

Custom Genotyping

Illumina's custom genotyping options offer the broadest range of marker density and the most flexible content to deliver uncompromised data quality for any genome.

Custom Genotyping Highlights

- Industry-Leading Data Quality
 Accurate and reproducible data with proven assays
- Flexible Content
 Custom assays in the widest range of multiplex levels for any SNP and any genome
- Broad Range of Throughput Capabilities
 Multi-sample formats and automation-compatible, rapid scanning technology

Custom Genotyping Options

Illumina's custom genotyping options offer the broadest range of marker density, unlimited flexibility, and highest data quality for assaying focused genomic regions of interest, low- to high-plexity, or organisms for which there are no standard products. Custom genotyping assays are available for several Illumina technologies including BeadArray™ technology for Infinium®, GoldenGate®, and GoldenGate Indexing™ assays; VeraCode® technology for GoldenGate assays; and the Eco™ Real-Time PCR System for qPCR assays.

Genotyping assays enable a wide range of experimental designs, depending on your needs (Figure 1). Custom iSelect® HD BeadChips or the semi-custom HumanOmniExpress+ BeadChip allow addition of the highest density of genetic markers. Custom GoldenGate Assays are ideal for focused studies of up to 32 samples per array. The GoldenGate Indexing Assay allows pooling multiple samples, dramatically increasing the number of samples that can be analyzed in a single day. For high-throughput processing of mid- to low-plex samples, the GoldenGate Assay using VeraCode technology enables up to 384-plex genotyping of hundreds of samples per day. The allele-specific primer extension (ASPE) assay provides a flexible and economical solution for low-plex genotyping studies. The Eco Real-Time PCR System delivers low-plex, high-performance genotyping applications using allelic discrimination or High Resolution Melt (HRM) analysis.

Custom iSelect HD BeadChips

iSelect HD BeadChips enable creation of custom genotyping panels containing up to one million markers. Flexible content combined with high plexity and throughput makes iSelect HD BeadChips ideal for association studies, genomic scans, or fine-mapping studies of any species. iSelect HD BeadChips are available in several formats, depending on the number of samples and genetic markers (24 samples, ~3K–90K attempted bead types; 12 samples, ~90K–250K attempted bead types; or 4 samples, ~250K–1M attempted bead types). By requesting an iSelect+ project, researchers can add additional content up to the maximum number of attempted bead types on the format used for the original iSelect project.

Semi-Custom Infinium HD BeadChips

For human studies, the HumanOmniExpress+ BeadChip can be customized to meet your research needs. The fixed content portion of the chip contains proven whole-genome SNP genotyping content, comprising ~733K markers. Expand this industry-leading genomic coverage by adding ~200K additional markers for specialized association studies with the semi-custom option.

Custom GoldenGate Assays

Illumina GoldenGate genotyping is a flexible, pre-optimized assay that uses a discriminatory DNA polymerase and ligase to interrogate 96, or from 384 to 3,072 SNP loci, simultaneously. GoldenGate genotyping can be performed using either the BeadArray or VeraCode technologies.

BeadArray Technology

Designed for the iScan and HiScan™SQ platforms, the GoldenGate Assay using BeadArray technology is a bead-based planar array substrate supporting a highly specific and accurate assay for highly multiplexed (up to 3,072-plex) focused genotyping panels. GoldenGate 12- or 32-sample Universal BeadChips are available in multiple formats, depending on the desired workflow and study design. Optional assay automation supports efficient high-throughput screening.

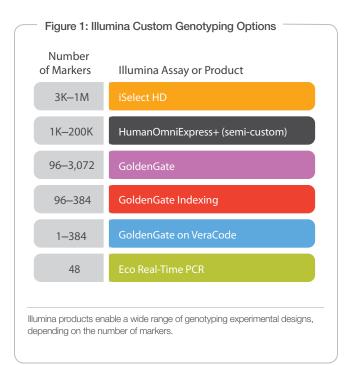


Table 1: Custom Genotyping Assay Details

	Custom Infinium	Semi-Custom Infinium	GoldenGate (BeadArray)	GoldenGate Indexing	GoldenGate (VeraCode)	Real-Time qPCR
Number of Markers Assayed	~3K—1 million	Up to ~900K*	96-3,072	96-384	48, 96,144, 192, or 384	48
Chip Format	4, 12, or 24 samples [†]	8 samples	12 or 32 samples [†]	4, 8, or 16 samples [†]	N/A	N/A
Sample Throughput	Up to 288 samples/day§	Up to 288 samples/day§	Up to 288 samples/day§	Up to 2,000 samples/day§	Up to 288 samples/day	48 samples/run, 384 samples/day
FFPE Compatible			✓		✓	✓
LIMS Support	✓	✓	✓	✓		N/A
Automation Support	✓	✓	✓	✓	✓	N/A

