

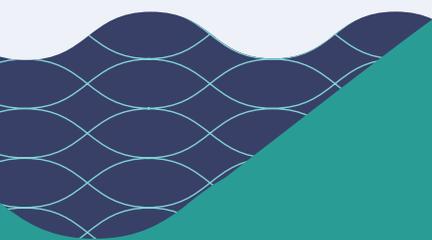
# CytoSure



A Sysmex Group Company

## Application Note

# Interpret NGS Analysis Software – Seamless transition from microarray to NGS in constitutional cytogenetics



### Introduction

At OGT we understand that making the change from microarrays to NGS is a daunting prospect, particularly when it comes to data analysis.

That's why we've developed the Interpret NGS analysis software for use in conjunction with the CytoSure® Constitutional NGS panel for constitutional cytogenetics research. To facilitate the easy transition for copy number variant (CNV) and loss of heterozygosity (LOH) analysis as well as streamlining single nucleotide variant (SNV) and insertion/deletion (indel) calling, we've provided familiar visualisation and interpretation tools alongside an intuitive web interface.

This application note outlines some of the key features we've incorporated to help make the change from arrays to NGS as painless as possible.

#### 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

### CNV and LOH Data Visualisation

Interpret replicates the copy number and LOH data visualisation approach used in OGT's microarray analysis platform, CytoSure Interpret, including the  $\log^2$  ratio/B-Allele Frequency scatter plots, segmentation lines and highlighted CNV and LOH calls to ensure users immediately understand the results (Figure 1).

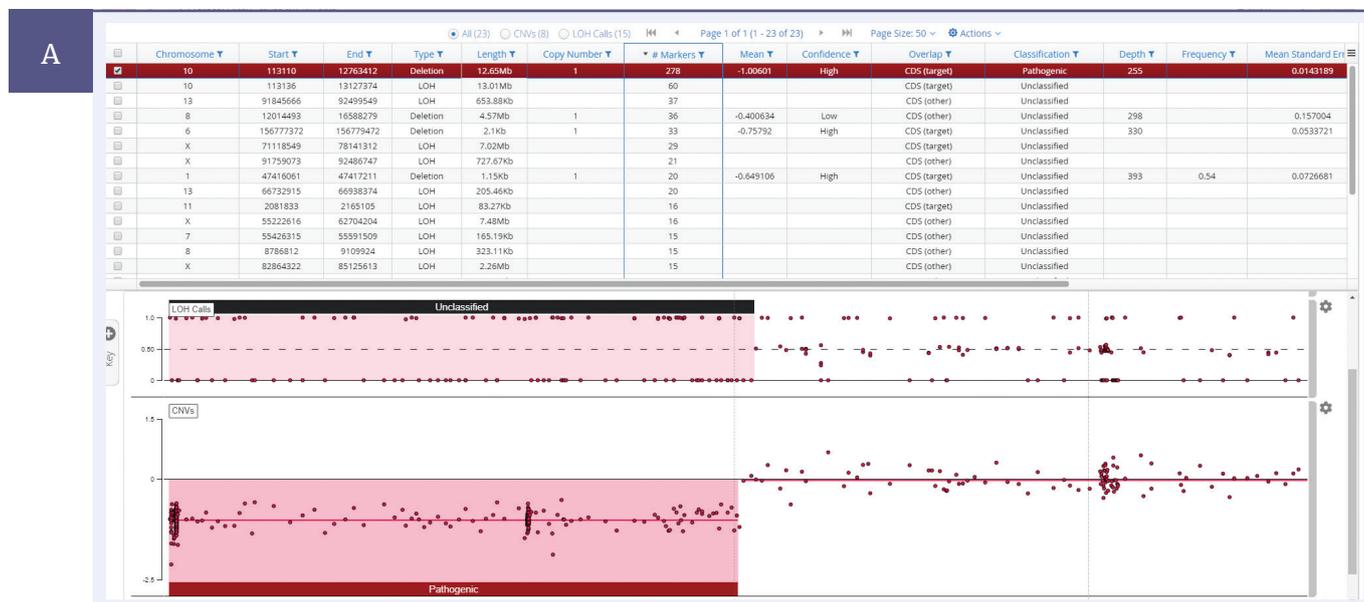




Figure 1: **A** 10p15.3-p13 deletion and associated LOH viewed in Interpret NGS Analysis Software, **B** the same deletion viewed in CytoSure Interpret.

