CytoSure

A Sysmex Group Company

Comprehensive FH NGS Panel

Features

Detection of CNVs as well as SNVs with a single assay

- Enabled by the exon resolution of the targeted genes

Pre-optimised content that meets your technical requirements

- No more laborious in-house optimisation, decreasing assay development time

Bespoke panel content

- Sequence only what's relevant for your research

Interpret, OGT's complimentary analysis software

- Designed to give unparalleled CNV and SNV calling

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Introduction

Familial Hypercholesterolaemia (FH) is a genetic condition which results in a high cholesterol level and subsequently leads to a higher risk of early heart disease. It affects approximately 1 in 250 people with around 34 million cases worldwide[1].

OGT is offering an optimised NGS panel which has selected the most relevant genes and SNPs implicated in FH, for your research needs. Together with the complimentary Interpret analytical software, the CytoSure® Comprehensive FH NGS platform provides the optimal solution for FH research.

Genes at exon-level resolution

ABCG5	*ABCG8*	*APOB*	*APOE*	*LDLR*	*LDLRAP1*	*LIPA*	*PCSK9*

SNPs associated with FH

- rs10401969 SUGP1 Intron Variant
- rs10455872 LPA Intron Variant
- rs11206510 Ch1
- rs11220462 ST3GAL4 intron variant
- rs121909548 SERPINC1 : Missense Variant
- rs12740374 CELSR2 UTR variant
- rs1346268 GATM : Intron Variant
- rs1367117 APOB : Missense Variant
- rs1501908
- rs1564348 SLC22A1 : Intron Variant
- rs1719247
- rs17244841 HMGCR : Intron Variant
- rs1799768 SERPINE1 : 2KB Upstream Variant
- rs1799963 F2 : 3 Prime UTR Variant
- rs1800562 HFE : Missense Variant, LOC108783645 : 2KB Upstream Variant
- rs1801131 MTHFR : Missense Variant
- rs1801133 MTHFR : Missense Variant
- rs2032582 ABCB1 : Missense Variant
- rs2231142 ABCG2 : Missense Variant
- rs2306283 SLCO1B1 : Missense Variant
- rs2479409 PCSK9 : 2KB Upstream Variant
- rs2650000
- rs3757354 MYLIP : 2KB Upstream Variant
- rs3798220 LPA : Missense Variant
- rs3846663 HMGCR : Intron Variant
- rs4149015 SLCO1B1 : 2KB Upstream Variant
- rs4149056 SLCO1B1 : Missense Variant
- rs428785 ADAMTS1 : Missense Variant
- rs429358 APOE : Missense Variant
- rs4299376 ABCG8 : Intron Variant
- rs4693075 COQ2 : Intron Variant
- rs515135
- rs6025 F5 : Missense Variant
- rs6102059 LOC102724968 : Intron Variant
- rs629301 CELSR2 : 3 Prime UTR Variant
- rs6511720 LDLR : Intron Variant, LDLR-AS1 : 2KB Upstream Variant
- rs6544713 ABCG8 : Intron Variant
- rs7412 APOE : Missense Variant
- rs8017377 NYNRIN : Missense Variant

Some genes/SNPs may not be available in your region - contact OGT for more details.

CNV and SNV detection from a single assay

The hybridisation enrichment methodology, combined with our bait design expertise, allows generation of panels with outstanding completeness and coverage uniformity. Together, this allows the areas of CNV to be easily identified within each sample using our proprietary algorithm — delivering an increased understanding of the sample without an increase in cost or time (Figure 1).

