Illumina Custom Genotyping Options

The broadest range of marker density and the most flexible content, delivering uncompromised data quality for any genome, in any species.

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, any genome, in any species.
- **Broad Range of Throughput Capabilities**
  Multi-sample formats and automation-compatible, rapid scanning technologies.

Custom Genotyping Options

For studies focused on specific genomic regions of interest, or organisms for which there are no standard products, Illumina offers a broad range of custom genotyping options. Three Illumina technologies provide the foundation to support a wide variety of marker densities (Figure 1). BeadArray™ technology on the iScan, HiScan® and HiScanSQ™ systems supports Custom Infinium (iSelectHD) and GoldenGate BeadChip assays. VeraCode® technology on the BeadXpress® system supports GoldenGate (on VeraCode beads) and allele-specific primer extension (ASPE) assays, and the Eco™ Real-Time PCR System supports qPCR assays.

While each Illumina technology supports multiple genotyping assays, enabling a wide range of experimental designs, researchers can also develop their own custom genotyping panels for a spectrum of applications (Table 1).

Custom Infinium iSelect®HD BeadChips

The Infinium iSelectHD platform accelerates the cycle of innovation to publication, providing access to virtually any marker in the genome to fit research needs. Custom genotyping panels of 3,072 to one million markers per sample can be created in several multi-sample BeadChip formats, depending upon the number of total designed markers. Flexible content design, combined with industry-leading Infinium data quality and versatile throughput formats, make this platform ideal for fine mapping and validation studies, association studies, genomic scans, and marker-assisted breeding for any species.

Custom Infinium Add-On Content

Infinium Add-On Content enables researchers to keep up to date with recent discoveries through the addition of new custom content to existing Custom iSelect, off-the-shelf Infinium products, or Consortia-developed products. Whether content comes from publicly available databases or recent discoveries, Add-On Content allows researchers to combine existing marker sets with new, unique content on a single BeadChip for increased efficiency and cost effectiveness in study design. It also provides core labs and researchers with the flexibility to create highly differentiated service offerings or optimized content

Figure 1: Illumina Genotyping Platforms

Platform	iScan System	HiScanSQ System	BeadXpress Reader	Eco Real-Time PCR System
Technology	BeadArray		VeraCode	Real-Time PCR
Assay	InfiniumHD GoldenGate		GoldenGate ASPE	Allelic Discrimination/ High Resolution Melt (HRM)
Product	iSelectHD BeadChips; Custom and Semi-Custom Add-On; GoldenGate Genotyping Assay Kit		VeraCode GoldenGate Genotyping Assay Kit; Universal Capture Beads	Open Platform

ASPE: Allele-Specific Primer Extension

Illumina 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 Ordering Information).

Table 1: Custom Genotyping Assay Details

	Custom Infinium iSelectHD	Semi-Custom Infinium	GoldenGate (BeadArray)	GoldenGate (VeraCode)	Real-Time qPCR
Number of Markers Assayed	~3K—1 million	Up to 4.8 million*	96—3,072	48, 96,144,192, or 384	48
Format	4, 12, or 24 samples†	4, 8, or 12 samples	12 or 32 samples†	96 samples	48-well plate
Sample Throughput	Up to 288 samples/day§	Up to 288 samples/day§	Up to 288 samples/day§	Up to 288 samples/day**	48 samples/run 384 samples/day
FFPE Compatible			✓	✓	✓
LIMS Support	✓	✓	✓		N/A
Automation Support	✓	✓	✓	✓	N/A

*4.3M markers base content, 500K additional custom content.
†Dependent on the number of custom markers selected.
§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.
FFPE = Formalin-Fixed Paraffin-Embedded LIMS = Laboratory Information Management System

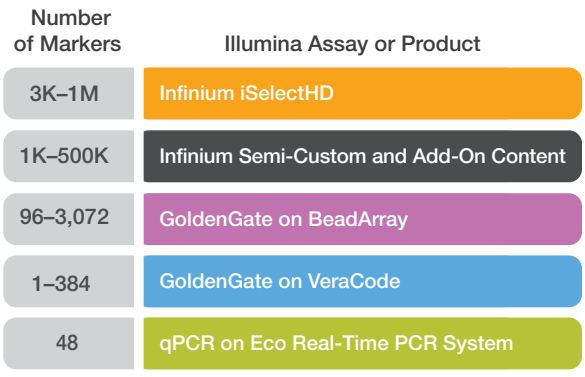
specific to their studies. The amount of Add-On content supported is dependent on the base content and associated BeadChip and will vary for each product (see Ordering Information).

