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Australian population data for the twenty Promega PowerPlex 21 short tandem repeat loci

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This paper describes the analysis of population data typed using the Promega PowerPlex 21 multiplex for the three major sub populations within Australia. Samples from 1427 declared Australian Aboriginal, 546 Pure Aboriginals from the Northern Territory, 990 Asian, and 1707 Caucasian individuals representing were analysed. Departures from Hardy–Weinberg equilibrium (HWE) and linkage equilibrium (LE) were assessed using exact tests. The Aboriginal populations were shown to display significant departures from equilibrium. All four subpopulation databases are of suitable size for the purpose of estimating allele frequencies.

Keywords: forensic DNA; PowerPlex 21; population data

Announcement of population data

Population

DNA profiles from individuals of Aboriginal, Asian or Caucasian descent from across the eight states and territories within Australia were compiled. In addition, a Pure Aboriginal dataset collected from individuals known to be Indigenous Australians and residing in remote tribal communities from the Northern Territory was analysed. The majority of Caucasian and Asian samples were collected from individuals of self-declared ethnicity. The ethnicity of some sample donors was inferred based on donor surnames. The Asian dataset included individuals from South East Asia. The Caucasian dataset included individuals from Australia, Europe and the Middle East.

All duplicate profiles were removed. A summary of the submitting agencies and numbers of profiles is provided in Table 1. Samples were collected under relevant local

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Table 1. Summary of participating laboratories and sample numbers submitted.

Laboratory	Number of samples submitted			
	Aboriginal declared	Aboriginal NT pure	Asian	Caucasian
NSW Forensic & Analytical Science Service (New South Wales)	200	-	200	200
Forensic Science Branch NT Police (Northern Territory)	197	546	176	190
Forensic & Scientific Services (Queensland)	309	-	126	139
Australian Federal Police (Australian Capital Territory)	-	-	-	223
Forensic Science South Australia	249	-	261	355
PathWest, Western Australia	198	-	199	200
Forensic Science Service Tasmania	-	-	-	200
Victoria Police Forensic Services Department, Victoria	274	-	28	200

government legislation. This paper describes a summary of data analysis. The complete dataset is not available owing to legislative reasons.

Laboratory methods

All submitting agencies are accredited by the National Association of Testing Authorities (http://www.nata.asn.au/) and participate in annual external proficiency testing programmes.

Amplification: Extracted DNA was amplified using Promega's PowerPlex 21 testing kit (Madison, WI) following the manufacturer's recommended guidelines (or as half volume reactions).

Typing: Amplified DNA was separated on either an Applied Biosystems' 3130*xl* or 3500*xl*. Profiles were analysed using Applied Biosystems' GeneMapper ID or ID-X (Life Technologies, Carlsbad, CA) using the individual laboratory's analysis settings.

Results

All variant and microvariant alleles assigned with confidence were retained in the dataset. Ambiguous off-ladder alleles and tri-allelic loci were removed from the dataset and appear as 'R' alleles in the allele frequency tables (refer to online supplementary material). Allele frequencies and estimated values for observed and expected heterozygosity (H_O and H_E, respectively) for each population are available in the online supplementary material.

Table 2. Results of the truncated product method for assessing independence testing of each of the Australian subpopulation datasets.

Subpopulation	HWE		LE	
	Sum[-2ln(p)]	p-value	Sum[-2ln(p)]	p-value
Aboriginal - declared	69.99	2.33E-03	573.65	4.73E-10
NT Aboriginal - pure	139.05	7.20E-13	831.62	8.74E-36
Asian	66.41	5.43E-03	444.47	1.25E-02
Caucasian	50.44	1.25E-01	422.28	6.63E-02

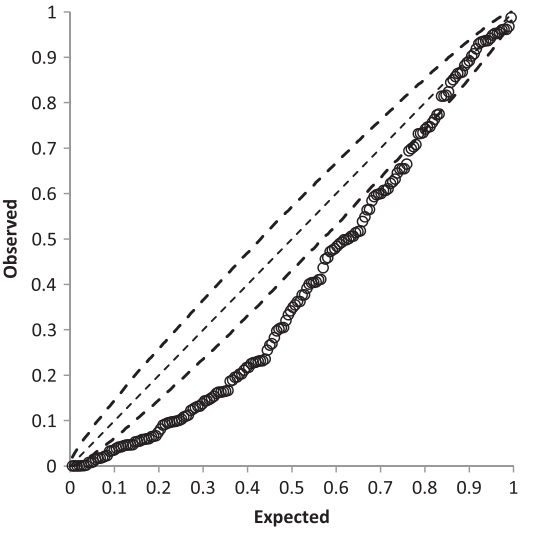

Figure 1. *p–p* plot for the Australian Aboriginal declared dataset.

Genetic Data Analysis software (courtesy of Paul Lewis, available at http://www.eeb.uconn.edu/people/plewis/software.php) was used to calculate *p* values for allelic association obtained from Fisher's exact test. Data were shuffled 10,000 times. Results of these tests show that there were significant deviations (at the 5% level) from Hardy-Weinberg equilibrium and linkage equilibrium at some loci for each subpopulation (appearing as bold values in the supplementary material).

