

An Overview of Illumina's DNA Analysis Products – From Genotyping to CNV

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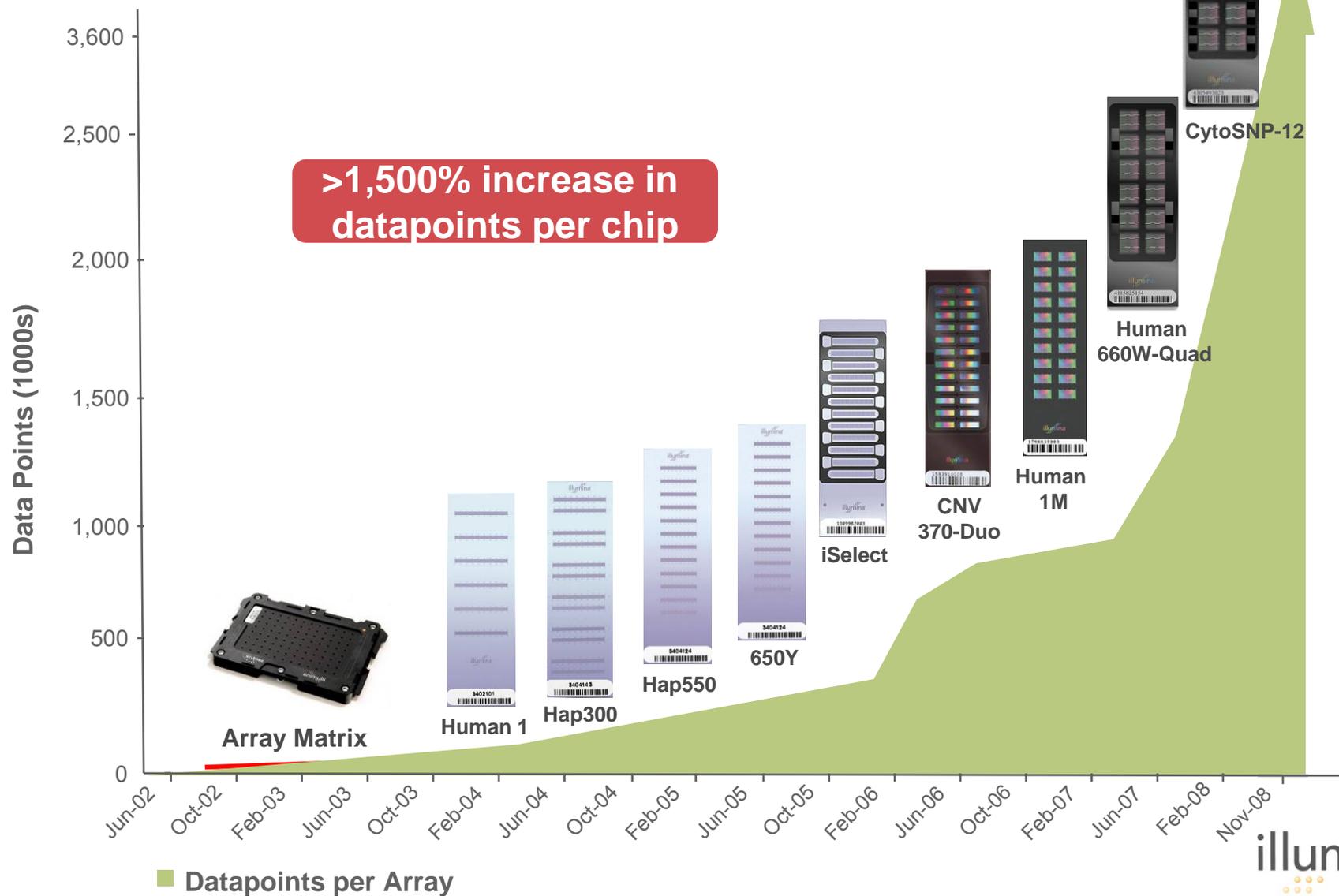


