Forensic DNA profiling:
Establishing allele frequencies for South Africa
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Background and motivation

• Data in South Africa to date:
  – Lucassen et al. 2014 (15 STRs, AmpFISTR® Identifiler Plus™)
  – Ristow et al. 2016 (22 STRs, GlobalFiler™ Express)

• Research and development at Salt River Mortuary
  – Challenging samples

• Investigator 24plex GO! Kit (QIAGEN)
  – Quality sensor
Aims and objectives

Aim
To generate forensic DNA profile data for South Africa using the QIAGEN Investigator 24plex GO! Kit

Objectives
• Collect biological samples from South African individuals
• Establish a forensic DNA profiling workflow
• Determine the alleles and frequencies of the STR loci
• Disseminate the results for use in the broader forensic community
Methods overview

- Ethics approval
- Participant recruitment and sample collection
- Sample preparation and processing
- Data analysis and statistics
• Ethics approval
  – This study obtained ethics approval from the UCT Faculty of Health Science, Human Research Ethics Committee (HREC REF: 342/2016)

• Informed consent
• Questionnaire
• Considerations
• Population groups (ancestry)
  - African
  - European
  - Indian/Asian
  - Mixed

• Recruitment: ~800 individuals
• Sample collection
  – Buccal swab (and extracted DNA)
• Optimisation of workflow
  – Establishment of thresholds (SWGDAM)
• Sample preparation and amplification
  – Lysed sample: Direct PCR
  – Extracted DNA: qPCR + PCR
• Capillary electrophoresis

All according to manufacturer's instructions
- Analyse and interpret electropherograms
  - GeneMapper versus GeneMarker
- Frequency tables
- Hardy-Weinberg equilibrium
- Differences between population groups

Data analysis and statistics was performed using Microsoft Excel and STATA
Optimisation: PCR cycle number

Extracted DNA: 27 cycles

Lysates:
- 24 cycles
- First time success: 98.62%

Allelic ladder

Lysate (27 cycles)

Lysate (24 cycles)
Tri-allelic patterns

- TPOX, allele 10
- Lane (2008)
  - African: 2.4%
- Ristow et al. (2016)
  - African: 1.5%
Additional sex marker: DYS391

Null allele in Ame (Butler, 2009)

Artefact in DYS391 (Moore et al., 2016)
Allele frequencies

- Significant differences between population groups
Novel alleles

- 16 new alleles in 27 individuals
- SE33, D13S317, D2S441, D22S1045, D19S433, FGA, D5S818
Limitations and way forward

• Cohort: demographics were skewed towards the Western Cape
• Limited sample size for Indian/Asian population group
• Ethnic data for some indigenous Black African individuals were missing
• Verify novel alleles by sequencing
Conclusion

• QIAGEN Investigator® 24 PLEX GO! kit was used to successfully generate DNA profiles
• Novel alleles accentuates importance of local database
• Second sex marker is important
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References


• Lane, A.B. 2008 The nature of tri-allelic TPOX genotypes in African populations. Forensic Science International: Genetics 2 134-137

• Lucassen, A., Ehlers, K., Grobler, P.J. and Shezi, A.L. 2014 Allele frequency data of 15 autosomal STR loci in four major population groups of South Africa. Int. J. Legal Med. 128 275–276

• Moore, D., Clayton, T. and Thomson, J. 2016 Description of artefact in the PowerPlex Y23® system associated with excessive quantities of background female DNA. Forensic Science International: Genetics 24 44-50

• National Institute of Standards and Technology (NIST) website (www.cstl.nist.gov/biotech/strbase/). Date accessed; 2017.05.07.


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