

Research article

Report on ISFG SNP Panel Discussion

J.M. Butler^{a,*}, B. Budowle^b, P. Gill^c, K.K. Kidd^d, C. Phillips^e,
P.M. Schneider^f, P.M. Vallone^a, N. Morling^g

^a National Institute of Standards and Technology, Gaithersburg, MD, USA

^b FBI Laboratory, Quantico, VA, USA

^c Forensic Science Service, Birmingham, UK

^d Yale University, New Haven, CT, USA

^e University of Santiago de Compostela, Spain

^f University of Cologne, Germany

^g University of Copenhagen, Denmark

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Abstract

Six scientists presented their views and experience with single nucleotide polymorphism (SNP) markers, multiplexes, and methods regarding their potential application in forensic identity and relationship testing. Benefits and limitations of SNPs were reviewed, as were different SNP marker categories and assays available.

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The final session of the 22nd Congress of the International Society of Forensic Genetics (ISFG) held in Copenhagen, Denmark on August 25, 2007 was a panel discussion on single nucleotide polymorphisms (SNPs) and their application in forensic identity and relationship testing. Six distinguished scientists with experience in SNPs were asked to present their views on the subject for 5 min each and then participation from the audience was encouraged with panelists commenting on issues raised.

The presentations began with two of the thought leaders in the field, *Bruce Budowle* from the FBI Laboratory and *Peter Gill* of the Forensic Science Service, both of whom have written previously on SNPs in forensics [1–3]. They discussed the benefits and limitations of SNPs and requirements for SNP implementation and interpretation (see Table 1). *Ken Kidd* from Yale University, who has been performing population genetic studies and selecting SNP candidate loci for potential forensic applications, defined several categories for SNP markers

(Table 2) and his criteria used in selecting SNPs [4,5]. Thus far, his team's efforts have selected 108 candidate SNPs that have been evaluated in a global set of >40 populations. *Chris Phillips* from the University of Santiago de Compostela addressed some lessons learned during the SNPforID project [6,7], which was a European Union effort spanning 2002–2005 that resulted in an autosomal SNP 52plex multiplex PCR and typing assay for forensic applications [8]. The European Union analyses found that in order to reach a 1 in 10 billion random match probability a minimum of 26 SNPs were required for Europeans, 29 SNPs for Africans, and 31 SNPs for Asians. *Peter Schneider* from the University of Cologne, who led the SNPforID project, delineated problems that could be resolved by SNP typing and that specific applications will require different sets of SNPs. He underscored that support from commercial suppliers providing reagents for standardized SNP sets is desirable. *Peter Vallone* from the U.S. National Institute of Standards and Technologies, who has developed a number of multiplex SNP assays (e.g. [9]), rounded out the session by re-emphasizing many of the points already made regarding specific SNP applications and the variety of currently available SNP typing technologies. All the speakers encouraged further studies and compilation of population data and other information on SNP markers.

* Corresponding author at: NIST, Biochemical Science Division, 100 Bureau Drive, M/S 8311, Building 227, Room A243, Gaithersburg, MD 20899-8311, USA. Tel.: +1 301 975 4049; fax: +1 301 975 8505.

E-mail address: john.butler@nist.gov (J.M. Butler).

Table 1
SNP advantages and disadvantages

Advantages/Benefits: Small amplicon size better for analyzing degraded samples, lower mutation rate compared with STRs (10^{-8} vs. 10^{-3}), amenable to high throughput analysis (automation), abundant in the human genome, can provide specific information (ancestry, lineage, evolution, or phenotype), multiple typing platforms provide assay design flexibility
Limitations/Challenges: No commercial kits available, no widely established core loci, mixture resolution issues/interpretation, large multiplexing assays required, SNPs are not likely to replace core STRs currently used in national DNA databases, linkage, substructure due to low mutation rate, multiple typing platforms make universal SNP selection difficult

Table 2
Categories for SNP markers classified by Ken Kidd

Individual Identification SNPs (IISNPs): SNPs that collectively give very low probabilities of two individuals having the same multi-locus genotype
Ancestry Informative SNPs (AISNPs): SNPs that collectively give a high probability of an individual's ancestry being from one part of the world or being derived from two or more areas of the world
Lineage Informative SNPs (LISNPs): Sets of tightly linked SNPs that function as multi-allelic markers that can serve to identify relatives with higher probabilities than simple bi-allelic SNPs
Phenotype Informative SNPs (PISNPs): SNPs that provide a high probability that the individual has particular phenotypes, such as a particular skin color, hair color, eye color, etc.

