



# Forensic DNA profiling:

## Establishing allele frequencies for South Africa

7 March 2018

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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

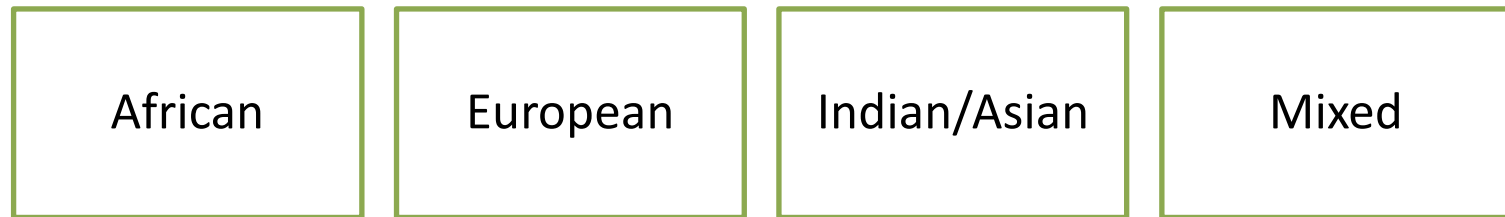




- Ethics approval
  - This study obtained ethics approval from the UCT Faculty of Health Science, Human Research Ethics Committee (HREC REF: 342/2016)
- Informed consent
- Questionnaire
- Considerations



- Population groups (ancestry)



- Recruitment: ~800 individuals
- Sample collection
  - Buccal swab (and extracted DNA)



- Optimisation of workflow
  - Establishment of thresholds (SWGDM)
- Sample preparation and amplification
  - Lysed sample: Direct PCR
  - Extracted DNA: qPCR + PCR
- Capillary electrophoresis

