

Genotyping Microarrays: Leading the Way in Genomic Research

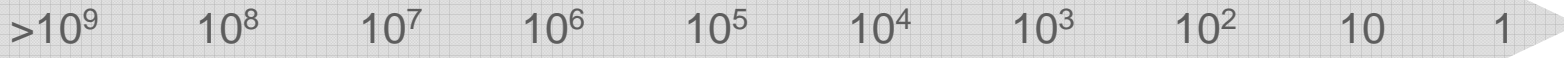
Jasmine Pritchard
Product Manager
Illumina DNA Analysis Products

Addressing Breadth of Customer Needs

Discovery

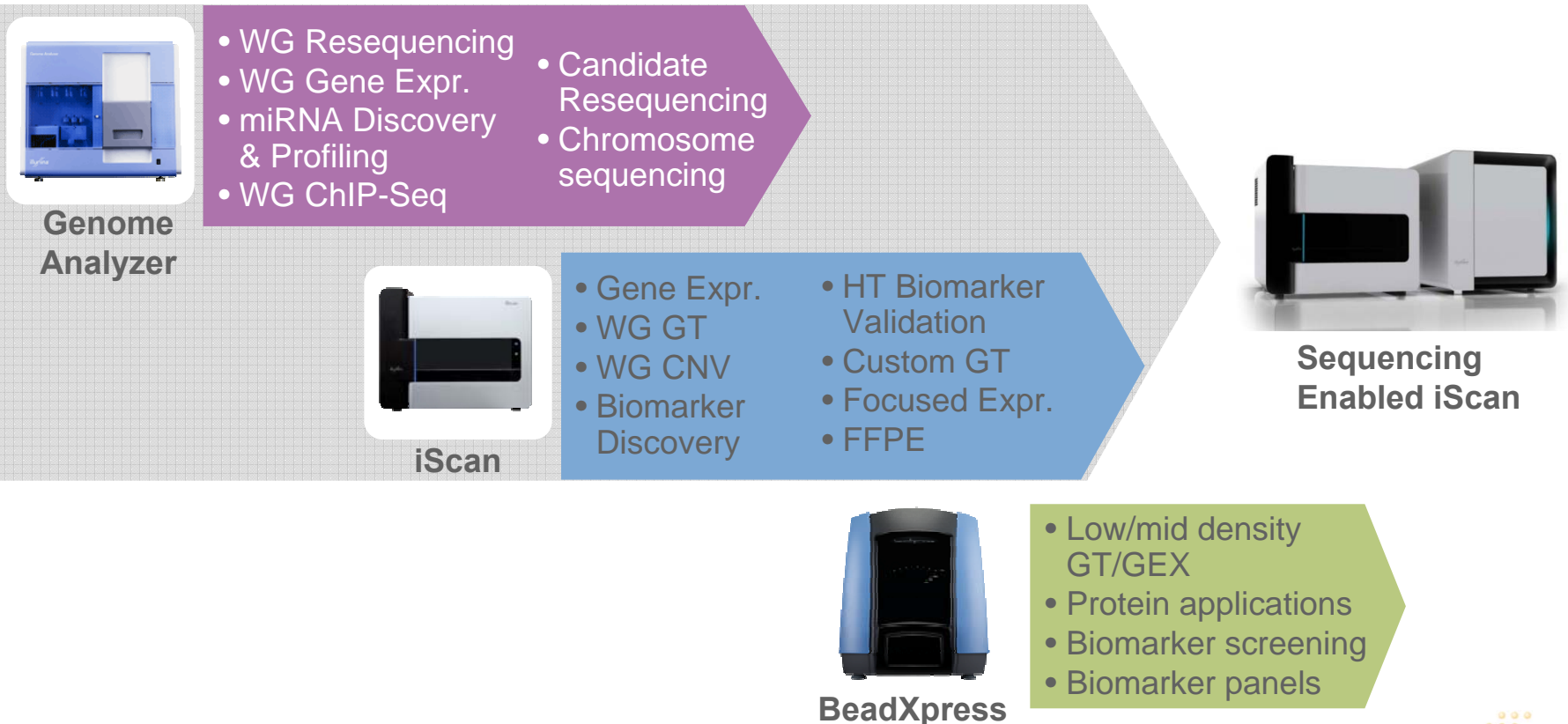
Focused Research/Validation

Screening

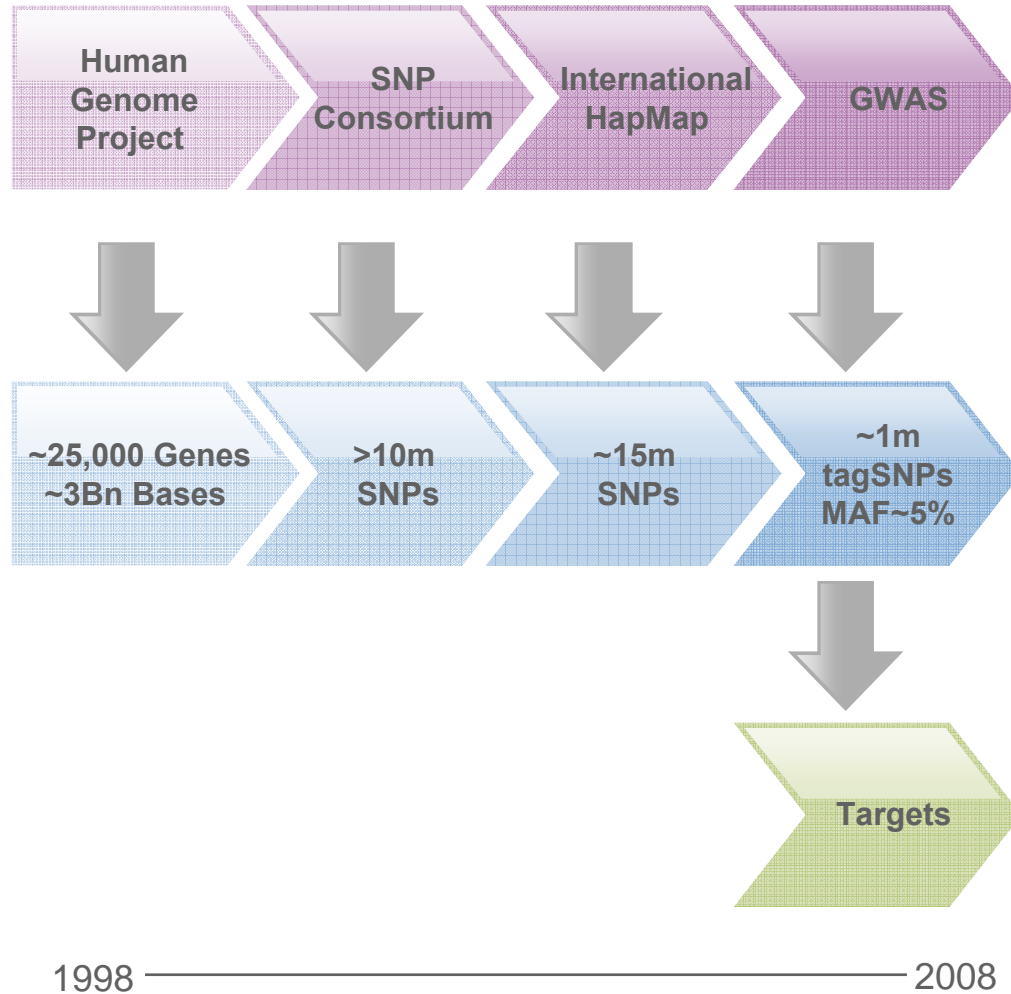


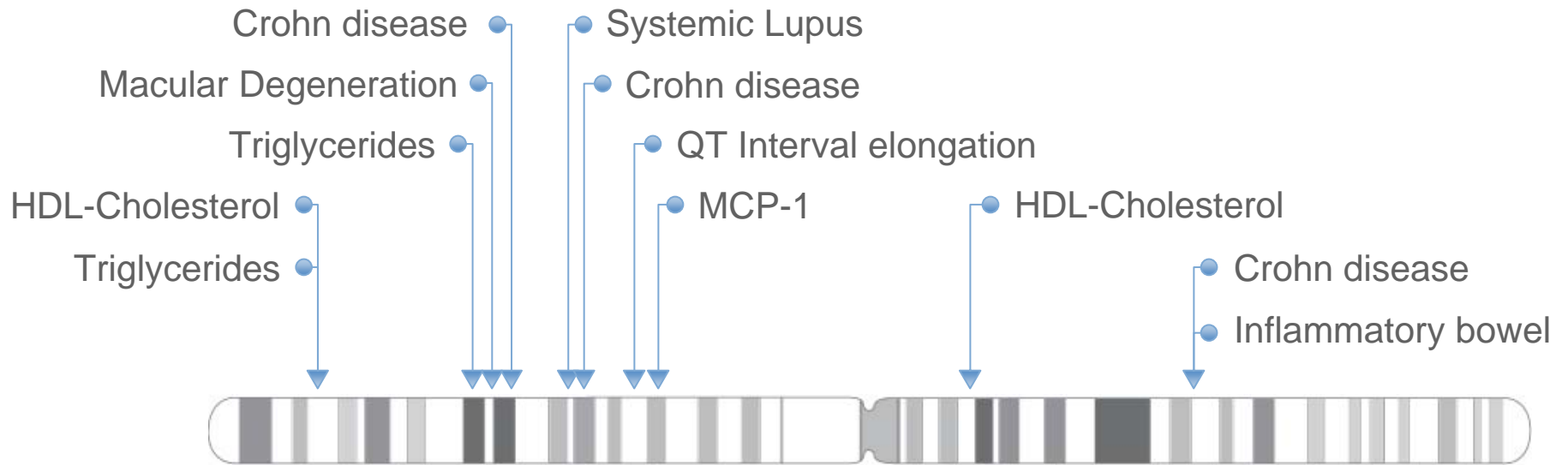
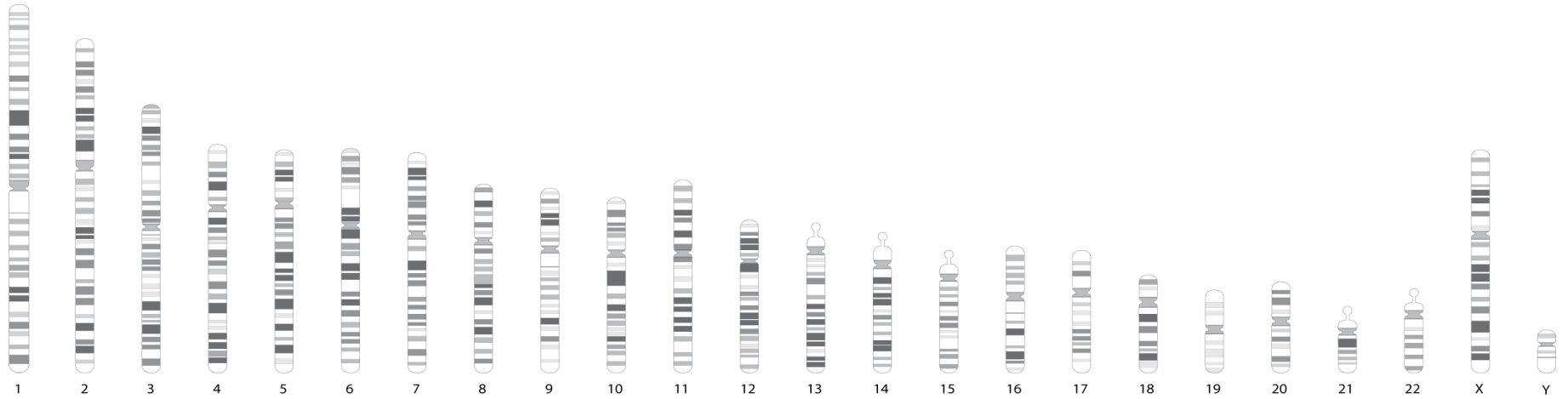
Whole Genome

