Interpret

A Sysmex Group Company

NGS Analysis Software

Features

Detection of a wide range of aberrations

- Reliably call variants ranging from low-frequency SNVs and indels to large structural deletions including CNVs and translocations

Extensive customisation options

- Easily customise variant and batch reports and database links to meet the exact needs of your laboratory

Comprehensive range of filtering options

- Standardise your analysis workflow and overlay bespoke variant filtering to meet your analytical criteria

Security and control

- Log and track user activity and standardise analysis protocols through multiple access permission levels

Powerful and complimentary with OGT's NGS panels

- Optimised for use with SureSeq and CytoSure NGS panels to detect all aberrations covered by your panel

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Introduction

Interpret is OGT's powerful and easy-to-use next-generation sequencing analysis solution, facilitating analysis and visualisation of a wide range of variants and structural aberrations. Coupled with a comprehensive and powerful filtering framework, the software delivers accurate calling of SNVs and indels, as well as structural aberrations, including ITDs, PTDs, CNVs, LOH and translocations. Interpret is designed to work seamlessly with all CytoSure® and SureSeq™ NGS panels and offers flexible accessibility for data analysis; whether through a stand-alone computer*, laboratory server or another web-enabled device. With a wide range of customisation options and links to various mutation databases, Interpret provides effortless translation of all your NGS data into meaningful results.

Accurate detection of a wide range of aberrations

Used in conjunction with CytoSure and SureSeq NGS panels, Interpret facilitates the analysis and visualisation of a wide range of mutation types and structural variants. Complementing the expert panel design and hybridisation-based approach of our NGS panels to deliver unparalleled coverage uniformity, Interpret is integral in facilitating the detection of low-frequency variants consistently and with confidence. Whether your input DNA is high-quality or formalincompromised, or your research focuses on oncology or rare diseases, Interpret delivers fast and accurate detection of all aberrations covered by your panel (Figures 1-9).

Figure 1: Following analysis, all variants are displayed in a table, below which is an Integrative Genomics Viewer (IGV)[i] window allowing a more detailed review of the data and additional verification. In this example a low-frequency *JAK2* V617F SNV has been selected and the user is able to view the aligned reads generated by the pipeline.

