Seraseq® Inherited Cardiovascular DNA Mix

Assay validation and daily-run QC material for testing of various hereditary cardiovascular conditions such as cardiomyopathies and arrhythmias

INTRODUCTION

Inherited cardiovascular disorders, including cardiomyopathies and arrhythmias, are significant causes of sudden cardiac arrest, heart failure, and other life-threatening conditions. These disorders often result from pathogenic genetic mutations that can be passed through families, making early diagnosis and intervention critical.

Genetic testing, particularly with next-generation sequencing (NGS), has become a cornerstone in identifying these mutations and guiding patient management and family risk assessments. However, the detection of complex variants, such as structural changes and mutations in repetitive regions, poses challenges for clinical laboratories.

Reference materials like the Seraseq® Inherited Cardiovascular DNA Mix provide a standardized means to evaluate and validate the performance of genetic tests. They offer:

- Invaluable alternative to hard-to-source clinical samples, providing labs with consistent, reliable, and high-quality controls to simulate real-world genomic complexities, including structural variations and mutations in challenging regions.
- Reproducible quality control, enabling consistent and reliable assay results.
- Regulatory compliance and accreditation requirements by demonstrating test accuracy and reliability.

By integrating these high-quality reference materials into their workflows, clinical laboratories can ensure the robustness of their assays and improve confidence in their genetic test results.

FEATURES AND BENEFITS

- Targeted for Inherited Cardiovascular Diseases Genetic Testing: Covers 56 genes linked to all major cardiomyopathy types, including hypertrophic, dilated, restrictive, and arrhythmogenic cardiomyopathies, as well as arrhythmias and other syndromes.
- Comprehensive Variant Coverage: Includes 60 clinically-relevant mutations, all classified as pathogenic or likely pathogenic according to ACMG recommendations, the FDA-recognized database, and/or an expert-reviewed panel.
- Expertly Designed: Crafted to mimic challenging genomic regions, providing a robust tool for clinical laboratories working on next-generation sequencing (NGS) assays for inherited cardiovascular conditions.
- High Variant Diversity: Features a wide variety of mutations, including single nucleotide variants (SNVs), deletions, duplications, microsatellite instability, large deletions in repetitive regions, and large duplications.
- Single-Vial Convenience: Simplify workflows with a highly multiplexed reference material packaged in a single vial, reducing costs and saving time while boosting QC consistency.

HIGHLIGHTS

High-quality third-party QC material in mutation mix format to develop, validate, monitor, and troubleshoot your assay

Convenient highly multiplexed material in a single vial format to save time, cost, and increase QC consistency.

Designed to support compliance with regulatory standards like CAP, CLIA ensures your lab meets performance, quality, and reporting requirements effortlessly.

ORDERING INFORMATION

Material #	Product	Concentration	Fill Size	Total Mass
0740-0132	Seraseq® Inherited Cardiovascular DNA Mix	25 ng/µL	1 vial x 20µL	500 ng

Not for In Vitro Diagnostic Use. Research Use Only.

