FAST DESKTOP APP FOR INFERENCE OF BIO-GEOGRAPHIC ANCESTRY, HAPLOGROUPS, APPEARANCE AND RELATEDNESS FROM DNA

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In missing persons investigations, and when a crime-scene DNA sample cannot be directly matched to a known, previously-genotyped individual, avenues of investigation may be costly and time-consuming. We report on a rapid and easy-to-use desktop application, which generates a clear, succinct visual profile for each DNA sample. Based upon a panel of 200,000 genome-wide markers, the app provides inferences of bio-geographic origin, mitochondrial haplogroup, Y-chromosome haplogroup, coefficient of inbreeding, hair and eye color. It also provides inference of familial relatedness, up to third degree relationships, with high confidence.

Bio-geographic ancestry from genome-wide data, together with the geographic origin of mitochondrial and Y-chromosome haplogroups, allow the characterisation of samples of mixed racial origin. The coefficient of inbreeding gives an insight into time since immigration. We previously reported on a pilot study of the underlying methodology: for 3196 DNA samples, trait-predictions were performed blind to the true characteristics of the individuals. Correct predictions were obtained across a range of sample-types, from DNA amounts as low as 1.75ng. A total of 95% of samples passed quality checks and for those, prediction accuracy was high.

The new app allows rapid, on-site, automated analysis of the genotype data, and it has utility in crime-scene, missing persons and mass disaster investigations, as well as matters of homeland (national) security.