Illumina Custom Genotyping Options

Broad range marker density and flexible content, delivering exceptional data quality for any genome, in any species.

Custom Genotyping Highlights

- **High Quality**
  - Accurate and reproducible data with proven assays
- **Flexible Content**
  - Custom assays in a wide range of multiplex levels for any SNP, any genome, in any species
- **Broad Range of Throughput Capabilities**
  - Multisample 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. Illumina technologies provide the foundation to support a wide variety of marker densities (Table 1). Infinium® technology on the iScan® (Figure 1) and HiScan® systems support Custom Infinium iSelect® BeadChip assays.

Although 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 BeadChips

The Infinium iSelect portfolio 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 700,000 markers per sample can be created in a 24-sample format. Flexible content design, combined with industry-leading Infinium data quality and versatile throughput configurations, make this platform ideal for fine mapping and validation studies, association studies, genomic scans, and marker-assisted breeding for any species (Figure 2).

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, 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 specific to their studies. The amount of Add-On content supported depends on the base content and associated BeadChip and will vary for each product.

Table 1: Custom Genotyping Assay Details.

<table>
<thead>
<tr>
<th>Product</th>
<th>Custom Infinium iSelect</th>
<th>Semi-Custom Infinium</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number of Markers Assayed</td>
<td>~ 3K - 700K</td>
<td>Up to 4.8 million^</td>
</tr>
<tr>
<td>Format</td>
<td>24 samples^</td>
<td>Variable</td>
</tr>
<tr>
<td>Sample Throughput</td>
<td>Up to 5760 samples/week^</td>
<td>Up to 5760 samples/week^</td>
</tr>
<tr>
<td>FFPE Compatible</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>LIMS Support</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Automation Support</td>
<td>✓</td>
<td>✓</td>
</tr>
</tbody>
</table>

^ a. 4.3M markers base content, 500K additional custom content.
b. Dependent on the number of custom markers selected.
c. Assuming 2 iScan systems, 1 AutoLoader 2x, and 2 Tecan robots over an 8-hour day.

Abbreviations: FFPE, formalin-fixed, paraffin-embedded; LIMS, Laboratory Information Management System

Illumina Solutions for Genotyping

High Data Quality

High call rates and accurate genotype calls are critical for successful studies. Because 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.

**User-Friendly Software for Streamlined Data Analysis**

With the Infinium workflow, data are processed directly in Illumina 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.

**Efficient Automation and Tracking**

For researchers who require the highest throughput, Illumina offers several options 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 workflow, allowing labs to maximize their throughput with an integrated microarray processing solution.

**Custom Services**

Researchers can also choose the 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/applications/microarrays.html to learn more about custom, focused, and whole-genome genotyping and CNV analysis solutions.

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**Figure 2: Illumina Custom Genotyping Options** — Illumina products enable a wide range of genotyping experimental designs, depending upon the number of markers.

**Ordering Information - Custom Infinium Genotyping Products**

<table>
<thead>
<tr>
<th>Product</th>
<th>Marker Density</th>
<th>Add-on Content Amount Supported</th>
<th>Samples Per BeadChip</th>
<th>Minimum Sample Order</th>
<th>Samples</th>
<th>Catalog No. for Standard Product</th>
<th>Catalog No. for Add-on Product</th>
</tr>
</thead>
<tbody>
<tr>
<td>iSelect HD 3-90K</td>
<td>3 - 90K</td>
<td>Up to 90K*</td>
<td>24</td>
<td>1152</td>
<td>48</td>
<td>WG-401-1001</td>
<td>WG-401-1014</td>
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<tr>
<td></td>
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<td></td>
<td></td>
<td>288</td>
<td>WG-401-1002</td>
<td></td>
</tr>
<tr>
<td></td>
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<td></td>
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<td>1152</td>
<td>WG-401-1003</td>
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</tr>
<tr>
<td>iSelect HTS 90,001-700K</td>
<td>90,001 - 700K</td>
<td>Up to 700K*</td>
<td>24</td>
<td>1152</td>
<td>48</td>
<td>WG-405-1001</td>
<td>WG-405-1014</td>
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<td>WG-405-1003</td>
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*Amount of Add-On content supported is the maximum amount minus the amount of initial iSelect product.