For human and agrigenomics applications, Add-On Content enables researchers to deploy custom content onto an existing Consortium product. Most consortium products use a 24-sample format that allows space for up to 90,000 total attempted beadtypes.

Custom GoldenGate Assays

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. The GoldenGate assay using BeadArray technology is a bead-based planar array substrate supporting a highly specific and accurate assay for highly multiplexed, focused genotyping panels. GoldenGate Universal BeadChips are available in 12 or 32 sample formats, depending on the desired workflow and study design. Optional assay automation supports efficient high-throughput screening.

Figure 2: Illumina Custom Genotyping Options



Illumina products enable a wide range of genotying experimental designs, depending upon the number of markers.

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 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 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.

qPCR Technology

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 ΔΔCq 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 Probe Design

Researchers can design probes for SNPs or indels for any species and any technology using the Assay Design Tool (ADT), available through MyIllumina, Illumina Technical Support, or FastTrack Services. The ADT scores each submitted design to provide a relative confidence level that a probe will successfully query a particular SNP or indel based on the surrounding sequence. The output file finalized by content contributors can then be submitted to the Illumina orders group to place the final order.

User-Friendly Software for Streamlined Data Analysis

With the Infinium and GoldenGate Assay workflows, data are processed directly in 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.

Efficient 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.

Custom 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.

Ordering Information—Semi-Custom Infinium DNA Analysis Products (Agrigenomics)

Product	Marker Density	Add-On Content Amount Supported	Samples Per Chip	Minimum Sample Order	Samples	Catalog No. for Add-On Product	Catalog No. for Standard Product
BovineHD BeadChip	777,962	85,000	8	48	48	WG-450-1011	WG-450-1002
					96	WG-450-1012	WG-450-1003
					384	WG-450-1013	WG-450-1004
BovineSNP50 BeadChip	54,609	31,000	24	48	48	WG-450-2006	WG-450-2001
					288	WG-450-2007	WG-450-2002
					1152	WG-450-2008	WG-450-2003
BovineLD BeadChip	6,909	80,000	24	48	48	WG-450-1010	WG-450-1007
					288	WG-450-1011	WG-450-1008
					1152	WG-450-1012	WG-450-1009
CanineHD BeadChip	173,662	58,000	12	48	48	WG-440-1005	WG-440-1001
					288	WG-440-1006	WG-440-1002
					1152	WG-440-1007	WG-440-1003
MaizeSNP50 BeadChip	56,110	29,000	24	48	48	WG-500-1011	WG-500-1001
					288	WG-500-1012	WG-500-1002
					1152	WG-500-1013	WG-500-1003
OvineSNP50 BeadChip	52,241	183,000	12	48	48	WG-420-1011	WG-420-1001
					288	WG-420-1012	WG-420-1002
					1152	WG-420-1013	WG-420-1003
PorcineSNP60 BeadChip*	62,163	193,000	12	48	48	WG-410-1011	WG-410-1001
					288	WG-410-1012	WG-410-1002
					1152	WG-410-1013	WG-410-1003

*This product is transitioning to a 24-sample BeadChip, supporting 25K ABT of add-on content.