## Annotation Tracks

Comprehensive annotation of genomic features (for example genes, exons, segmental duplications) from local and external CNV databases (such as DGV, ClinGen, DECIPHER), is an essential tool of CNV and LOH interpretation. Interpret displays a number of annotation tracks and provides a variety of mechanisms for importing new tracks, including from an existing CytoSure Interpret database (Figure 2).

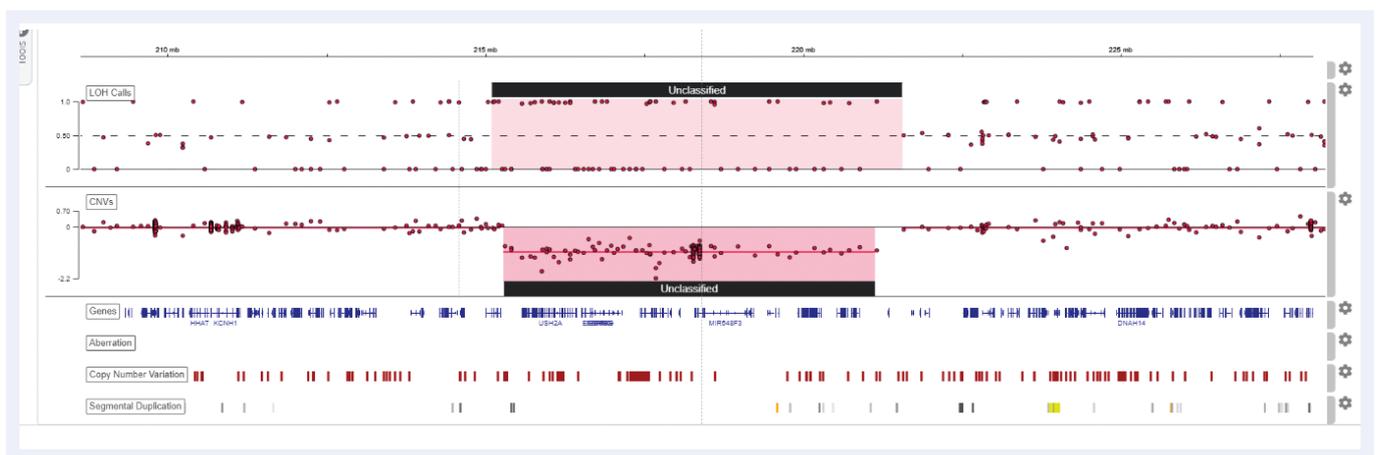


Figure 2. Annotation tracks providing context to a deletion in Interpret.

### Links to External Resources

Interpret also provides customisable links to external web resources, such as Ensembl, UCSC and DGV, to enable easy access to information relevant to the results and further streamline interpretation (Figure 3).

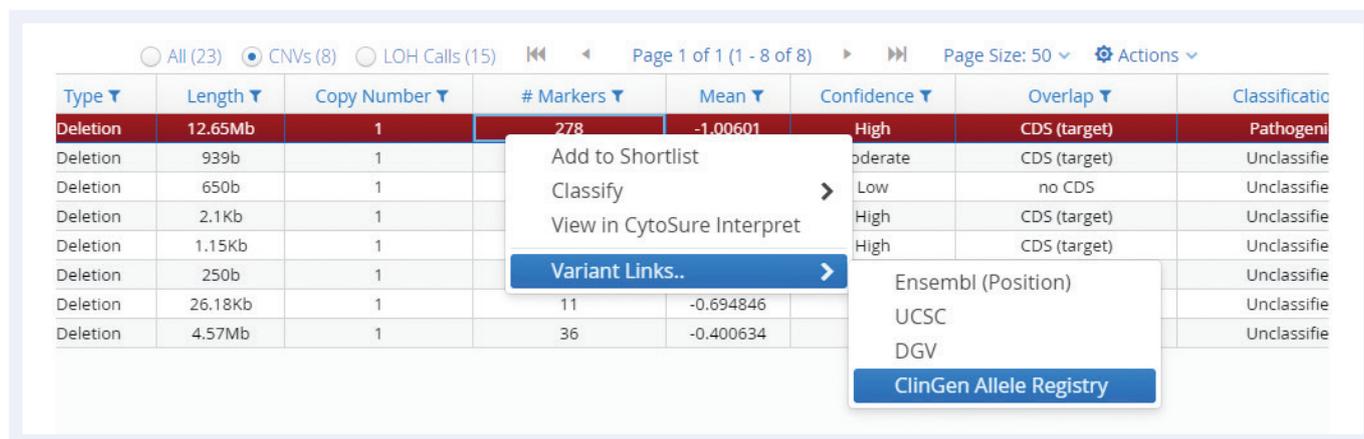


Figure 3: Links to external web resources specific to the selected variant.