All according to manufacturer's instructions

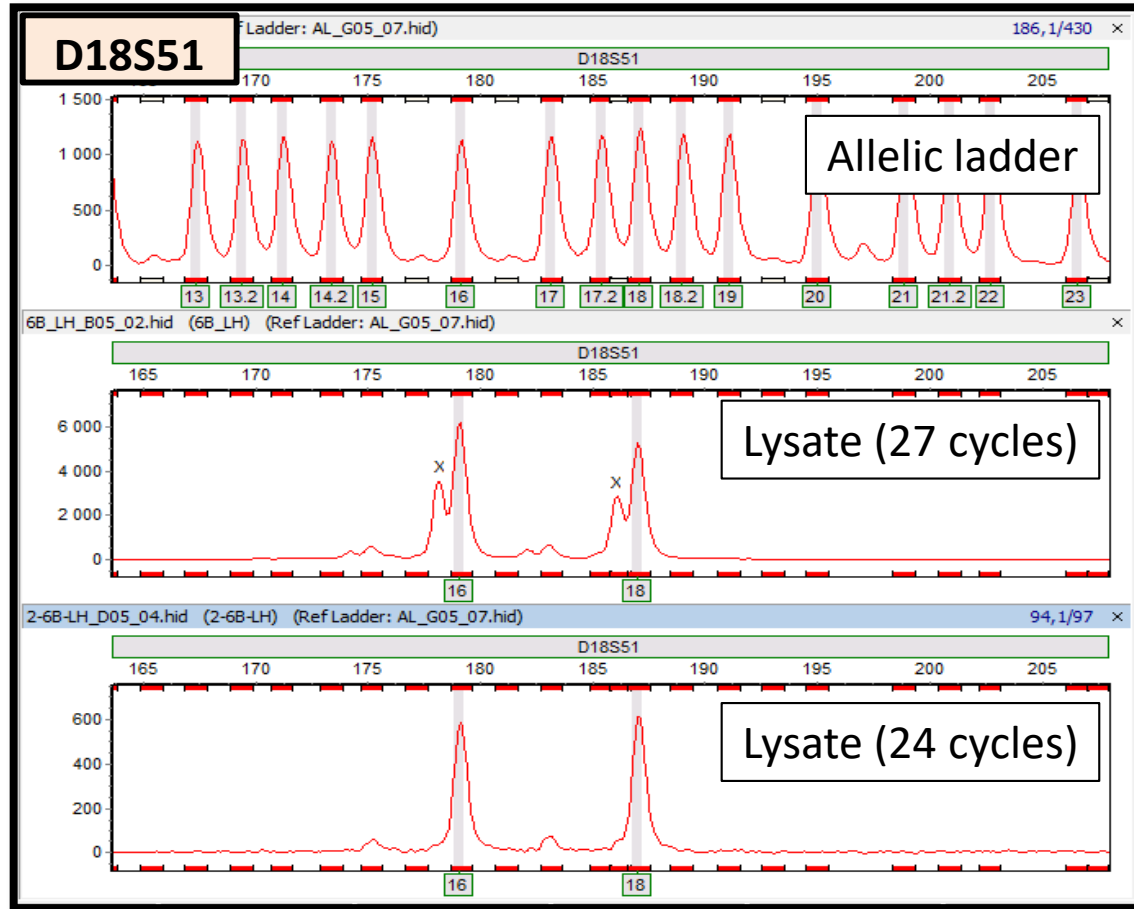




- Analyse and interpret electropherograms
  - GeneMapper versus GeneMarker
- Frequency tables
- Hardy-Weinberg equilibrium
- Differences between population groups

