

Exome as a Solution

Hello V8.

Exome V8

Toward an Integrated Exome Sequencing Solution

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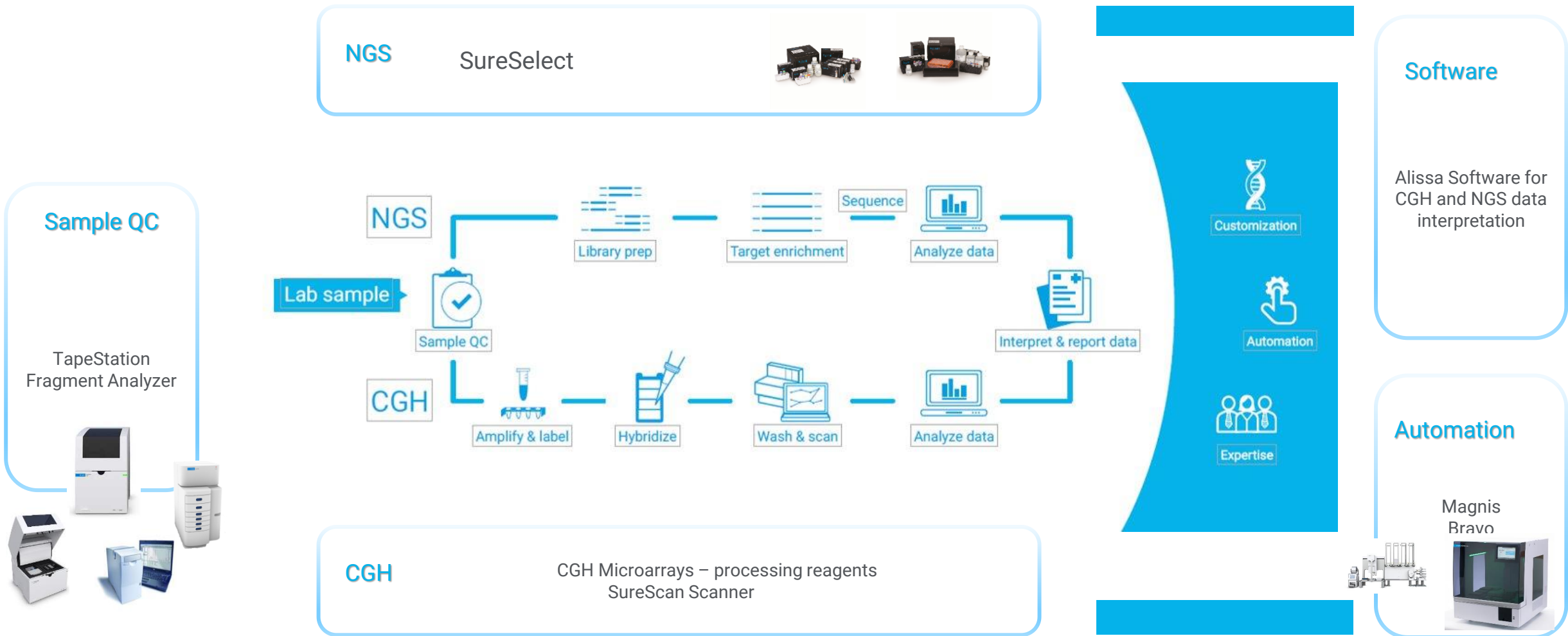
Journée AChroPuce
17/11/2021

Title
Confidentiality label
Regulatory statement (if applicable)



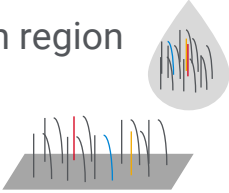


NGS-CGH



Complete workflow





Exome V8 overview

Best-in-class performance & sequencing economy. Versatile content. Enabling workflow.