Most Complete Genomics Assay Portfolio

APPLICATION	Sequence-Based Discovery	Whole-Genome	Targeted Analysis		Validation, Screening & Standard Tests
TECHNOLOGY	Sequencing	BeadArray	BeadArray		VeraCode
PLATFORM	 Genome Analyzer	 iScan	 iScan		 BeadXpress
ASSAY	Sequencing by Synthesis	Infinium Assay Direct Hyb	Infinium Assay GoldenGate & DASL Assay		GoldenGate & DASL Assay
MARKER DENSITY	1base to >1GB	240K to 1M	3K to 200K 96 to 1536 Custom Standard		1 to 384-plex Custom
PRODUCT		Human1M-Duo Human660W-Quad HumanCytoSNP-12	Infinium iSelect Genotyping Panels 6K-200K GoldenGate Genotyping Panels 96-1536	HumanCVD BovineSNP50 EquineSNP50 CanineSNP20 PorcineSNP50 OvineSNP50	VeraCode GoldenGate 96- & 384-plex

Forward Compatibility

From 1,536 to 3.6 Million Data Points per Chip in 6 Years

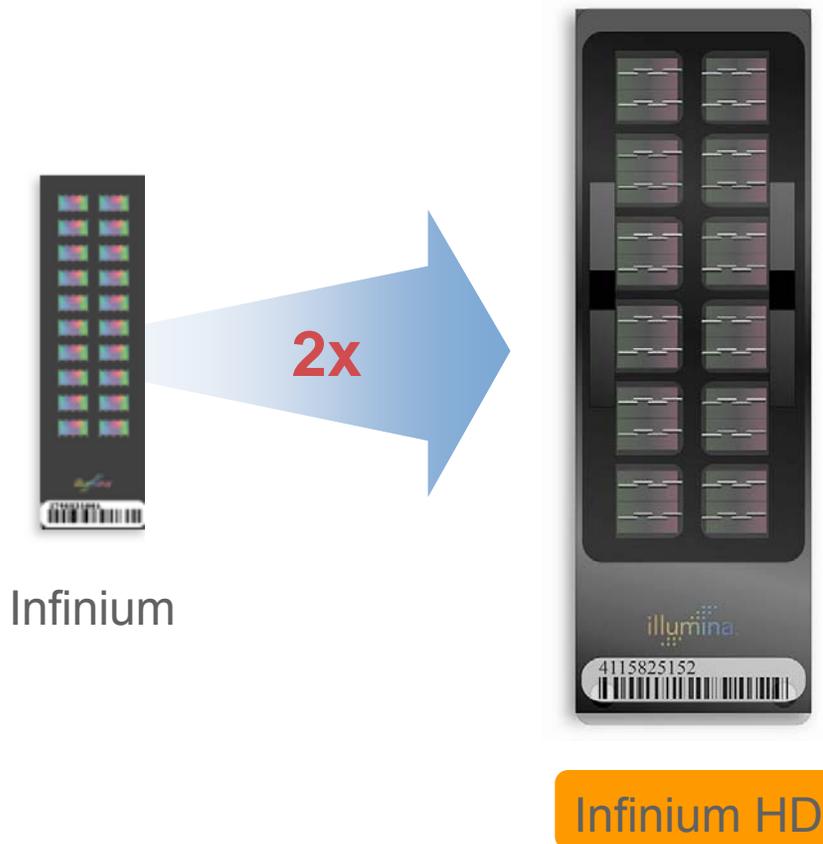




Infinium Whole-Genome Genotyping Solutions

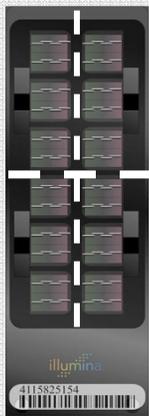
The Evolution of the Infinium HD Product Line

More Features, Higher Quality, Price Flexibility

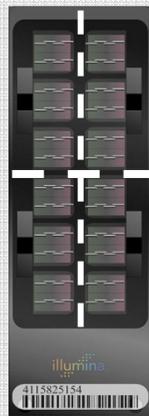


- Improved Assay Chemistry
- New Substrate Density
- Higher Quality Data
- New Decoding Process
- New Decoding Instrument
- Increased Capacity
- Flexible Price Structure

