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

Sample Type	Infinium Genotyping (200 ng*)	Infinium cnv Analysis (200 ng*)	GoldenGate (250 ng*)
Blood	Yes	Yes	Yes
Solid Tumors	Yes	Yes	Yes
Whole-Genome Amplified (WGA)‡	Yes	Yes	Yes
Fresh Frozen	Yes	Yes	Yes
Lower Input Amounts	Yes	Yes	Yes
Formalin-Fixed, Paraffin Embedded (FFPE)	No	No	Yes
Single Cells	No	No	No
Saliva	Yes	Not Validated	Yes
Buccal Swabs (> 3 per sample)	Yes	Not Validated	Yes

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

‡Input 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.

AGAATGATAACAGTAACACACTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTCTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAAC
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CGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTGCAACGAAAAGAATGATAACAGTA

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