MUTATIONS PRESENT IN THE SERASEQ® INHERITED CARDIOVASCULAR DNA MIX

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	Variant size (bp)	Classification	Conditions
ABCC9	c.3347G>A (p.Arg1116His)	SNV	Missense variant	1	** Pathogenic	Hypertrichotic osteochondrodysplasia Cantu type; Dilated cardiomyopathy 1O
ACTC1	c.301G>A (p.Glu101Lys)	SNV	Missense variant	1	** Pathogenic/Likely pathogenic	Dilated cardiomyopathy 1R; Hypertrophic cardiomyopathy 11; Atrial septal defect 5
ACTN2	c.355G>A (p.Ala119Thr)	SNV	Missense variant | 5 prime UTR variant	1	** Pathogenic	Primary dilated cardiomyopathy; Ventricular fibrillation; Noncompaction cardiomyopathy; Atrial septal defect 7; Primary dilated cardiomyopathy; Hypertrophic; Dilated cardiomyopathy 1AA; ACTN2-related disorder
ALPK3	c.2043_2044delinsCT (p.Gln681_Glu682delinsHisTer)	Indel	Nonsense	2	** Pathogenic/Likely pathogenic	Cardiomyopathy; Cardiovascular phenotype
BAG3	c.351_352insTGGATGCAGCG ATTCCGAACTGAGGCGGCAG CAGCGGCTCCTCAGAGGTCC CAGTCACCTCTGCGGGGCA TGCCAGAAACCACTCAGCCA GATAAACAGCGTGGACAGGT GGCAGCGGCGGCGGCACCC AGCCCCCAGCCT (p.Gly118fs)	Large Microsatellite	Frameshift variant	142	* Pathogenic	Dilated cardiomyopathy 1HH; Myofibrillar myopathy 6
CACNA1C	c.1553G>A (p.Arg518Hi	SNV	Missense variant	1	** Pathogenic/Likely pathogenic	Hypertrophic cardiomyopathy 1, Long QT syndrome 8, Cardiac arrhythmia; Timothy syndrome
CALM1	c.293A>G (p.Asn98Ser)	SNV	Missense variant	1	** Pathogenic/Likely pathogenic	Catecholaminergic polymorphic ventricular tachycardia 4, Long QT syndrome 14
CALM2	c.293A>G (p.Asn98Ser)	SNV	Missense variant	1	** Pathogenic	Long QT syndrome 15
CASQ2	c.339_354del (p.Ser113fs)	Deletion	Frameshift variant	16	** Pathogenic	Catecholaminergic polymorphic ventricular tachycardia 1 & 2
CDH2	c.1344+1G>A	SNV	Splice donor variant	1	** Pathogenic	Arrhythmogenic right ventricular dysplasia, familial, 14; Agenesis of corpus callosum
CTNNA3	c.2293TTG[1] (p.Leu766del)	Microsatellite	Inframe deletion	3	Pathogenic	Arrhythmogenic right ventricular cardiomyopathy
CRYAB	c.145del (p.Leu49fs)	Deletion	Frameshift variant	1	* Pathogenic	Dilated cardiomyopathy 1II
DES	c.1216C>T (p.Arg406Trp)	SNV	Missense variant	1	** Pathogenic/Likely pathogenic	Desmin-related myofibrillar myopathy; Arrhythmogenic right ventricular cardiomyopathy
DMD	c.6613_6614del (p.Arg2205fs)	Microsatellite	Frameshift variant | 5 prime UTR variant	2	** Pathogenic	Duchenne muscular dystrophy; Becker muscular dystrophy; Dilated cardiomyopathy 3B
DSC2	c.2186del (p.Pro729fs)	Deletion	Frameshift variant	1	** Pathogenic	Arrhythmogenic right ventricular dysplasia 11
DSG2	c.1038_1040del (p.Lys346del)	Microsatellite	Inframe deletion - Analytic detection difficult	3	** Pathogenic	Arrhythmogenic right ventricular dysplasia 10; Dilated cardiomyopathy 1BB
DSP	c.2131_2132del	Microsatellite	Genic downstream transcript variant	2	** Pathogenic	Arrhythmogenic cardiomyopathy with wooly hair and keratoderma; Arrhythmogenic right ventricular dysplasia 8; Primary dilated cardiomyopathy
	c.5671_*835+957del4738ins AGAACAGTCTT	Large Indel	Genic downstream transcript variant	4,738	** Pathogenic/Likely pathogenic	Arrhythmogenic cardiomyopathy with wooly hair and keratoderm; Arrhythmogenic right ventricular dysplasia 8
EMD	c.153dup (p.Ser52fs)	Duplication	Frameshift variant	1	** Pathogenic	X-linked Emery-Dreifuss muscular dystrophy

MUTATIONS PRESENT IN THE SERASEQ® INHERITED CARDIOVASCULAR DNA MIX (continued)

