Illumina Custom Infinium® Genotyping

Illumina Custom Infinium Genotyping BeadChips and the Infinium Assay deliver high-quality data and enable whole-genome genotyping with the flexibility to add up to 121,600 additional custom SNP assays.

Highlights of Custom Infinium Genotyping Products

- **High-Quality Data:**
  Infinium Assays provide high reproducibility and call rates
- **Custom SNP Selection:**
  Choose up to 60,800 SNPs for iSelect Infinium content in a 12-sample format
- **Add SNPs to Trusted Content:**
  Up to 60,800 additional SNPs to the HumanHap300-Duo+ BeadChip; up to 121,600 additional SNPs to the HumanHap550+ BeadChip
- **Worry-Free Lab Workflow:**
  Run the PCR-free assay with optional automation and LIMS tracking

Figure 1: Illumina Custom Infinium Genotyping Beadchips

HumanHap300-Duo+ HumanHap550+ iSelect

The two-sample HumanHap300-Duo+ allows the addition of up to 60,800 custom SNPs to the content on the HumanHap300 Genotyping BeadChip. The Human-Hap550+ Genotyping BeadChip interrogates >683,000 SNPs with the flexibility to add up to 121,600 additional SNPs. The iSelect Infinium Custom Genotyping BeadChip allows simultaneous 12-sample interrogation of up to 60,800 SNPs per sample.

Introduction

Illumina offers several Custom Infinium Genotyping solutions, with arrays permitting the analysis of a single sample to arrays allowing simultaneous analysis of up to 12 samples. The iSelect™ Genotyping BeadChip gives scientists the greatest degree of flexibility in both content and format. Custom Infinium Genotyping provides a streamlined solution for performing fine-mapping studies and other large-scale custom genotyping applications. The iSelect and HumanHap300-Duo+ Genotyping BeadChips support the analysis of multiple samples on a single array and enable scientists to generate the highest quality genotyping data in the industry with a simpler workflow and in significantly less time.

Custom SNP content can be added to Illumina Genotyping BeadChips. The HumanHap300-Duo+, which contains the same SNPs as the HumanHap300-Duo BeadChip, allows the ability to add up to 60,800 custom SNPs per sample for a total of >758,000 SNPs per BeadChip and >379,000 SNPs per sample. This format allows the analysis of two samples on a single array, which makes it an ideal platform for DNA copy number analysis.

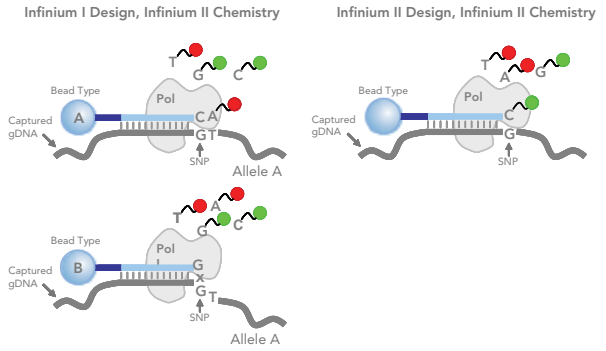
The single-sample Human-Hap550+ Genotyping BeadChip uses the content of the current HumanHap550 Genotyping BeadChip and allows researchers to add up to 121,600 custom SNPs for a total of >683,000 SNPs.

Applications

Large-scale, whole-genome disease association studies make up a major portion of current genotyping research. These studies are designed to facilitate the discovery of regions in the genome associated with pre-defined phenotypes or diseases.

The HumanHap300-Duo+ and the HumanHap550+ let researchers increase genome coverage in a specific region of the genome or in the genome of specific populations (see the *HumanHap650Y Genotyping BeadChip* data sheet for a product description for the Yoruban population1). The iSelect Genotyping BeadChip gives researchers the ability to add up to 60,800 attempted custom SNPs to focus on specific regions of the genome (candidate regions, target genes, protein coding SNPs). The iSelect Genotyping BeadChip is the Illumina custom solution for fine-mapping applications for follow-up linkage, association, or Loss of Heterozygosity (LOH)/DNA copy number studies.

Figure 2: Assay Design for Iselect Whole-Genome Genotyping



Infinium II Assay chemistry is used for all iSelect Custom Infinium Assays with either Infinium I or Infinium II probe design. Infinium I probe design includes two bead types per SNP locus. Infinium I probe design requires just one bead type per locus. Infinium I probes end at the queried SNP base. Infinium II probes end at the base proceeding the queried SNP. This method allows unlimited access for SNP interrogation.

As the genotyping field moves towards the study of a broader set of organisms, Custom Infinium Genotyping allows researchers to investigate new genetic models. Geneticists have been working to identify target SNPs for organisms such as bovine, canine, Arabidopsis, zebrafish, and Drosophila. The Custom Infinium Genotyping program affords researchers the ability to study SNPs in these organisms on the Illumina platform using the flexibility of the Infinium Assay for unrestricted SNP selection.

