Infinium® Exome-24 v1.1 BeadChip

Access over 240,000 exonic variants to uncover biologically significant associations.

Overview

The Infinium Exome-24 v1.1 BeadChip delivers comprehensive coverage of putative functional exonic variants selected from over 12,000 individual human exome and whole-genome sequences. Markers were identified through a close collaboration with leading geneticists with the goal of developing an extensive catalog of exome variants. The exonic content consists of more than 240,000 markers representing diverse world populations, including European, African, Chinese, and Hispanic individuals. Exonic content also includes a range of common conditions, such as type 2 diabetes, cancer, metabolic, and psychiatric disorders. The Infinium Exome-24+ v1.1 BeadChip can be customized to include up to 400,000 additional markers. When combined with the iScan® or HiScan® System, the Infinium Exome-24 v1.1 BeadChip can be used to obtain new insights from previously genotyped cohorts, or run new studies focused on identifying functionally relevant associations.

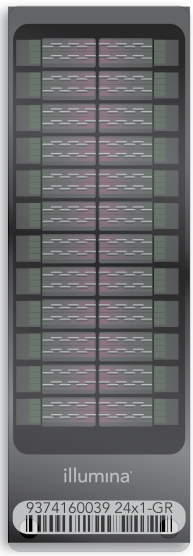

Figure 1: The Infinium Exome-24 v1.1 BeadChip— The Infinium Exome-24 v1.1 BeadChip provides comprehensive coverage of the human exome in multiple, diverse world populations.

High-Throughput Workflow

The Infinium Exome-24 v1.1 BeadChip uses the highly scalable 24-sample Infinium HTS format for high-throughput processing of thousands of samples per week for large, population-scale research and variant screening. The Infinium HTS format also provides a rapid three-day workflow that allows genotyping service providers and clinical researchers to gather data and advance studies quickly (Figure 2).

Optional integration of the Illumina Laboratory Information Management System (LIMS) into the workflow provides high laboratory efficiency with automation functionality, process tracking, and quality control (QC) data tracking. The Illumina ArrayLab Consulting Service offers customized solutions to high-throughput genotyping labs that desire increased efficiency and overall operational excellence.

Robust, High-Quality Assay

The Infinium Exome-24 v1.1 BeadChip uses proven Infinium assay chemistry to deliver the same high-quality, reproducible data (Table 1) that Illumina genotyping arrays have provided for over a decade. The Infinium product line provides high call rates and high reproducibility for numerous sample types including, saliva, blood, solid tumors, fresh frozen, and buccal swabs. It is compatible with the Infinium FFPE QC Kit and Infinium HD FFPE DNA Restore Kit, enabling genotyping of formalin-fixed, paraffin-embedded (FFPE) samples. In addition, the high signal-to-noise ratio of the individual genotyping calls from the Infinium assay provides researchers with access to genome-wide copy number variant (CNV) calling with a mean probe spacing of ~ 11.80 kb.