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	Variant size (bp)	Classification	Conditions
FKTN	c.454dup (p.Ser152fs)	Duplication	Frameshift variant | Non-coding transcript variant	1	** Pathogenic	Walker-Warburg congenital muscular dystroph; Dilated cardiomyopathy 1X; Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4
FLNC	c.4275del (p.Arg1426fs)	Deletion	Frameshift variant	1	** Pathogenic	Dilated Cardiomyopathy, Dominant Distal myopathy with posterior leg and anterior hand involvement; Hypertrophic cardiomyopathy 26; Myofibrillar myopathy 5
GAA	c.525del (p.Glu176fs)	Deletion	Frameshift variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Glycogen storage disease, type II; Dilated cardiomyopathy
GATA6	c.701del (p.Pro234fs)	Deletion	Frameshift variant	1	* Likely pathogenic	Primary dilated cardiomyopathy
GLA	c.644A>G (p.Asn215Ser)	SNV	Missense intronic variant| Non-coding transcript variant	1	** Pathogenic	Hypertrophic cardiomyopathy; Fabry disease
HCN4	c.1454C>T (p.Ala485Val)	SNV	Missense variant	1	** Pathogenic/ Likely pathogenic	Brugada syndrome 8; Left ventricular noncompaction cardiomyopathy
JPH2	c.482C>A (p.Thr161Lys)	SNV	Missense variant	1	** Pathogenic/ Likely pathogenic	Primary familial hypertrophic cardiomyopathy
JUP	c.2038_2039del (p.Trp680fs)	Deletion	Frameshift variant	2	** Pathogenic	Arrhythmogenic right ventricular dysplasia 1; Naxos disease
LAMP2	c.877C>T (p.Arg293Ter)	SNV	Nonsense	1	Pathogenic	Hypertrophic cardiomyopathy; Danon disease
LDB3	c.1723T>A (p.Phe575Ile)	SNV	Missense variant	1	** Pathogenic	Dilated cardiomyopathy 1C
LMNA	c.568C>T (p.Arg190Trp)	SNV	Missense variant	1	** Pathogenic	Primary dilated cardiomyopathy; LMNA-related disorder; Charcot-Marie-Tooth disease type 2
MYBPC3	c.2373dup	Duplication	Frameshift variant. Major contributor to disease as per Get-RM. Seen in >50 probands.	1	**Pathogenic.	Hypertrophic cardiomyopathy
MYBPC3	c.1224-52G>A	SNV	Intronic variant. Seen in >20 probands. Not difficult technically but often outside the region labs call variants in.	1	** Pathogenic/Likely pathogenic.	Hypertrophic cardiomyopathy; Left ventricular noncompaction 10
MYH6	c.1410+1G>A	SNV	Splice donor variant	1	** Pathogenic	Hypertrophic cardiomyopathy 14
MYH7	c.2513C>T (p.Pro838Leu)	SNV	Missense variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Restrictive cardiomyopathy
	c.2167C>T (p.Arg723Cys)	SNV	Missense variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Hypertrophic cardiomyopathy
	c.2711G>A (p.Arg904His)	SNV	Missense variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Dilated cardiomyopathy
MYL2	c.173G>A (p.Arg58Gln)	SNV	Missense variant	1	** Pathogenic	Hypertrophic cardiomyopathy 10

MUTATIONS PRESENT IN THE SERASEQ® INHERITED CARDIOVASCULAR DNA MIX (continued)