Ordering Information—Semi-Custom Infinium DNA Analysis Products (Human)

Product	Marker Density	Add-On Content Amount Supported	Samples Per Chip	Minimum Sample Order	Samples	Catalog No. for Add-On Product	Catalog No. for Standard Product
Cancer Beadchip*	> 200,000	25,000	12	48	48	WG-310-1011	WG-354-1002
					288	WG-310-1012	WG-354-1003
					1152	WG-310-1013	WG-354-1004
Cardio-Metabo BeadChip*	> 200,000	25,000	12	48	48	WG-310-1005	WG-310-1001
					288	WG-310-1006	WG-310-1002
					1152	WG-310-1007	WG-310-1003
HumanExome BeadChip	> 245,000	30,000	12	48	48	WG-353-1005	WG-353-1001
					288	WG-353-1006	WG-353-1002
					1152	WG-353-1007	WG-353-1003
Immuno BeadChip	> 200,000	25,000	12	48	48	WG-352-1011	WG-352-1001
					288	WG-352-1012	WG-352-1002
					1152	WG-352-1013	WG-352-1003
HumanOmniExpress BeadChip	> 700,000	200,000	12	48	48	WG-350-2202	WG-311-1120
					288	WG-350-2203	WG-311-1121
					1152	WG-350-2204	WG-311-1122
HumanOmniExpressExome BeadChip	> 900,000	30,000	8	16	16	WG-353-2210	WG-350-2206
					48	WG-353-2211	WG-350-2207
					96	WG-353-2212	WG-350-2208
					384	WG-353-2213	WG-350-2209
					FTS	FT-353-1004	FT-353-1003
Omni2.5–8 BeadChip	> 250,000	200,000	8	8	16	WG-311-2537	WG-311-2522
					48	WG-311-2538	WG-311-2512
					96	WG-311-2539	WG-311-2513
					384	WG-311-2540	WG-311-2514
					FTS	FT-311-1008	FT-311-1003
Omni5 BeadChip	~ 4.3 million	500,000	4	16	16	WG-311-5005	WG-311-5001
					48	WG-311-5006	WG-311-5002
					96	WG-311-5007	WG-311-5003
					384	WG-311-5008	WG-311-5004
					FTS	NA	FT-311-1003
Omni5Exome BeadChip	> 4.5 million	250,000	4	16	16	WG-311-5014	WG-311-5010
					48	WG-311-5015	WG-311-5011
					96	WG-311-5016	WG-311-5012
					384	WG-311-5017	WG-311-5013
					FTS	FT-311-1007	FT-311-1006

*These products are controlled sell, consortia products. Permission from the consoritum is required prior to purchase.
FastTrack Services (FTS)

Ordering Information—Custom Infinium DNA Analysis Products

Product	Marker Density	Add-On Content Amount Supported	Samples Per Chip	Minimum Sample Order	Samples	Catalog No. for Add-On Product	Catalog No. for Standard Product
iSelect 3–90k BeadChip	3–90k	Up to 90,000*	24	1152	24	WG-401-1014	WG-401-1004
					48		WG-401-1001
					288		WG-401-1002
					1152		WG-401-1003
iSelect 90–250k BeadChip	90–250k	Up to 250,000*	12		12	WG-400-1014	WG-400-1004
					48		WG-400-1001
					288		WG-400-1002
					1152		WG-400-1003
iSelect 250–1M BeadChip	250–1M	Up to 1M*	4		4	WG-404-1014	WG-404-1004
					48		WG-401-1001
					96		WG-401-1002
					384		WG-401-1003

*Amount of Add-On content supported is the maximum amount *minus* the amount of initial iSelect product.

Ordering Information—GoldenGate and VeraCode DNA Analysis Products*

Product	Marker Density	Samples Per Chip	Minimum Sample Order	Samples	Catalog No. for Standard Product
GoldenGate BeadChip	1536	32	480	96	GT-221-1100
				576	GT-221-1101
	768	32		96	GT-221-1104
				576	GT-221-1105
	384	32		96	GT-221-1108
				576	GT-221-1109
	96	32		96	GT-221-1112
				576	GT-221-1113
	3072	32		96	GT-221-1120
				576	GT-221-1121
	1536	12		96	GT-221-1116
				576	GT-221-1117
VeraCode	48	96	480	480	VC-201-0048
				96	VC-202-0048
	96	96		480	VC-201-0096
				96	VC-202-0096
	144	96		480	VC-201-0144
				96	VC-202-0144
	192	96		480	VC-201-0192
				96	VC-202-0192
	384	96		480	VC-201-0384
				96	VC-202-0384

* GoldenGate and VeraCode do not support Add-on Content

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