Data analysis and statistics was performed using Microsoft Excel and STATA

# Optimisation: PCR cycle number



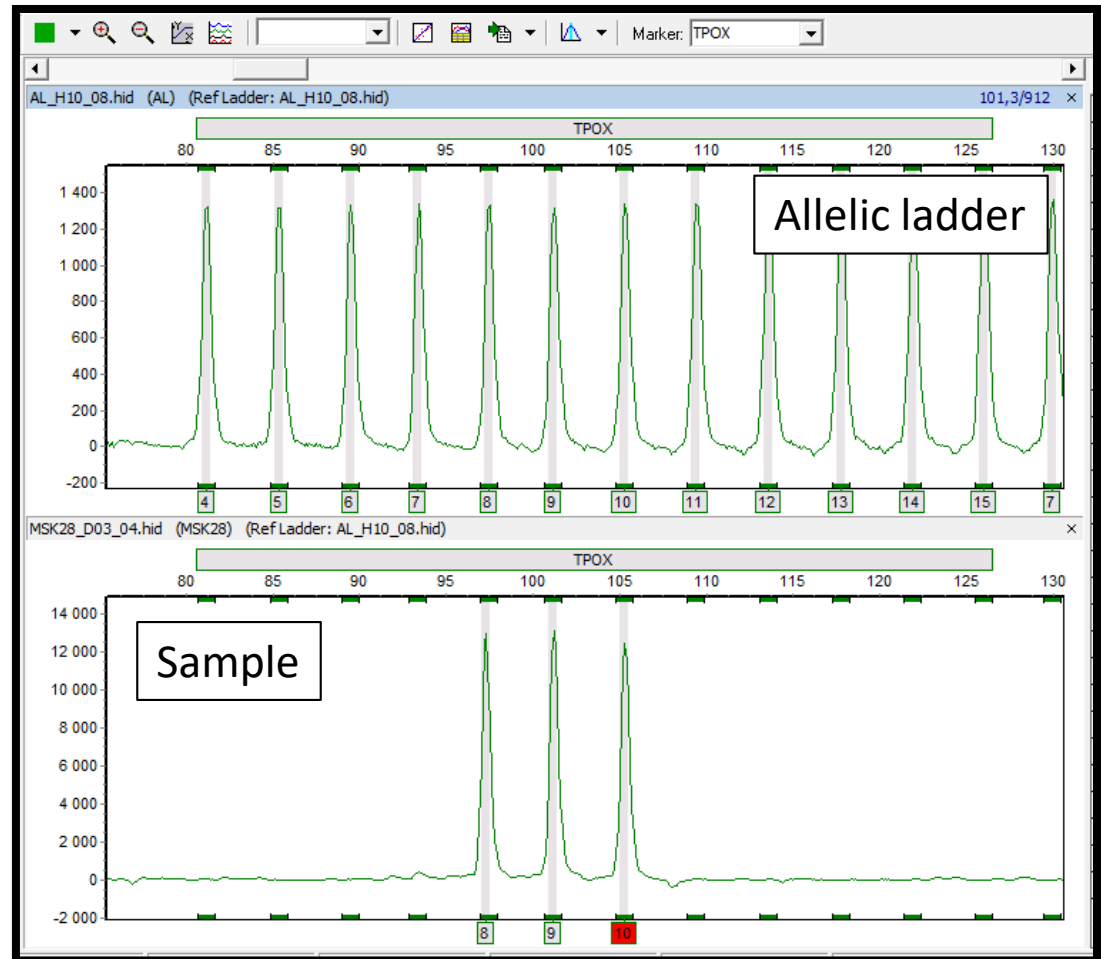
Extracted DNA:  
27 cycles

Lysates:  
24 cycles

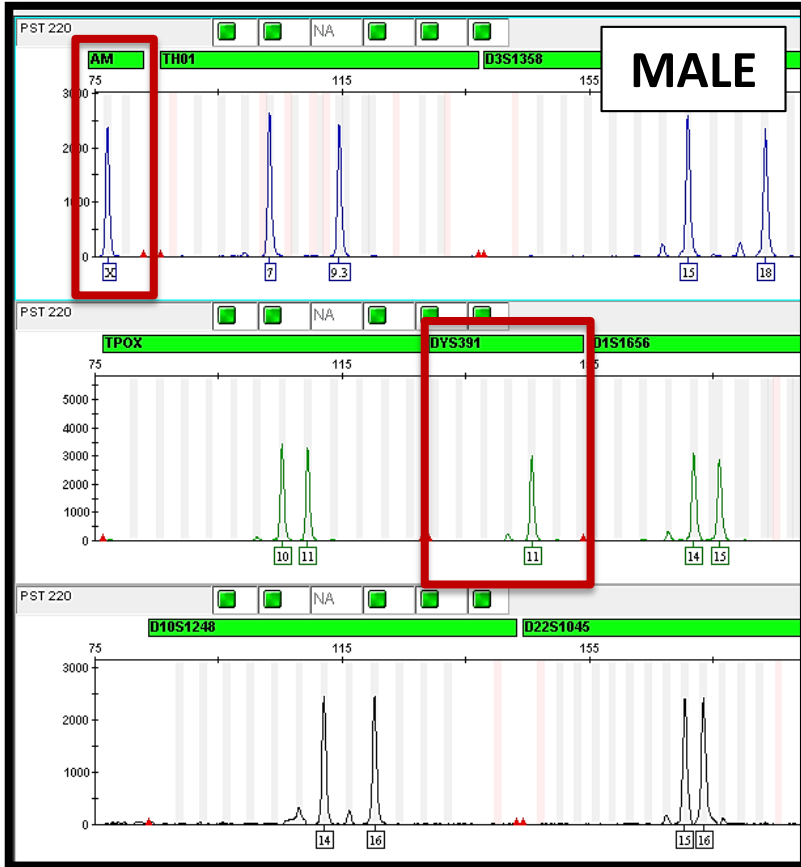
First time  