### Automated Workflow

All NGS analysis pipeline and variant detection settings are encapsulated in a customisable analysis protocol, ensuring minimal hands-on time and consistent results generation – simply upload your FASTQ files and select a protocol to produce CNVs, LOH calls and SNVs/indels ready for visualisation and interpretation (Figure 4). In addition, there is no need to process each sample separately, upload a complete run or multiple runs as a single batch for seamless processing (Figure 5).

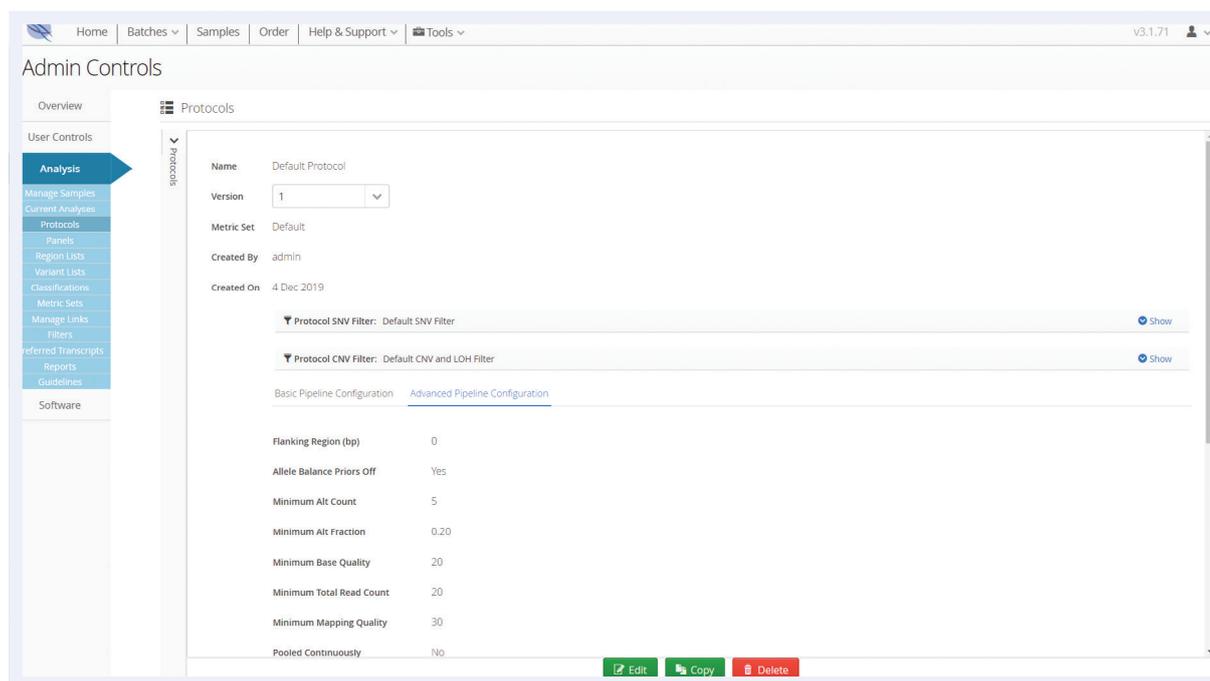


Figure 4: Analysis protocol settings.