Advanced Technology First catalog panel to leverage new production process and design algorithm	Improved Manufacturing Process Probe printing targeting design region and reverse complement New probe recovery process 	Powered by Machine Learning Machine learning-based probe selection and placement. 	Consistent Results Over Time Large scale production of oligos that will last many years 
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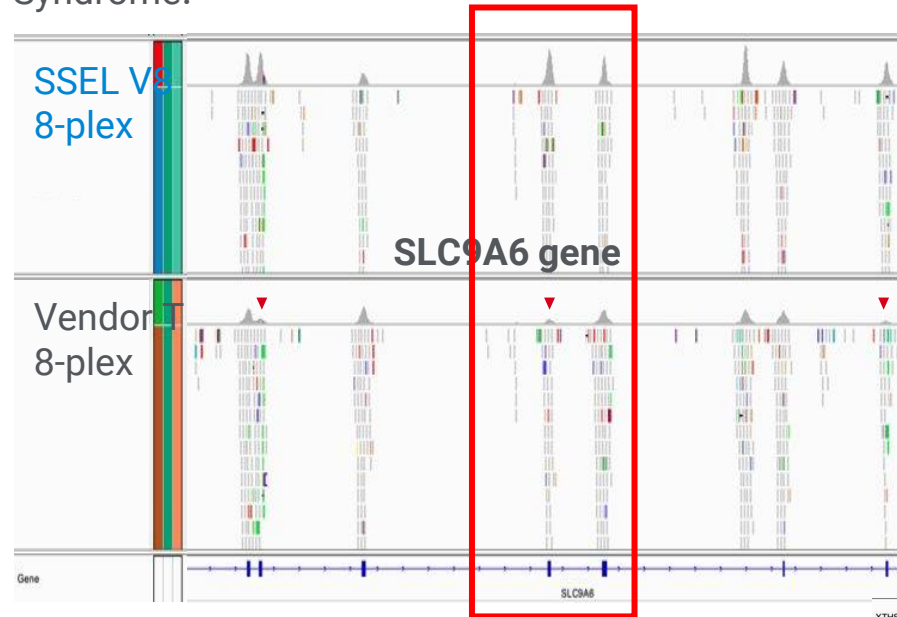
Versatile Content
Comprehensive Exome Targets protein coding regions in major gene definition databases (CCDS, RefSeq, GENCODE) 
Relevant Plus Content V8 UTR Plus V8 Clinical Plus V8 + custom content 

Excellent Performance
Small Design Footprint Efficient probe design that is more economical to sequence 
High-Performance Enrichment Deeper and more uniform coverage for confident results & efficient sequencing 

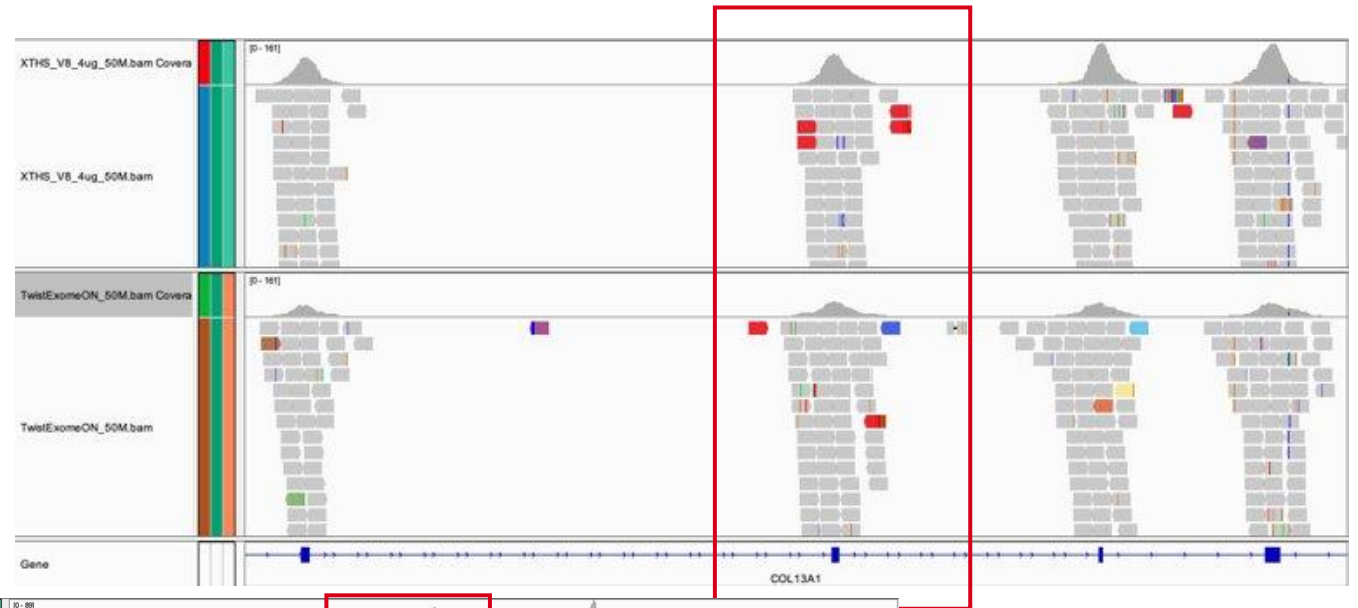
Enabling Workflow
Natively Automated Magnis walkaway automation and Bravo high-throughput sample preparation for increased productivity 
Optimized Informatics Alissa Reporter and Alissa Interpret for efficient variant analysis and reporting 