success: 98.62%

# Tri-allelic patterns

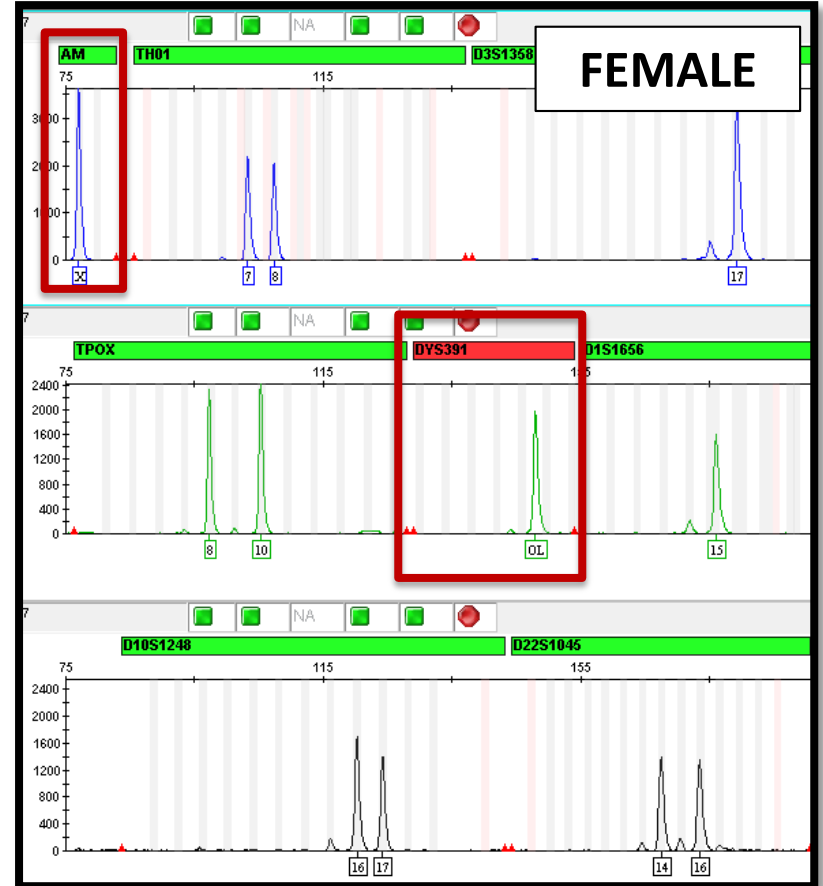
- TPOX, allele 10
- Lane (2008)
  - African: 2.4%
- Ristow *et al.* (2016)
  - African: 1.5%



# Additional sex marker: DYS391



Null allele in Ame (Butler, 2009)