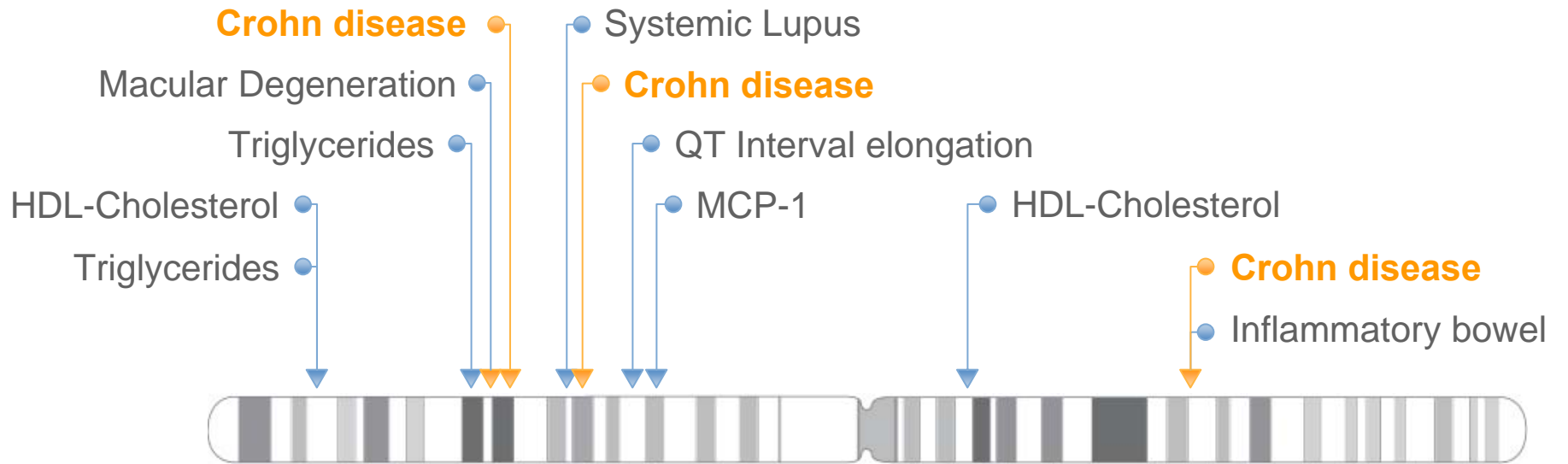
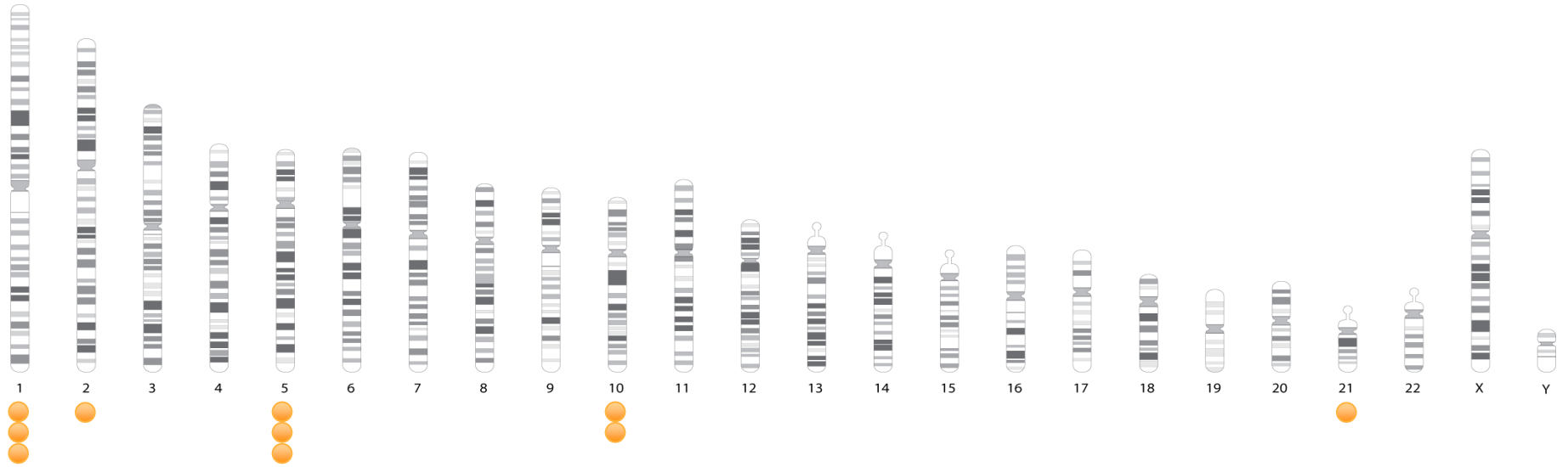
Single Marker

