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Infinium™ Japanese Screening Array-24 v1.0 Consortium BeadChip

A powerful, high-quality, cost-effective array for genetic studies in Japanese populations.

Highlights

- **Optimized, exclusive Japanese content**
  Features expert-selected content, including ~100K Japanese-specific markers and the option to add custom content
- **Broad clinical research applications**
  Enables genotyping of clinical research variants for a broad range of applications, including complex disease studies, pharmacogenomics research, and more
- **Robust, high-quality assay**
  Maintains the same data quality of Illumina genotyping arrays with call rates > 99% and reproducibility > 99.9%

Introduction

The Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip provides a high-value, scalable, and cost-effective solution for variant screening and precision medicine research in Japanese populations (Table 1). Designed in collaboration with a Japanese consortium, it harnesses content from the Infinium Asian Screening Array-24 v1.0 BeadChip and new tag single nucleotide polymorphisms (SNPs) based on Japanese whole-genome sequencing (WGS) data. Using the iScan™ System, integrated analysis software, and the Infinium high-throughput screening (HTS) assay, this high-density, 24-sample BeadChip (Figure 1) provides optimized content with the same high-quality, reproducible data that Illumina genotyping arrays have provided for over a decade. The Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip will power growing biobank and translational research studies in the Japanese population.

Table 1: Product information[a]

Feature	Description
Species	Human
Total number of markers[b]	736,847
Capacity for custom bead types	50,000
Number of samples per BeadChip	24
Input requirement	200 ng genomic DNA
Assay chemistry	Infinium HTS
Instrument	iScan System
Maximum sample throughput[a]	~2304 samples/week
Scan time per sample	2.5 minutes

a. Approximate values, scan times, and maximum throughput may vary depending on laboratory and system configurations.
b. Total number of markers calculated from the consortium manifest.

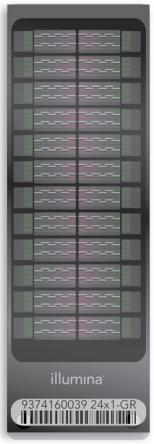

Figure 1: Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip—Built on the trusted 24-sample Infinium HTS platform.

Optimized Japanese content from WGS studies

The Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip contains highly informative tag SNPs in Japanese populations (Table 2), including > 490K genome-wide backbone markers from the Infinium Asian Screening Array-24 v1.0 BeadChip and ~100K Japanese-specific markers contributed by expert human disease researchers and genomic service providers in Japan. Exclusive to this BeadChip, the clinical research content contains ~33K regional markers from various consortia members in Japan. The array combines curated clinical research variants and quality control (QC) markers for a broad range of clinical research and variant screening applications. These applications include disease association and risk profiling studies, pharmacogenomics research, disease characterization, lifestyle and wellness characterization, and marker discovery in complex disease research.[1-4]

Table 2: Marker information

Marker categories[a]			No. of markers
Exonic markers[b]			73,737
Nonsense markers[c]			5608
Missense markers[c]			50,775
Synonymous markers[c]			11,636
MItochondrial markers[c]			1262
Indels[d]			9286
Sex chromosomes[c]	X	Y	PAR/homologous
	27,445	6869	721

a. Number of markers are calculated from the consortium manifest.
b. RefSeq: NCBI Reference Sequence Database. Accessed August 30, 2020.[18]
c. Compared against the UCSC Genome Browser. Accessed August 30, 2020.[6]
d. NCBI Genome Reference Consortium, Version GRCh37. Accessed August 30, 2020.[19]
Abbreviations: indel: insertion/deletion, PAR: pseudoautosomal region.

