

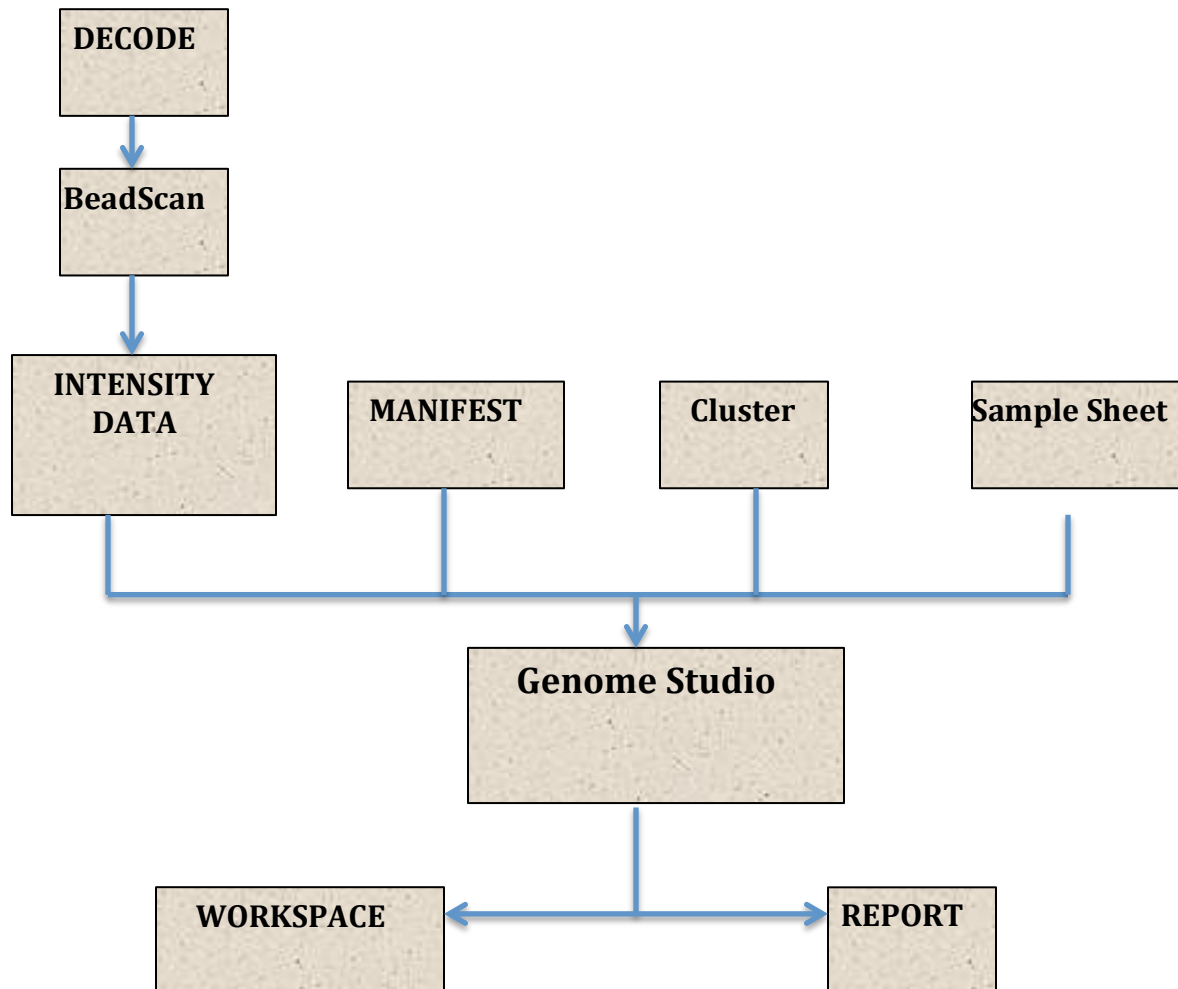
## 1). Illumina's Infinium Beadchips

The Infinium II assay is for analysis for amount of 100K up to 5M SNPs.

First, a whole genome amplification step with the following hybridization to bead array chips capture probes. Each 2-m( $\mu$ )m bead carries a unique probe, and allelic discrimination happens when the single-base primer extension process. After the extension, the beads are labeled with green and/or red fluorescence. The fluorescent emission specific bead is then used to determine the genotype at that locus: red or green indicates a homozygote and yellow indicates a heterozygote state. There are many allele-calling algorithms that can interpret the red and green fluorescence intensities associated with a locus-specific probe and convert this information to a DNA allele. Illumina uses the GeneCall algorithm, which is based on prior training datasets and the Illumina algorithm, a model-based approach that pools information from individual sample data to improved call rates. An advantage of the Beadchip assay is that open positions on the bead plate allow addition of custom SNPs. Small amounts of DNA are required: 200 ng for the one million SNPs HumanOmni\_Quand v1. The two-color system restricts the technology to biallelic SNP calls. Data for the Infinium II assay indicates high pass rates and accuracy (>99.9) <sup>13</sup>



## Data Analysis of Illumina



## 2).If you are interested, here is some about Affymetrix - GeneChip

The GeneChip 6.0 assays are based on allelic discrimination by direct hybridization of genomic DNA to arrays containing locus and allele-specific oligonucleotides (25 mers). These oligonucleotides represent either perfect match or mismatch probes to each SNP. This assay is designed for analysis of ~900K SNPs and 900K copy number polymorphisms (CNPs) per chip. The complexity of the DNA sample is first reduced via restriction endonuclease digestion and DNA fragmentation. Following a PCR amplification step, the products are end-labeled and hybridized to the chip array. In this single-color assay, allele calling is based on fluorescent intensity at each allele-specific position. Affymetrix and the Wellcome Trust Case Control Consortium

have introduced powerful allele-calling algorithms resulting in call rates greater than 99%. Allele-calling techniques are dealt with more thoroughly in the following section on data analysis. Affymetrix also uses a CNP calling algorithm developed by the Broad Institute. Data for the GeneChip SNP array indicates pass rates greater than 95% and accuracy greater than 99%.<sup>13</sup>