Assay Design for Iselect Infinium Custom Genotyping

The iSelect Infinium BeadChip uses standard Infinium II Assay chemistry (two fluorophore colors)[2,3] and has been adapted to function either with Infinium I or Infinium II probe designs (Figure 2, Table 1). This innovation makes it possible for researchers to target any SNP in the genome without restriction.

Iselect infinium content ordering and design process

1. To order Custom Infinium Genotyping products, send preliminary SNP files to techsupport@illumina.com. Accepted file formats include gene symbols (e.g., HUGO gene names or RefSeq accession numbers), RS SNP IDs, regions (coordinates or marker to marker), and sequences. Illumina scientists will review the requested assays and return SNP Score files to you.
2. Select desired SNPs from the returned list of scored SNPs.
3. Repeat steps 1 and 2 as necessary.
4. Select the final SNP list for Custom Infinium BeadChips.
5. Send PO#, quote, and final SNP list to orders@illumina.com. Illumina will send you an order confirmation. The first product shipment will arrive in approximately eight to ten weeks.

Summary

The ability to choose custom SNPs adds valuable additional information to both the HumanHap300-Duo+ and the HumanHap550+ BeadChips. The iSelect and HumanHap300-Duo+ Genotyping BeadChips allow you to simultaneously interrogate multiple samples. The combination of Illumina proprietary assay technologies and flexible content deployment delivers the most comprehensive solution available for genotyping. In addition, the optional automation and Laboratory information Management System (LIMS) solutions lower costs by eliminating errors associated with manual processing. Illumina genotyping products, with a combination of standard and custom content, can be accessed via Illumina FastTrack Genotyping Services or they can be purchased separately for use with one's own Illumina BeadStation. Illumina solutions provide industry-leading levels of accuracy, flexibility, and affordability.

Additional Information

Visit our website or contact us at the address below to learn more about Custom Infinium Genotyping or other Illumina products or services.

REFERENCES

1. HumanHap650Y Genotyping BeadChip Data Sheet. Illumina Pub. No. 370-2006-019.
2. Steemers FJ, Chang W, Lee G, Barker DL, Shen R, Gunderson KL (2006) Whole-genome genotyping with the single-base extension assay. Nat Methods 3(1): 31-33.
3. Gunderson KL, Steemers FJ, Ren H, Ng P, Zhou L, et al. (2006) Whole-genome genotyping. Methods Enzymol 410: 359-76.

Table 1: Product Specifications for the Infinium Custom Genotyping Program

PARAMETER	SPECIFICATION	NOTES
Probe Design	Infinium I or Infinium II	Unlimited SNP selection
Assay Chemistry	Infinium II	Both Infinium I and II probe designs use Infinium II Assay chemistry
% Accessibility to the Genome	99.9%	Nearly any SNP can be targeted
Call Rate	98.5–99.5%	
Reproducibility	>99.5%	
Heritability	>99.5%	
Number of Samples per BeadChip iSelect HumanHap300-Duo+ HumanHap550+	 12 2 1	 Uses IntelliHyb™ seal technology Uses IntelliHyb seal technology Uses standard single-sample seal
Range of Custom Bead Types Attempted iSelect HumanHap300-Duo+ HumanHap550+	 7,600–60,800 7,600–60,800 7,600–121,600	 Two custom bead pools (60,800 each) can be added to the HumanHap550+
Software	BeadStudio	Full suite of analysis options for genotyping copy number analysis
LIMS	Yes	Enables positive sample tracking
Automation iSelect HumanHap300-Duo+ HumanHap550+	Up to 24 BeadChips per X-Stain run (288 samples) (48 samples) (24 samples)	Equivalent capability to standard Infinium products
Packaging iSelect HumanHap300-Duo+ HumanHap550+	 4-, 24-, or 96-BeadChip packs 4-, 24-, or 48-BeadChip packs 24- or 96-BeadChip packs	 Packaging to meet your throughput requirements
Shelf Life	Six months	

Ordering Information

CATALOG NO.	PRODUCT	DESCRIPTION
WG-35-311	HumanHap300-Duo+ Custom Genotyping BeadChip	Each HumanHap300-Duo+ Genotyping BeadChip can process two samples and genotype >318,000 standard loci plus an additional 7,500 to 60,800 custom SNP loci.
WG-35-521	HumanHap550+ Custom Genotyping BeadChip	Each HumanHap550+ Genotyping BeadChip contains >561,000 standard loci and an additional 7,500 to 121,600 custom SNP loci.
WG-15-303	iSelect Infinium Automation Option Package (24)	iSelect Infinium Automation Option for processing 24 BeadChips at a time includes robot BeadChip alignment fixtures (4), BeadChip Hyb Chamber inserts (24), a multi-sample BeadChip alignment fixture (1), robot automation software, and documentation.
WG-15-302	iSelect Infinium Manual Option Package (8)	iSelect Infinium Manual Option for processing 8 BeadChips at a time includes BeadChip Hyb Chamber inserts (8), multi-sample BeadChip alignment fixture (1), and documentation.
WG-15-301	iSelect Infinium Hyb Chamber Inserts (8)	iSelect Infinium Hyb Chamber inserts (8) for increased sample processing.

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