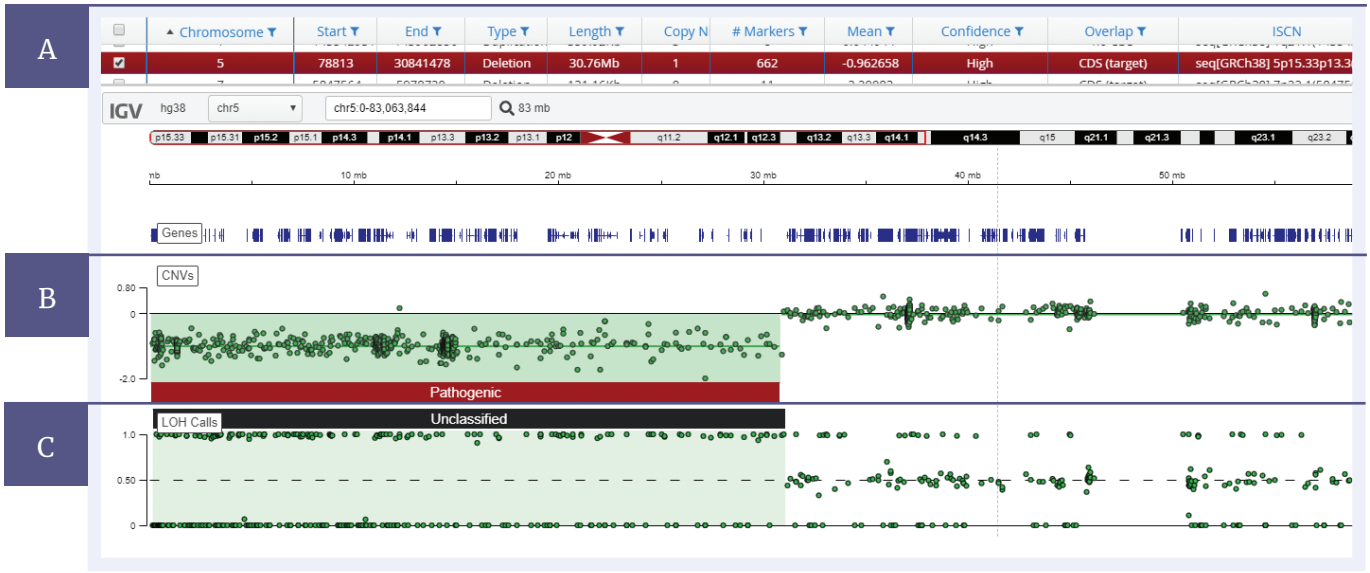

Figure 2: CytoSure Constitutional NGS panel analysed using Interpret, with a 30 Mb deletion on chromosome 5 and LOH across the deletion. A Details of the samples and mutations are displayed in a table format. B The CNV data is displayed in a log² ratio plot and C a b-allele plot shows the LOH present within the sample.

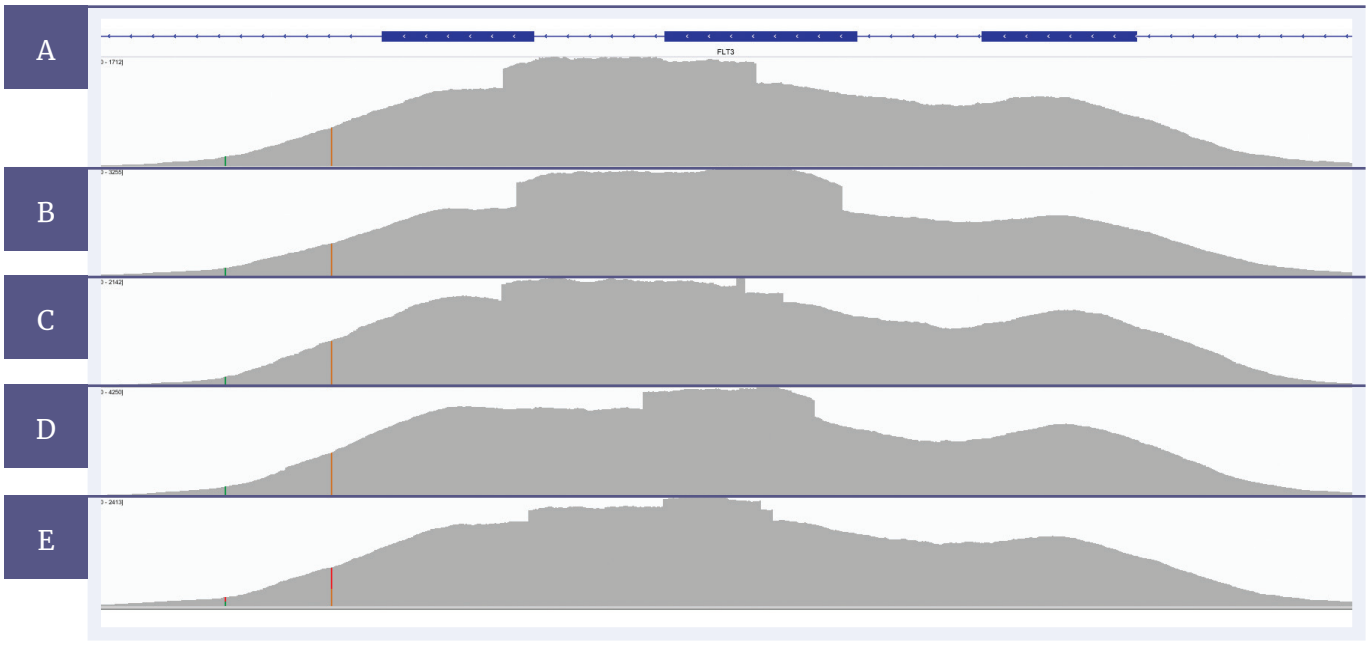

Figure 3: Detection of *FLT3*-ITDs of various sizes, including regions containing multiple ITDs. ITD sizes are A 174 bp, B 225 bp, C 195 bp with an additional 6 bp, D 120 bp and E 168 bp with an additional 69 bp. Note how Interpret can confidently identify even ITDs much longer than the sequencing read length of 150 bp.