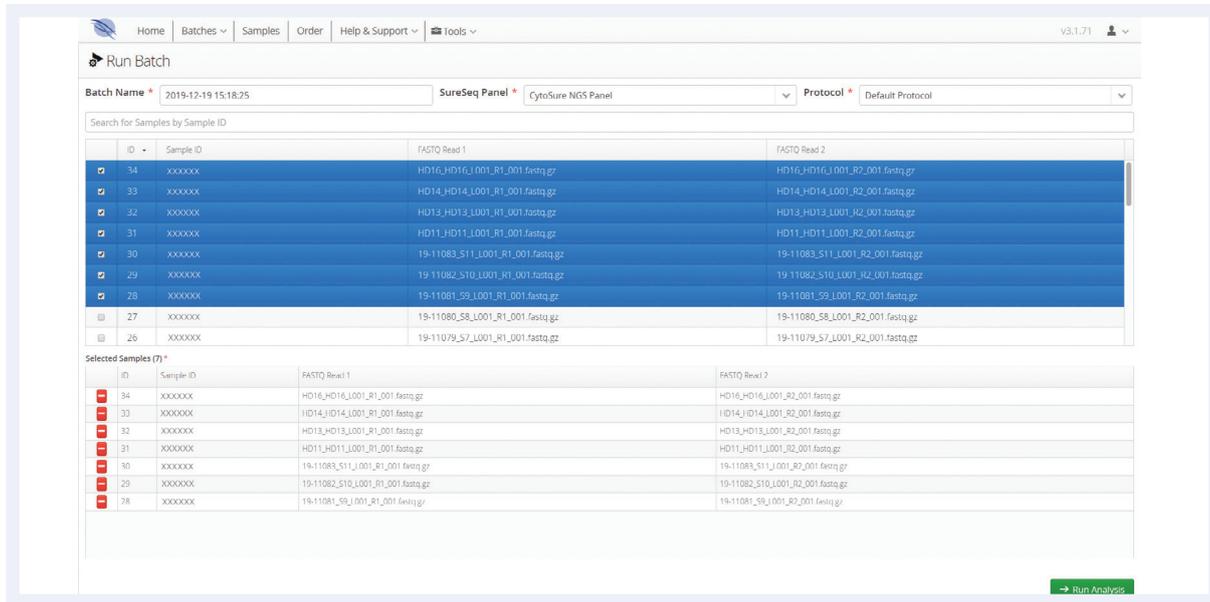


Figure 5: Creation of a batch by selecting samples and an analysis protocol.

## Easy Results Filtering

Given the relative size of the CytoSure Constitutional NGS panel, it is inevitable that a large number of calls will be generated by the data analysis pipeline. Simplify your analysis by using predefined protocol filters (Figure 6) or by creating your own. This is especially useful for SNV and indel call filtering and can be achieved through detailed annotation and a highly configurable and user-friendly filtering interface. Easily reduce the number of variants requiring interpretation based on information from external databases such as ExAC, dbSNP and ClinVar, in silico consequence predictions from SIFT and PolyPhen-2 and your own regions of interest (Figure 7).