Infinium HD Product Line – Whole-Genome GT



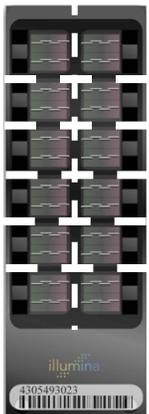
HumanCNV370-Quad



Human610-Quad

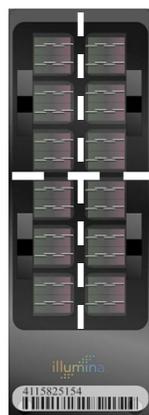


Human1M-Duo



HumanCytoSNP-12

Low-Cost GT



Human660W-Quad

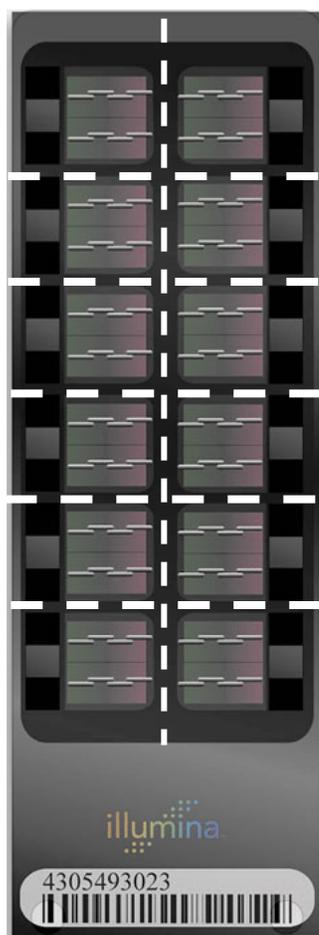
Common SNPs & CNVs



Human1M-Duo

Premium GT

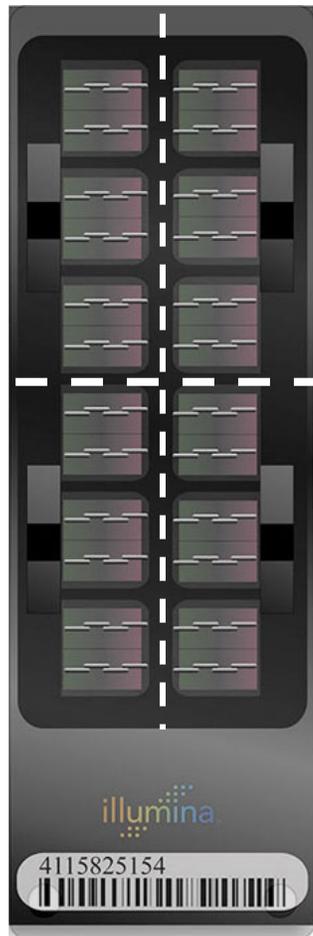
Infinium HD HumanCytoSNP-12 BeadChip: newest SNP chip for cost-effective GWAS and Molecular Cytogenetics



HumanCytoSNP-12

- ~300k genetic variations per sample
- 12 samples per BeadChip
- Coverage of all known cytogenetic regions
- Comprehensive genomic coverage
 - CEU (72% at $r^2=0.8$)
 - CHB/JPT (74% at $r^2=0.8$)
- Infinium HD Assay
- Lowest DNA input in the industry (200ng)

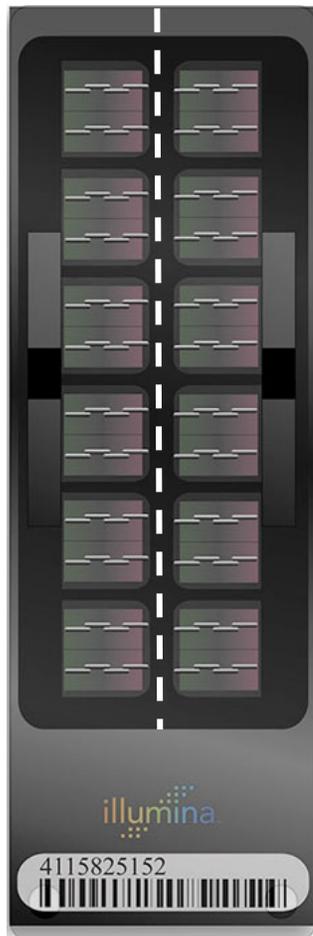
Infinium HD Human660W-Quad BeadChip



Human660W-Quad

- ~660K genetic variations per sample
- 4 Samples per Chip
- HumanHap550-2 SNP content plus 110,000 new common CNV probes (6000 regions)
- Developed with Wellcome Trust Sanger, Sick Kids Hospital Toronto, Brigham and Women's Boston
- Comprehensive genomic coverage
 - CEU (90% at $r^2=0.8$)
 - CHB/JPT (86% at $r^2=0.8$)
- Infinium HD Assay
- Lowest DNA input in the industry (200ng)

