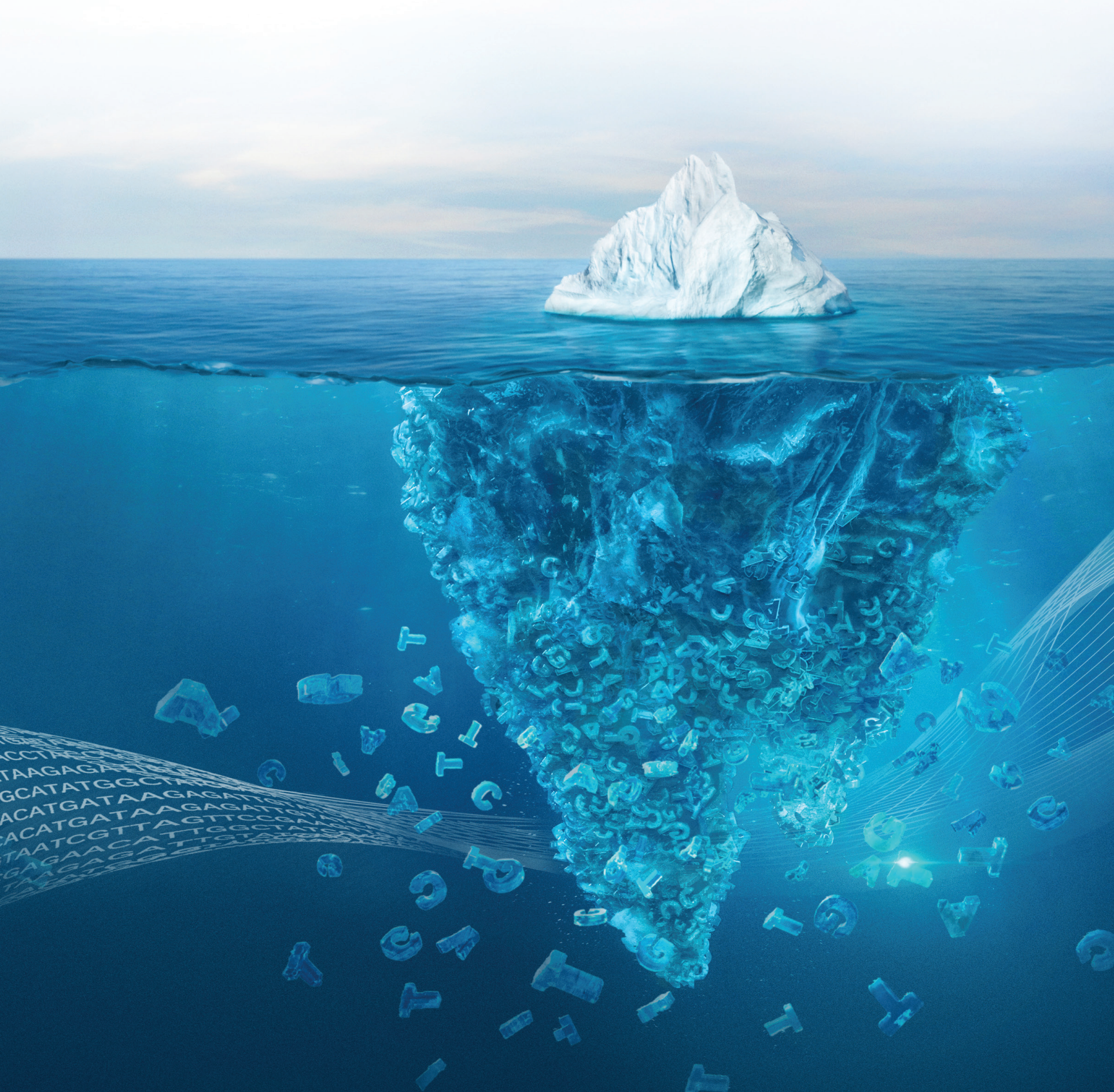


Genomic solutions for complex disease



Power your discovery with our genomic solutions

Access a breadth of applications.
Gain a depth of insights.

To enhance their understanding of complex disease, researchers are applying multiple genomic approaches to gain a more complete picture of biology. Our powerful methods and streamlined workflows equip you to discover more.

