Data Sheet

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SureMASTR ADH

A ready-to-use research assay that offers robust performance with minimum hands-on time for the identification of all SNVs and CNVs associated with Autosomal Dominant Hypercholesterolemia. The assay is compatible with all current Next-Generation Sequencing (NGS) systems, providing the flexibility to choose your preferred method.

Research application

- Identification of all SNVs and CNVs in *LDLR, PCSK9, APOE,* part of exon 26 (c.10200 to c.11100) of *APOB*
- Identification of 12 common LDL-C raising SNPs

Table 1. Assay characteristics.

Genes analyzed	*LDLR, PCSK9, APOE,* part of exon 26 (c.10200 to c.11100) of *APOB* (SNVs + CNVs), 12 LDL-C raising SNPs
Genomic region analyzed	16.6 kb
Number of amplicons	76 including 17 control amplicons
Amplicon length	300-430 bp
Number of plexes	5
Designed to be compatible with	Illumina MiSeq

Table 2. Plexes 1-4.

Gene	Coding sequences	Promoter region	5'UTR regions
LDLR	All exons	chr19:11 199 687-11 200 037 (350 bp)	c.-187 to c.-1 (NM_000527.4) = chr19:11 200 038-11 200 224
PCSK9	All exons	chr1:55 504 598-55 505 148 (550 bp)	c.-362 to c.-1 (NM_174936.3) = chr1:55 505 149-55 505 510
APOE	All exons	chr19:45 408 688-45 409 038 (350 bp)	c.-83 to c.-24 (NM_000041.2) = chr19:45 409 039-45 409 098 and c.-23 to c.-1(NM_000041.2) = chr19:45 409 859-45 409 881
APOB	Exon 26 (c.10200 to c.11100)		

Table 3. Plex 5: common LDL-C raising SNPs.

Gene	Reported SNP	Minor allele	Common allele
PCSK9	*rs2479409*	*G**	*A*
CELSR2	*rs629301*	*G*	*T**
APOB	*rs1367117*	*A**	*G*
ABCG8	*rs4299376*	*G**	*T*
SLC22A1	*rs1564348*	*C*	*T**
HFE	*rs1800562*	*A*	*G**
MYLIP	*rs3757354*	*T*	*C**
ST3GAL4	*rs11220462*	*A**	*G*
NYNRIN	*rs8017377*	*A**	*G*
LDLR	*rs6511720*	*T*	*G**
APOE	*rs429358*	*C*	*T*
APOE	*rs7412*	*T*	*C*

Table 4. Performance.

Uniformity of amplification (≥ 0.2x mean coverage)	99 %
On target read count	>96 %
DNA input	20 ng per plex reaction

Table 5. SNV and CNV variant calling: advised maximum number of samples per run.

Sequencing system	Illumina MiSeq Reagent kit		
Flow cell	**Nano v2**	**Kit v2**	**Kit v3**
SNV variant calling (20 reads/allele)	53	814	1492
SNV and CNV variant calling (200 reads/amplicon)	11	166	305

