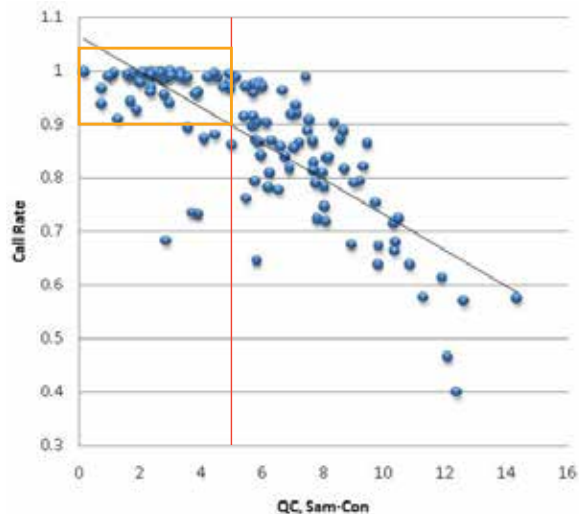


Figure 2: QC of FFPE Samples



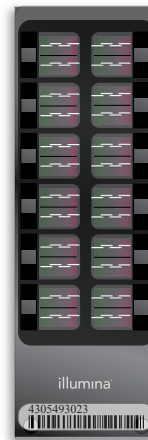
DNA from 130 FFPE samples of varying quality and control DNA (2 ng each) were analyzed in triplicate by real-time PCR. The average Cq value for the control DNA was subtracted from Ct values for FFPE DNA. The resulting Delta Ct values were plotted against the Infinium HD Assay call rates for the same samples after restoration. Greater than 80% of samples with a QC value below 5 were successfully genotyped (orange box). Depending on sample origin and age (1–35 years), 35% to 95% of samples passed QC.

Each of these BeadChips runs on the proven Infinium HD Assay, consistently producing high call rates (> 95%), very high reproducibility, and extremely low error rates¹. This assay technology has enabled many revolutionary discoveries in disease research, resulting in a vast publication record.

Structural Variation Analysis

Structural variation is thought to be a significant contributor to the genetic basis of many human diseases. Dense marker spacing on the whole-genome BeadChips coupled with the sensitive

Figure 3: Infinium HD Twelve-Sample BeadChip



Researchers with FFPE samples can choose from several Infinium BeadChips to perform applications ranging from whole-genome genotyping and methylation studies, to fully custom genotyping.

Infinium HD Assay offers a powerful tool for structural variation analysis. Researchers can profile virtually any chromosomal aberration such as amplifications, deletions, rearrangements, point mutations, copy number changes, and copy-neutral loss of heterozygosity (LOH) events. The assay delivers very high signal-to-noise ratios and low overall noise levels, which are ideal for precise structural variation analysis.

Exceptional Data Quality

To evaluate the data quality produced from degraded DNA, a series of FFPE samples were treated with the Infinium HD DNA Restore Kit and evaluated on the HumanCytoSNP-FFPE-12 BeadChip. The assay signal intensities were referenced against canonical genotype cluster files. The clusters were generated from artificially degraded Coriell DNA samples that were processed with the Infinium HD FFPE DNA Restore Kit and evaluated using the Infinium HD Assay.

Table 1: FFPE-Enabled BeadChip Specifications

	HumanOmniExpress FFPE BeadChip	HumanCytoSNP-FFPE BeadChip	HumanMethylation450 BeadChip	Infinium iSelect BeadChips
Number of Markers	> 693,000	> 262,000	450,000	3,072–1,000,000
Number of Samples/BeadChip	12	12	12	4–24
DNA Requirement (ng)	100	100	250	100
Assay	Infinium HD	Infinium HD	Infinium HD	Infinium HD
Instrument Support	HiScanSQ™ or iScan	HiScanSQ, iScan, BeadArray™ Reader	HiScanSQ, iScan	HiScanSQ, iScan, BeadArray Reader
Applications	Whole-Genome Genotyping and CNV Analysis	Whole-Genome Genotyping and CNV Analysis	Epigenetic Studies	Custom Genotyping