- Identify and validate the relationship of genetic profiles to disease phenotypes
- Investigate gene expression and regulation profiles throughout entire pathways
- Experience 1-stop support

Applications

		Common variant discovery in defined and admixed populations	Rare variant discovery	Rare variant discovery in coding regions	eQTL discovery	Methylation and expression correlations	Epigenetic variant discovery	Epigenetic variant validation	Gene variant validation	Novel variant discovery in known genes
Discovery ↓ Methods ↓ Validation	Whole-genome array	●			●					
	Whole-genome sequencing	●	●	●						
	Whole-exome sequencing			●						
	RNA sequencing				●	●				
	Methylation array					●	●			
	Methyl capture sequencing					●	●	●		
	Targeted genotyping array								●	
	Targeted resequencing								●	●

Explore a variety of methods and workflows

Whole-genome sequencing

Prepare library



TruSeq[®] DNA
PCR-Free Kit,
Nextera[™] Flex DNA

Sequence



NovaSeq[™] 6000
Sequencing System

Analyze data



2^o: Whole Genome Sequencing v6.0
3^o: BaseSpace[®] Variant Interpreter,
Correlation Engine, Cohort Analyzer

Whole-exome sequencing

Prepare library



TruSeq workflow with
IDT Exome

Sequence



NextSeq[®] Series

Analyze data



2^o: Whole Exome Sequencing v3.0
3^o: BaseSpace Variant Interpreter,
Correlation Engine, Cohort Analyzer

RNA sequencing

Prepare library



TruSeq Stranded
Total RNA Kit

Sequence



NextSeq Series

Analyze data



2^o: RNASeq v1.1
3^o: Correlation Engine

Methyl capture sequencing

Prepare library



TruSeq Methyl
Capture EPIC Kit

Sequence



NextSeq Series

Analyze data



2^o: MethylSeq v2 App,
MethylKit v2 App

Targeted resequencing

Prepare library



TruSeq Custom Amplicon Kit, TruSeq Neurodegeneration Panel, TruSight Panels, Nextera Rapid Capture Custom Enrichment

Sequence



NextSeq System, MiSeq™ System, MiniSeq™ System

Analyze data



2°: TruSeq Custom Amplicon v3.0
3° : BaseSpace Variant Interpreter, Correlation Engine

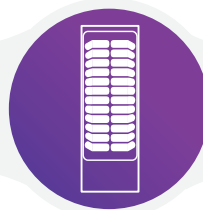
Whole-genome array

Prepare BeadChip



Infinium® Multi Ethnic Array family, Human Omni arrays, Infinium Global Screening Array