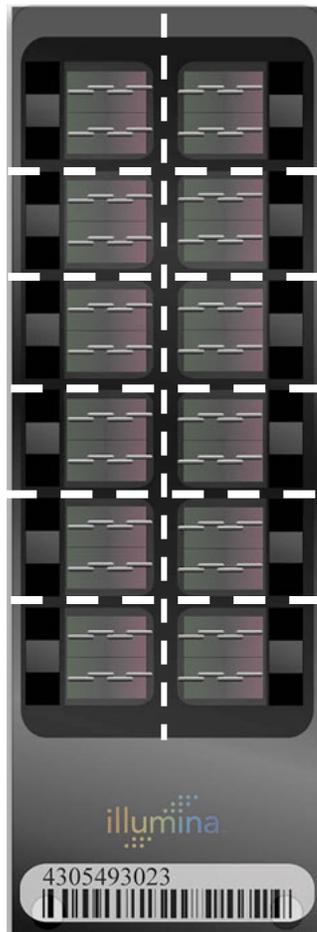
Infinium HD Human1M-Duo BeadChip



Human1M-Duo

- >1 million genetic variations per sample
- 2 Samples per Chip
- Additional high-value content:
 - gene centric SNPs
 - CNV probes
 - SNPs associated with diseases
 - Additional TagSNPs
- Industry-best genomic coverage
 - CEU (96% at $r^2=0.8$)
 - CHB/JPT (93% at $r^2=0.8$)
 - YRI (75% at $r^2=0.8$)
- CNV Discovery – lowest number of large gaps
- Infinium HD Assay
- Low DNA input (400ng)

Infinium HD HumanCNV100W BeadChip



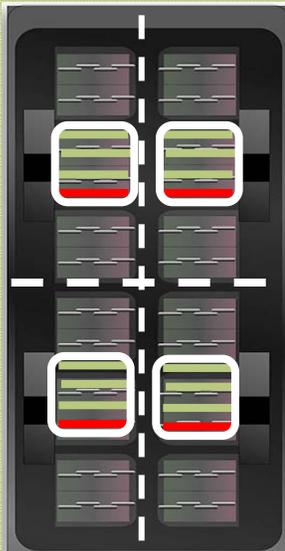
HumanCNV100W

- 12 samples per BeadChip
- ~100k genetic variations per sample
- Targets over 6,000 common CNV regions identified by Wellcome Trust Sanger, Sick Kids Hospital Toronto, Brigham and Women's Boston
 - Regions identified in high-density screen of HapMap samples
 - Average CNV size: 15kb
 - Average markers per region: 16
- Infinium HD Assay
- Lowest DNA input in the industry (200ng)

Infinium Human Semi-Custom BeadChip Offerings

Hap550-Quad+

4 x 60,800 SNPs



illumina

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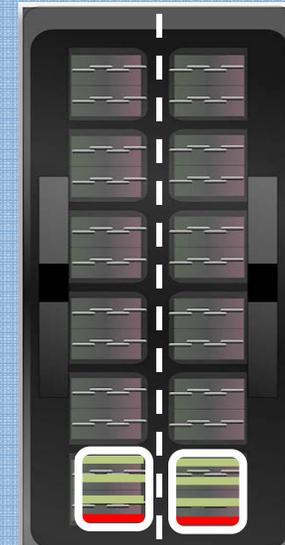
Applications

- Focused GWAS
 - CNV
 - Cardiovascular
 - MHC
- Consumer Genomics

- 3,000 to 60,800 attempted BTs
- >80% BeadType Success Rate
- 250ng input dna for 550-Quad+
- 400ng input dna for 1M-Duo+