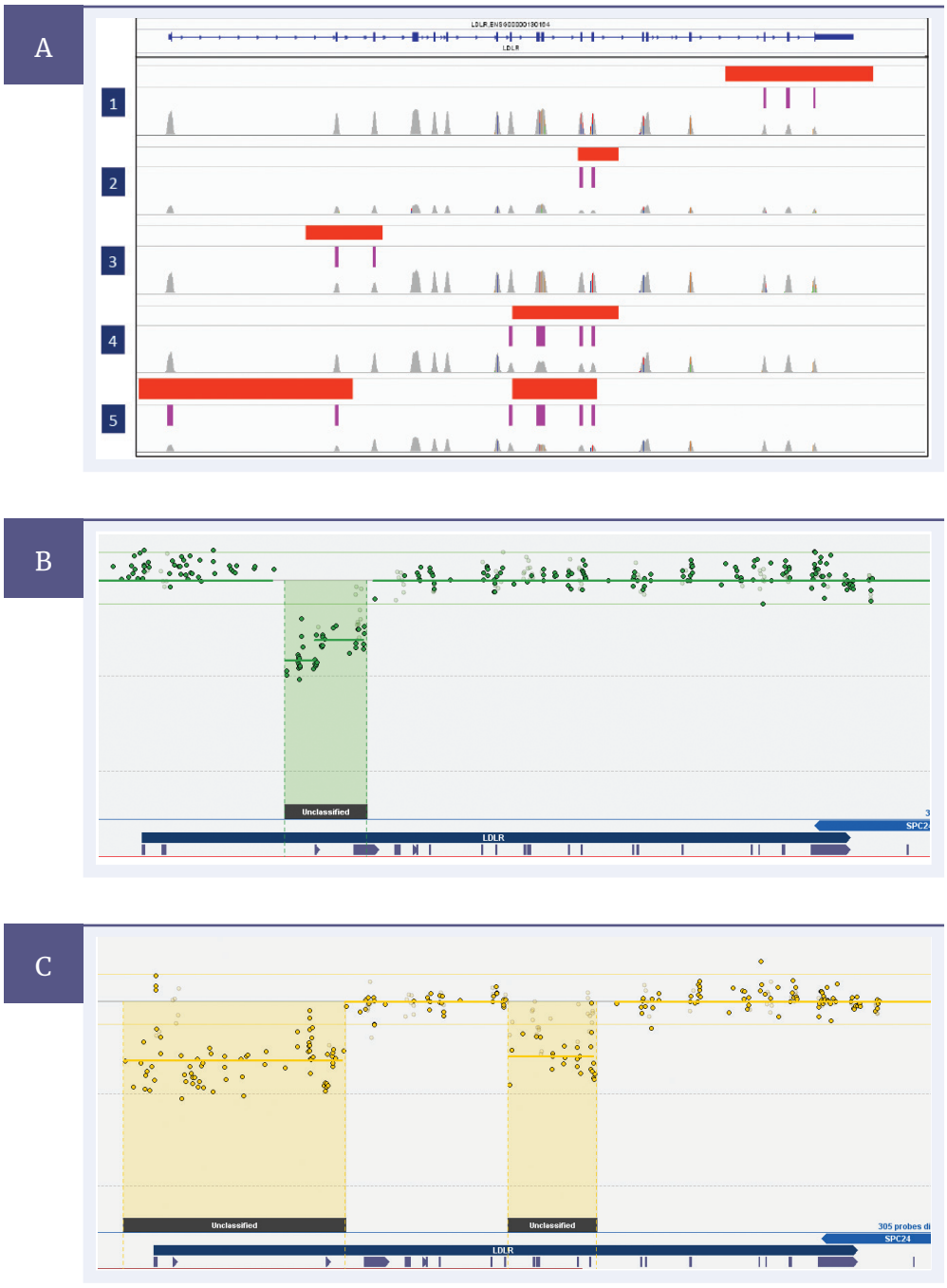

Figure 1: CNV in *LDLR* gene shown using IGV from the Broad Institute A: Red bars indicate areas of CNV (data from aCGH), purple bars represent deleted exons (data from NGS): 5 samples are shown, each with at least one area of CNV. There is complete concordance between the aCGH and NGS data. Note the evenness of the NGS coverage (even peak height) across each exon, allowing the areas of CNV to be easily identified. The data from the custom CytoSure aCGH array, confirms the deletions in *LDLR*. B: A 2 exon deletion and C: deletion of 2 exons and 4 exons, corresponding to samples 3 and 5 in A respectively.