Several audience members including Angel Carracedo, John Buckleton, Antonio Amorim, and Manfred Kayser posed questions of the panel members and/or provided helpful insights. Niels Morling also mentioned that his laboratory in Copenhagen was close to obtaining accreditation under ISO17025 for use of the SNPforID 52plex, suggesting that it was a reliable technique in their hands.

1. Conclusions

The panelists were in agreement that while SNPs would not replace STRs for most forensic applications anytime soon, SNP markers and assays should continue to be explored. There was a common view that SNPs may serve as an adjunct to STRs for solving special problems in forensic genetics and that new areas of application will be identified when more laboratories start researching SNPs for forensic applications. At the close of the session, a number of attendees expressed interest in commercial

kits becoming available to further spur research or application with SNP markers.

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Conflict of interest

None.

References

- [1] R. Chakraborty, D.N. Stivers, Y. Su, B. Budowle, The utility of short tandem repeat loci beyond human identification: implications for development of new DNA typing systems, *Electrophoresis* 20 (8) (1999) 1682–1696.
- [2] P. Gill, An assessment of the utility of single nucleotide polymorphisms (SNPs) for forensic purposes, *International Journal of Legal Medicine* 114 (4–5) (2001) 204–210.
- [3] P. Gill, D.J. Werrett, B. Budowle, R. Guerrieri, An assessment of whether SNPs will replace STRs in national DNA databases—joint considerations of the DNA working group of the European Network of Forensic Science Institutes (ENFSI) and the Scientific Working Group on DNA Analysis Methods (SWGDM), *Science and Justice* 44 (1) (2004) 51–53.
- [4] K.K. Kidd, A.J. Pakstis, W.C. Speed, E.L. Grigorenko, S.L. Kajuna, N.J. Karoma, S. Kungulilo, J.J. Kim, R.B. Lu, A. Odunsi, F. Okonofua, J. Parnas, L.O. Schulz, O.V. Zhukova, J.R. Kidd, Developing a SNP panel for forensic identification of individuals, *Forensic Science International* 164 (1) (2006) 20–32.
- [5] A.J. Pakstis, W.C. Speed, J.R. Kidd, K.K. Kidd, Candidate SNPs for a universal individual identification panel, *Human Genetics* 121 (2007) 305–317 (see also <http://info.med.yale.edu/genetics/kkidd> and <http://alfred.med.yale.edu>).
- [6] <http://www.snpforid.org>.
- [7] C. Phillips, M. Lareu, J. Sanchez, M. Brion, M. Sobrino, N. Morling, P. Schneider, D. Syndercombe Court, A. Carracedo, Selecting single nucleotide polymorphisms for forensic applications, in: C. Doutremépuich, N. Morling (Eds.), *Progress in Forensic Genetics. International Congress Series* 10, 1261, 2004, pp. 18–20.
- [8] J.J. Sanchez, C. Phillips, C. Borsting, K. Balogh, M. Bogus, M. Fondevila, C.D. Harrison, E. Musgrave-Brown, A. Salas, Syndercombe-Court, P.M. Schneider, A. Carracedo, N. Morling, A multiplex assay with 52 single nucleotide polymorphisms for human identification, *Electrophoresis* 27 (9) (2006) 1713–1724.
- [9] P.M. Vallone, R.S. Just, M.D. Coble, J.M. Butler, T.J. Parsons, A multiplex allele-specific primer extension assay for forensically informative SNPs distributed throughout the mitochondrial genome, *International Journal of Legal Medicine* 118 (2004) 147–157.