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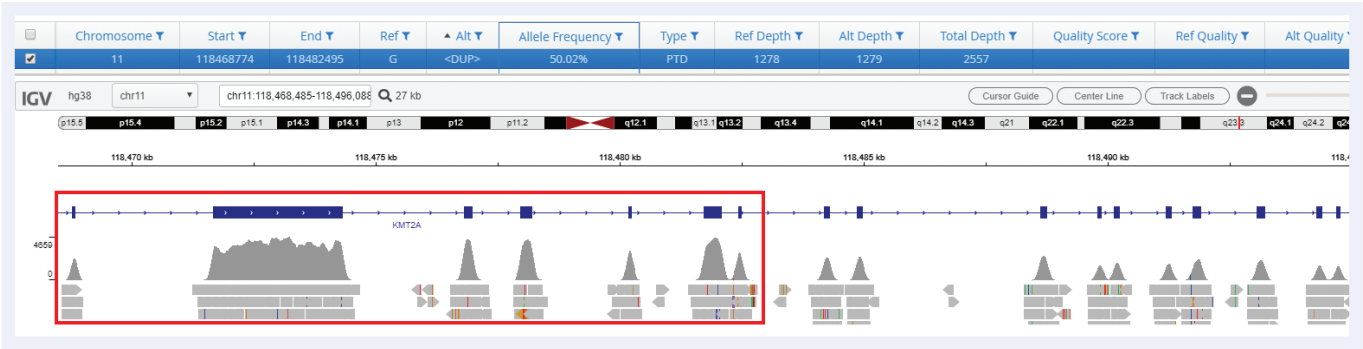

Figure 4: Detection of a *KMT2A*-PTD spanning exons 2-8. In conjunction with OGT's expert panel design, Interpret offers robust detection of all sizes of PTDs in *KMT2A*.

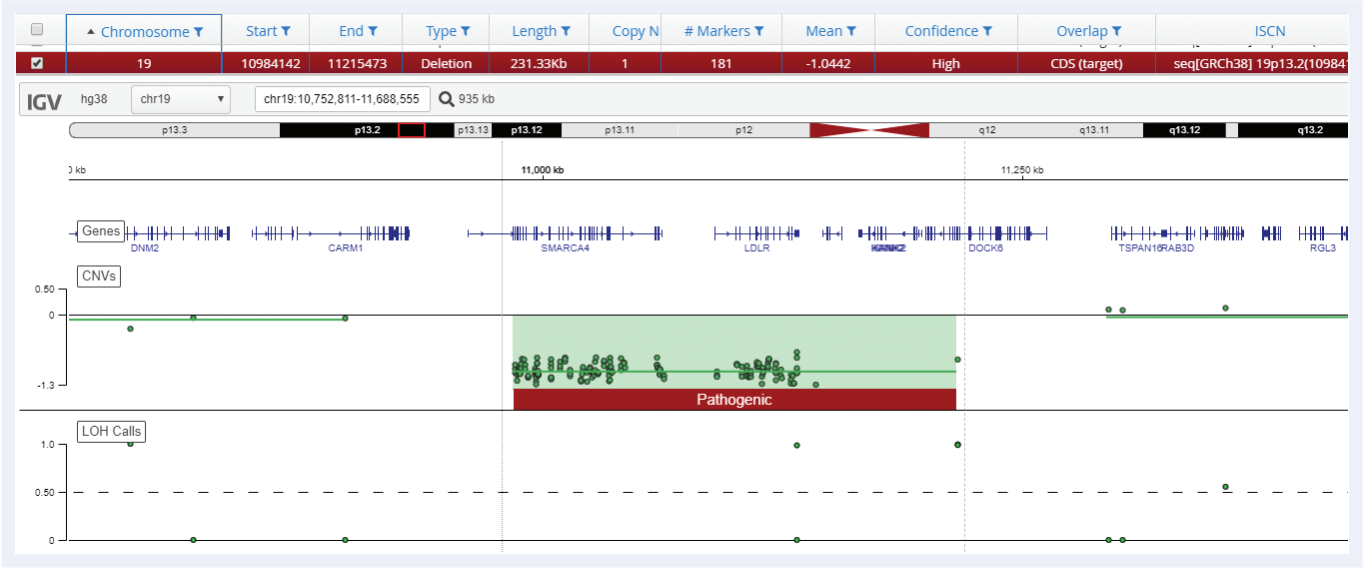

Figure 5: Small 231 kb deletion detected on chromosome 19 using Interpret with CytoSure Constitutional NGS.

