

EVALUATION OF THE PRECISION ID ANCESTRY PANEL USING ION TORRENT™ SEQUENCING TECHNOLOGY

Amy D. Roeder, Paul Elsmore and Matthew J. Greenhalgh, Orchid Cellmark Ltd

Although Massively Parallel Sequencing (MPS) is unlikely to replace CE based STR typing methodology in the near future, there may be circumstances where the use of MPS could be advantageous over current techniques. Due to the potential for genetic ancestry information to generate leads in cold cases, we have evaluated the Precision ID Ancestry Panel (Life Technologies) using the Ion Torrent™ Personal Genome Machine (PGM™) System (Life Technologies). SNP based ancestry estimations obtained using the ancestry panel were compared with the self-reported ancestries of the sample donors. The genetic admixture results were generally comparable to the reported ancestries, however, it was not uncommon for the SNP data to indicate genetic contributions from geographic areas that were not indicated by the sample donor. A series of DNA dilutions (down to 31 pg) were also analyzed. The amount of DNA used as template for library preparation could be reduced from 1 ng to 250 pg without substantially affecting the quality of the results. In the samples containing <250 pg of DNA, the number of failed SNPs increased as the amount of template DNA was reduced. Allelic drop-out was only observed in the sample amplified using 31 pg of DNA. The majority of the SNPs (92/165) failed in a sample that was exposed to high temperature. The partial SNP profile produced using MPS was, however, more informative than the single unconfirmed allele observed in the STR profile generated for this sample using CE based methodology. As expected, the read lengths for the degraded sample were shorter than the other samples tested. Based on this study, the ancestry panel has potential for use as an intelligence tool for predicting the broad geographic ancestry (e.g. Europe, Africa, East Asia) of a sample donor but when using only the manufacturer provided population databases, it is of limited use for more precise ancestry predictions.