Scan BeadChip



iScan®, HiScan®

Analyze data



GenomeStudio®, Genotyping module, GTC to VCF file converter, Correlation Engine

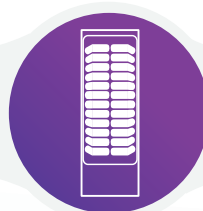
Targeted genotyping array

Prepare BeadChip



Infinium ImmunoArray v2 BeadChip, Infinium Neuro Consortium Array

Scan BeadChip



iScan, HiScan

Analyze data



GenomeStudio, Genotyping module, GTC to VCF file converter, Correlation Engine

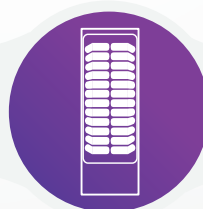
Methylation array

Prepare BeadChip



Infinium MethylationEPIC BeadChip

Scan BeadChip



iScan, HiScan

Analyze data



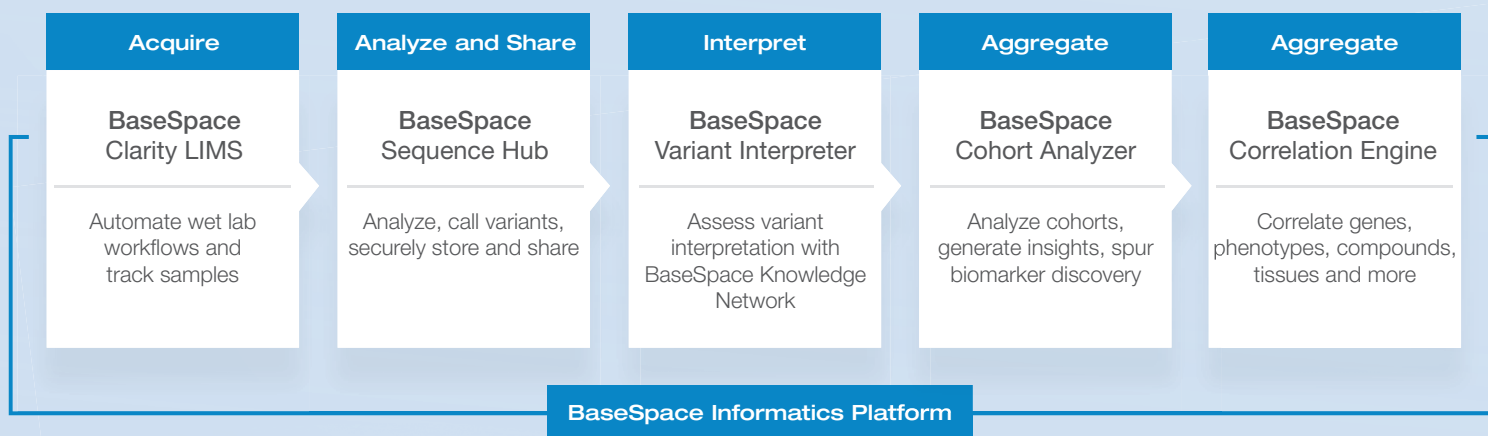
GenomeStudio, Methylation Module, Publicly available third-party bioconductor tools for more sophisticated analyses

Genomic data analysis made simple

BaseSpace Informatics Suite: Comprehensive, cloud-based informatics solutions

- Rely on a software platform that provides common capabilities across the suite, simplifying and expediting your next generation sequencing (NGS) workflows
- Tap into key functionality such as laboratory information management, data storage, analysis, and interpretation
- Obtain high-quality genomic data quickly and apply it immediately

BaseSpace Informatics Suite



Streamline data analysis with BaseSpace Informatics Suite

Easily share and store genomic information

Track samples and optimize lab workflows

- Access preconfigured whole genome sequencing (WGS), whole exome sequencing (WES), RNA, and targeted protocols library prep protocols
- Integrate with out-of-the box lab instruments, including the NovaSeq 6000 System
- Simplify sample management with intuitive reporting module, including sample history, reagent usage, and turnaround time

Analyze, store, and share genomic data

- Instantly upload data and automatically start analysis
- Access single nucleotide variant (SNV), indel, structural variation (SV), copy number variant (CNV), repeat expansion, human leukocyte antigen (HLA), and runs of homozygosity (ROH) results
- Analyze data quickly

Assess variant significance

- Access high-quality curated content of public literature sources
- View underlying aligned reads in BaseSpace Sequence Hub that support a variant call
- Construct gene panels using genotype/phenotype associations
- Increase understanding of clinically and biologically significant markers
- Perform cohort analysis based on molecular or phenotypic features
- Compare somatic mutation information and CNVs
- Import WGS variant call formats or other popular variant callers generated in BaseSpace Sequence Hub

Use data-driven answers to understand genes, variants, and signatures

- Upload a list of variants or genes from your NGS experiments
- Find which diseases are deregulated for your gene
- Combine NGS data with other genomic methods

Discover more with our comprehensive genomic solutions for complex disease. Contact your Illumina representative or visit www.illumina.com/complexdisease

A global leader in DNA sequencing and microarray-based solutions, Illumina is dedicated to improving human health by unlocking the power of the genome. Serving customers in the research, clinical, and applied markets, Illumina technology is responsible for generating more than 90% of the world's sequencing data.* Through collaborative innovation, Illumina is fueling groundbreaking advancements in oncology, reproductive health, genetic disease, agriculture, microbiology, forensic science, and beyond. By empowering large-scale analysis of genetic variation and function, Illumina is enabling studies that were not imaginable just a few years ago, moving us closer to the realization of precision medicine.

*Data calculations on file. Illumina, Inc., 2015.

Illumina • 1.800.809.4566 toll-free (US) • +1.858.202.4566 tel • techsupport@illumina.com • www.illumina.com

For Research Use Only. Not for use in diagnostic procedures.

© 2017 Illumina, Inc. All rights reserved. Pub No. 1070-2016-010

illumina[®]