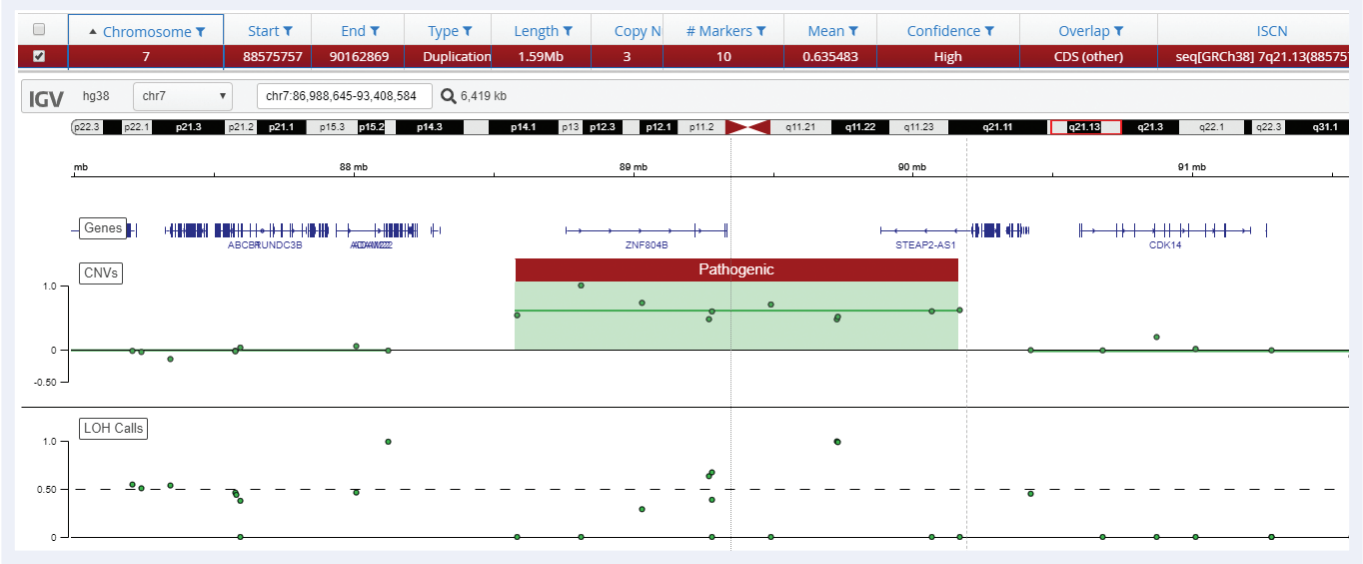

Figure 6: Interpret is able to call duplications with the same precision as microarrays, in this example a 1.59 Mb duplication on chromosome 7 is detected using the CytoSure Constitutional NGS Panel.