Table 3: High-value content

Content[a]	No. of markers[b]	Research application/note	Content[a]	No. of markers[b]	Research application/note
ACMG[5] 59 2016 gene coverage	15,156	Variants with known clinical significance identified from clinical WGS and WES samples	GO[10] CVS genes	106,209	Cardiovascular conditions
ACMG[5] 59 all annotations	13,115		Database of Genomic Variants[11]	573,087	Genomic structural variation
ACMG[5] 59 pathogenic	5903		eQTLs[12]	3982	Genomic loci regulating mRNA expression levels
ACMG[5] 59 likely pathogenic	1953		Fingerprint SNPs[13]	378	Human identification
ACMG[5] 59 benign	580		gnomAD[14] exome	73,737	WGS and WES results from unrelated individuals from various studies
ACMG[5] 59 likely benign	888		HLA genes[15]	1103	Disease defense, transplant rejection, and autoimmune disorders
ACMG[5] 59 VUS	2272		Extended MHC[15d]	12,668	Disease defense, transplant rejection, and autoimmune disorders
AIMs[c]	2595	Ancestry-informative markers	KIR genes[6]	80	Autoimmune disorders and disease defense
APOE[6]	16	Cardiovascular disease, Alzheimer's disease, and cognition	Neanderthal SNPs[16]	1651	Neanderthal ancestry and human population migration
Blood phenotype genes[7]	1889	Blood phenotypes	Newborn/carrier screening gene coverage	26,303	Genes associated childhood diseases included in the TruSight™ Inherited Disease Sequencing Panel[19]
ClinVar[1] variants	50,223	Relationships among variation, phenotypes, and human health	NHGRI-EBI GWAS catalog[17]	22,103	Markers from published GWAS
ClinVar[1] pathogenic	19,432		NHGRI diseases	19,492	Markers related to various diseases from published studies
ClinVar[1] likely pathogenic	7684		PharmGKB[2,17]	4287	Human genetic variation associated with drug responses
ClinVar[1] benign	13,134		RefSeq[18] 3' UTRs	16,350	3' untranslated regions
ClinVar[1] likely benign	6516		RefSeq[18] 5' UTRs	7450	5' untranslated regions
COSMIC[8] genes	323,620	Somatic mutations in cancer	RefSeq[18] All UTRs	23,073	Untranslated regions
CPIC[9] all	250	Variants with potential guidelines to optimize drug therapy	RefSeq[18]	362,588	All known genes
CPIC[9]-A/B	140		RefSeq[18] +/- 10 kb	427,037	Regulatory regions
CPIC[9]-B	17		RefSeq[18] Promoters	17,248	2 kb upstream to include promoter regions
CPIC[9]-C	14		RefSeq[18] Splice Regions	2696	Variants at splice sites
CPIC[9]-C/D	109				
CPIC[9]-D	76				

a. Content are derived from the consortium manifest.
b. The number of markers for each category may be subject to change.
c. Based on internal calculations.
d. Extended MHC is a 8 Mb region.

Abbreviations: ACMG: American College of Medical Genetics; AIM: ancestry-informative marker; APOE: apolipoprotein E; COSMIC: catalog of somatic mutations in cancer; CPIC: Clinical Pharmacogenetics Implementation Consortium; EBI: European Bioinformatics Institute; eQTL: expression quantitative trait loci; gnomAD: Genome Aggregation Database; GO CVS: gene ontology annotation of the cardiovascular system; GWAS: genome-wide association study; HLA: human leukocyte antigen; KIR: killer cell immunoglobulin-like receptor; MHC: major histocompatibility complex; NHGRI: national human genome research institute; PharmGKB: Pharmacogenomics Knowledgebase; RefSeq: NCBI Reference Sequence Database; UTR: untranslated region, WES: whole-exome sequencing.

Broad clinical research applications

The clinical research content of the Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip was designed through collaboration with medical genomics experts using multiple annotation databases[1-4] to create an informative, cost-effective panel for clinical research applications (Figure 2 and Table 3).

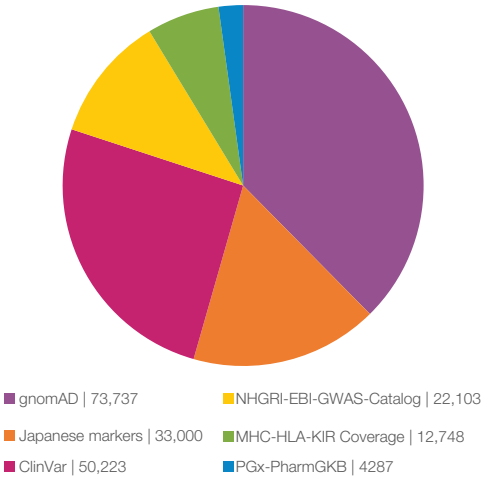

Figure 2: Clinical research content—Content was expertly selected from scientifically recognized databases to create a highly informative array for clinical research applications. Variant counts may be subject to change.

Extensive range of disease categories covered

Including over 18K variants with established clinical associations based on the ClinVar database,[1] clinical research content on the Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip enables validation of disease associations, risk profiling, preemptive screening research, and pharmacogenomics studies. Variant selection includes a range of pathology classifications based on ClinVar American College of Medical Genetics and Genomics (ACMG) annotations (Figure 3A).[5] There are over 7K disease and trait associations from the ClinVar database (Figure 3B) and over 12K variants selected from the NHGRI-GWAS catalog[3] (Figure 4), representing a broad range of phenotypes and disease classifications.