Better coverage of clinically relevant genes

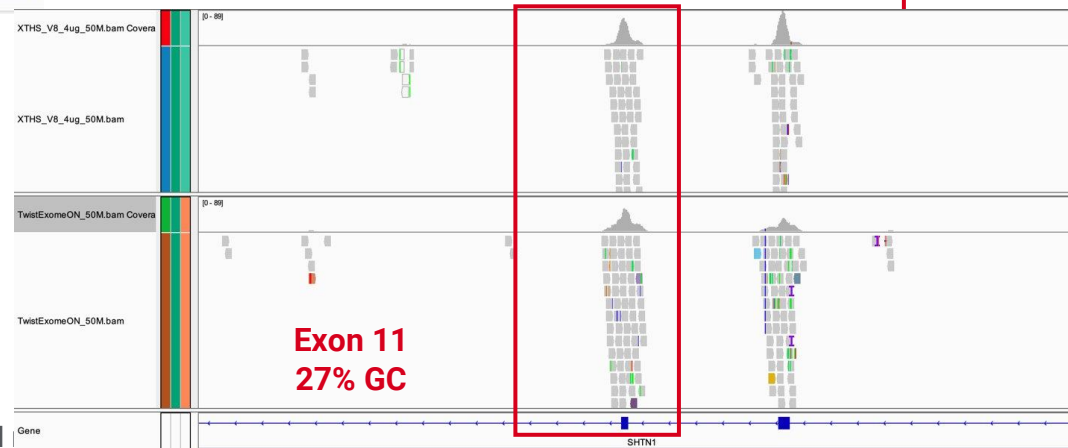
Diseases associated with **SLC9A6** include Intellectual Disability, X-Linked, Syndromic, Christianson Type and Christianson Syndrome.



Diseases associated with **COL13A1** include Myasthenic Syndrome, Congenital, 19 and Presynaptic Congenital Myasthenic Syndromes.



Diseases associated with **SHTN1** include Epileptic Encephalopathy, Early Infantile, 2 (EIEE2).



Exon 31
76% GC

All samples sequenced using 2x100 bp reads and normalized to 50M

Exome Workflow Solution for Any Lab

SureSelect Target Enrichment

Library Prep & Target Enrichment

Exome Probes



Library Prep Solution for Every Application

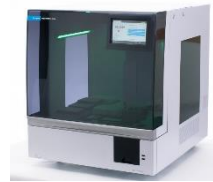
- **Parallel DNA/RNA workflow**
- Mechanical or enzymatic shearing (DNA)
- Pre-capture pooling
- Low input, FFPE samples
- Duplex molecular barcodes

Best-in-Class Exome Enrichment

- Up-to-date exome with excellent enrichment performance
- Content for every project:
V8: routine WES
V8+NCV: clinical WES
V8+UTR: translational
V8+custom content

Magnis

Rapid, Complete Automation



Bravo

High-Throughput Automation



Natively Supported Automation

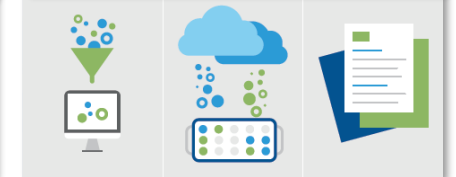
- **Magnis – fully automated system with pre-aliquoted reagents**
- Bravo – automated liquid handling robot for increased throughput