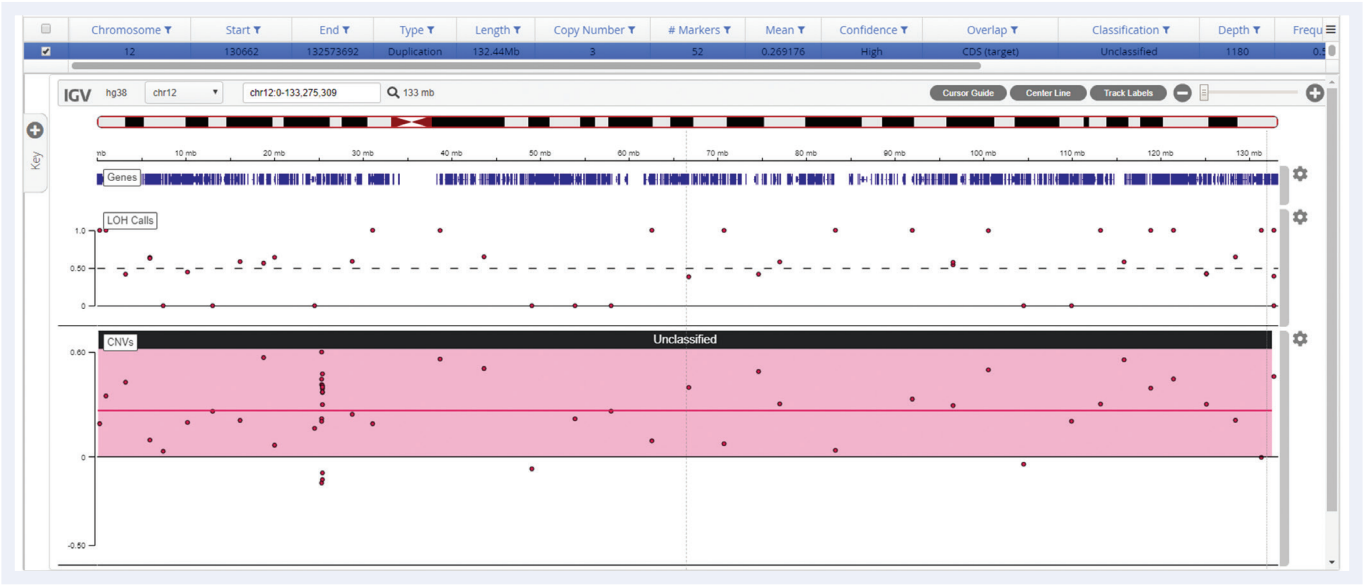

Figure 7: Detection of trisomy 12 using the SureSeq CLL + CNV Panel, showing a reliable gain call across the whole chromosome. Interpret enables CNV detection ranging from loss of a single exon to full chromosomal arms and trisomies.

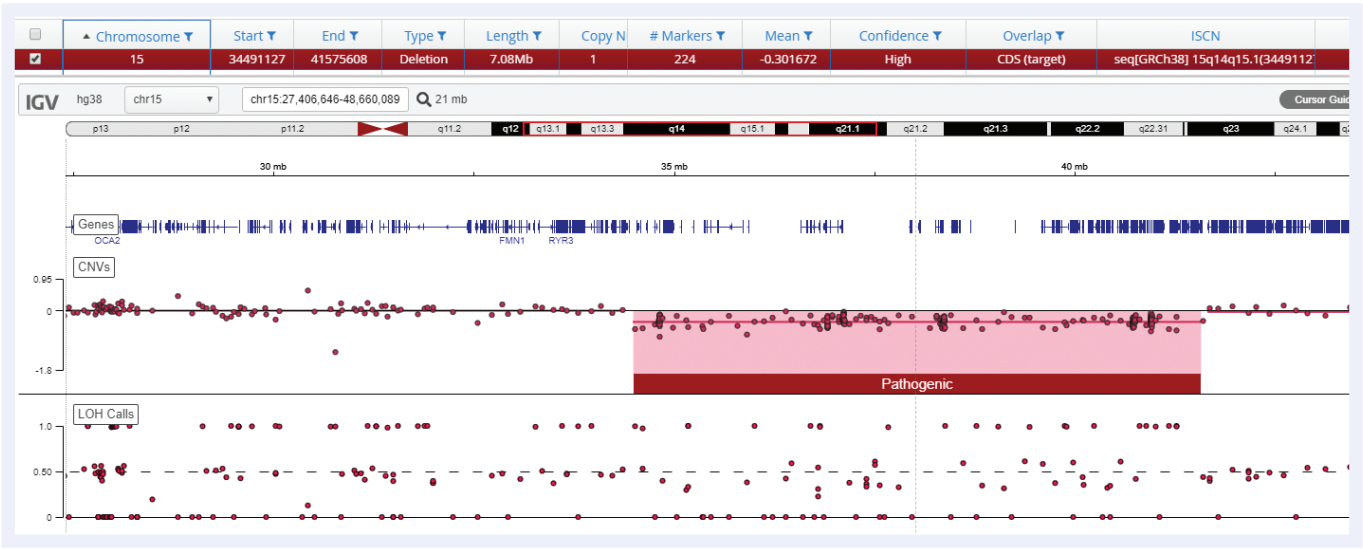

Figure 8: Detection of a 7 Mb deletion on a >50% mosaic sample using CytoSure Constitutional NGS.

Figure 9: *BCR-ABL* translocation. Split-reads covering both *BCR* (left panel) and *ABL1* (right panel) are detected, indicative of the *BCR-ABL* gene fusion. Interpret agnostically detects split-reads across the genome, enabling detection of both known and unknown translocation partners.