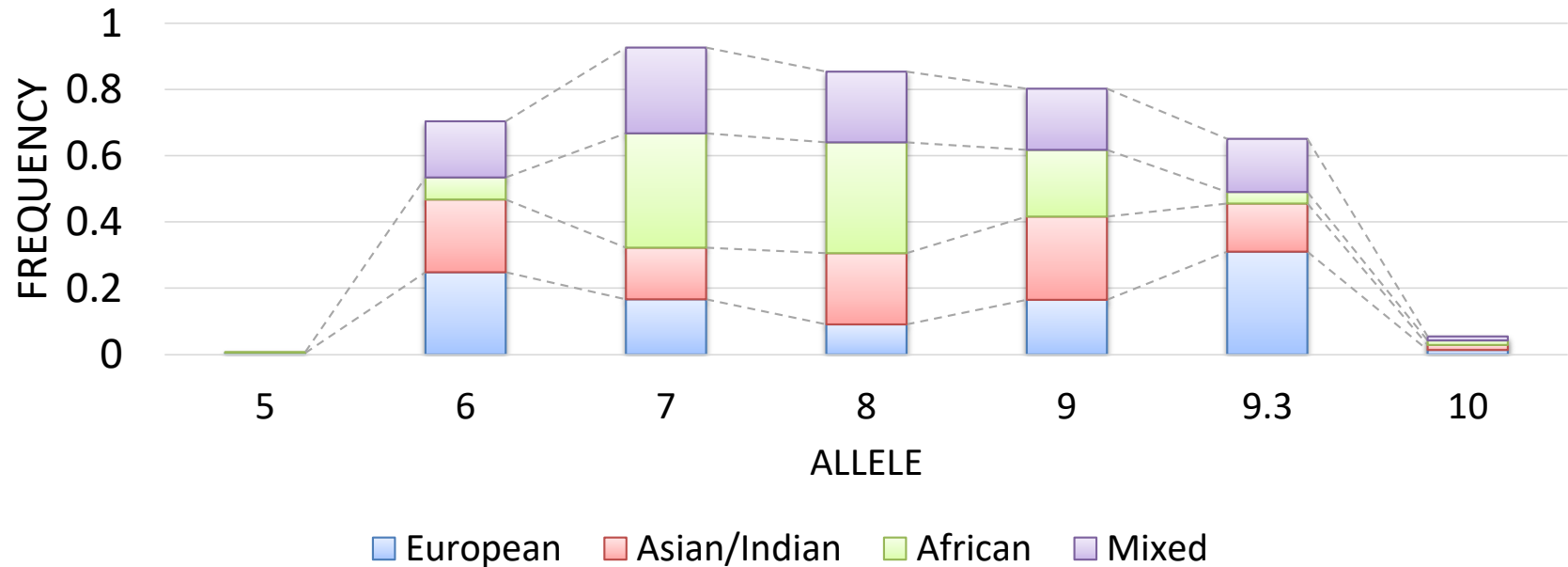


Artefact in DYS391 (Moore *et al.*, 2016)

# Allele frequencies

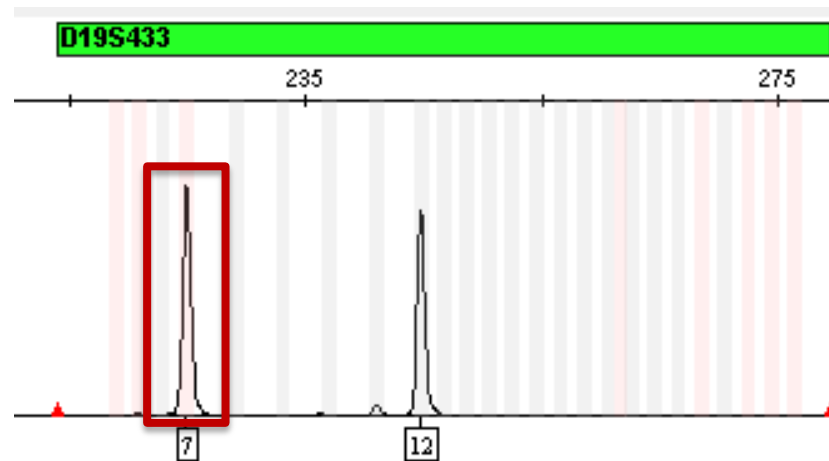
- Significant differences between population groups

Allele frequencies for THO1



# 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<sup>®</sup> 24 PLEX GO! kit was used to successfully generate DNA profiles
- Novel alleles accentuates importance of local database
- Second sex marker is important



# Acknowledgements



- **Lorraine Nel** (University of Cape Town)
- Kate Reid (University of Cape Town)
- Calvin Mole (University of Cape Town)
- Laurent Moncomble (QIAGEN)
- Ryan Vogt (Whitehead Scientific)
- Maryvonne Abrahams (Unistel Medical Laboratories)
- Kayla Hendricks (SoftGenetics LLC)

# References

- Butler, J.M. 2009: 167-168. Fundamentals of Forensic DNA Typing. Elsevier, London, United Kingdom. [ISBN: 978-0-12-374999-4]
- 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/](http://www.cstl.nist.gov/biotech/strbase/)). Date accessed; 2017.05.07.
- Promega Reference Manual. Internal validation guide of autosomal STR systems for forensic laboratories. Available online. <https://www.promega.jp/-/media/files/resources/validation-guides/internal-validation-of-autosomal-str-systems.pdf?la=ja-jp> [Date accessed: 2015.08.03]
- SWGDAM Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories. Available online. <https://www.fbi.gov/about-us/lab/biometric-analysis/codis/swgdam-interpretation-guidelines> [Date accessed 2015.08.03]

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