Human1M-Duo+

2 x 60,800 SNPs



illumina

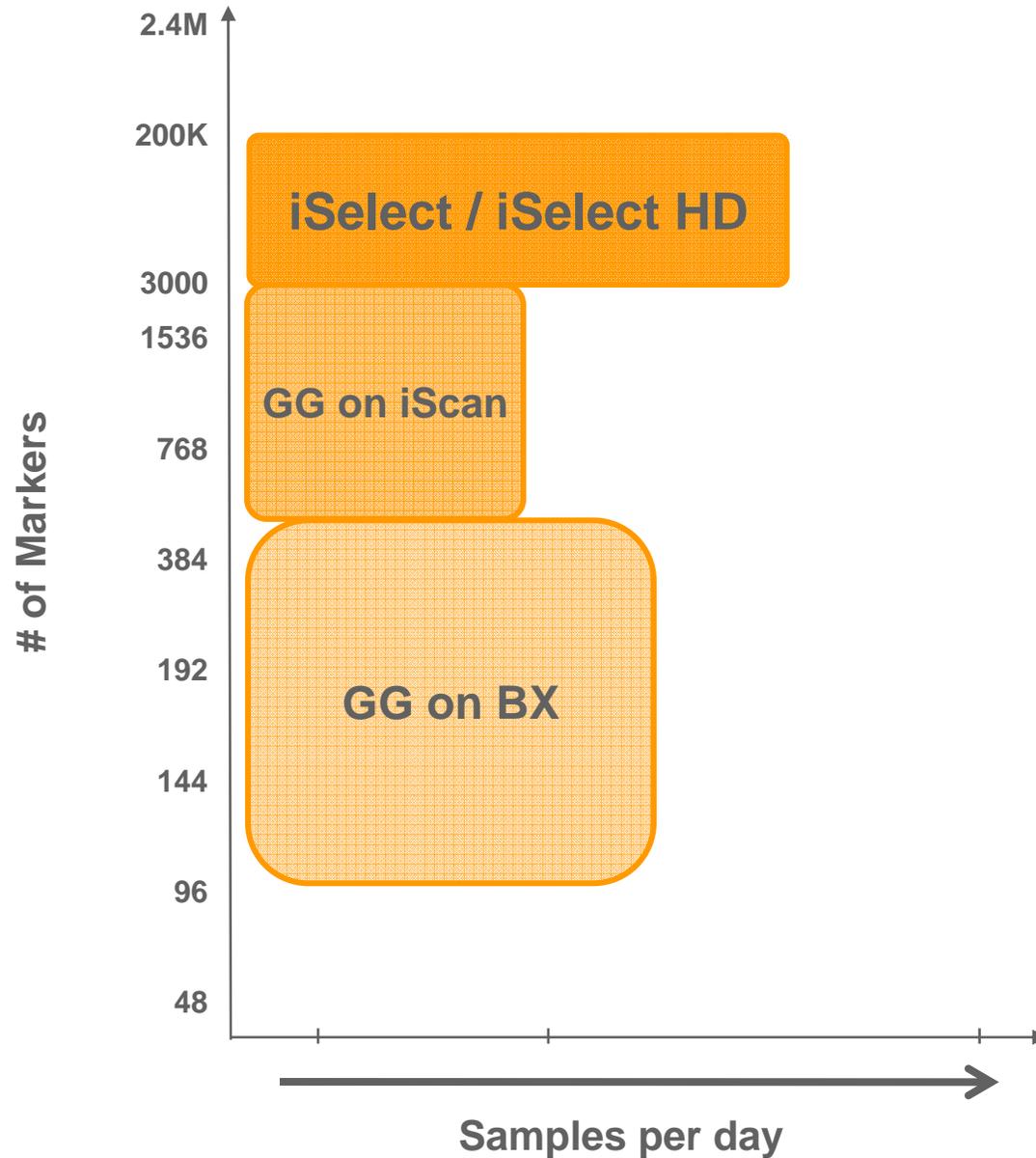
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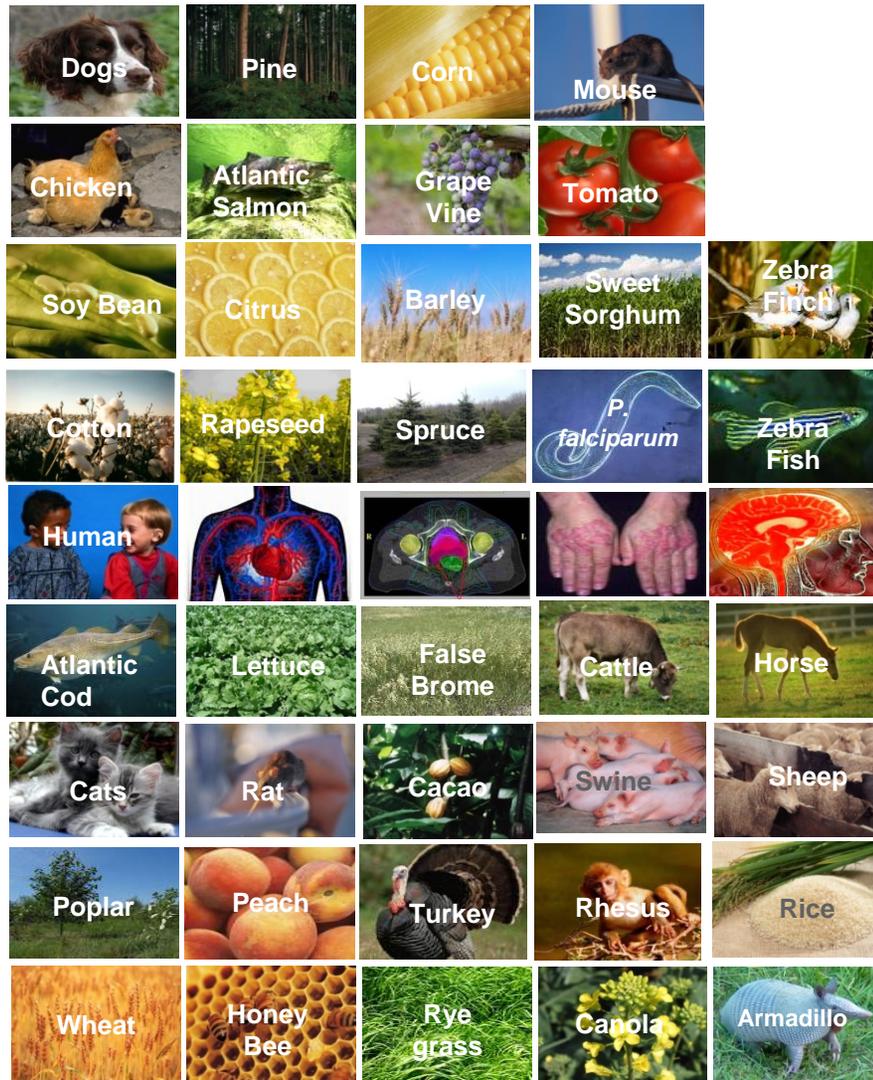


