# SureSelect CD Heidelberg CardioGenetics Panel

Innovation powered by you



Jan Haas, Ph.D.

Head of Cardiogenetics,
Institute of Cardiomyopathies Heidelberg,
University Hospital Heidelberg, Germany

"For more than 10 years, we have a strong interaction with Agilent, which helped us to continuously develop our SureSelect target enrichment-based cardiomyopathy panel.

Workflows for both low- and highthroughput custom kits result in comprehensive, reproducible coverage, enabling precise detection of variants."

# Targeted Next-Generation Sequencing to Identify Cardiomyopathy Mutations

Agilent Community Designs for next-generation sequencing (NGS) are targeted sequencing panels established in collaboration with subject matter experts in different research fields. These NGS designs are available as made-to-order panels that provide you with robust and cost-effective sequencing results that focus only on your genes of interest.

Benjamin Meder, Ph.D. and Jan Haas, Ph.D., from the Institute of Cardiomyopathies (ICH), Heidelberg, Germany, are experts in the application of NGS technologies for clinical research of cardiomyopathy. They developed the SureSelect CD Heidelberg CardioGenetics panel now offered by Agilent.

Familial clusters among cardiomyopathy patients are not uncommon because genetic factors play a fundamental role in the etiology of the diseases.

Genetic testing can help clinical researchers find the causative gene in 25 to 60% of individuals tested for cardiomyopathy.¹ By detecting mutations in genes associated with cardiomyopathies, researchers can make strides toward improved patient therapy, aimed at increasing the quality of life, and identify any family members who may also carry the mutation. A well-curated, targeted panel of cardiomyopathy gene candidates is crucial when investigating the cause of cardiomyopathies.

#### **Advantages of the Design**

- Features 1.246 Mb designed to genome build Hg38, covering all coding exons and UTRs
- Includes 117 clinically relevant genes, curated by ICH experts
- Contains 25 bp of flanking sequence into each intron to identify variants that may influence splicing
- Includes 24 Pengelly SNPs for sample tracking<sup>2</sup>



### **Targeted NGS Performance at ICH**

The ICH has shown that with improved accuracy and efficiency, targeted NGS is the technique for selected high-throughput gene sequencing in their laboratory. Consistent performance in a large clinical research cohort demonstrated that targeted NGS can be well-suited to routine cardiac diagnostics, substantiating the ongoing paradigm shift from low- to high-throughput genomics in medicine.<sup>3</sup>

 Table 1. Complete 117 gene list included in the SureSelect CD Heidelberg CardioGenetics panel

Genes	Genes	Genes	Genes	Genes	Genes
ABCC9	CRYAB	KCND3	MLYCD	PTPN11	TAZ
ACTC1	CSRP3	KCNE1	MMACHC	RAF1	TCAP
ACTN2	CTNNA3	KCNE2	MYBPC3	RBM20	TECRL
ADRB3	DES	KCNE3	MYH6	RBM24	TGFB3
AKAP9	DMD	KCNE5	MYH7	RPS6KA3	TMEM43
ALPK3	DNAJC19	KCNH2	MYL1	RYR2	TMPO
ANK2	DSC2	KCNJ2	MYL2	SCN10A	TNNC1
ARFGEF2	DSG2	KCNJ5	MYL3	SCN1B	TNNI3
ATP2A2	DSP	KCNJ8	MYPN	SCN2B	TNNT2
BAG3	DTNA	KCNQ1	NEBL	SCN3B	TPM1
CACNA1C	EMD	KCNQ2	NEXN	SCN4B	TRDN
CACNA2D1	EYA4	KRAS	NKX2-5	SCN5A	TRPM4
CACNB2	FLNC	LAMA2	NNT	SGCB	TTN
CALM1	GLA	LAMA4	NSD1	SGCD	TTR
CALM2	GPD1L	LAMP2	PGM1	SH0C2	TUBB
CALM3	HCN4	LDB3	PKP2	SLMAP	VCL
CASQ2	HRAS	LIMS1	PLEKHM2	SNTA1	YWHAE
CAV3	ILK	LIMS2	PLN	SOS1	
COA5	JPH2	LMNA	PRDM16	SYNE1	
CPT2	JUP	MIB1	PRKAG2	SYNE2	

**Table 2.** Core genes and database representation within the SureSelect CD Heidelberg CardioGenetics panel

	DCM	HCM	ARVC
MYH7	•	•	•
TTN	•	•	•
LMNA	•	•	•
SCN5A	•	•	•
DES	•	•	•
TNNT2	•	•	•
TNNC1	•	•	•
BAG3	•	•	•
FLNC	•	•	•
RBM20	•	•	•
DSP	•	•	•
DSG2	•	•	•
LDB3	•	•	•
МҮВРС3	•	•	•
MYL2	•	•	•
CSRP3	•	•	•
ACTC1	•	•	•
TNNI3	•	•	•
TPM1	•	•	•
NEXN	•	•	•
VCL	•	•	•
PKP2	•	•	•
MYL3	•	•	•
JUP	•	•	•
CTNNA3	•	•	•
TGFB3	•	•	•
PLN	•	•	•

**Table 3.** Ordering information for the SureSelect Community Design Heidelberg CardioGenetics panel. Note: part numbers cover the capture probe libraries only. Library prep and target enrichment kits must be purchased separately.

Product	Part Number
SureSelect CD Heidelberg CardioGen 16	5282-0022
SureSelect CD Heidelberg CardioGen 96	5282-0023
SureSelect CD Heidelberg CardioGen 96A	5282-0024

Gene-Disease Validity **DCM** (Clingen) Gene-Disease Validity **HCM** (Clingen) Gene-Disease Validity **ARVC** (Clingen)

Definitive	
Strong	
Limited	
Moderate	
Disputed	
No Known Disease Relationship/#NA	

## References

- Cardiomyopathy.org. https://www.cardiomyopathy.org/sites/default/ files/2021-04/genetic-testing-in-cardiomyopathy-october-2017.pdf (accessed 2022-09-19).
- 2. Pengelly, R. J.; Gibson, J.; Andreoletti, G.; Collins, A.; Mattocks, C. J.; Ennis, S. A SNP Profiling Panel for Sample Tracking in Whole-Exome Sequencing Studies. Genome Med. **2013**, 5 (9), 89. https://doi.org/10.1186/gm492
- 3. Haas, J.; Frese, K.S.; Peil, B.; et al. Atlas of the Clinical Genetics of Human Dilated Cardiomyopathy. European Heart Journal. **2015**, 36 (18), 1123-1135. https://doi.org/10.1093/eurheartj/ehu301

#### www.agilent.com

 $\label{eq:Agilent} \mbox{ Agilent has not performed verification and validation on these panels.}$ 

For Research Use Only. Not for use in diagnostic procedures.

PR7001-0156

This information is subject to change without notice.

