Sample Types For Illumina® Genotyping Assays

The Infinium® HD and GoldenGate® Assays support genotyping and copy number analysis using a range of sample types.

Introduction
Numerous sample types have been shown to yield high-quality data for genotyping and copy number variation analysis with Illumina Assays. Both the Infinium HD and GoldenGate assays have low DNA loading requirements and offer the flexibility to support diverse experimental designs (Table 1).

Blood
Blood samples produce high-quality genotypes and copy number variation (CNV) data. Immortalized lymphoblastoid cell lines are likely to have high call rates but may show a low level of passage-induced aberrations.

Solid Tumors
Tumor samples produce high-quality genotypes and CNV data. However, call rates are lower due to unpredictable chromosomal aberrations.

Whole-Genome Amplified (WGA)
Most WGA samples produce high-quality genotypes and CNV data. Cluster with similarly prepared WGA samples to maintain call rates, as the intensity is often lower than in genomic controls.

Fresh Frozen
Fresh frozen samples produce high call rates and precise CNV measurements.

Lower Input Amounts
Lower input amounts can be genotyped, but yield lower call rates and higher CNV noise compared to amounts over 50 ng/µl. Paired samples with similar input amounts can be effectively analyzed for CNV, because the samples are compared to each other.

Formalin-Fixed, Paraffin-Embedded (FFPE)
FFPE samples work very well in the GoldenGate Assay but are not ideal for the Infinium HD Assay.

Single Cells
Amplified single cells are expected to have low call rates and show a bias towards heterozygotes. Pre-amplifying the sample with a single-cell amplification protocol may generate usable genotypes.

Saliva
Saliva samples isolated using the Oragene Kit from DNA Genotek and genotyped using the Infinium HD Assay produce high-quality genotypes and CNV data.

Buccal Swabs
Several preparation methods for buccal swabs generate high-quality genotype data using both the Infinium HD and GoldenGate Assays.

Table 1: Sample Type Recommendations

<table>
<thead>
<tr>
<th>Sample Type</th>
<th>Infinium Genotyping (200 ng*)</th>
<th>Infinium cnv Analysis (200 ng*)</th>
<th>GoldenGate (250 ng*)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Solid Tumors</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Whole-Genome Amplified (WGA)</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Fresh Frozen</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Lower Input Amounts</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Formalin-Fixed, Paraffin Embedded (FFPE)</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Single Cells</td>
<td>No</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>Saliva</td>
<td>Yes</td>
<td>Not Validated</td>
<td>Yes</td>
</tr>
<tr>
<td>Buccal Swabs (&gt; 3 per sample)</td>
<td>Yes</td>
<td>Not Validated</td>
<td>Yes</td>
</tr>
</tbody>
</table>

*FastTrack Services require larger input amounts than those recommended here, due to plate format, robot processing, and potential sample requeues.

1Input of as little as 50 ng is acceptable. The sample can be amplified to 200 ng using the Qiagen Repli-G Mini (catalog # 150023), but this is not required.
Technical Note: DNA Analysis