0% (17/17)	99.98%
Heteroplasmic (50%)	100.0% (17/17)	99.99%
Heteroplasmic (25%)	100.0% (17/17)	100.0%
Heteroplasmic (20%)	100.0% (17/17)	100.0%
Heteroplasmic (15%)	100.0% (17/17)	100.0%
Heteroplasmic (10%)	94.1% (16/17)	100.0%
Heteroplasmic (5%)	94.1% (16/17)	100.0%
Copy number variants (separate artifical mutations; n=1500)		
Homoplasmic (100%) 500 bp, 1kb, 5 kb	100.0%	100.0%
Heteroplasmic (50%) 500 bp, 1kb, 5 kb	100.0%	100.0%
Heteroplasmic (30%) 500 bp, 1kb, 5 kb	100.0%	100.0%
Heteroplasmic (20%) 500 bp, 1kb, 5 kb	99.7%	100.0%
Heteroplasmic (10%) 500 bp, 1kb, 5 kb	99.0%	100.0%
The performance presented above reached by following coverage metrics at assay level (n=66)		

	Mean of medians	Median of medians
Mean sequencing depth MQ0 (clinical)	18224X	17366X
Nucleotides with >1000x MQ0 sequencing coverage (%) (clinical)	100%	
rho zero cell line (=no mtDNA), mean sequencing depth	12X	

Bioinformatics

The target region for each gene includes coding exons and ±20 base pairs from the exon-intron boundary. In addition, the panel includes non-coding and regulatory variants if listed above (Non-coding variants covered by the panel). Some regions of the gene(s) may be removed from the panel if specifically mentioned in the 'Test limitations" section above. If the test includes the mitochondrial genome the target region gene list contains the mitochondrial genes. The sequencing data generated in our laboratory is analyzed with our proprietary data analysis and annotation pipeline, integrating state-of-the art algorithms and industry-standard software solutions. Incorporation of rigorous quality control steps throughout the workflow of the pipeline ensures the consistency, validity and accuracy of results. Our pipeline is streamlined to maximize sensitivity without sacrificing specificity. We have incorporated a number of reference population databases and mutation databases including, but not limited, to 1000 Genomes Project, gnomAD, ClinVar and HGMD into our clinical interpretation software to make the process effective and efficient. For missense variants, *in silico* variant prediction tools such as SIFT, PolyPhen,MutationTaster are used to assist with variant classification. Through our online ordering and statement reporting system, Nucleus, ordering providers have access to the details of the analysis, including patient specific sequencing metrics, a gene level coverage plot and a list of regions with suboptimal coverage (<20X for nuclear genes and <1000X for mtDNA) if applicable. This reflects our mission to build fully transparent diagnostics where ordering providers can easily visualize the crucial details of the analysis process.

Clinical Interpretation

We provide customers with the most comprehensive clinical report available on the market. Clinical interpretation requires a fundamental understanding of clinical genetics and genetic principles. At Blueprint Genetics, our PhD molecular geneticists, medical geneticists and clinical consultants prepare the clinical statement together by evaluating the identified variants in the context of the phenotypic information provided in the requisition form. Our goal is to provide clinically meaningful statements that are understandable for all medical professionals regardless of whether they have formal training in genetics.

Variant classification is the corner stone of clinical interpretation and resulting patient management decisions. Our classifications follow the ACMG guideline 2015.

The final step in the analysis is orthogonal confirmation. Sequence and copy number variants classified as pathogenic, likely pathogenic and variants of uncertain significance (VUS) are confirmed using bi-directional Sanger sequencing or by orthogonal methods such as qPCR/ddPCR when they do not meet our stringent NGS quality metrics for a true positive call.

Our clinical statement includes tables for sequencing and copy number variants that include basic variant information (genomic coordinates, HGVS nomenclature, zygosity, allele frequencies, in silico predictions, OMIM phenotypes and classification of the variant). In addition, the statement includes detailed descriptions of the variant, gene and phenotype(s) including the role of the specific gene in human disease, the mutation profile, information about the gene's variation in population cohorts and detailed information about related phenotypes. We also provide links to the references, abstracts and variant databases used to help ordering providers further evaluate the reported findings if desired. The conclusion summarizes all of the existing information and provides our rationale for the classification of the variant.

Identification of pathogenic or likely pathogenic variants in dominant disorders or their combinations in different alleles in recessive disorders are considered molecular confirmation of the clinical diagnosis. In these cases, family member testing can be used for risk stratification. We do not recommend using variants of uncertain significance (VUS) for family member risk stratification or patient management. Genetic counseling is recommended.

Our interpretation team analyzes millions of variants from thousands of individuals with rare diseases. Our internal database and our understanding of variants and related phenotypes increases with every case analyzed. Our laboratory is therefore

well-positioned to re-classify previously reported variants as new information becomes available. If a variant previously reported by Blueprint Genetics is re-classified, our laboratory will issue a follow-up statement to the original ordering health care provider at no additional cost.

CPT code(s) *

81439(1)

* The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.

ICD Codes

Refer to the most current version of ICD-10-CM manual for a complete list of ICD-10 codes.

Sample Requirements

- Blood (min. 1ml) in an EDTA tube
- Extracted DNA, min. 2 µg in TE buffer or equivalent
- Saliva (Please see Sample Requirements for accepted saliva kits)

Label the sample tube with your patient's name, date of birth and the date of sample collection.

We do not accept DNA samples isolated from formalin-fixed paraffin-embedded (FFPE) tissue. In addition, if the patient is affected with a hematological malignancy, DNA extracted from a non-hematological source (e.g. skin fibroblasts) is strongly recommended.

Please note that, in rare cases, mitochondrial genome (mtDNA) variants may not be detectable in blood or saliva in which case DNA extracted from post-mitotic tissue such as skeletal muscle may be a better option.

Read more about our sample requirements here.

For Patients

Other

- American Foundation for Cardiomyopathy
- Ashley EA et al. Genetics and cardiovascular disease: a policy statement from the American Heart Association. Circulation. 2012 Jul 3;126(1):142-57.
- Bozkurt B et al. Current Diagnostic and Treatment Strategies for Specific Dilated Cardiomyopathies: A Scientific Statement From the American Heart Association. Circulation. 2016 Dec 6;134(23):e579-e646.
- Cardiomyopathy Association Australia
- Cardiomyopathy UK - DCM
- Cardiomyopathy UK - HCM
- Cardiomyopathy UK - LVNC
- GeneReviews - DCM
- GeneReviews - HCM
- Genetic and Rare Diseases Information Center - LVNC
- Hypertrophic Cardiomyopathy Association
- Ingles J et al. Genetic testing for inherited heart diseases: longitudinal impact on health-related quality of life. Genet Med. 2012 May 3.
- NORD - Pediatric Cardiomyopathy