Selection and Use of SNP Markers for Human Identification and Paternity Analysis in Koreans

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SNPs merit attention for human Identification and paternity analysis

- **STR-typing methods** are amenable to partial automation and easy to collect data from markers

  For a faster turnaround time and facility of massive sample processing, the development of new technologies is necessary

- **DNA microarrays** are notable **for high throughput and its accuracy**, and **SNP markers** are considered **the preferred target DNA probes for microarrays**

SNPs are valuable for forensic applications

Because they are

- **Most common** sequence differences between individuals
- Genetically **stable**
- Amenable to **high-throughput automated analysis** using microarray technologies
- Better suited for the analysis of **highly degraded DNA**

Twenty-four highly informative SNP markers were selected

- Twenty-four SNPs representing **22 autosomes** and **both sex chromosomes** were selected
- SNP data in **the SNP Consortium (TSC)** were employed
- In order to increase the theoretical power of discrimination, we screened SNPs with **a common 50:50 or 45:55 allelic distribution** in **Caucasians, Asians, and African Americans**

Selection of candidate SNP markers from public database

APBiotech - AstraZeneca - Aventis - Bayer - Bristol-Myers Squib - F.Hoffman-La Roche - Glaxo Wellcome

THE SNP CONSORTIUM LTD

IBM - Motorola - Novartis - Pfizer - Searle - SmithKline Beecham - Wellcome Trust

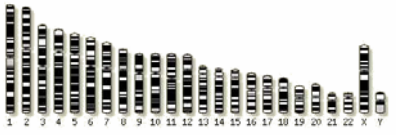


Image courtesy of http://www.ensembl.org, slightly modified from the original

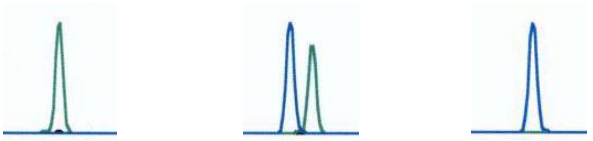
SNP report for TSC0513851

- Population frequency data (Genomic location: Chr5: 155,397,742)

Lab	A	C	G	T	Rev.	# Indiv.	Panel	Protocol
Kwok	**50%**	0%	**50%**	0%	N	42 (pooled)	TSC 42 **AA**	TSCM0036
Kwok	**49%**	0%	**51%**	0%	N	42 (pooled)	TSC 42 **A**	TSCM0036
Kwok	**50%**	0%	**50%**	0%	N	42 (pooled)	TSC 42 **C**	TSCM0036

SNP scoring was performed in 30 Koreans

- **SNP scoring** was carried out by using **single-base extension method**



- SNPs with **allele distributions** in a range of **30:70 to 50:50 in Koreans** were selected as "highly informative" SNP markers

Statistical analysis

Chr.	TSC accession number	Alleles (1,2)	Allele Frequencies 1	2	Genotype Frequencies 1,1	1,2	2,2	P_I	P_E
21	TSC0096586	G, A	0.467	0.533	0.200	0.533	0.267	**0.375**	**0.187**
22	TSC0217548	A, C	0.600	0.400	0.300	0.600	0.100	**0.441**	**0.182**
X	TSC0001989	C, T[a]	0.500	0.500	0.267	0.467	0.267	**0.375**	**na**
		C, T[b]	0.533	0.467	na[c]	na	na	**0.502**	**na**
Y	TSC1248559	A, G	0.133	0.867	na	na	na	**0.752**	**na**

[a]Alleles for female; [b]Alleles for male; [c]na, not applicable

22 autosomal markers combined P_I = 1.905E-10
P_E = 0.989

Whether SNPs will replace STRs

- Population data in Koreans

	22 autosomal SNPs	AmpF/STR Profiler Plus (9 STRs)
P_I	1.905 X 10^{-10}	2.31 X 10^{-12}
P_E	0.989	0.999

Han et al. 2000, Int J Legal Med

- According to the calculation, the **addition of three unlinked SNPs with a P_I of 0.3** to the selected SNP marker set would make **a drop in the combined probability of identity by 2.7 X 10^{-2}**
- If **50 highly informative SNP markers** were combined, the resultant power of discrimination would be **comparable to** those of **AmpF/STR Identifiler** or **PowerPlex16**

Concluding remarks

- **The more SNPs of high informativity** that are added to the marker set, **the higher the power of discrimination**
- The SNPs in this study offers **a small but highly accurate database** that provides **an important reference for SNP-based human identification in three world major populations**

Our Lab Members

At Jeju island in April 2003