Extensive customisation options

OGT has extensive experience in providing individualised options through its established plug-in infrastructure. This enables the software to be tailored to your laboratory's specific requirements, whether it be report formats, user controls or variant annotation. Additional outputs from the pipeline include a range of publicly available data resources for annotation of variants detected, including:

ClinVar | **COSMIC** | **dbSNP** | **Ensembl** | **gnomAD** | **PolyPhen** | **SIFT**

Additional licence-based sources, such as HGMD, can be incorporated on provision of suitable credentials. In Interpret, samples are rapidly processed through customisable protocols and optimised settings in order to generate the variant lists. Your laboratory has the option to use the protocols provided with the software or develop your own through the intuitive user interface. Personal modifications are available for:

- Hardware settings
- Quality metrics
- Variant calling parameters
- Variant filtering parameters

Following analysis, results can be viewed in the user-friendly variant browser showing a tabular display of the calls and an IGV window, which can be maximised in a separate window for greater visibility.

Report generation is implemented through a templating system, prepared by OGT following discussion with your laboratory. Our report templates are highly customisable and designed to be modified to individual requirements. For example, one template could simply provide an overview of user activity in the sequencing analysis while another could provide detailed sample or batch analysis results (Figure 10).

Gene	Chr	Start	End	Alt	Ref	HGVSc (Gene Symbol)	Zygosity	Total Depth	Ref Depth	Alt Depth	Allele Frequency	Genotype	Type
ATR	3	142178144	142178144	C	T	BRCA1:c.1067A>G	Heterozygous	530	273	275	48.49%	0/1	snp
ATR	11	108183167	108183167	A	G	BRCA1:c.1067A>G	Heterozygous	546	0	546	100%	1/1	snp
BRCA2	13	32907420	32907421	GA	G	BRCA1:c.1067A>G	Heterozygous	502	269	229	45.98%	0/1	del
BRCA2	13	32911888	32911888	A	G	BRCA1:c.1067A>G	Heterozygous	588	261	327	55.61%	0/1	snp
BRCA2	13	32913055	32913055	A	G	BRCA1:c.1067A>G	Heterozygous	574	1	572	100%	1/1	snp
BRCA2	13	32915005	32915005	G	C	BRCA1:c.1067A>G	Heterozygous	593	0	593	100%	1/1	snp
BRCA2	13	32929387	32929387	T	C	BRCA1:c.1067A>G	Heterozygous	616	599	17	2.76%	0/0	snp
BRCA2	13	41246481	41246481	T	C	BRCA1:c.1067A>G	Heterozygous	567	0	565	100%	1/1	snp
TP53	17	7579472	7579472	G	C	BRCA1:c.1067A>G	Heterozygous	514	0	514	100%	1/1	snp
NF1	17	29705947	29705947	T	C	BRCA1:c.1067A>G	Heterozygous	534	19	515	96.44%	1/1	snp
BRCA1	17	41246481	41246481	T	C	BRCA1:c.1067A>G	Heterozygous	542	287	254	46.95%	0/1	snp

Figure 10: An example batch analysis report. Let OGT customise your report to meet your exact requirements.

Comprehensive range of filtering options

An extensive range of dynamic filtering options are available, which allow you to filter your data to meet your exact analytical criteria (Table 1).

Basic Variant Attributes	Variant Call Attributes	Consequence/ Severity Predictions	Population Frequencies	Classification/ Pathogenicity	Genes, Exons and Proteins	Region/ Variant Lists
Chromosome	Total Depth	Most Severe Consequence	rsID	ClinVar Significance	Gene ID	Region Lists
Start	Ref / Alt Depth	Impact	Minor Allele / Freq		Gene Symbol	Variant Lists
End	Allele Frequency	Consequence	American Minor Allele / Freq		Transcript ID	
Genome Build	Quality Score	Terms	European Minor Allele / Freq		Protein ID	
Ref	Ref / Alt Quality	PolyPhen Prediction	African Minor Allele / Freq		Exon ID	
Alt	Log Ratio	PolyPhen Score	South Asian Minor Allele / Freq		Exon Number	
Genotype	Ref Reads (+) / (-)	SIFT Prediction	East Asian Minor Allele / Freq			
	Alt Reads (+) / (-)	SIFT Score				
	Ref Strand Bias					
	Reads Placed Left / Right					

Table 1: Overview of the wide range of filtering options available in Interpret.