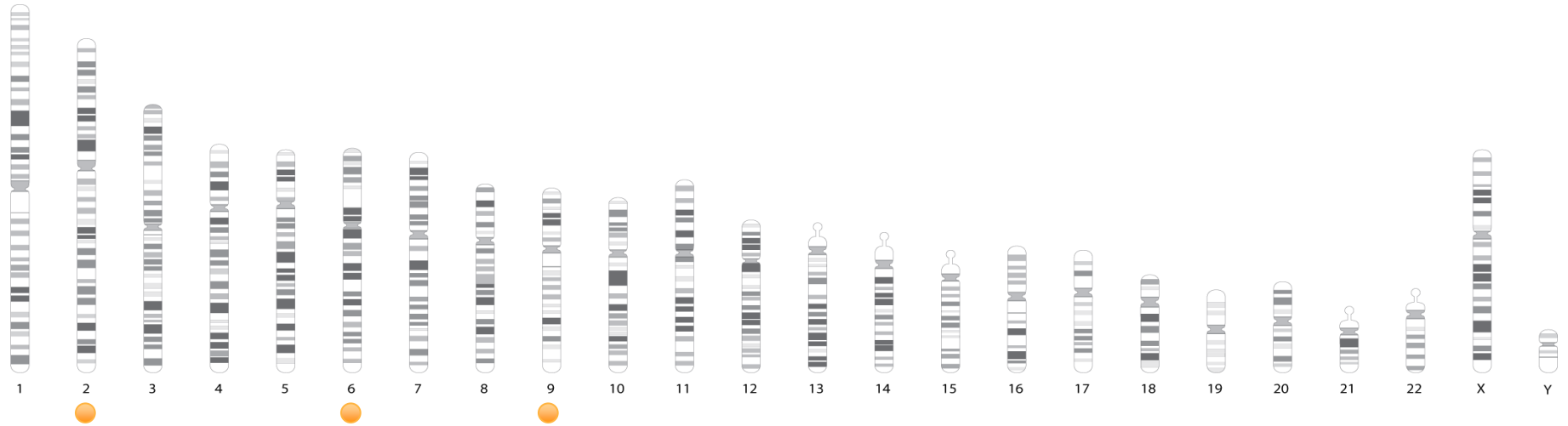


The Genetic Discovery Timeline









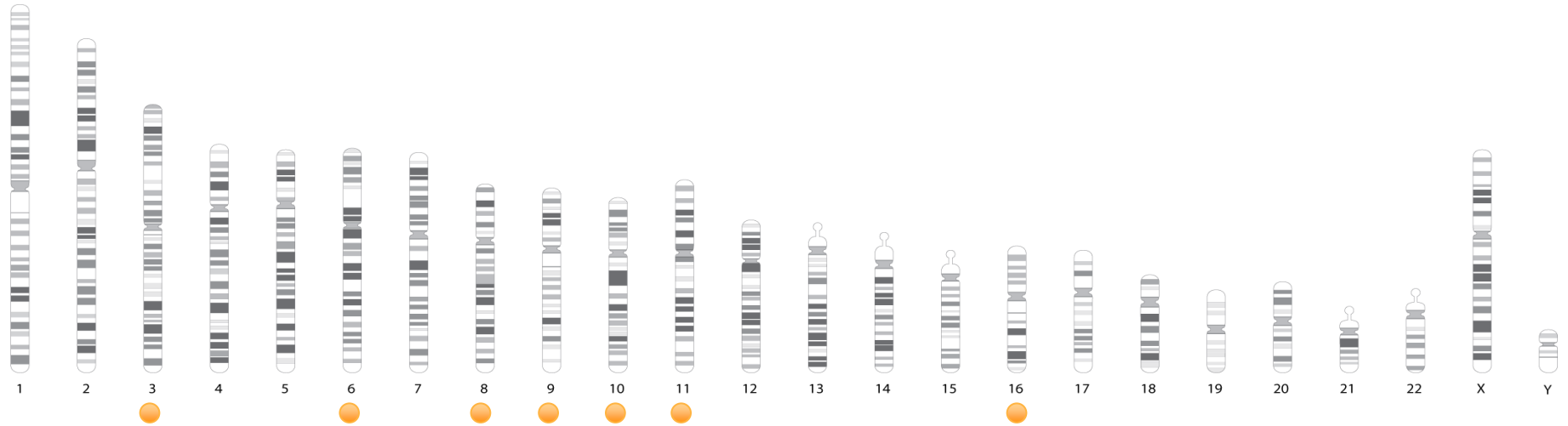
Coronary disease

Type II diabetes

Height

Prostate cancer

Breast cancer



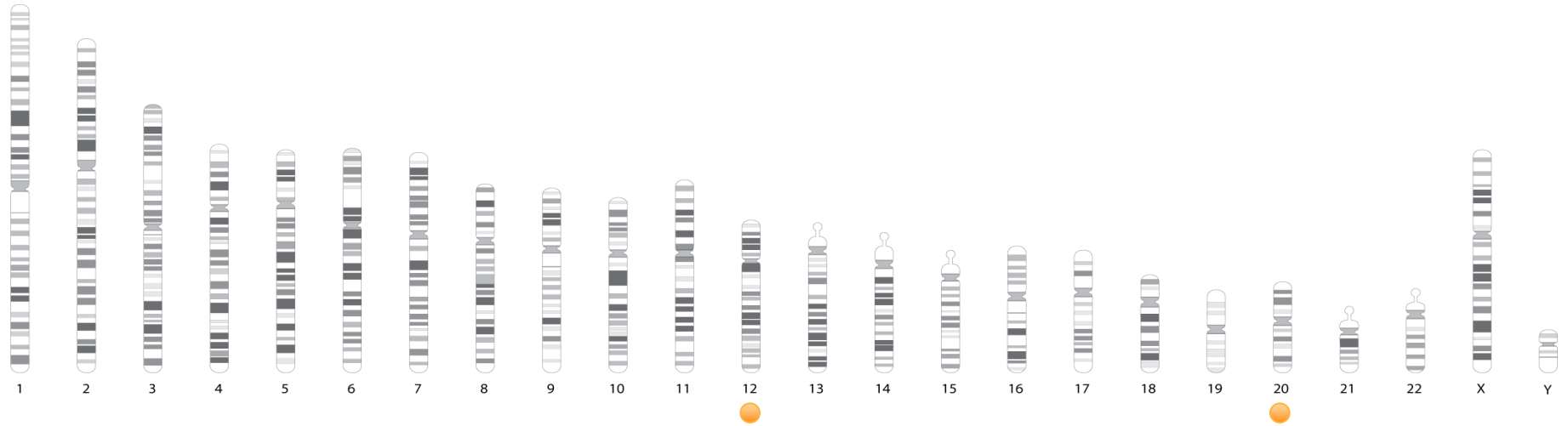
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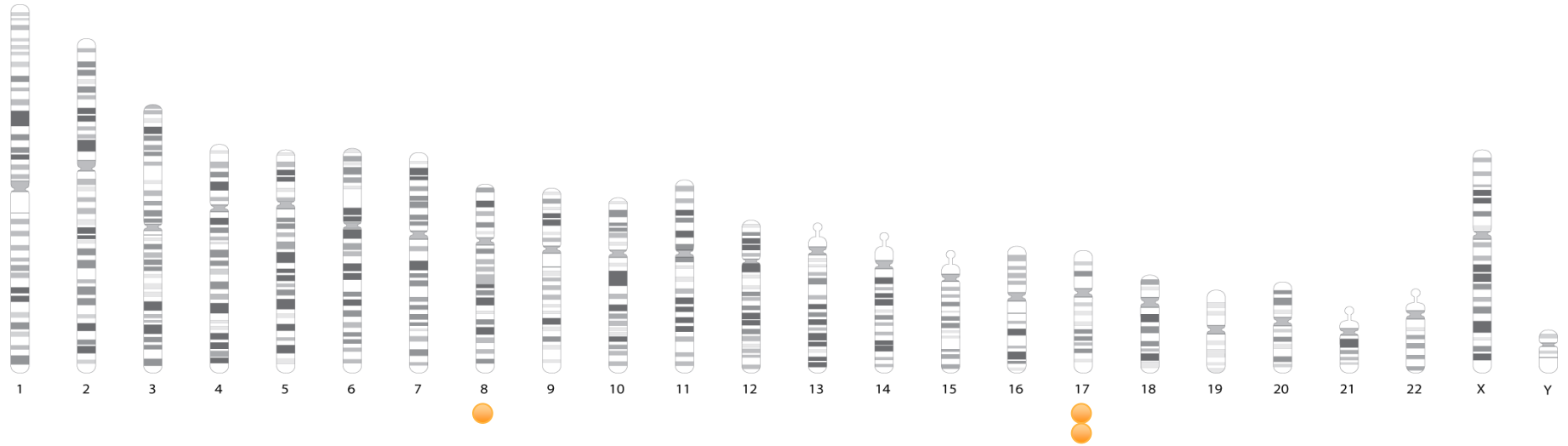
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Type II diabetes