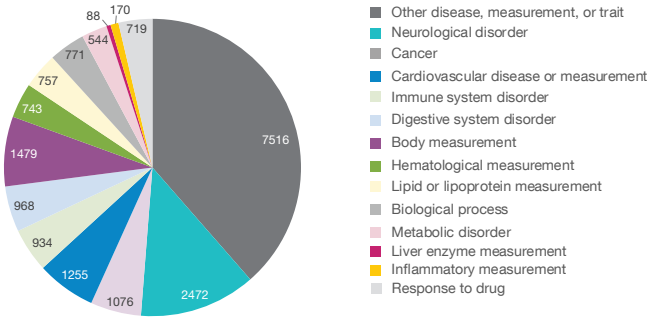

Figure 4: NHGRI disease categories—Clinical research content on the array features > 12,000 markers across a broad range of disease categories based on the NHGRI database. Variant counts are derived from consortium manifest and may be subject to change.

QC markers for sample identification, tracking, and stratification

The Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip includes QC and high-value markers for large-scale studies, enabling sample identification, tracking, ancestry determination, and stratification (Figure 5).

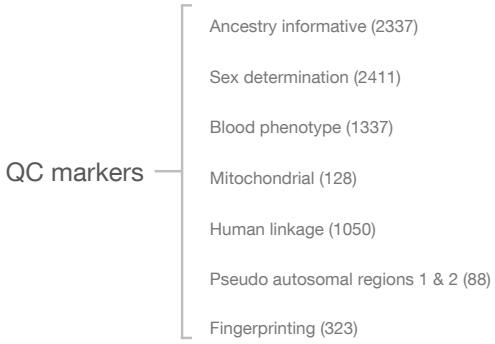

Figure 5: QC markers—QC variants on the array enable various capabilities for sample tracking such as sex determination, continental ancestry, and human identification, and more. Data are derived from the consortium manifest.

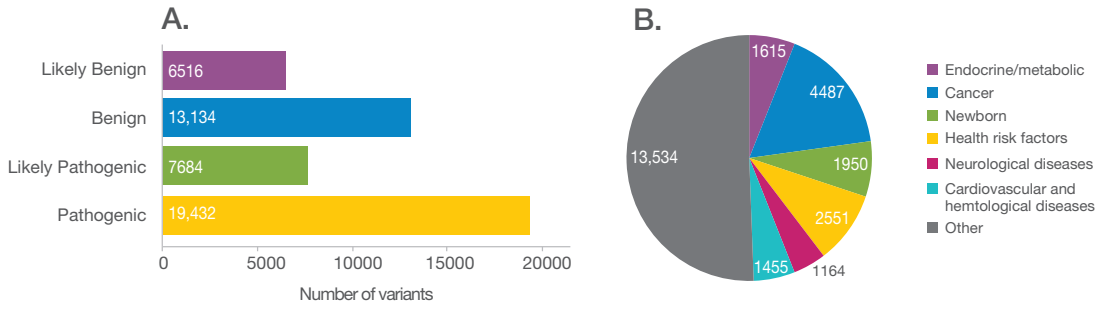

Figure 3: Broad coverage of disease categories—(A) Variants sorted by range of pathology classifications according to ClinVar ACMG annotations. (B) Infinium Japanese Screening Array-24 v1.0 clinical research content features > 7000 disease traits and associations based on categories within the ClinVar database. Variant counts may be subject to change.

Flexible content options

The Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip can be customized to incorporate up to 50K custom bead types or a predesigned content panel (Table 4). The DesignStudio™ Microarray Assay Designer can be used to design targets such as SNPs, copy number variants (CNVs), and indels.

Table 4: Flexible content options

Compatible content	No. of markers	Description
Custom content	≤ 50,000	Custom design virtually any target (eg, SNP, CNV, indel) using the DesignStudio Microarray Assay Designer
Multi-disease drop-in panel	~50,000	Fine-mapping content derived from exome sequencing and meta analysis of phenotype-specific consortia focused on the following traits: psychiatric, neurological, cancer, cardiometabolic, autoimmune, and anthropometric

Summary

The Infinium Japanese Screening Array-24 v1.0 Consortium BeadChip provides a cost-effective solution for population-scale genetic studies, variant screening, and precision medicine research focusing on the Japanese population. The array builds on the success of the widely adopted Infinium Global Screening Array and Infinium Asian Screening Array. Using the iScan System, Infinium HTS Assay, and integrated analysis software, this high-density, 24-sample BeadChip provides optimized content for a broad range of clinical research applications.

Learn more

Learn more about the Infinium Japanese Screening Array-24 v1.0 BeadChip and other Illumina genotyping products and services at www.illumina.com/techniques/microarrays.html.

For labs interested in higher throughput processing, contact your local account manager for more information about Infinium HTS Extra high-throughput kit configurations.

Ordering Information

Infinium Japanese Screening Array-24 v1.0 BeadChip Kit	Catalog no.
48 samples	20040743
288 samples	20040744
1152 samples	20040745
Infinium Japanese Screening Array-24+ v1.0 BeadChip Kit[a]	**Catalog no.**
48 samples	20040746
288 samples	20040747
1152 samples	20040748

a. Enabled for custom content

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