THE VALIDATION ANALYSIS OF THE 27-PLEX SNP PANEL FOR ANCESTRY INFERENCE (P)

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Previously, our lab set up a single-tube 27-plex Single Nucleotide Polymorphisms (SNPs) panel to infer ancestry components of unknown individuals in a three-continental level. Here we enhanced the accuracy of the system by establishing an analysis pipeline, including likelihood ratio, ancestry composition and individual ancestry assignment, based on a reference database of 3081 individuals from 33 global populations. The accuracy of the pipeline was evaluated by cross validation study using data from the database. The results show that the analysis pipeline was over 99% accurate when analyzing unknown individual's ancestry origin from East Asia, Europe, Africa and the admixture population. A test sample group contained 1096 individuals of 11 populations were analyzed using this pipeline. In total, 99.19%, 78.79%, 99.09% and 85.67% of East Asian, European, African and Admixture individuals were assigned consistently with their predefined geographical regions, respectively. While the inconclusive rate corresponding to the above mentioned regions are 0.81%, 21.21%, 0.92% and 13.62%, respectively. Most of the inconclusive result happened between Admixtures and European, or between Admixtures and East Asian, with a likelihood ratio within one order of magnitude, in this case, the ancestry composition provide a good indicator of ancestry origin of an individual.