The Interpret software has been designed to easily visualise CNVs and SNVs, with an intuitive interface to switch between different sets of results. Interpret also has simple to use protocols and filtering options, to easily target the results of interest.

The CytoSure Comprehensive FH NGS panel has the ability to detect CNVs in whole genes, at exon resolution (Figures 2-3) and can target select SNPs that have been implicated in FH (Figure 4). The CytoSure Comprehensive FH panel can also detect SNVs and Indels within genes, as demonstrated by Figure 5.

Figure 2: Double deletion on the *LDLR* gene, as visualised by Interpret software.

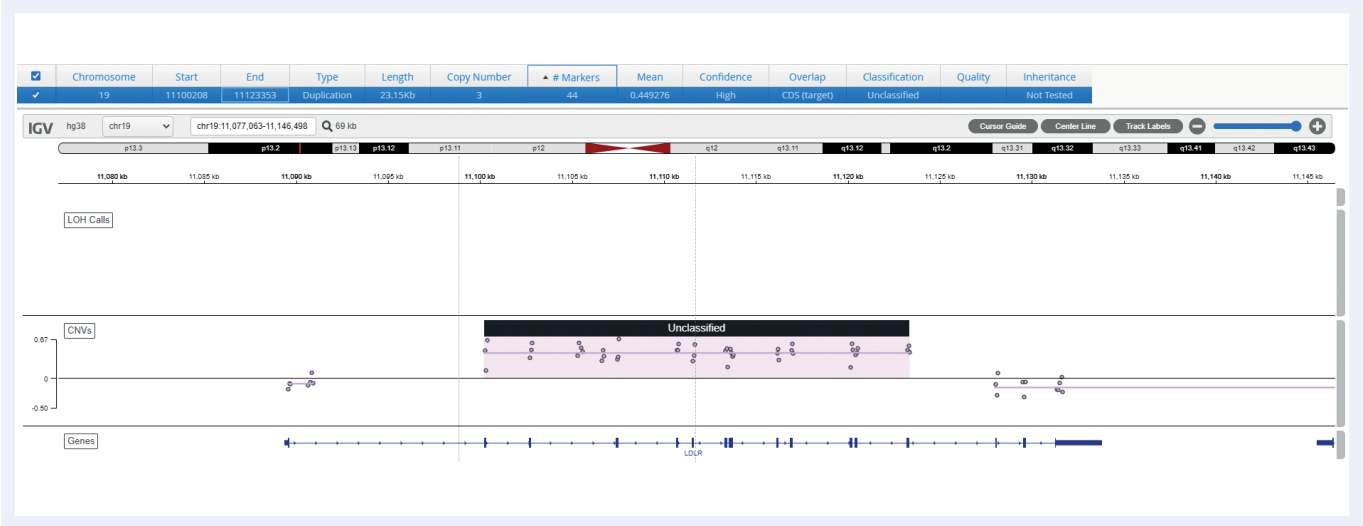

Figure 3: Duplication on the *LDLR* gene, as visualised by Interpret software.

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Figure 4: Deletion on the *SLOCO1B1* gene, as visualised by Interpret software.