^{*733}K markers base content, 200K additional custom content

GoldenGate Indexing

The GoldenGate Indexing assay enables sample multiplexing, dramatically increasing the number of samples that can be analyzed per run. Samples are tagged with illumiCodes, unique 23-bp single-stranded DNA oligos that identify the DNA samples and loci being interrogated, allowing multiple samples to be pooled. Current plexity ranges are 96- to 384-plex, making GoldenGate Indexing an attractive option for affordable, high-throughout genotyping of many samples.

VeraCode Technology

Illumina's proprietary VeraCode technology provides a robust detection method for multiplex assays requiring high precision, accuracy, and speed. Digital holographic elements are embedded within glass microbeads, creating unique, trackable bead types and providing an ideal surface for genotyping assays. VeraCode assays are conducted completely in solution, providing an easy and consistent workflow.

GoldenGate Genotyping on VeraCode

Combining the flexibility of the GoldenGate Genotyping Assay with VeraCode technology provides a robust and flexible method for genotyping up to 384 SNP loci within a single well of a standard microplate. With consistent performance, flexible assay content and rapid turnaround, performing low- to mid-plex GoldenGate Genotyping using VeraCode technology is ideal for biomarker validation, studies involving large samples and routine testing.

ASPE Assay

The allele-specific primer extension (ASPE) assay is a flexible, easy-to-design, and economical solution for low-multiplex genotyping. Individual assays at 1–72-plex are designed with the web-based VeraCode Assay Designer. The Assay Designer accounts for multiplex compatibility of all primers and universal capture sequences, and then generates a file ready to use for ordering. Since multiplex assays are generated by mixing pools of individual SNP assays, each multiplex assay can be customized by including or excluding specific SNPs.

Eco Real-Time PCR

The Eco Real-Time PCR System delivers highly accurate Real-Time PCR results in an open platform that supports all chemistries. In addition to traditional qPCR applications such as absolute quantification by standard curve and relative quantification using the $\Delta\Delta$ Cq method with support for multiple reference gene normalization, the 48-well Eco system allows for genotyping experiments such as allelic discrimination by endpoint fluorescence or High Resolution Melt (HRM) curve analysis. HRM is the most affordable Real-Time PCR approach to genotyping; even a class IV SNP (A/T) can be detected using DNA binding dyes and monitoring the amplicon fluorescence. The Eco Real-Time PCR system is a cost-efficient and easy-to-use genotyping tool for low throughput experiments.

Illumina Solutions for Genotyping Industry-Leading Data Quality

High call rates and accurate genotype calls are critical for successful studies. Since complex traits often have relatively small gene effects, potential associations can be missed if the assayed SNP in linkage disequilibrium with the SNP of interest has a low call rate, or incorrect genotype call. All Illumina genotyping products have undergone rigorous quality control to ensure strong and reproducible performance.

Easy Design and Ordering

Customers can design custom Infinium or GoldenGate assays to target specific SNP loci of interest using the online Assay Design Tool (ADT). The ADT provides a simple portal for evaluating loci and creating accurate custom genotyping assays. Metrics returned by ADT provide predicted success information, validation status and minor allele frequencies from published studies. For more information, refer to the Technical Notes on designing custom iSelect HD and GoldenGate assays, available on www.illumina.com/literature.

[†]Based on marker density

[§]Assuming one Tecan robot, one Autoloader2, and one iScan reader over an eight hour day

^{**}Assuming one Tecan robot and one BeadXpress reader over an eight hour day

Data Sheet: DNA Analysis

Systems

Illumina's genotyping assays run on four instrument platforms (Figure 2). The iSelect HD, HumanOmniExpress+ and GoldenGate BeadChips are compatible with both the iScan and HiScanSQ platforms. These cutting-edge array scanners feature high-performance lasers and powerful optical systems that deliver rapid scan times and precise assay detection. The HiScanSQ can also perform Illumina sequencing by synthesis chemistry, the world's most widely adopted next-generation sequencing. The GoldenGate Assay using VeraCode technology runs on the BeadXpress® platform, which uses a dual color laser to identify the signal intensity and unique holographic codes embedded in the VeraCode microbeads, providing a streamlined workflow for high-throughput processing of hundreds of samples per day. The Eco Real-Time PCR System combines a proprietary thermal system for unrivaled temperature control and a sensitive optical system to facilitate all four-color multiplex applications.