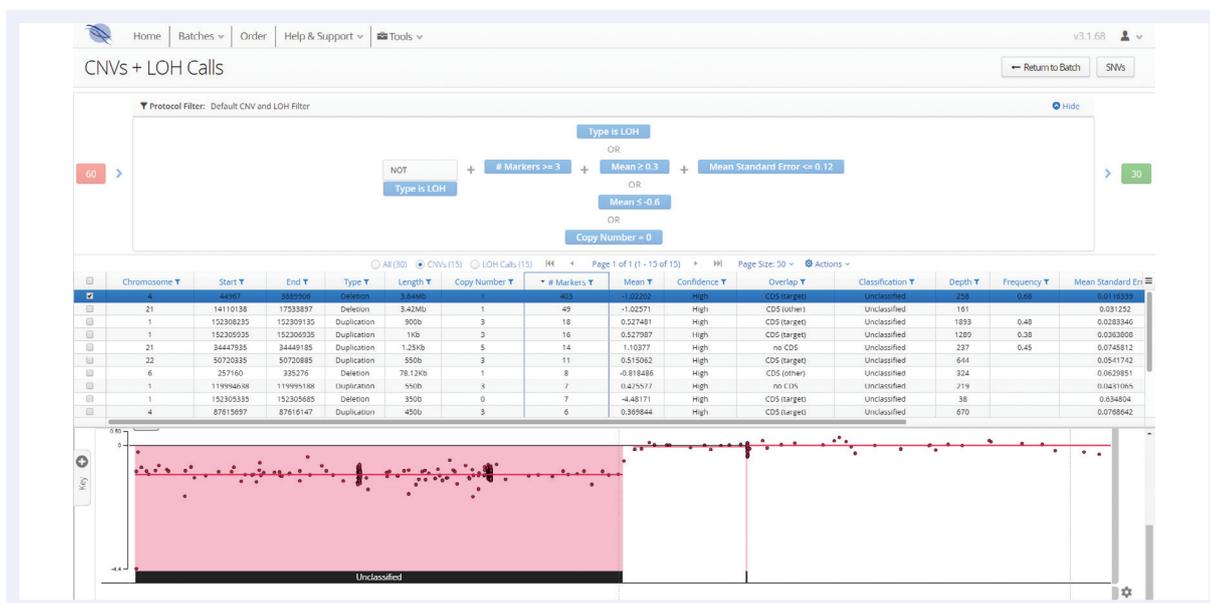


Figure 6: CNV and LOH results generated automatically by the analysis pipeline, including filtering according to predefined criteria in the analysis protocol (60 CNVs/LOH regions reduced to 15 CNVs and 15 LOH regions).

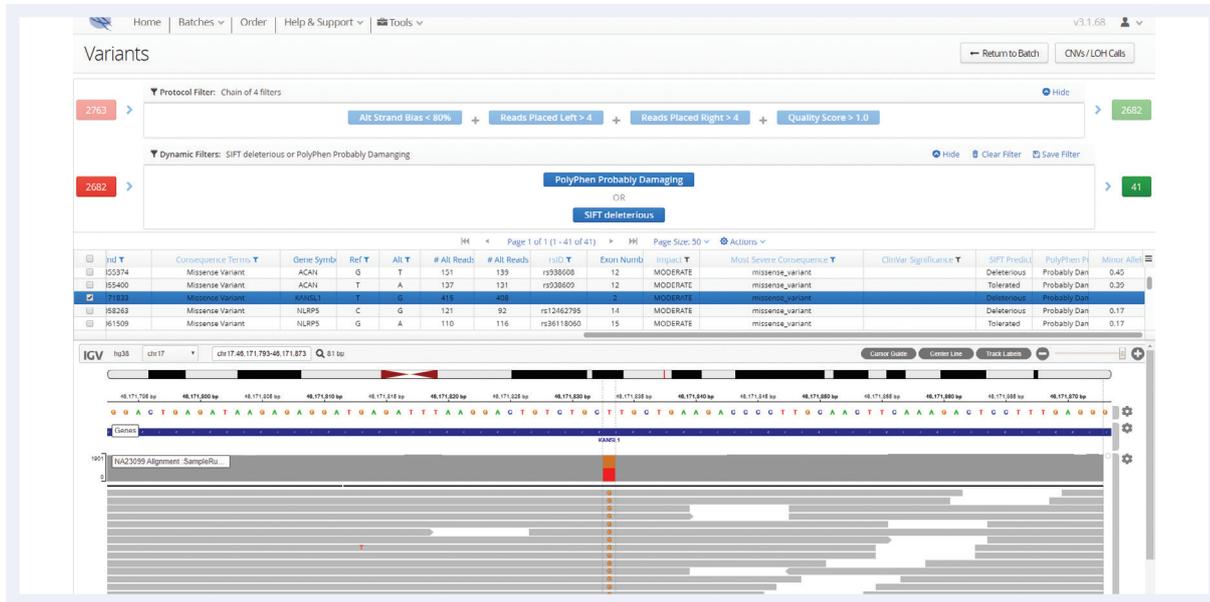


Figure 7: Filtering of SNVs based on in silico prediction tools to identify causative variants.

## Flexible reporting

Interpret enables users to fully customise the layout and format of batch, sample and variant reports through its template-based reporting and plug-in system. Given an example report, OGT can generate a template or plug-in to enable seamless integration with your existing reporting framework (Figure 8).