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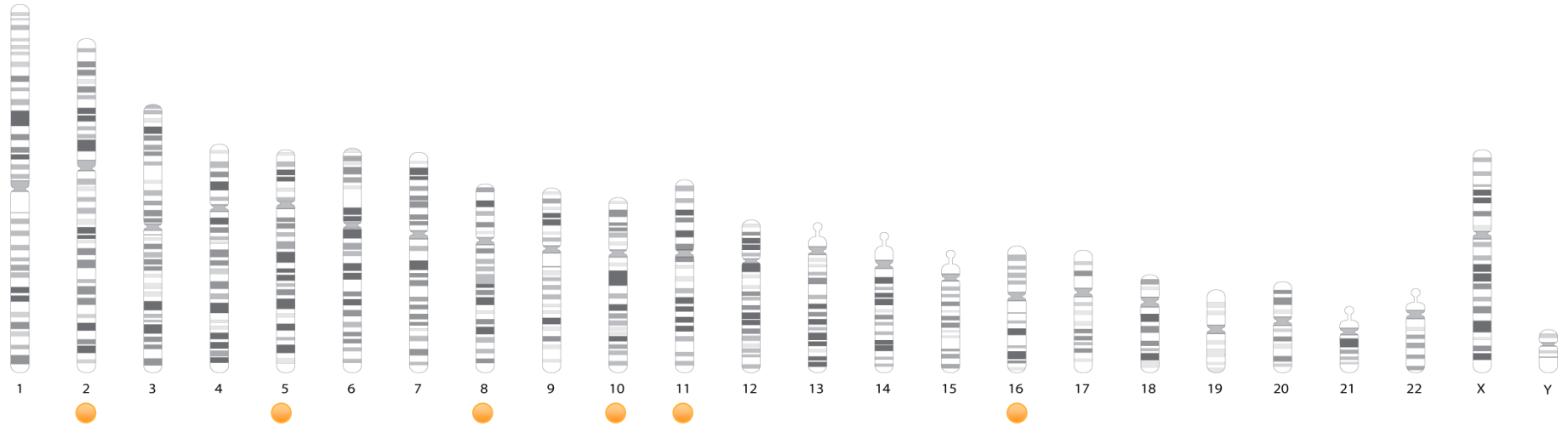
Coronary disease