Custom Genotyping Solutions

Custom Genotyping Plexity Ranges



Focused and Custom Genotyping: Infinium and GoldenGate



Applications

- Targeted follow-up studies after GWAS
- Targeted disease panels
- Targeted rare variant panels
- SNP Discovery/validation in non-humans
- Agriculture fingerprinting panels
- Commercial Agriculture screening panels
- Consumer Genomics

The Right Content Lets you Capture More Biology



Infinium iSelect® Custom Genotyping

3,000

6,000

60,800

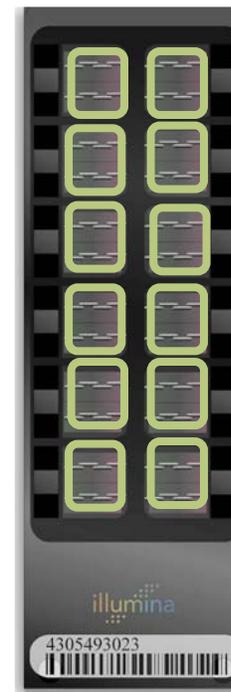
200,000

iSelect



- Target any variant, any species
- Low dna input (200ng)
- >80% BeadType Success Rate
- 12-samples per chip

iSelect HD



Expanding the Market with Focused Content

Leveraging the 12 Sample iSelect Platform to Deliver Innovative Content



● HumanCVD

- Candidate diseases include myocardial infarction, heart failure, stroke, insulin resistance, metabolic disorders, dyslipidemia and inflammation



● BovineSNP50

- Collaboration with University of Missouri, University of Alberta, USDA-ARS US Meat Animal Research Center



● CanineSNP20

- 22,000 validated SNP probes derived from the CamFam2.0 assembly



● EquineSNP50

- Developed in collaboration with: International Equine Genome Mapping Workshop and the Morris Animal Foundation's Equine Genome Consortium