Alissa Clinical Informatics Platform

Alignment & Variant Calling



Interpretation & Reporting



Streamlined Variant Analysis

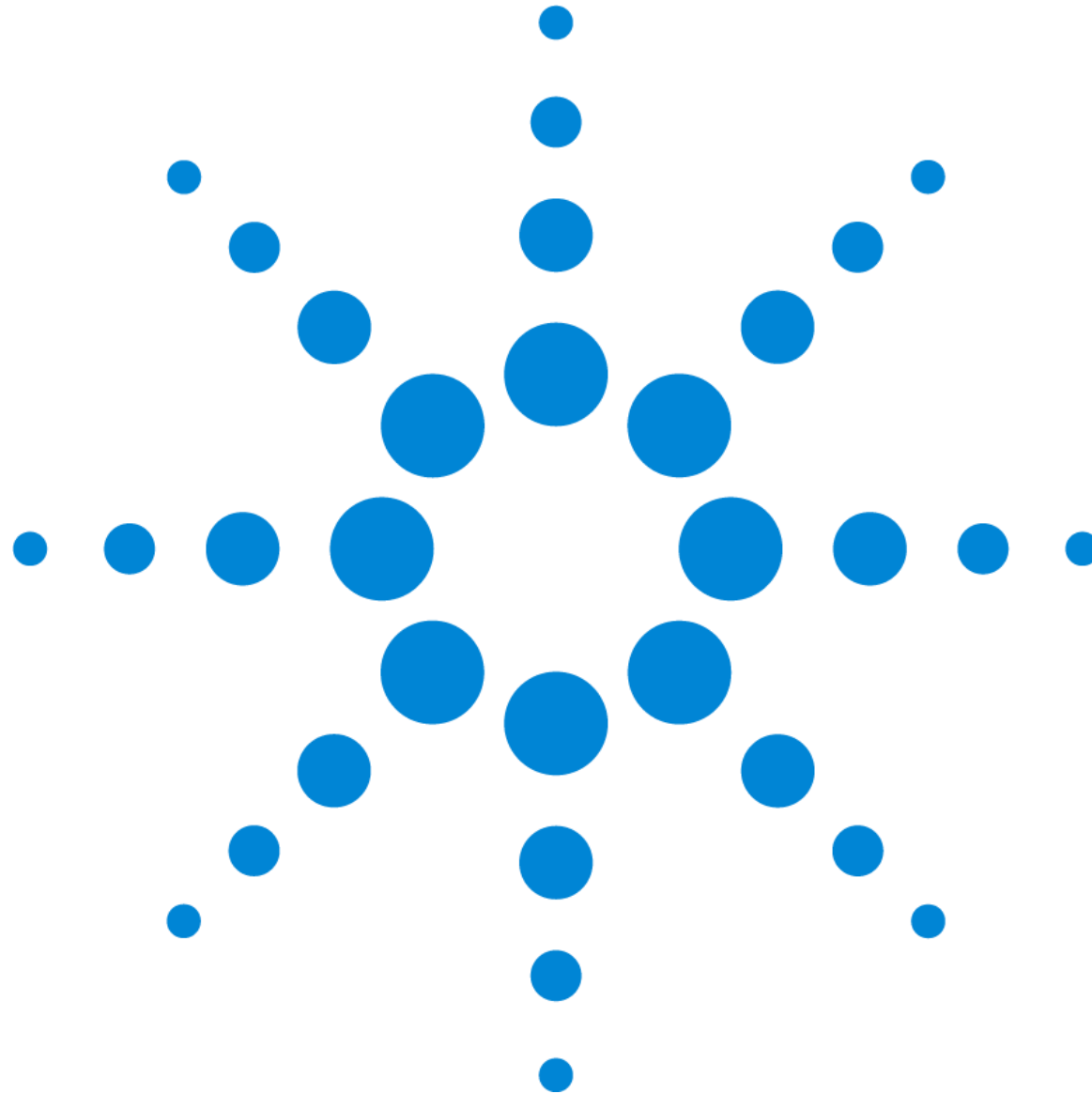
- Scalable data analysis platform from raw data to VCF
- Optimized variant calling algorithms for SureSelect products
- Seamless integration Alissa Interpret
CNV : Alissa Reporter

Efficient Variant Interpretation and Reporting

- Efficient variant triage, classification, and reporting
- Single-workflow variant assessment for both inherited disease and somatic assays
- Scalable, highly secure cloud-based solution

* Alissa Interpret is a Class I Exempt Medical Device in the US, a CE IVD in the EU, and a Class I IVD in Canada and Australia.

Questions?



MERCI