Type II diabetes

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Coronary disease

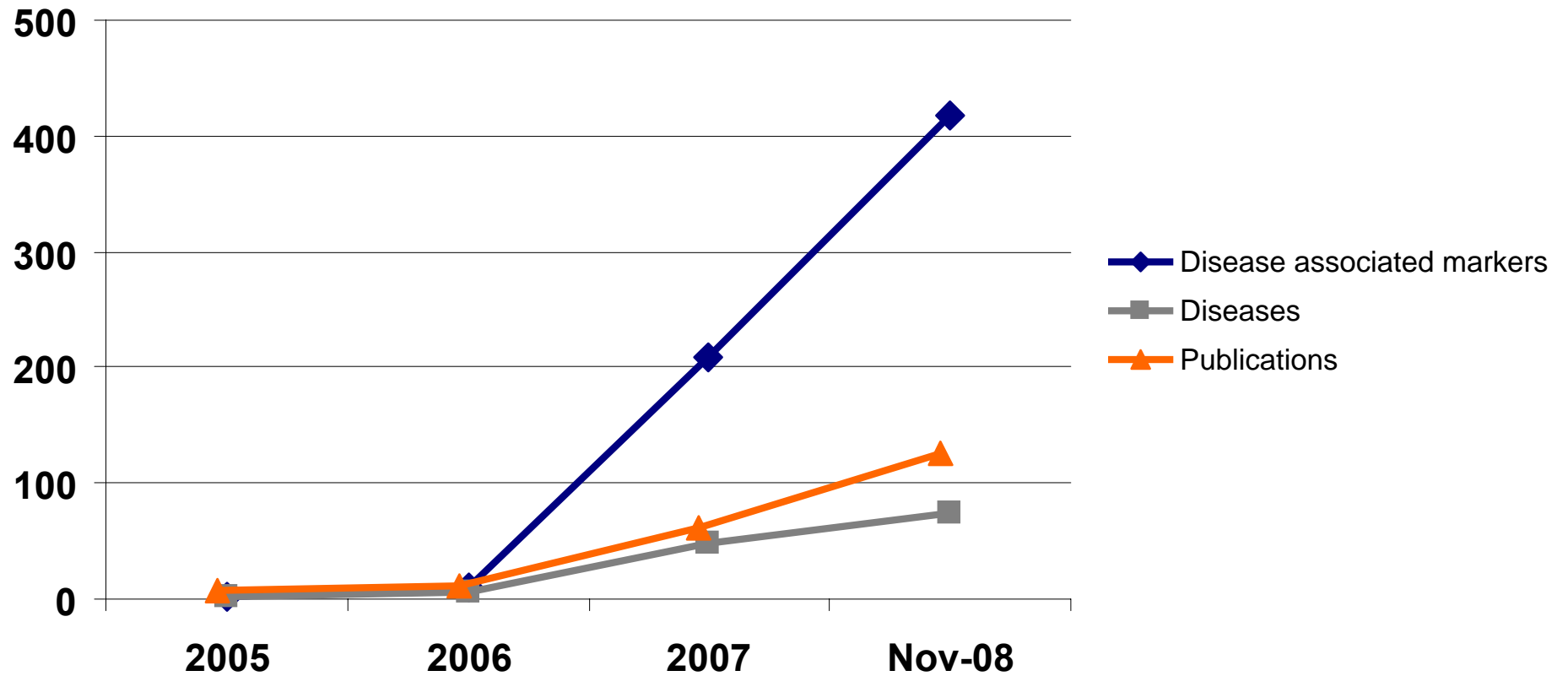
Type II diabetes

Height

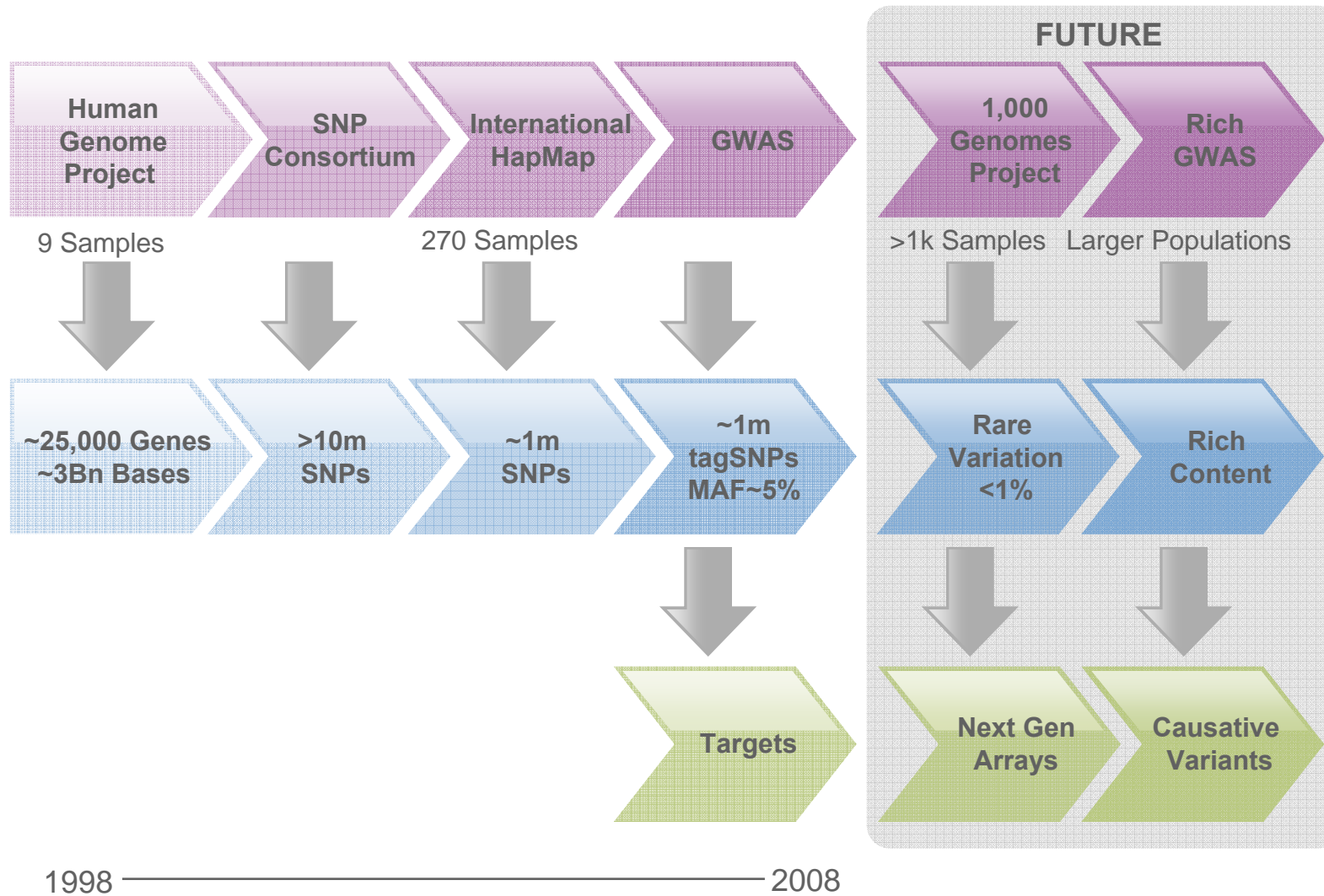
Prostate cancer

Breast cancer

Many Significant Discoveries in the last 2 Years



The Genetic Discovery Timeline



ARRAYS

*Targeted
resequencing*

Next-Gen Sequencing
Identifying Variants

High Density Arrays
Deploying Variants

Content Development is Accelerating

High Density Custom Arrays
Confirming Variants

HumanOmni1-Quad

Introducing the latest Whole-Genome
BeadChip from Illumina

Contains the most up-to-date content for premier coverage of common variation (SNP & CNV) as well as the largest amount of biologically-relevant content, including new discoveries from the 1kGP

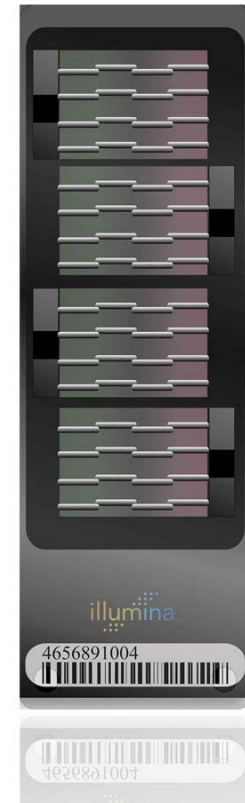


The HumanOmni1-Quad targets common SNPs, CNVs, and High Value Regions of the genome

Three areas of focus:

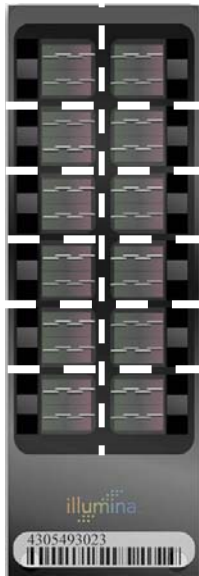
1. Intelligent tagSNPs from all 3 phases of HapMap
 - Industry leading coverage across all 3 HapMap populations
 - Redesigned algorithm to ID most efficient tagSNPs
2. Rare and Common CNVs (discovery and screening)
 - Dense and uniform grid of markers across the genome
 - Partnered to target regions of known CNV with HD probes
3. Most valuable, cutting edge content:
 - Newly discovered SNPs from the 1000 Genomes Project
 - Gene-centric SNPs, indels
 - 1000+ Disease associated SNPs

Estimated % coverage at $r^2 \geq 0.8$	CEU	CHB+JPT	YRI
HumanOmni1-Quad*	94	93	76



Illumina's Whole-genome Genotyping Family of Products

CytoSNP-12



SNP chip for cost-effective GWAS and Molecular Cytogenetics

660W-Quad



Excellent tagSNP coverage of common variation and probes designed to target known regions of CNV

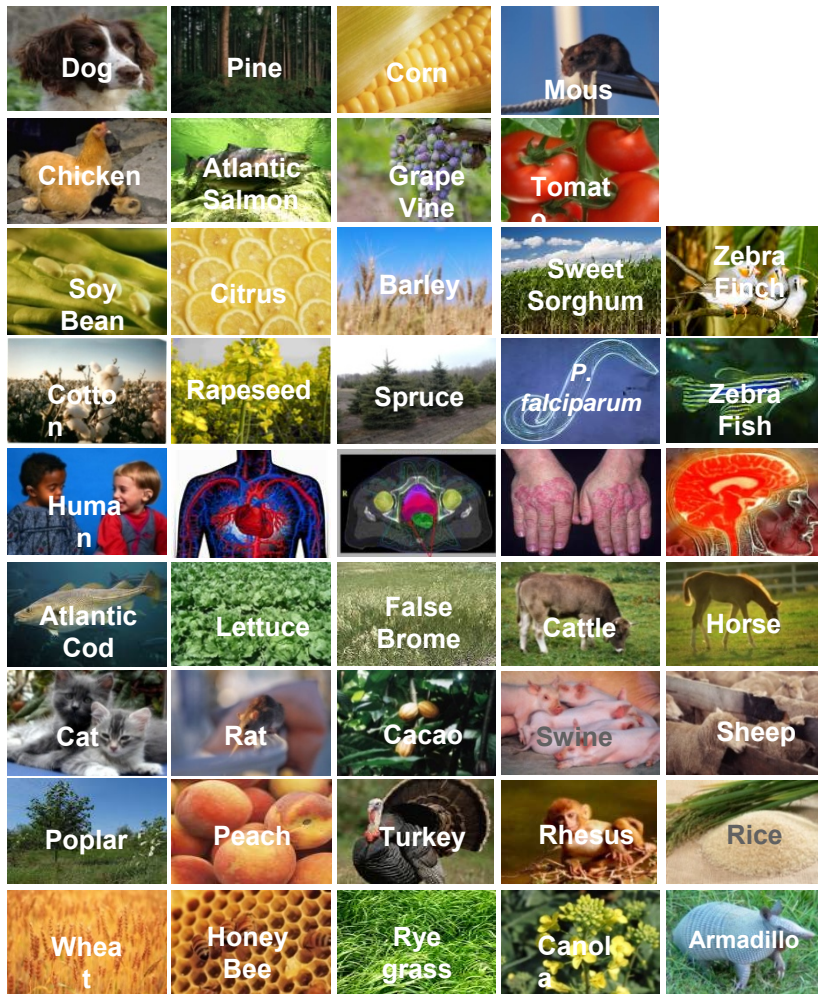
HumanOmni1-Quad



Superior tagSNP coverage of common variation, CNV discovery and screening, and high-value regions



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®HD Custom Genotyping

3,000 ← 6,000 ————— 60,800 ————— 200,000

- Target any variant, any species
- >80% Beadtype success rate
- Infinium assay provides high reproducibility and call rates
- Low DNA input (200ng)
- 12-samples per chip
- Design and order on-line or use in-house support to assist with content design
- CNVs and indels



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



- **PorcineSNP60**

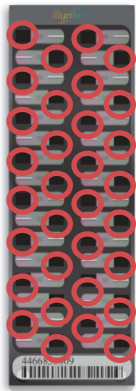
- **OvineSNP50**



GoldenGate[®] Custom Genotyping

BeadArray Technology

Universal-32

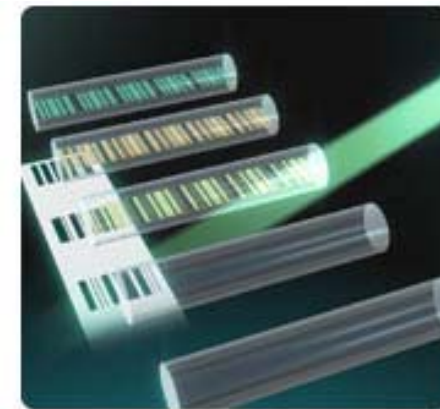


SAM



- Target any variant, any species
- 96-plex to 1536-plex
- 3 day assay
- Up to 288 samples per day*
- Low DNA input (250ng)

Veracode Technology



- Target any variant, any species
- 48, 96, 144, 192 or 384-plex
- 2 day assay
- Up to 768 samples per day*
- Low DNA input (250ng)

Introducing GoldenGate® Indexing

The next evolution of GoldenGate®

**GoldenGate® Indexing Technology Offers Optimal
Combination of Throughput and Cost for Low Complexity
Sample Screening**