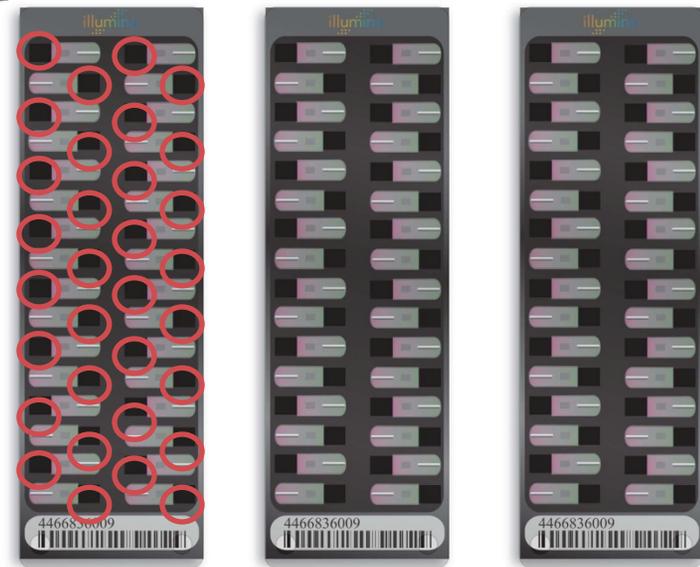
● PorcineSNP50



● OvineSNP50

Universal-32 BeadChip for the GoldenGate Assay

- Designed for the flexible, pre-optimized GoldenGate Assay
 - Available for 96 to 1536 SNPs
- Compatible with the iScan and BeadArray
 - Up to 128 samples per iScan run
 - 96 samples scan in less than 30 min.
- Designed for 8 channel manual loading
- Autoloader capability
- Ideal for streaming with Infinium processes
- Automation and LIMS support coming in 2009



GenomeStudio Overview



- GenomeStudio™ analyzes data generated from all of Illumina's technologies
- Simple, intuitive project creation and data management tools
- Ability to combine data from more than one assay type within same project
 - Differential expression levels in mRNA versus microRNA
 - Gene regulation (DNA Methylation) versus differential expression (mRNA)
- Can also combine data from different versions of same product for research continuity
 - E.g. Human1M + Human1M-Duo



BeadArray Instrumentation



Sequencing Extension



iScan



**iScan
Sequencing
Module**



iScan Sequencing Module* Product Description

- **Add-on to enable sequencing on the iScan**

- Uses lasers and optics of iScan for imaging of flow cell
- Fluidics module holding sequencing reagents, pumps and reagent chiller

- **Sequencing specs**

- Throughput: 0.3 GB per day, up to 3GB per run
- Data density: > 32Million clusters per run, > 4 million clusters per lane
- Flexible read length: from short single reads to 2x50pb reads
- 8 lanes, 1-12 samples per lane
- Includes paired-end capability
- Multiplexing enabled

- **Applications supported**

- smallRNA, ChIP-Seq & mRNA seq
- Targeted re-sequencing & sequencing of small genomes



