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Infinium™ CoreExome-24 v1.4 BeadChip

Customizable, high-density array for cost-effective, large-scale genotyping and screening studies.

Overview

The customizable Infinium CoreExome-24 v1.4 BeadChip offers an economical way to perform and support large genetic studies, especially large-scale genotyping studies. Developed in collaboration with several leading research institutions, the Infinium CoreExome-24 v1.4 BeadChip includes all the tag single nucleotide polymorphisms (SNPs) found on the Infinium Core-24 BeadChip, plus over 240,000 markers from the Infinium HumanExome BeadChip (Table 5, Table 6). The Infinium CoreExome-24+ v1.4 BeadChip has the added capacity to include up to 100,000 semicustom markers. In addition to performing cost-effective large-scale genotyping studies, the Infinium CoreExome-24 v1.4 BeadChip can be used to obtain baseline sample data sets for various downstream applications quickly and easily.

These applications include common variant, mitochondrial DNA (mtDNA), ancestry, sex confirmation, loss of-variant, and insertion/deletion (indel) detection studies (Table 2). Infinium CoreExome-24 v1.4 BeadChips use the trusted Infinium high-throughput screening (HTS) Assay. When combined with the iScan™ or HiScan™ System, this high-density, 24-sample BeadChip (Figure 1) delivers affordable, high-quality, genome-wide information across diverse world populations.

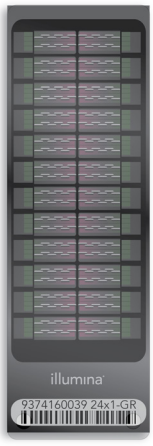

Figure 1: Infinium CoreExome-24 v1.4 BeadChip—The 24-sample Infinium CoreExome-24 v1.4 BeadChip enables informative genotyping of tag SNP and exome-focused markers across diverse world populations, delivering high-quality data that can be used for various downstream applications.

High-throughput workflow

The Infinium CoreExome-24 v1.4 BeadChip uses the highly scalable 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 to 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 CoreExome-24 v1.4 BeadChip uses 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 reproducibility for numerous sample types including saliva, blood, solid tumors, fresh/frozen, and buccal swabs (Table 3). 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 ~ 5.27 kb

Table 1: Product information

Feature	Description		
Species	Human		
Total number of markers	567,218		
Capacity for custom bead types	100,000		
Number of samples per BeadChip	24		
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 2.5 minutes	HiScan System 2.0 minutes	
Data performance	**Value[b]**	**Product specification[c]**	
Call rate	99.8%	> 99% average	
Reproducibility	99.99%	> 99.9%	
Log R deviation	0.09	< 0.30	
Spacing			
Spacing (kb)	Mean 5.27	Median 1.82	90th%[d] 14.30

a. Assumes 1 iScan System, 1 AutoLoader 2.x, 2 Tecan robots, and a 5-day work week.
b. Values are derived from genotyping 270 HapMap reference samples.
c. Excludes Y chromosome markers for female samples.
d. Value expected for typical projects using standard Illumina protocols. Tumor samples and samples prepared by methods other than standard Illumina protocols are excluded.

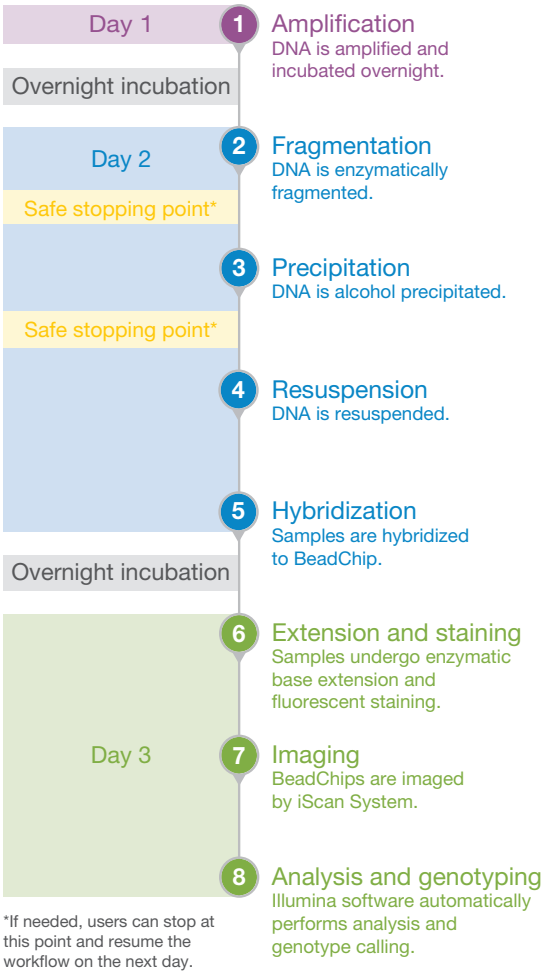

Figure 2: The Infinium HTS workflow—The Infinium HTS format provides rapid 3-day workflow with minimal hands-on time.

Table 2: Marker information

Marker categories			No. of markers
Exonic markers[a]			268,631
Intronic markers[a]			152,454
Nonsense markers[b]			15,040
Missense markers[b]			219,228
Synonymous markers[b]			14,774
MItochondrial markers[c]			369
Indels[c]			12,451
Sex chromosomes[c]	X	Y	PAR/homologous
	13,115	2118	256

a. RefSeq - NCBI Reference Sequence Database. Accessed May 2020.
b. Compared against the UCSC Genome Broswer. Accessed May 2020.
c. NCBI Genome Reference Consortium, Version GRCh37. Accessed May 2020.
Abbreviations: indel, insertion/deletion; PAR, pseudoautosomal region.

Table 3: Imputation accuracy from 1000G[a] at various MAF thresholds

Population[b]	Imputation accuracy		
	MAF ≥ 5%	MAF ≥ 1%	MAF 1–5%
AFR	0.90	0.84	0.76
AMR	0.94	0.89	0.80
EAS	0.93	0.86	0.66
EUR	0.94	0.89	0.76
SAS	0.93	0.86	0.71

LD r^2 ≥ 0.80 from 1000G[a] at various MAF thresholds

Population[b]	LD coverage (r^2 ≥ 0.80)	
	MAF ≥ 5%	MAF ≥ 1%
AFR	0.29	0.18
AMR	0.57	0.40
EAS	0.66	0.54
EUR	0.63	0.49
SAS	0.58	0.44

LD mean from 1000G[a] at various MAF thresholds

Population[b]	LD coverage (r^2 ≥ 0.80)	
	MAF ≥ 5%	MAF ≥ 1%
AFR	0.47	0.31
AMR	0.71	0.53
EAS	0.77	0.64
EUR	0.74	0.59
SAS	0.72	0.56

a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). www.internationalgenome.org. Accessed May 2020.
b. See www.internationalgenome.org/faq/which-populations-are-part-your-study.
Abbreviations: MAF, minor allele frequency; AFR, African; AMR, Ad Mixed American; EAS, East Asian; EUR, European; SAS, South Asian; LD, linkage disequilibrium.

Ordering information

Infinium CoreExome-24 v1.4 Kit	Catalog no.
48 samples	20039222
288 samples	20039223
1152 samples	20039224
Infinium CoreExome-24+ v1.4 Kit[a]	**Catalog no.**
48 samples	20039214
288 samples	20039215
1152 samples	20039216

a. Enabled for additional custom content.

Learn more

To learn more about the Infinium CoreExome-24 v1.4 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/genotyping

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

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