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Infinium® Global Screening Array

A high-powered, economical array for population-scale genomics and screening.

Overview

The Infinium Global Screening Array combines a highly optimized, universal genome-wide backbone, hand curated clinical research variants, and sample tracking content to create a highly economical array for population-scale genomics and screening (Table 1). Using current information from Phase III of the 1000 Genomes Project (1KGP) and population-specific reference databases, the universal genome-wide backbone has been optimized for high imputation accuracy for minor allele frequencies (MAF) of > 1% across all continental populations (Tables 2 and 3).

The hand curated clinical research variants have been designed against the breadth of functional variation and include known associations, pharmacogenomics content, and curated exonic content (Tables 4 and 5). Also, quality control (QC) and high value content has been added to enable the sample identification, tracking, and stratification necessary for large-scale genomics and screening applications.

The Infinium Global Screening Array uses the 24-sample Infinium HTS format (Figure 1), enabling high content flexibility, throughput capacity, and genotyping accuracy. When combined with the iScan® or HiScan® systems, the Infinium Global Screening Array delivers high-throughput sample processing and high-quality data, providing a highly economical tool for population-scale genomics and screening. The Infinium Global Screening Array is also capable of performing copy number analysis and is compatible with the Illumina FFPE QC Kit and Infinium HD FFPE DNA Restore Kit.

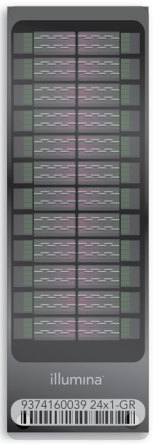

Figure 1: Infinium Global Screening Array—The Infinium Global Screening Array provides a highly economical tool for population-scale genomics and screening.

Table 1: Infinium Global Screening Array Product Information

Feature	Description	
Total Number of Markers	~ 660,000	
Capacity for Custom Markers*	50,000	
Number of Samples per BeadChip	24	
DNA Input Requirement	200 ng	
Assay	Infinium HTS	
Instrument Support	iScan or HiScan	
Spacing	**Mean**	**Median**
Spacing (kb)	4.2	2.0

*Assumes predefined supplemental content is not added. With predefined supplemental content, no add-on capacity is available.

Table 2: Imputation Accuracy for 5 Populations from 1KGP at Varying MAF Thresholds

Feature	Description		
	Imputation Accuracy		
Population[a]	1KGP[b] MAF > 1%	1KGP[b] MAF 1–5%	1KGP[b] MAF > 5%
AFR	0.88	0.82	0.92
AMR	0.93	0.87	0.96
EAS	0.91	0.80	0.95
EUR	0.94	0.88	0.96
SAS	0.90	0.80	0.95

a. See www.1000genomes.org/category/frequently-asked-questions/population
b. Compared against the Phase 3 1000 Genomes Project (1KGP) data release (2014).
Abbreviations: 1kGP, 1000 Genomes Project; MAF, minor allele frequency.

Table 3: Number of Imputated Markers from 1KGP at Varying MAF Thresholds

Feature	Description		
	Number of Imputed Markers at Accuracy Score > 0.80 (% of Total Markers)		
Population[a]	1KGP[b] MAF > 1%	1KGP[b] MAF 1–5%	1KGP[b] MAF > 5%
AFR	12.6 M (86%)	5.1 M (81%)	7.6 M (89%)
AMR	10.0 M (89%)	5.4 M (88%)	5.4 M (88%)
EAS	6.5 M (83%)	2.2 M (79%)	4.8 M (87%)
EUR	7.5 M (87%)	3.0 M (87%)	5.3 M (89%)
SAS	7.7 M (84%)	3.0 M (80%)	5.4 M (88%)

a. See www.1000genomes.org/category/frequently-asked-questions/population
b. Compared against the Phase 3 1000 Genomes Project (1KGP) data release (2014).
Abbreviations: 1kGP, 1000 Genomes Project; MAF, minor allele frequency.

This data sheet contains preliminary information and is subject to change.

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Table 4: Infinium Global Screening Array Marker Information

Marker Categories			Number of Markers[a]
In RefSeq[b] Genes			339,094
In RefSeq Exons			72,474
In RefSeq Promoter Regions			15,058
In ADME Genes			6668
In ADME Exons			2787
MHC[c]			9797
Overlap with Genes in COSMIC[d]			300,733
Overlap with Genes in Gene Ontology[e]			90,628
Mitochondrial Markers			164
Indels			4196
Sex Chromosomes	X 19,660	Y 1681	PAR 547

a. Compared against the June 2011 1kGP data release. www.1000genomes.org
b. RefSeq - NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq
c. MHC is a ~ 4 Mb region.
d. Catalog of somatic mutations in cancer. cancer.sanger.ac.uk/cosmic
e. Gene Ontology Consortium. www.geneontology.org
Abbreviations: ADME, absorption, distribution, metabolism, and excretion; MHC, Major Histocompatibility Complex.

Table 5: Infinium Global Screening Array Clinical Research Content

Known Associations	Number of Markers
PharmGKB	3367
ClinVar[a]	17,020
Types of Functional Variation[b]	**Number of Markers**
SNPs	16,545
Indels	323
CNV	729
Structural	4838
Genic	21,094
Intergenic	1666
Disease Categories Covered	**Number of Markers**
Malformations/chromosomal abnormalities	2113
Cancer	2033
Circulatory	1826
Endocrine/metabolic	1460
Nervous system	1113
Other categories (15)	1700
Pharmacogenomics	**Number of Markers**
High evidence[c]	62
Moderate evidence[c]	205
Low evidence[c]	1869
Preliminary[c]	243
Unknown[d]	1943
Putative Associations	**Number of Markers**
NHGRI-GWAS	7000
ClinVar	6249
eMERGE	1551
Curated Exonic Content	**Number of Markers**
ExAC	28,190
Tracking and Stratification	**Number of Markers**
Blood phenotype	2003
Sex determination	3101
Ancestry informative	3212
Fingerprinting	480
Mitochondrial	155
Forensics	173

a. Known associations from ClinVAR exclude somatic mutations, variants missing clinical annotation, variants missing review status, and variants classified as benign or likely benign.
b. Categories are not mutually exclusive.
c. Classification based on PharmGKB classification.
d. Classification based on putative Pharmacogenomic associations either missing annotation or not classified in PharmGKB.

Learn More

To learn more about the Infinium Global Screening Array and other Illumina genotyping products and services, visit: www.illumina.com/applications/genotyping.html

Ordering Information

To place an order, contact consortiamanager@illumina.com or your local genotyping specialist:
North America: 800.809.4566
Europe, Middle East, Africa: +44.1799.534000
Other regions: www.illumina.com/company/contact-us.html

This data sheet contains preliminary information and is subject to change.

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