Interpret provides a sophisticated user interface which facilitates, through the comprehensive range of filtering options available, the standardisation of your laboratory workflow. These filters can be incorporated into any analysis protocol and are automatically deployed when a particular protocol is selected. This facilitates the building of more complex filter sets to optimise the search and identification of your variants to your exact requirements (Figures 11 and 12).

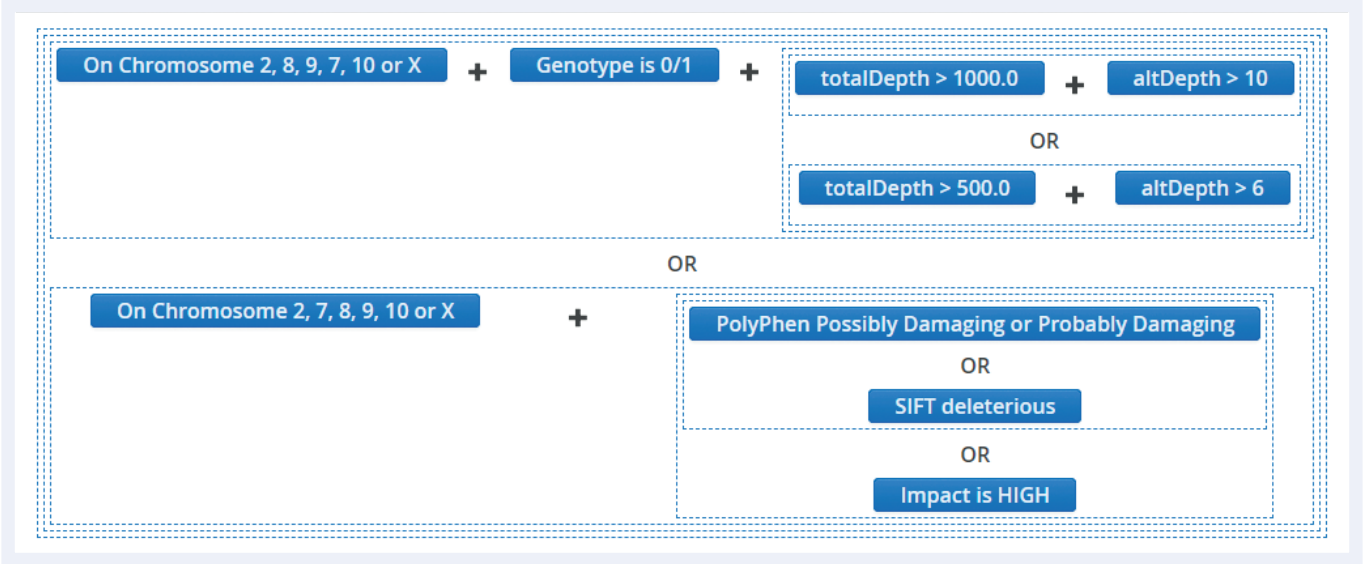

Figure 11: An example of the type of filters that can be easily generated for use within Interpret.

Figure 12: Filtering exists on two levels, firstly within the protocol selected for the analysis and secondly, users are able to filter results dynamically within the variants page.

Security and control

A relational database stores all activities conducted using the software, from the loading of the samples to the variant calls made in those samples. This facilitates the logging and tracking of individual user activity for consistent data processing and laboratory monitoring.

The database enables implementation of security protocols through multiple access permission levels, allowing users with administrator rights to control all functions of the software and the actions of users based on their roles within the laboratory. Additionally, data stored within the relational database can be easily backed up or ported.

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Ordering information

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ogt.com

Product	Contents	Cat. No.
Interpret	Powerful and easy-to-use NGS analysis software. Complimentary with all CytoSure and SureSeq NGS panels.	500076

* Computer specification: RAM: min 16 GB, suggested 24 GB; HDD: min 500 GB, suggested 2TB; CPU: 8+ logical cores at 2+ GHz; OS: Windows (7 or newer; Virtualization (VT-x) needs to be enabled in the BIOS) or Unix (any flavour supporting Docker CE).

† Helga Thorvaldsdóttir, James T. Robinson and Jill P. Mesirov. Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration. Briefings in Bioinformatics 14(2), 178-192 (2013).

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What binds us, makes us.

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