Comparison of sequencing platforms for single nucleotide variant calls in a human Sample

Next-generation sequencing platforms coupled with advanced bioinformatic tools enable re-sequencing of the human genome at high-speed and large cost savings. We compare sequencing platforms from Roche/454, Illumina/HiSeq, and Life Technologies/SOLiD for their ability to identify single nucleotide substitutions in whole genome sequences from the same human sample. The differences in the variant calls were investigated with regards to coverage, GC content and sequencing error. Some of the variants called by only one or two of the platforms were experimentally tested using mass spectrometry; a method that is independent of DNA sequencing. We establish several causes why variants remained unreported, specific to each platform. We report the indel called using the three sequencing technologies and from the obtained results we conclude that sequencing human genomes with more than a single platform and multiple libraries is beneficial when high level of accuracy is required.