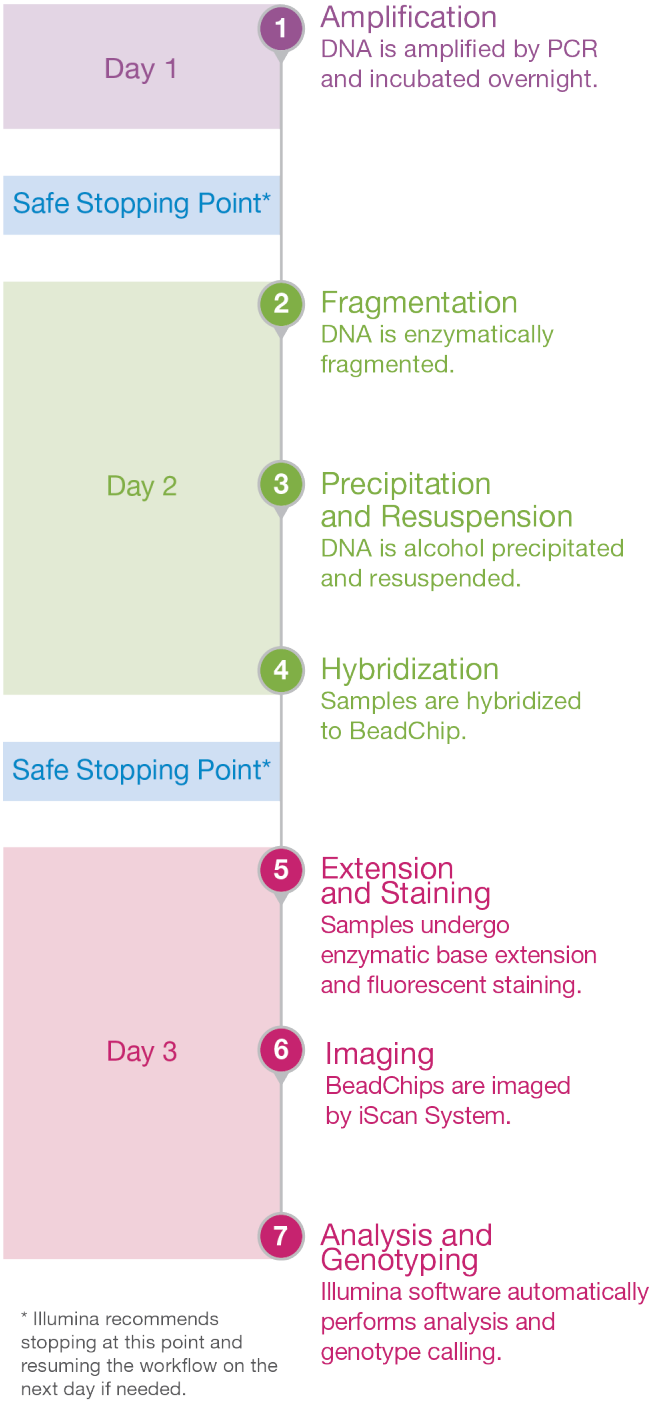

Figure 2: The Infinium HTS Workflow–The Infinium HTS format provides rapid 3-day workflow with minimal hands-on time.

Table 1: Product Information

Feature	Description		
Species	Human		
Total Number of Markers	244,883		
Capacity for Custom Bead Types	400,000		
Number of Samples per BeadChip	24 Samples		
DNA Input Requirement	200 ng		
Assay Chemistry	Infinium HTS		
Instrument Support	iScan or HiScan System		
Sample Throughput[a]	~ 2304 samples/week		
Scan Time per Sample	iScan System	HiScan System	
	2.5 min	2.0 min	
Data Performance	**Value[b]**	**Product Specification**	
Call Rate	99.96%	> 99% avg.	
Reproducibility	99.99%	> 99.9%	
Log R Deviation	0.12	< 0.30[c]	
Spacing			
Spacing (kb)	Mean	Median	90th%[c]
	11.80	0.21	22.86

a. Estimate assumes 1 iScan System, 1 AutoLoader 2.x, 2 Tecan robots, and a 5-day work week.
b. Values are derived from genotyping 330 HapMap reference samples.
c. Value expected for typical projects using standard Illumina protocols. Tumor samples and samples prepared by methods other than standard Illumina protocols are excluded.

Table 2: Marker Information

Marker Categories			No. of Markers
Exonic Markers[a]			227,570
Intronic Markers[a]			31,781
Nonsense Markers[b]			5197
Missense Markers[b]			211,874
Synonymous Markers[b]			9284
Mitochondrial Markers[c]			200
Indels[c]			139
Sex Chromosomes[c]	X	Y	PAR/Homologous
	5015	105	104

a. RefSeq - NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed September 2016.
b. Compared against the University of California, Santa Cruz (UCSC) Genome Browser. genome.ucsd.edu. Accessed August 2014.
c. NCBI Genome Reference Consortium, Version GRCh37. www.ncbi.nlm.nih.gov/grc/human. Accessed July 2016.

Abbreviations: indel: insertion/deletion; PAR: pseudoautosomal region.