**Available Fall 2009*



VeraCode®



Illumina's Solution for Diverse Applications

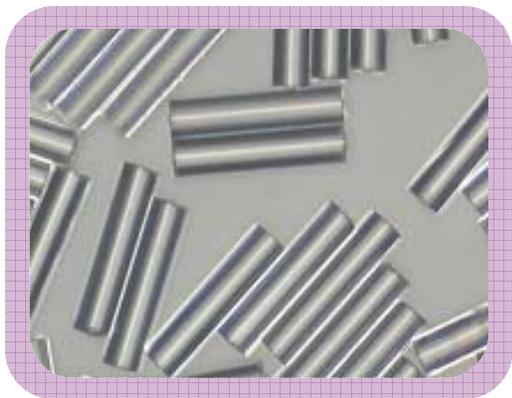
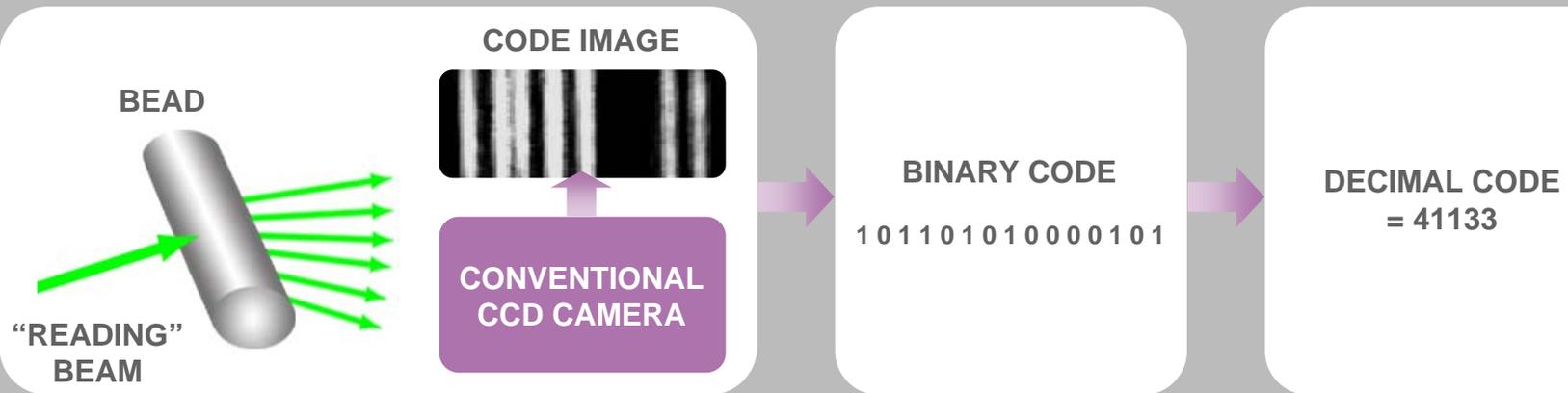
VeraCode® Technology



- Cylindrical glass microbeads
- 240 μm length x 28 μm diameter

VeraCode Technology

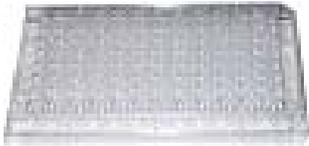
CODE GENERATED BY CREATING EMBEDDED HOLOGRAPHIC DIFFRACTIVE ELEMENTS



- High Density Codes Easily Imprinted (24 bit)
- Virtually Unlimited Unique Bead Codes Available – 626 unique codes used in launched products
- Glass Surface of Beads Ideal for Bioassays – numerous surface chemistries available
- Specific panels created easily with addition or removal of specific bead types

Easily adaptable to changing content requirements

**VeraCode Beads
Pre-plated into standard
product**



**Beads Pooled in
96-well Plates
(96 & 384-plex/well)**



**Individual Tubes of
VeraCode Beads**



**Customized panels
created by pooling
different tubes of beads**

Comprehensive VeraCode product portfolio

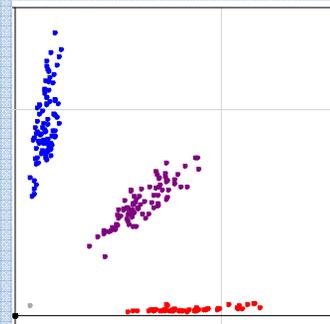
- VeraCode Carboxyl Bead Sets
- VeraCode Universal Capture Bead Sets
- Custom GoldenGate Genotyping, 48, 96, 144, 192, & 384-plex
- DASL Custom Gene Expression, 32 to 384-plex
- Custom GoldenGate Methylation, 48 to 384-plex



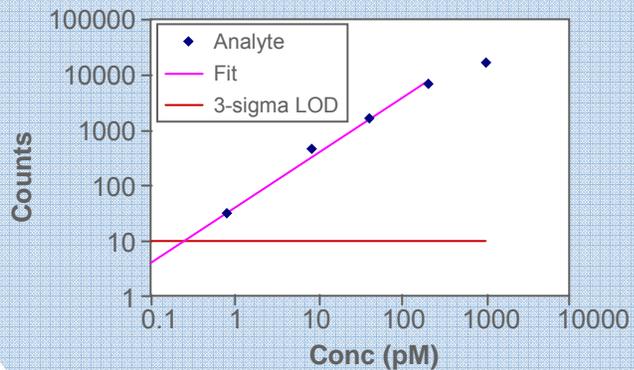
Analyzer for Multiple Applications



GENOTYPING



GENE REGULATION



PROTEIN EXPRESSION

