**Evaluation of the Illumina Infinium Omni Express Exome Bead chip for forensic testing**

Jon Davoren, MS*, Giovanna Vidoli, PhD**, Amy Mundorff, PhD**

*Bode Technology. 10430 Furnace Road Suite 107, Lorton, VA 22079
**University of Tennessee - Anthropology, Knoxville, TN 37996-1525

**Learning Overview**: After attending this presentation, attendees will understand the performance of the Infinium™ OmniExpressExome-8 BeadChip on DNA from blood and bone samples.

**Impact on the Forensic Science Community**: This presentation will impact the forensic science community by providing information on the current benefits and limitations for SNP chip genotyping of forensic samples.

Single nucleotide polymorphism (SNP) chips provide hundreds of thousands to millions of loci in a single test. The large number of loci can provide prediction of ancestry, phenotypic information and relatedness well beyond the limits of short tandem repeat (STR) technology. The main limitation to using Infinium SNP chips is that they are designed for use on much higher levels of relatively intact DNA than is found in most forensic samples. Research into the effects of running lower template levels and degraded template on SNP chips is currently lacking. This study aims to evaluate the performance of the ~960,000 SNPs on the Infinium™ OmniExpressExome-8 BeadChip across a range of template amounts from human blood and bone samples.

Blood samples were collected approximately 1 week post-mortem and bones were sampled at 4-8 years post-mortem. All samples were extracted using Qiagen chemistry, showed minimal inhibition and degradation when tested with Quantifiler™ Trio, and gave full or nearly full STR profiles when tested with the Globalfiler™ system. A DNA dilution series from 250 Ng to 10 Ng, for blood and bone samples, was tested using the OmniExpressExome system. The chips were read on an Illumina iScan® instrument, analyzed in the GenomeStudio® software using the manufacturer recommended settings, and then compared for accuracy, reproducibility and sensitivity.

The SNP call rates varied by individual tested, were lower for the DNA from bones and were reduced as the template levels were lowered. The blood and bone DNA at 10 Ng of template had ~72% and ~30% call rates, respectively. At the lowest call rate approximately 300,000 SNPs were called. The accuracy of the SNP calls fell dramatically when the call rates were below 70% with up to 50% error rates. Re-analysis in the genome studio software with higher stringency settings lowered error rates but also lowered call rates.

Phenotype and ancestry predictions, using the Parabon® Snapshot® system, for samples with at least a 70% SNP call rate were consistent with the known hair color, eye color, skin color and ancestry of the individual. For the samples with less than a 70% SNP call rate the confidence levels of the predictions were too low to be significant.

The results of this evaluation suggest that the Infinium Omni express exome SNP chip has potential to be useful for forensic-type sample testing, however additional development work is needed to increase call rates and accuracy for samples with lower levels of DNA and/or degraded DNA.

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