+

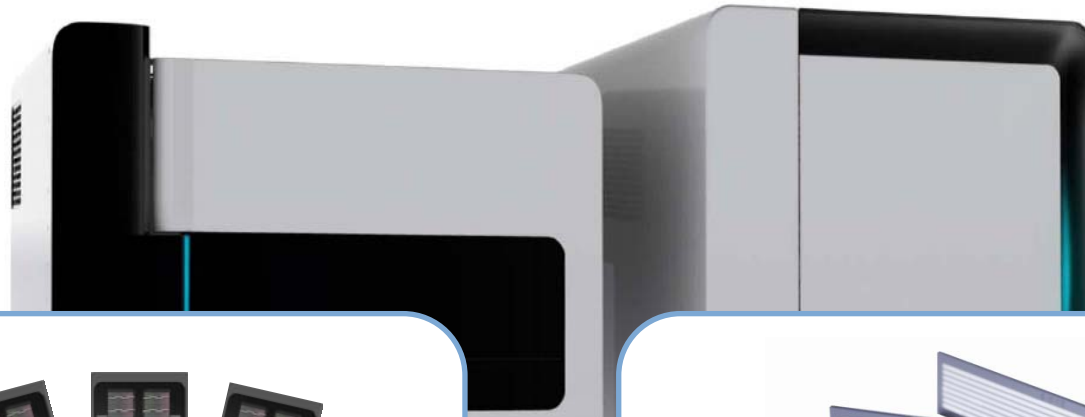


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- **Highest Throughput:** Greater than 2,000 samples per day
- **Highly Flexible:** Advanced multiplexing enables analysis of 96 to 384 loci per sample
- **Fully Integrated:** Automated platform incorporating LIMS
- **Proven Technology:** Robust assay used in genotyping centers worldwide with average call rates > 99%

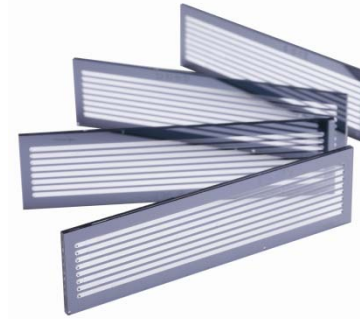
iScan with Sequencing Module



iScan

- Scans HD BeadChips
- Multi-sample formats
- Infinium & Golden Gate assays
- Up to 4800 samples a day
- Seamless automation integration
- Available since April 2008

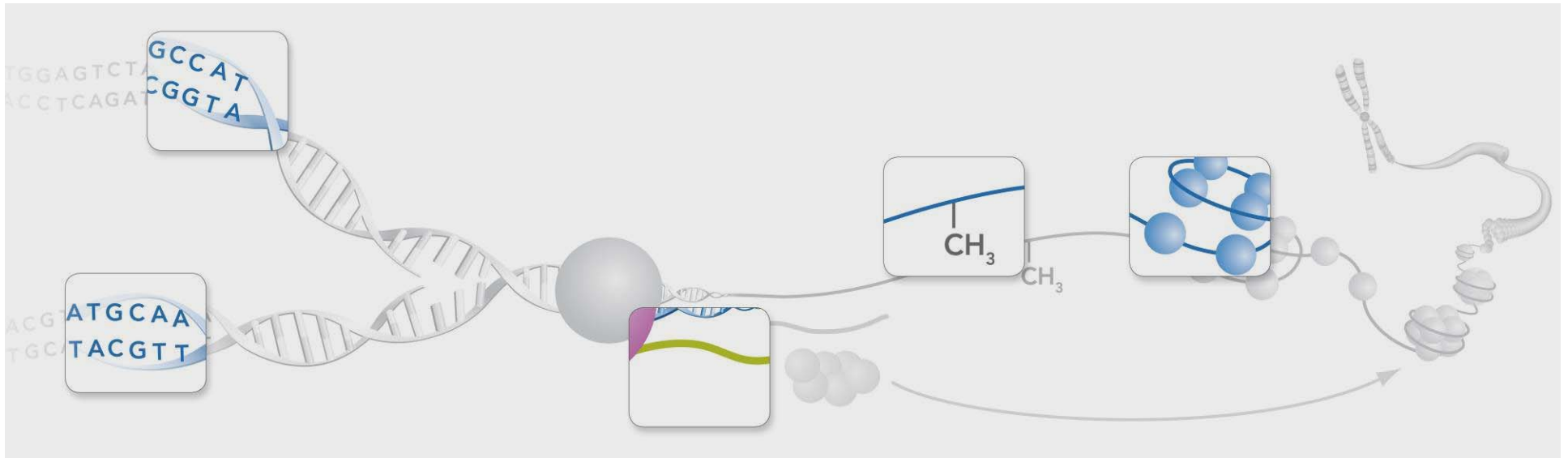
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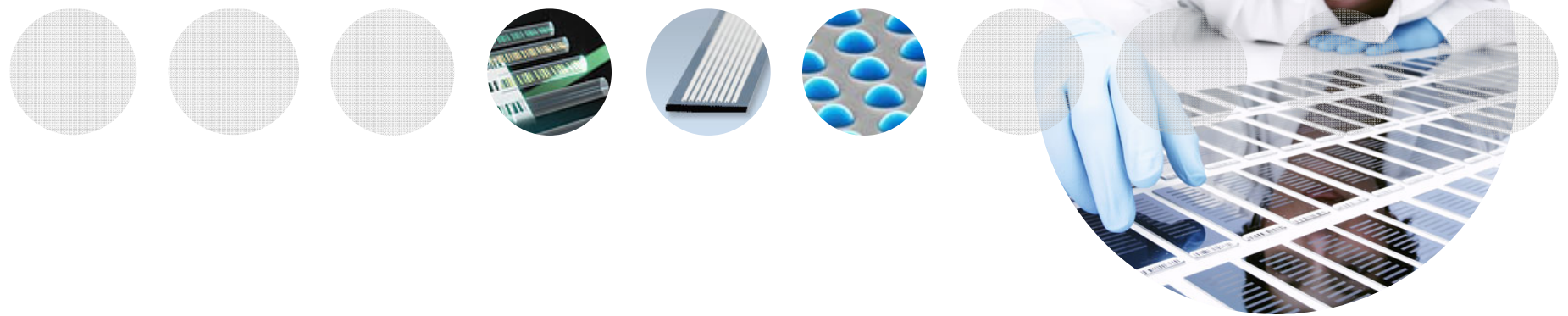
iScan Sequencing Module

- Scans flow cells
- Uses SBS reagents
- ~ 10% GAIIx output
- Paired end capability
- > 2 x 50bp
- > 32 M clusters per run

Go where the biology takes you



Thank You



Complex Diseases: A Mouthful of Scope. The Power of Illumina Platforms

Annette Lee, PhD,
The Feinstein Institute for Medical Research, North Shore