illumina®

Benefits of whole-genome sequencing

Human whole-genome sequencing (WGS) has been instrumental in identifying genomic causes of rare disease, understanding variation in complex disease, and characterizing mutations that drive cancer progression. Advances in sequencing technology have led to lower sequencing costs and the ability to produce large volumes of data, enabling WGS to be a powerful, broadly used tool for genomics research. The following are key benefits of human WGS.

Simplify your workflow

- Go from sample to prepared library with a minimal number of steps using the new DNA extraction-free Nextera™ DNA Library Preparation Kit or traditional TruSeqTM DNA PCR-Free HT Library Preparation Kit
- Take advantage of a single comprehensive assay that lets you interrogate regions across the whole genome
- Return to genomic regions of emerging importance for further investigation without the need to redesign, reprep, or resequence

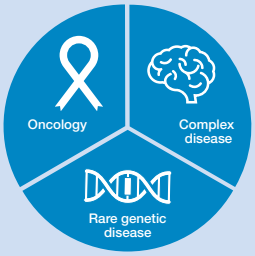
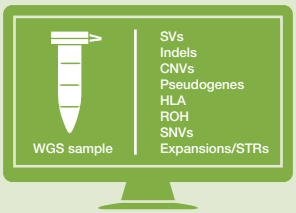
Detect a broad range of variants

- Identify single nucleotide variants (SNVs), indels, structural variants (SVs), copy number variants (CNVs), and repeat expansions all with a single assay
- Get a less biased view of the genome with a simple, streamlined assay

	Variant types							
Features Detected Method	Exonic SNVs–common and rare	Intronic SNVs–common and rare	Indels common and rare	Structural variants	Copy number variants	Expansion repeats/STRs	Pseudogenes	Haplotyping
Arrays	X	X	–	X	X	–	–	–
Exome	X	–	X	–	X*	–	–	–
WGS	X	X	X	X	X	X	X	X

*Requires a control sample/s to normalize coverage

Get a comprehensive view of the genome

Save time using a single assay	Investigate a range of applications with one method	Learn more from your sample	Achieve maximum sensitivity
			

Streamline data analysis with BaseSpace® Suite

Clarity LIMS	**Track samples and optimize lab workflows** • Access pre-configured WGS protocols including TruSeq PCR Free Kit and Nextera DNA Library Preparation Kit • Easily integrate with lab instruments including the NovaSeq™ Series • Simplify sample management with intuitive reporting module, including sample history, reagent usage, and turnaround time
Sequence Hub	**Analyze, store, and share genomic data** • Instantly upload data and automatically start analysis • Access SNV indel, SV, CNV, repeat expansion, HLA, and ROH results • Analyze data quickly: ~3 hours for 30X samples
Variant Interpreter	**Assess variant significance** • Access high-quality curated content of public literature sources • View underlying aligned reads that support a variant call in BaseSpace Hub • Construct virtual gene panels using genotype/phenotype associations
Cohort Analyzer	**Increase understanding of phenotypical markers** • Perform cohort analysis based on molecular or phenotypic features • Compare somatic mutation information and CNVs • Import WGS variant call files or other popular variant callers generated in BaseSpace Sequence Hub
Correlation Engine	**Use data-driven answers to understand genes, variants, and signatures** • Upload a list of variants or genes from your WGS experiments • Find which additional diseases are deregulated for genes of interest • Combine WGS data with other genomic methods

See a detailed report of your findings in Variant Interpreter

- View variants ordered by availability of guidance information
- Interrogate underlying variant details using literature-based evidence
- Track genome-wide copy number, allelic imbalance, and rearrangements
- Visualize variants of interest in a genome browser

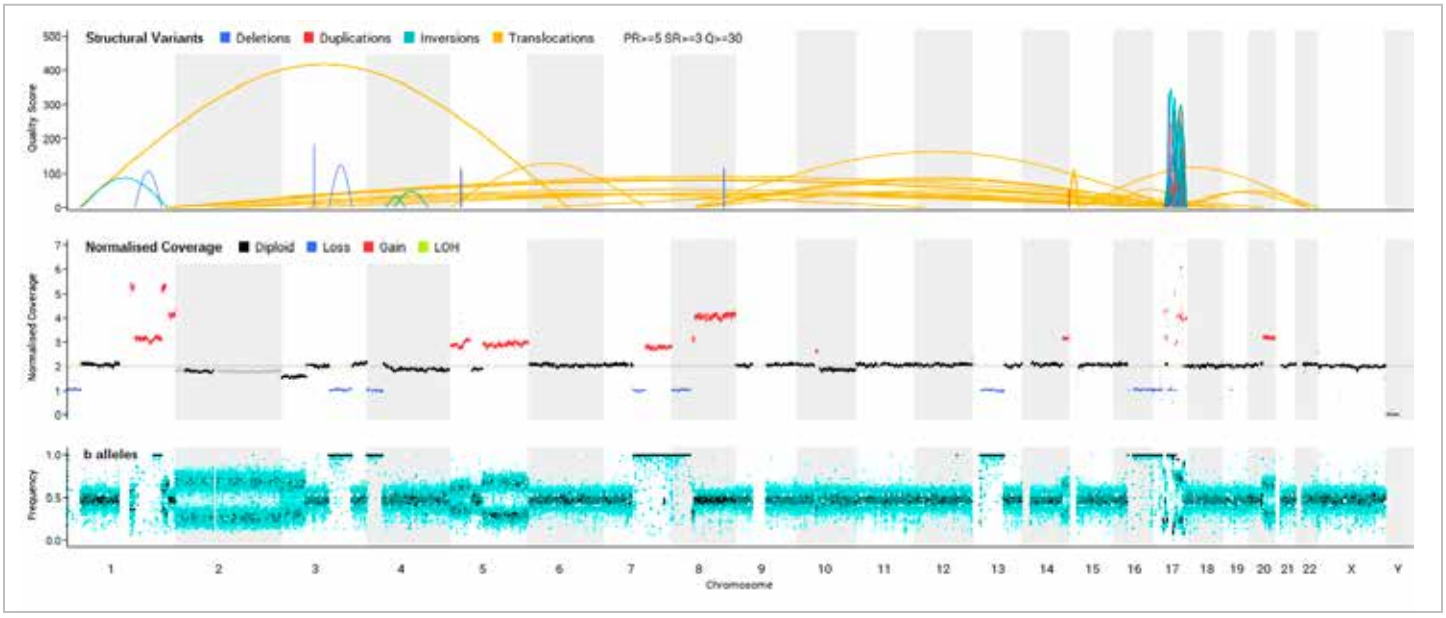


Figure 1: Genome-Wide Landscape of Copy Number Variation and Rearrangements

Resource

Turner TN, Hormozdiari F, Duyzend M. Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA. *Am J Hum Genet*. 2016;98(1):58-74.

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