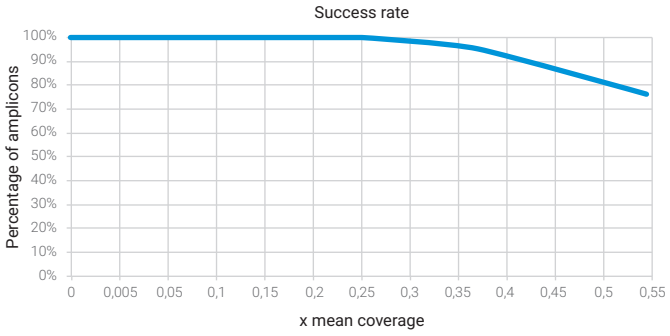
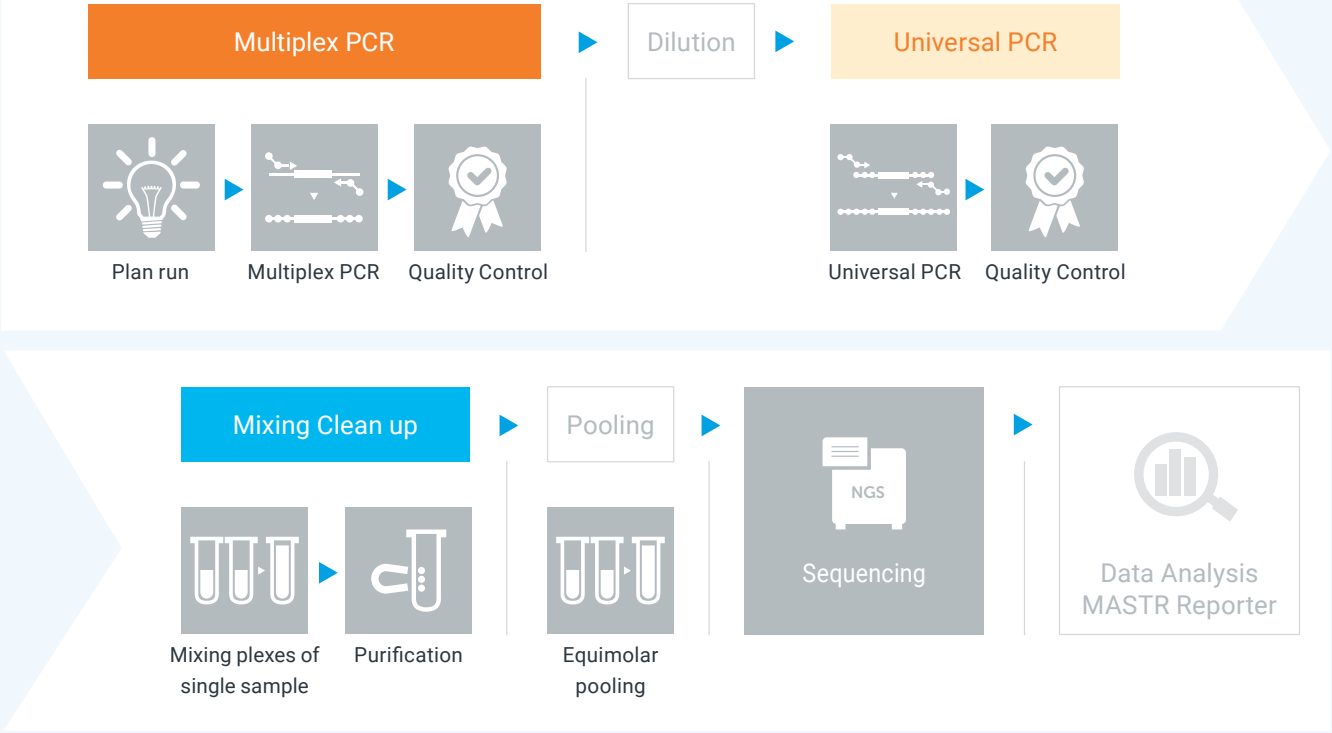


Figure 1. Graph representing the read counts for all 76 SureMASTR ADH amplicons, showing their uniform representation. To allow comparison between samples, the read counts were normalized.

Workflow



Publications

- Use of low-density lipoprotein cholesterol gene score to distinguish patients with polygenic and monogenic familial hypercholesterolaemia: a case-control study. Talmud PJ et al. The Lancet (2013) 381,1293-1301. DOI: http://dx.doi.org/10.1016/S0140-6736(12)62127-8
- Universal Screening for Familial Hypercholesterolemia in Children. Gasper Klancar et al. (2015) http://dx.doi.org/10.1016/j.jacc.2015.07.017
- Identification and molecular characterisation of Lausanne Institutional Biobank participants with familial hypercholesterolaemia – a proof-of-concept study. Fabienne Maurer et al. (2016) Swiss Med Wkly. 2016;146:w14326 doi:10.4414/smw.2016.14326
- Familial hypercholesterolemia: experience from France. Jean-Pierre Babès, Sophie Béliard and Alain Carrié. Curr Opin Lipidol (2018). DOI: 10.1097/MOL.0000000000000496

Ordering information

Cat. No.	Product Name	Samples
MR-0141.024	SureMASTR ADH	24

MID (Molecular Identifiers) kits are necessary to complete the workflow

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Rest of World:
https://www.agilent.com/en/contact-us/page

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