The truncated product method of Fisher was used to test the hypotheses that none of the loci departed from Hardy-Weinberg and linkage equilibrium[1]. The results are presented in Table 2. Representation of the results of independence testing for each of the subpopulations were also undertaken graphically, using *p-p* plots, in Figures 1

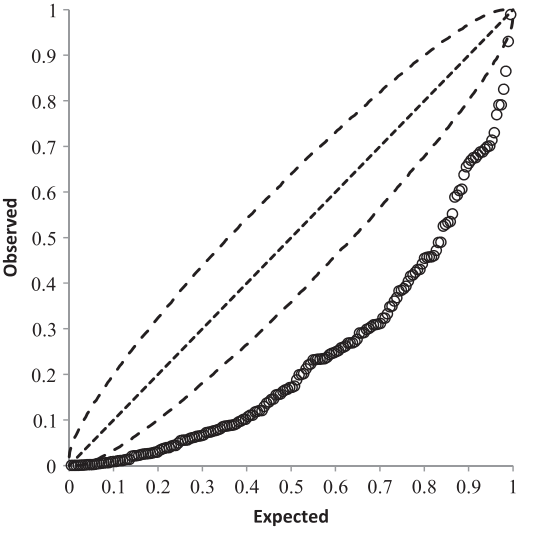

Figure 2. *p–p* plot for the Northern Territory Pure Aboriginal dataset.

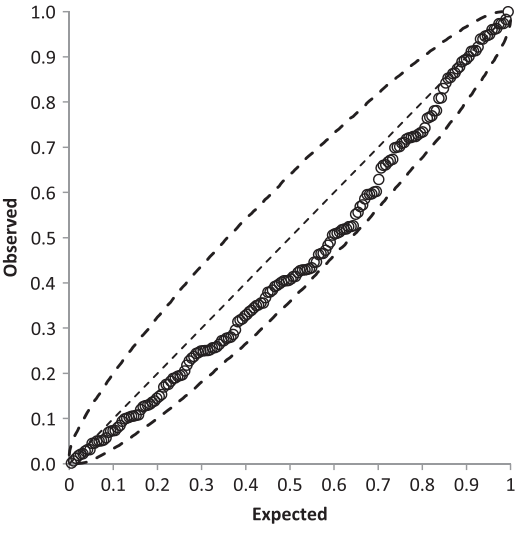

Figure 3. *p–p* plot for the Australian Asian dataset.

through 4, following Buckleton[2]. These allow a level of visual comparison of the results. If the hypotheses of Hardy-Weinberg and linkage equilibrium were true then the *p*-values should be distributed uniformly between 0 and 1; $p \sim U[0, 1]$. As an example, the $x = y$ line in the *p-p* plots represents equilibrium and deviations from that line can be seen as departures from equilibrium. The 95% confidence limit is also displayed on the *p-p* plots as the region within the two curved lines.

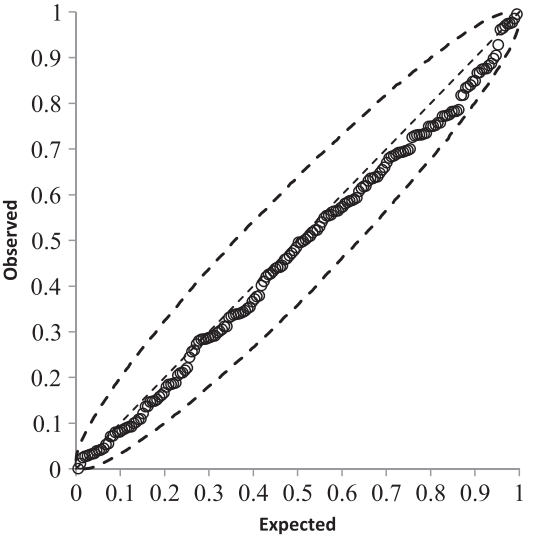

Figure 4. *p–p* plot for the Australian Caucasian dataset.

Conclusions

Within the dataset we have found evidence of the 'multi-testing problem'; 20 Hardy-Weinberg and 190 linkage-equilibrium tests have been undertaken for each subpopulation. About 5% of these are expected to give significant results by chance alone. The *p*-values for the truncated product method (Table 2) for the HWE tests would be deemed significant at the 5% significance level for each subpopulation and as such would be taken as evidence for departure from HW.

In the *p*-*p* plot for the Australian Asian and Caucasian subpopulations the data remains within the boundaries of the 95% confidence limits. The Aboriginal populations exhibit clear departures from expected *p*-values, and hence are neither in Hardy-Weinberg nor linkage equilibrium. Regardless of the results of independence testing, our knowledge of the history of human populations leads us to expect that some level of admixture or substructure exists in all populations.

The Australian Aboriginal (declared and pure), Asian and Caucasian subpopulation databases are of suitable size for the purpose of estimating allele frequencies. For criminal investigations, it is recommended that multi-locus profile probabilities be calculated by the method of Balding and Nichols[3] with conservative values of the inbreeding coefficient (θ or Fst). The Fst values recommended are 0.05, 0.03, and 0.02 respectively for the Aboriginal, Asian and Caucasian datasets based on the work undertaken by Curran et al.[4].

Supplemental material

Allele frequency data and the results of tests for linkage disequilibrium for all four sub-populations are available as supplemental material online.

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