Table 3: High-Value Content

Content	No. of Markers	Research Application/Note
ADME Core and Extended Genes[1]	4278	Drug metabolism and excretion
ADME Core and Extended Genes +/- 10 kb	4913	Drug metabolism and excretion (plus regulatory regions)
APOE[2]	6	Cardiovascular disease, Alzheimer's disease, immunoregulation, and cognition
Blood Phenotype Genes[3]	544	Blood phenotypes
COSMIC[4] Genes	226,546	Somatic mutations in cancer
GO[5] CVS Genes	59,043	Cardiovascular conditions
Database of Genomic Variants[6]	195,985	Genomic structural variation
eQTLs[7]	724	Genomic loci regulating mRNA expression levels
Fingerprint SNPs[8]	286	Human identification
HLA Genes[2]	246	Disease defense, transplant rejection, and autoimmune disorders
Extended MHC[a9]	4218	Disease defense, transplant rejection, and autoimmune disorders
KIR Genes[2]	16	Autoimmune disorders and disease defense
Neanderthal SNPs[10]	253	Neanderthal ancestry and human population migration
NHGRI GWAS Catalog[11]	4654	Markers from published genome-wide association studies
RefSeq[12] 3' UTRs	11,191	3' untranslated regions of known genes
RefSeq 5' UTRs	11,365	5' untranslated regions of known genes
RefSeq All UTRs	21,941	All untranslated regions of known genes
RefSeq	236,086	All known genes
RefSeq +/- 10 kb	238,233	All known genes plus regulatory regions
RefSeq Promoters	6812	2 kb upstream of all known genes to include promoter regions
RefSeq Splice Regions	2084	Variants at splice sites in all known genes

a. Extended MHC is a ~ 8 Mb region.

Abbreviations: ADME: absorption, distribution, metabolism, and excretion; APOE: apolipoprotein E; COSMIC: catalog of somatic mutations in cancer; GO CVS: gene ontology annotation of the cardiovascular system; eQTL: expression quantitative trait loci; HLA: human leukocyte antigen; KIR: killer cell immunoglobulin-like receptor; MHC: major histocompatibility complex; NHGRI: national human genome research institute; GWAS: genome-wide association study; UTR: untranslated region; RefSeq: reference sequence.

Ordering Information

Infinium Exome-24 v1.1 Kit	Catalog No.
48 Samples	20015246
288 Samples	20015247
1152 Samples	20015248
Infinium Exome-24+ v1.1 Kit[a]	**Catalog No.**
48 Samples	20015249
288 Samples	20015250
1152 Samples	20015251

a. Enabled for additional custom content.

Learn More

To learn more about the Infinium Exome-24 v1.1 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/genotyping.

References

1. PharmaADME Gene List. www.pharmaadme.org. Accessed August 2014.
2. University of California, Santa Cruz (UCSC) Genome Browser. genome.ucsc.edu. Accessed August 2014.
3. NCBI Reference Sequence Blood Group Antigen Gene Mutation Database. www.ncbi.nlm.nih.gov/projects/gv/rbc/xslcgi.fcgi?cmd=bgmut/systems. Accessed July 2016.
4. Catalog of somatic mutations in cancer. cancer.sanger.uk/cosmic. Accessed July 2016.
5. Gene Ontology Consortium. www.geneontology.org. Accessed July 2016.
6. Database of Genomic Variants. dgv.tcag.ca/dgv/app/home. Accessed July 2016.
7. NCBI eQTL Database. www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi. Accessed July 2016.
8. The Allele Frequency Database. alfred.med.yale.edu/alfred/snpSets.asp. Accessed July 2016.
9. de Bakker PIW, McVean G, Sabeti PC, et al. A high-resolution HLA and SNP haplotype map for disease association studies in the extended human MHC. *Nat Genet*. 2006;38:1166–1172.
10. Neanderthal Genome Browser. neandertal.ensemblgenomes.org/index.html. Accessed July 2016.
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12. NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed July 2016.

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

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