

HIGH-THROUGHPUT GENOTYPING SERVICES

ICBR, University of Florida

Personnel

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Description of the instrumentation

We perform high-throughput genotyping using the *Illumina BeadStation 550GX*. This powerful genotyping system includes a high resolution (0.8 microns) laser scanner (BeadScanner) and the BeadStudio software for low-to-high throughput SNP analysis. .

List of Genotyping and Gene Expression Core Services using the Illumina Platform

Currently available*

- Whole genome SNP genotyping (from 10^5 - 10^6 tag SNPs per chip)
- Copy number/Loss of heterozygosity genotyping
- Focused content SNP genotyping
 - Standard panels (cancer SNP, DNA test, non-synonymous human, MHC)
 - Human and mouse linkage analysis panels
 - GoldenGate custom panels
 - iSelect custom panels
- Gene expression analysis in a *multiplex format*

*Please visit the Illumina website (www.illumina.com) under the "products and services" link for more details.

Coming soon!

- DNA methylation profiling
- DASL assay for expression studies on formalin-fixed *paraffin-embedded* samples

Sample/Project Submission

Researchers who wish to submit large genotyping projects are advised to meet with personnel from the ICBR and the Center for Pharmacogenomics. The goal of this discussion will be to establish a

consensus regarding the technical logistics and choice of chemistry to be used for sample analysis. To initiate a project submission, please contact: Dr. David Moraga, moraga@biotech.ufl.edu, or 352-273-8053. A typical workflow for sample submission and processing will be as follows:

1. Users (i.e., you) submit DNA samples in the quality and quantity required by the assay (see “sample quality” below). Quantitation must be done by the Pico Green assay or equivalent. This method quantifies preferentially double-stranded high MW DNA. Samples should have a A_{260} of 1.8 or above (indicative of purity).
2. We will further assess quantity, as well as quality of the DNA samples by gel analysis.
3. Users will be notified if we identify any samples that may not be suited for the genotyping assays.
4. We will order the required chips for the assay from Illumina.
5. We will proceed to perform the genotyping assay, and do the work in the shortest possible time, based on a first-come-first-serve bases. You will be informed of the approximate turnaround time at the time of sample submission.
6. You will be notified when assays have been finished, and will invite you to go through a preliminary analysis of the data using the BeadStudio software in order to generate the report outputs that you deem best for your purposes.

Projects involving gene expression analysis will be evaluated in coordination with the ICBR Microarray Core in order to provide researchers with advise regarding the best-suited platform to use based on the nature of the project.

All reagents (including Oligos, OPAs) and consumables must be placed through the Core facility. For any other arrangements, please contact the ICBR at 352-273-8053 or email: moraga@biotech.ufl.edu

Sample Quality and Quantity Requirements

Genomic DNA samples for genotyping analysis must be of **high quality** (i.e., >50 Kbp in size) **and purity** ($A_{260}/A_{280} > 1.8$). Samples must be accompanied by experimental data demonstrating good quality and purity. We cannot guarantee good quality data on impure and/or highly degraded DNA samples.

Our standard operating procedure includes our own pre-genotyping-assay sample assessment. We will use the Quant-iT PicoGreen dsDNA kit (Invitrogen) for quantification, and gel electrophoresis for quality assessment.

Minimum amount of sample/single assay to be submitted should be as followed: *GoldenGate* (500 ng @ 50 ng/ul), *Infinium* (1 microgram @ 50 ng/ul), *Whole Genome Expression* (100 ng @ 10 ng/ul), *DNA Methylation* (1 microgram @ 50 ng/ul).

Deliverables

Following completion of assays, users will receive a CD or DVD containing study-specific genotyping data output from the Illumina **BeadStudio** software. These data constitute a first-stage statistical

analysis of sample, per genotype (eg., evaluation of controls, filtering of failed samples, and clustering). Raw TIFF images for arrays graphs can be also provided upon request.

The ICBR's Bioinformatics Core can provide additional "genotyping analysis" services upon request.

All data generated by the SNP Genotyping Core laboratory will be kept strictly confidential. The data belongs solely to the submitting investigator. ICBR requests only an acknowledgment upon publication of any results: "*Genotyping services were provided by the SNP Genotyping Core, ICBR.*"

Literature References

BeadArray™-based solutions for enabling the promise of Pharmacogenomics. J.-B, Fang et al., *Biotechniques* **39**: 583-588 (October 2005)

Highly Parallel Genotyping. J.-B, Fang et al., *Cold Spring Harbor Symposia on Quantitative Biology*. **Vol. LXVIII**. 2003 Cold Spring Harbor Lab Press 0-87969-1/04