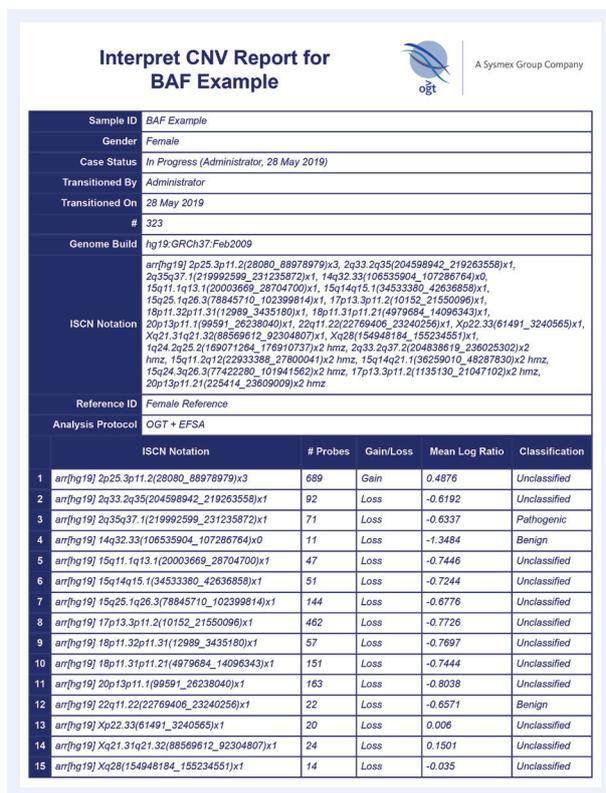
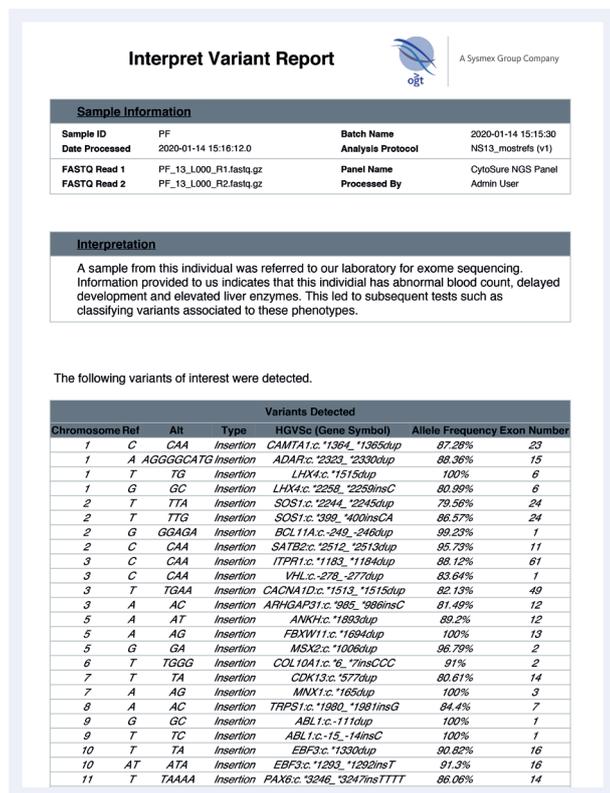


Figure 8: Examples of reports generated by Interpret's template and plug-in framework.

### CytoSure Interpret Integration

If you are a current user of OGT's CytoSure microarrays and CytoSure Interpret, you can also connect your Interpret NGS Analysis software with your existing CytoSure Interpret database to enable interpretation of your CytoSure NGS panel data in CytoSure Interpret:

- Automatically add cases processed in Interpret to your CytoSure Interpret database.
- Open and interpret these cases in the same way as your CytoSure microarrays.
- View variants detected in CytoSure NGS Panel data in CytoSure Interpret with a single click from within the Interpret user interface (Figure 9).