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	Variant size (bp)	Classification	Conditions
MYL3	c.445A>G (p.Met149Val)	SNV	Missense variant	1	** Pathogenic/Likely pathogenic	Hypertrophic cardiomyopathy 8
MYPN	c.3127del (p.Ser1043fs)	Deletion	Frameshift variant | Non-coding transcript variant	1	** Pathogenic/Likely pathogenic	Dilated cardiomyopathy 1KK
NEXN	c.1536dup (p.Met513fs)	Duplication	Frameshift variant	1	* Pathogenic	Dilated cardiomyopathy 1CC; Hypertrophic cardiomyopathy 21
NKX2-5	c.711C>A (p.Tyr237Ter)	SNV	3 prime UTR variant | Nonsense	1	** Pathogenic	Primary dilated cardiomyopathy; Ventricular fibrillation; Noncompaction cardiomyopathy; Atrial septal defect 7
PKP2	c.968_971delinsGCT (p.Gln323fs)	Indel	Frameshift variant	4	** Pathogenic/Likely pathogenic	Arrhythmogenic right ventricular dysplasia 9
PKP2	c.1217_1379-790inv	Inversion	Splice donor - Impacts exons 5-6. Analytic detection difficult	8,933	* Pathogenic	Arrhythmogenic right ventricular dysplasia 9
PLN	c.1020+6570_1020+6572del (p.Arg14del)	Microsatellite	Intronic variant	3	* Pathogenic - Major contributor to disease - Founder variant Europe/North America	Dilated cardiomyopathy 1P; Arrhythmogenic right ventricular dysplasia 9; Hypertrophic cardiomyopathy 18; Sudden Infant Deaths Syndrome
PPA2	c.514G>A (p.Glu172Lys)	SNV	Missense Intronic variant	1	** Pathogenic/Likely pathogenic	Inborn genetic diseases; Sudden cardiac failure, infantile
PRDM16	c.1573dup (p.Arg525fs)	Duplication	Frameshift variant	1	** Pathogenic	Left ventricular noncompaction cardiomyopathy 8
PRKAG2	c.905G>A (p.Arg302Gln)	SNV	Missense variant	1	** Pathogenic	Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome; Hypertrophic cardiomyopathy 6; Lethal congenital glycogen storage disease of heart
PTPN11	c.1403C>T (p.Thr468Met)	SNV	Missense variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Noonan syndrome with multiple lentigines; LEOPARD syndrome 1; RASopathy; Hypertrophic Cardiomyopathy
RAF1	c.1837C>G (p.Leu613Val)	SNV	Missense variant | Non-coding transcript variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Noonan syndrome 5 with multiple lentigines; LEOPARD syndrome 2; RASopathy; Hypertrophic cardiomyopathy 1
RBM20	c.1900C>T (p.Arg634Trp)	SNV	Missense variant	1	** Pathogenic	Dilated cardiomyopathy 1DD
RYR2	c.1069G>A (p.Gly357Ser)	SNV	Missense variant	1	** Pathogenic	Cardiomyopathy; Catecholaminergic polymorphic ventricular tachycardia 1
SCN5A	c.2550_2551dup (p.Phe851fs)	Microsatellite	Frameshift variant	2	** Pathogenic	Dilated cardiomyopathy 1E; Cardiac arrhythmia; Long QT syndrome 3|Atrial fibrillation, familial, 10; Sick sinus syndrome 1; Brugada syndrome 1; Progressive familial heart block, type 1A; SUDDEN INFANT DEATH SYNDROME; Ventricular fibrillation, paroxysmal familial, type 1
TMEM43	c.1073C>T (p.Ser358Leu)	SNV	Missense variant	1	** Pathogenic	Primary dilated cardiomyopathy; Arrhythmogenic right ventricular cardiomyopathy, type 5
TNNC1	c.23C>T (p.Ala8Val)	SNV	Missense variant	1	** Pathogenic/Likely pathogenic	Dilated cardiomyopathy 1Z; Hypertrophic cardiomyopathy 13
TNNI3	c.485G>A (p.Arg162Gln)	SNV	Missense variant	1	** Pathogenic -Major contributor to disease - Seen in >20 proband.	Hypertrophic cardiomyopathy 7; Dilated cardiomyopathy 1FF; Familial Restrictive Cardiomyopathy 1; TNNI3-related disorder

MUTATIONS PRESENT IN THE SERASEQ® INHERITED CARDIOVASCULAR DNA MIX (continued)

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	Variant size (bp)	Classification	Conditions
TNNI3K	c.2302G>A (p.Glu768Lys)	SNV	Missense Intronic variant	1	** Pathogenic/Likely pathogenic	Atrial conduction disease; Dilated cardiomyopathy
TNNT2	c.508GAG[3] (p.Glu173del)	Microsatellite	Inframe deletion in highly repetitive region - Analytic detection difficult	3	** Pathogenic/Likely pathogenic	Dilated cardiomyopathy 1D; Hypertrophic cardiomyopathy 2
TPM1	c.688G>A (p.Asp230Asn)	SNV	Missense variant	1	** Pathogenic	Hypertrophic cardiomyopathy 3; Dilated cardiomyopathy 1Y
TTR	c.148G>A (p.Val50Met)	SNV	Missense variant	1	** Pathogenic	TTR-related disorder; Familial amyloid neuropathy; Charcot-Marie-Tooth disease; Cardiomyopathy; Carpal tunnel syndrome 1;Hyperthyroxinemia, dystransthyretinemic; Familial amyloid neuropathy
TTN	c.80365_80432del (p.Leu26789fs)	Large Deletion	Frameshift variant	68	** Pathogenic/Likely pathogenic	Cardiovascular phenotype
SHOC2	c.4A>G (p.Ser2Gly)	SNV	Missense variant	1	*** Pathogenic Reviewed by expert panel. FDA RECOGNIZED DATABASE	Noonan syndrome-like disorder with loose anagen hair 1; RASopathy

NOTE: Above list does not include variants present in the GM24385 background

ABOUT US

SeraCare offers a comprehensive portfolio of reference materials for oncology, reproductive health, inherited diseases, and pharmacogenomics, designed and manufactured to meet the precision demanded by NGS assays. The portfolio includes high quality ground-truth RNA, ctDNA and genomic DNA-based reference materials that are NGS platform agnostic for tumor profiling, immuno-oncology, liquid biopsy, NIPT and germline cancer assay workflows. **For more information visit seracare.com**

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