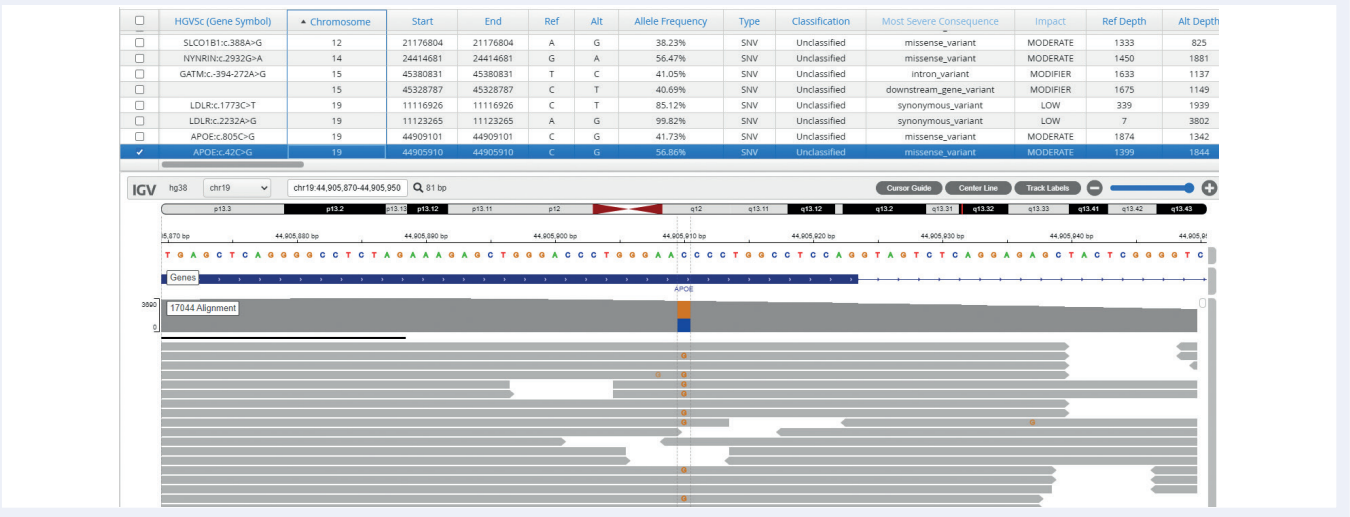

Figure 5: Missense variant on the *APOB* gene, as visualised by Interpret software.

Product Specifications	
Targeted Genes	8
Targeted SNPs	39
CNV Resolution	Exon Level
SNVs and Indels	Targeted Genes (8)
Mean Target Coverage	> 300x
Recommended DNA Input	> 500 ng high quality DNA
Panel Size	50Kb
Samples per MiSeq® v2 run	16

Ordering information

UK +44 (0) 1865 856800

US +1 914 467 5285

contact@ogt.com

ogt.com

Product	Contents	Cat. No.
CytoSure Comprehensive FH Panel (16 reactions)	Enrichment baits; Interpret Software	601004-16
SureSeq NGS Library Preparation Complete Solution (16)	Bundle of 1x SureSeq NGS Library Preparation Kit (16), containing adaptors, PCR primers and enzymes, 1x SureSeq NGS Index Kit – Collection A, 1x SureSeq NGS Hyb & Wash Kit (16), 1x Dynabeads M270 Streptavidin (2ml) and 1x AMPure XP beads (10ml). Sufficient for 16 samples	500084
SureSeq NGS Library Preparation Complete Solution (48)	Bundle of 1x SureSeq NGS Library Preparation Kit (48), containing adaptors, PCR primers and enzymes, 1x SureSeq NGS Index Kit – Collection B, 3x SureSeq NGS Hyb & Wash Kit (16), 3x Dynabeads M270 Streptavidin (2ml) and 3x AMPure XP beads (10ml). Sufficient for 48 samples	500085

Talk to us about your requirements, email contact@ogt.com or visit ogt.com/FH.

References

1. Goldberg AC, and Gidding, SS. Knowing the Prevalence of Familial Hypercholesterolemia Matters. Circulation, 2016; 133 (11).

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What binds us, makes us.

Oxford Gene Technology Ltd., Begbroke Science Park, Woodstock Road, Begbroke, Oxfordshire, OX5 1PF, UK