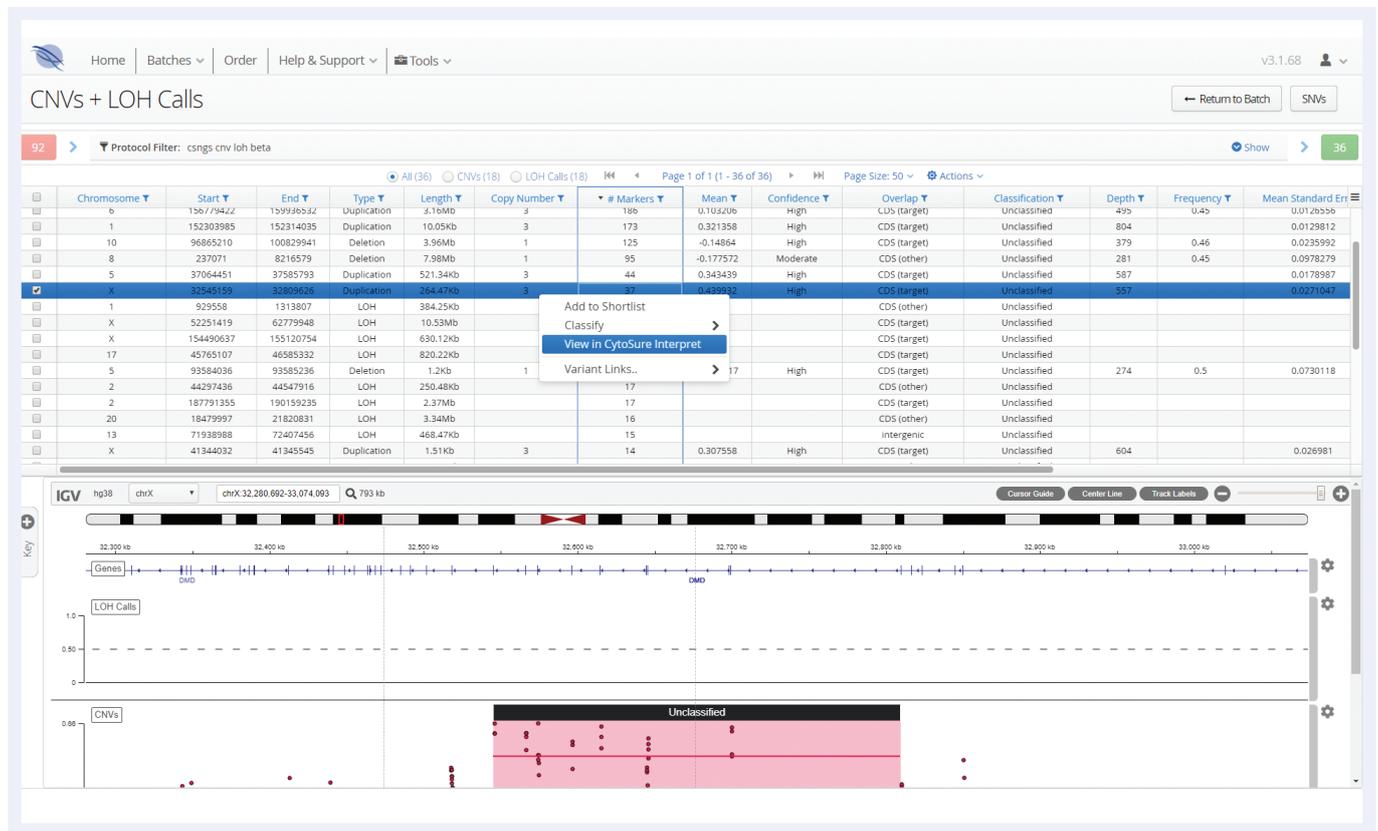


Figure 9: Easy link to open and view CytoSure NGS data in CytoSure Interpret software.

### Ordering information

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Product	Contents	Cat. No.
CytoSure Constitutional NGS Solution (24)	Bundle of 1x CytoSure Constitutional NGS Panel (24), 1x CytoSure NGS Library. Preparation Kit (24) and 1x CytoSure NGS Hybridisation & Wash Kit (24)	502005-B24
CytoSure Constitutional NGS Solution (96)	Bundle of 1x CytoSure Constitutional NGS Panel (96), 1x CytoSure NGS Library. Preparation Kit (96) and 1x CytoSure NGS Hybridisation & Wash Kit (96)	502005-B96

### References

1. Iglesias, A., Anyane-Yeboa, K., Wynn, J. *et al.*, (2014) The usefulness of whole-exome sequencing in routine clinical practice. *Genet Med* 16, 922–931 doi:10.1038/gim.2014.58
2. Yao, R., Zhang, C., Yu, T. *et al.*, (2017) Evaluation of three read-depth based CNV detection tools using whole-exome sequencing data. *Mol Cytogenet* 10: 30. <https://doi.org/10.1186/s13039-017-0333-5>
3. <https://clinicalgenome.org/> [Date accessed 26 September 2018]
4. <https://www.sanger.ac.uk/science/collaboration/deciphering-developmental-disorders-ddd> [Date accessed 26 September 2018]



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**What binds us,  
makes us.**

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