Software

With the Infinium and GoldenGate Assay workflows, data are processed directly into Illumina's GenomeStudio® software to provide streamlined genotype calling, analysis, and reporting. GenomeStudio software offers an open interface that encourages third-party applications integration to keep pace with evolving downstream data analysis options. The illumina • Connect program leverages this open architecture to support genotyping and copy number analysis. For VeraCode assays, the VeraScan software provides a user-friendly graphical interface for instrument control, enabling users to load assays, perform real-time scans, and view scan data. The software included with the Eco Real-Time PCR system provides instrument control, experimental setup, data collection and analysis in a single package that comes pre-loaded on the included netbook computer.

Automation and Tracking

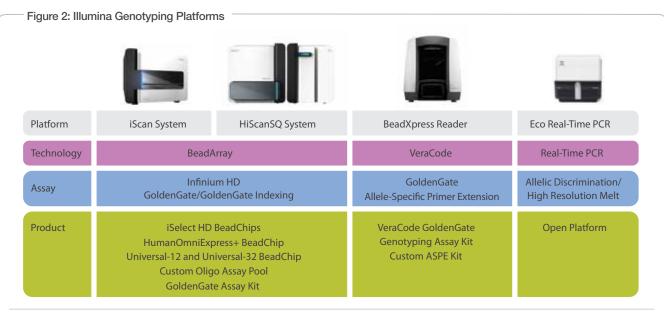
For researchers who require the highest throughput, Illumina offers several options exist to meet your genotyping needs. Robotic automation capabilities can be added to improve throughput for labs processing large numbers of samples. An optional Laboratory Information Management System (LIMS) is available to accurately and efficiently track samples. These systems are designed specifically for the Infinium and GoldenGate workflows, allowing labs to maximize their throughput with a completely integrated microarray processing solution.

Services

Researchers can also choose Illumina's convenient FastTrack Genotyping service to have samples genotyped and data delivered in a format suitable for GWAS or QTL analysis. For more information, visit www.illumina.com/services.

Additional Information

Visit www.illumina.com/dna to learn more about custom, focused, and whole-genome genotyping and CNV analysis solutions.



Illumina's genotyping assays are built on one of three technologies (BeadArray, VeraCode, and Real-Time PCR) and are designed to run on four unique instrument platforms. Perform a range of genotyping assays using different Illumina products (see back page for ordering information).

Ordering Information

Product*	Marker Density	Samples	Minimum Sample Order (Initial/Reorder)	Catalog No.
Custom Infinium DNA Analysis BeadChips	Donoity	Campico	(milai, nooraor)	outulog itol
	3K-90K	24	1,152/288	WG-401-1004
iSelect HD BeadChip	90K-250K	12	1,152/288	WG-400-1004
	250K-1M	4	1,152/288	WG-404-1004
Semi-Custom Infinium DNA Analysis BeadChips			·	
HumanOmniExpress+ BeadChip	200K	8	1,152/288	WG-350-2105
Custom GoldenGate Genotyping (BeadArray Technology)			·	
Custom Oligo Assay Pool (OPA)			custom	GT-17-110
Universal-12 BeadChip	1,536	96	480	GT-221-1008
	96	96	480	GT-221-1112
		576	480	GT-221-1113
		96	480	GT-221-1108
		576	480	GT-221-1109
	768	96	480	GT-221-1104
Universal-32 BeadChip		576	480	GT-221-1105
	1.500	96	480	GT-221-1100
	1,536	576	480	GT-221-1101
	3,072	96	480	GT-221-1120
		576	480	GT-221-1121
	96	1,536	10,752	GT-222-1005
GoldenGate Indexing Assay Kit, Custom	192	768	10,752	GT-222-1004
	384	768	10,752	GT-222-1003
Custom GoldenGate Genotyping (VeraCode Technology)				
	40	480	480	VC-201-0048
	48	96	480	VC-202-0048
	96	480	480	VC-201-0096
		96	480	VC-202-0096
VeraCode GoldenGate Genotyping Assay Kit	144	480	480	VC-201-0144
.s. access dollarisate denotyping notaly int		96	480	VC-202-0144
	196	480	480	VC-201-0196
		96	480	VC-202-0196
	384	480	480	VC-201-0384
		96	480	VC-202-0384
The Eco Real-Time PCR System				
Eco Real-Time PCR System, Netbook computer, HRM module, sample loading dock, USB flash drive, accessories kit (110 V)	1	48	N/A	EC-100-1000

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