

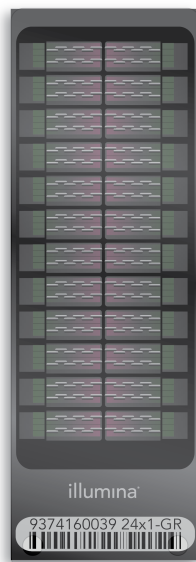
# Infinium® PsychArray-24 v1.2 BeadChip

Evaluate genetic variants associated with common psychiatric disorders using proven Infinium array technology.

## Overview

The Infinium PsychArray-24 v1.2 BeadChip is a cost-effective, high-density array, developed in collaboration with the Psychiatric Genomics Consortium<sup>1</sup> and several leading research institutions for large-scale genetic studies focused on psychiatric predisposition and risk. Content for the Infinium PsychArray-24 v1.2 BeadChip includes ~271,000 proven tag single nucleotide polymorphisms (SNPs) found on the Infinium Core-24 BeadChip, ~277,000 markers from the Infinium Exome-24 BeadChip, and ~50,000 markers associated with common psychiatric disorders. These markers include genetic variants associated with the research of common psychiatric conditions such as:

- Schizophrenia
- Bipolar disorder
- Autism-spectrum disorders
- Attention deficit hyperactivity disorder
- Major depressive disorder
- Obsessive compulsive disorder
- Anorexia nervosa
- Tourette's syndrome



**Figure 1: The Infinium PsychArray-24 v1.2 BeadChip**—The Infinium PsychArray-24 v1.2 BeadChip is a comprehensive array that provides excellent coverage of relevant, consortium-selected markers associated with common psychiatric disorders.

To support additional research requirements, the Infinium PsychArray-24+ v1.2 BeadChip can be tailored to incorporate up to 60,000 custom beadtypes. The combination of rare psychiatric-relevant markers, tag SNPs, and exome content make the Infinium PsychArray-24 v1.2 BeadChip the ideal starting point for genomic studies focused on psychiatric disease.

## High-Throughput Workflow

The Infinium PsychArray-24 v1.2 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 PsychArray-24 v1.2 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 ~4.90 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	593,260		
Capacity for Custom Bead Types	60,000		
Number of Samples per BeadChip	24 Samples		
DNA Input Requirement	200 ng		
Assay Chemistry	Infinium HTS		
Instrument Support	iScan or HiScan System		
Sample Throughput <sup>a</sup>	~ 2304 samples/week		
Scan Time per Sample	iScan System 2.5 min HiScan System 2.0 min		
Data Performance	Value <sup>b</sup>	Product Specification	
Call Rate	99.8%	> 99% avg.	
Reproducibility	99.99%	> 99.9%	
Log R Deviation	0.12	< 0.30 <sup>c</sup>	
Spacing			
Spacing (kb)	Mean	Median	90th% <sup>c</sup>
	4.90	1.75	13.23

- 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 324 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: LD  $r^2 \geq 0.80$  from 1000G<sup>a</sup> at Various MAF Thresholds**

1000G Population <sup>b</sup>	LD Coverage ( $r^2 \geq 0.80$ )	
	MAF $\geq 5\%$	MAF $\geq 1\%$
AFR	0.47	0.31
AMR	0.71	0.53
EAS	0.77	0.64
EUR	0.75	0.60
SAS	0.72	0.56

- a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). [www.1000genomes.org](http://www.1000genomes.org). Accessed July 2016.
  - b. See [www.1000genomes.org/category/frequently-asked-questions/population](http://www.1000genomes.org/category/frequently-asked-questions/population)
- Abbreviations: LD: linkage disequilibrium; AFR: African; AMR: Ad Mixed American; EAS: East Asian; EUR: European; SAS: South Asian.

**Table 3: LD Mean  $r^2$  from 1000G<sup>a</sup> at Various MAF Thresholds**

Population <sup>b</sup>	LD Coverage (Mean $r^2$ )	
	MAF $\geq 5\%$	MAF $\geq 1\%$
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

- a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). [www.1000genomes.org](http://www.1000genomes.org). Accessed July 2016.
  - b. See [www.1000genomes.org/category/frequently-asked-questions/population](http://www.1000genomes.org/category/frequently-asked-questions/population)
- Abbreviations: LD: linkage disequilibrium; MAF: minor allele frequency; AFR: African; AMR: Ad Mixed American; EAS: East Asian; EUR: European; SAS: South Asian.

**Table 4: Marker Information**

Marker Categories	No. of Markers		
Exonic Markers <sup>a</sup>	282,097		
Intronic Markers <sup>a</sup>	167,352		
Nonsense Markers <sup>b</sup>	25,901		
Missense Markers <sup>b</sup>	219,764		
Synonymous Markers <sup>b</sup>	14,896		
Mitochondrial Markers <sup>c</sup>	359		
Indels <sup>c</sup>	12,445		
Sex Chromosomes <sup>c</sup>	X	Y	PAR/Homologous
	14,139	1965	257

a. RefSeq - NCBI Reference Sequence Database.

[www.ncbi.nlm.nih.gov/refseq](http://www.ncbi.nlm.nih.gov/refseq). Accessed September 2016.

b. Compared against the University of California, Santa Cruz (UCSC) Genome Browser. [genome.ucsd.edu](http://genome.ucsd.edu). Accessed August 2014.

c. NCBI Genome Reference Consortium, Version GRCh37. [www.ncbi.nlm.nih.gov/grc/human](http://www.ncbi.nlm.nih.gov/grc/human). Accessed July 2016.

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

## Ordering Information

Infinium PsychArray-24 v1.2 Kit	Catalog No.
48 Samples	20015238
288 Samples	20015239
1152 Samples	20015240
Infinium PyschArray-24+ v1.2 Kit <sup>a</sup>	Catalog No.
48 Samples	20015241
288 Samples	20015242
1152 Samples	20015243

a. Enabled for additional custom content.

## Learn More

To learn more about the Infinium PsychArray-24 v1.2 BeadChip and other Illumina genotyping products and services, visit [www.illumina.com/genotyping](http://www.illumina.com/genotyping).

## References

1. Psychiatric Genomics Consortium. [www.med.unc.edu/pgc](http://www.med.unc.edu/pgc).

