

Forensic Science Regulator

Guidance

Y- STR Profiling

FSR-G-227

Issue 1

© Crown Copyright 2021

The text in this document (excluding the Forensic Science Regulator's logo, any other logo, and material quoted from other sources) may be reproduced free of charge in any format or medium providing it is reproduced accurately and not used in a misleading context. The material must be acknowledged as Crown copyright and its title specified.

This document is not subject to the Open Government Licence.

Guidance – Guidance – Guidance – Guidance – Guidance – Guidance – Guidance –

- 1. Introduction5
- 1.1 Background.....5
- 1.2 Y-STR profile considerations8
- 2. Scope.....10
- 3. Standards for Y-STR Profiling.....10
- 3.2 Existing Guidelines for Y-STR Profiling.....10
- 3.3 Guidelines Within the Forensic Science Regulator’s Codes11
- 4. Modification.....11
- 5. Terms and Definitions12
- 6. Implementation12
- 7. Y-STR Profile Evaluation12
- 7.1 Y-STR Profiles from Single Individuals (‘Single-Source Profiles’)12
- 7.2 Y-STR Profiles from More Than One Individual (‘Mixed Profiles’)13
- 8. Y-STR Statistical Evaluation14
- 8.1 Criteria for Suitability for Statistical Evaluation.....14
- 8.2 Haplotype (Profile) Frequency Databases14
- 8.3 Choice of Reference Population15
- 8.4 Choice of Method for Probability Estimation16
- 8.5 Choice of multiplex for evaluation purposes17
- 9. Reporting Outcomes of Statistical Evaluation18
- 9.2 Alternatives to Statistical Evaluation18
- 9.3 Future Challenges.....20
- 10. Quality Assurance for Y-STR Profiling.....20
- 10.1 Quality Assurance Checks.....20
- 10.2 Co-processing of Quality Assurance Controls21
- 11. Investigation of Potential Contamination in Y-STR Profiles22

Guidance – Guidance – Guidance – Guidance – Guidance – Guidance – Guidance –

11.2 Sharing Unsourced Contaminant Y-STR Profiles23

11.3 Y-STR Elimination Databases23

11.4 Creation of a Y-STR Elimination Database.....23

11.5 Searching Y-STR Elimination Databases25

12. Requirements for Forensic Unit Guidelines for Interpreting the Presence
and Designation of Peaks26

13. Acknowledgements.....27

14. Review28

15. References28

16. Abbreviations and Acronyms31

17. Glossary.....32

18. Further Reading.....35

1. Introduction

1.1 Background

- 1.1.1 Humans have 23 pairs of chromosomes, 22 of which are autosomes passed on with equal likelihood to both sons and daughters, the 23rd pair being the sex chromosomes. A child inheriting an X from their father will be genetically female (denoted 46, XX) while one inheriting a Y will be male (46, XY) as a consequence of the male-determining gene (*SRY*) located near the end of the short arm of the Y chromosome.¹ Most genes on the Y are unique to the chromosome and are associated with fertility and the production of sperm, whilst some share homology with the X chromosome, for example, the *Amelogenin* gene used in the DNA17 test to attribute chromosomal sex. Y-STRs have similar structures and levels of variability to autosomal STRs and share many of the profile characteristics such as broadly similar levels of stutter. However, they normally display only a single allele peak as males have only one Y chromosome copy.²
- 1.1.2 Unlike autosomal STRs, which are inherited independently of each other due to the random assortment of different chromosomes and recombination between paired chromosomes, the STRs on the Y chromosome are inherited from father to son as a single non-recombining set. This lack of independent inheritance means that the product rule cannot be applied to Y-STR allele frequencies. In addition, all patrilineally related males will have exactly the same combination of Y-STR alleles (known as a haplotype) unless a mutation occurs. The most common form of mutation is the alteration of the length of an STR (usually by the loss or gain of a single repeat unit).
- 1.1.3 Null alleles can arise in Y-STR profiles due to alteration or loss of the primer binding site, or loss of the entire Y-STR through deletion. Deletion and duplication events occur relatively frequently on the Y chromosome (Jobling,

¹ Although the *SRY* gene is usually on the Y chromosome, it occasionally gets transferred to the X, leading to 46,XX males, whilst inactivation of *SRY* by mutation leads to 46,XY females (Swyer Syndrome); however, both are rare occurrences (approximately 1 in 20,000 individuals).

² Approximately 1 in 1,000 males have 2 identical copies of their Y chromosome (47,XYY); this trait is not inherited.

2008) [1], promoted by rearrangements between the repeated structures that flank blocks of Y-STRs. As a consequence it is not uncommon for sets of adjacent STRs to be simultaneously lost or duplicated. Duplication events may only be detectable from a doubling of peak height of the duplicated STRs but eventually mutation will affect one copy and result in the appearance of a new allele, typically either one repeat larger or smaller than the original copy. Some deletions are also observed, for example, the Amelogenin Y (AMELY) deletions detected during DNA17 profiling are usually accompanied by the loss of several adjacent Y-STRs lying between the loci DYS456 and DYS19 (Figure 1); these are seen relatively commonly in males from the Indian subcontinent, at a frequency of approximately 0.02 (Jobling et al., 2007) [2].

1.1.4 Y-STR profiling has become well established in UK forensic casework since the introduction of a 12 Y-STR test (PowerPlex® Y System; Promega) in 2003, followed by a 17 Y-STR multiplex (AmpFLSTR™ Yfiler™; ThermoFisher) and subsequently by the 23 Y-STR PowerPlex® Y23 System (PPY23; Promega). Each new test added STRs while retaining those present in the preceding multiplex, the exception being Yfiler™ Plus (ThermoFisher), the most discriminating test currently (February 2021) widely available, which includes 27 Y-STRs but omits two that are present in PPY23. Early tests were restricted to the few Y-STRs that had been characterised at the time, and included some STRs with low mutation rates (less than approximately 0.001 per STR per generation) and consequent poor discrimination power. More recent multiplexes have incorporated rapidly mutating (RM) Y-STRs, which have mutation rates greater than 0.01 per generation (Ballantyne et al., 2010) [3]. With the inclusion of several of these in the PPY23 and Yfiler™ Plus kits it is unusual to observe matching profiles in males not known to be closely related. Figure 1 shows the physical order of the Y-STRs, and the multiplexes that detect them. Note that locus DYS385 (present in all the multiplexes) and the multi-copy locus DYS387S1 (included in Yfiler™ Plus only) are duplicated in the vast majority of males, displaying either two separate allele peaks or a single peak of approximately double the normal height.

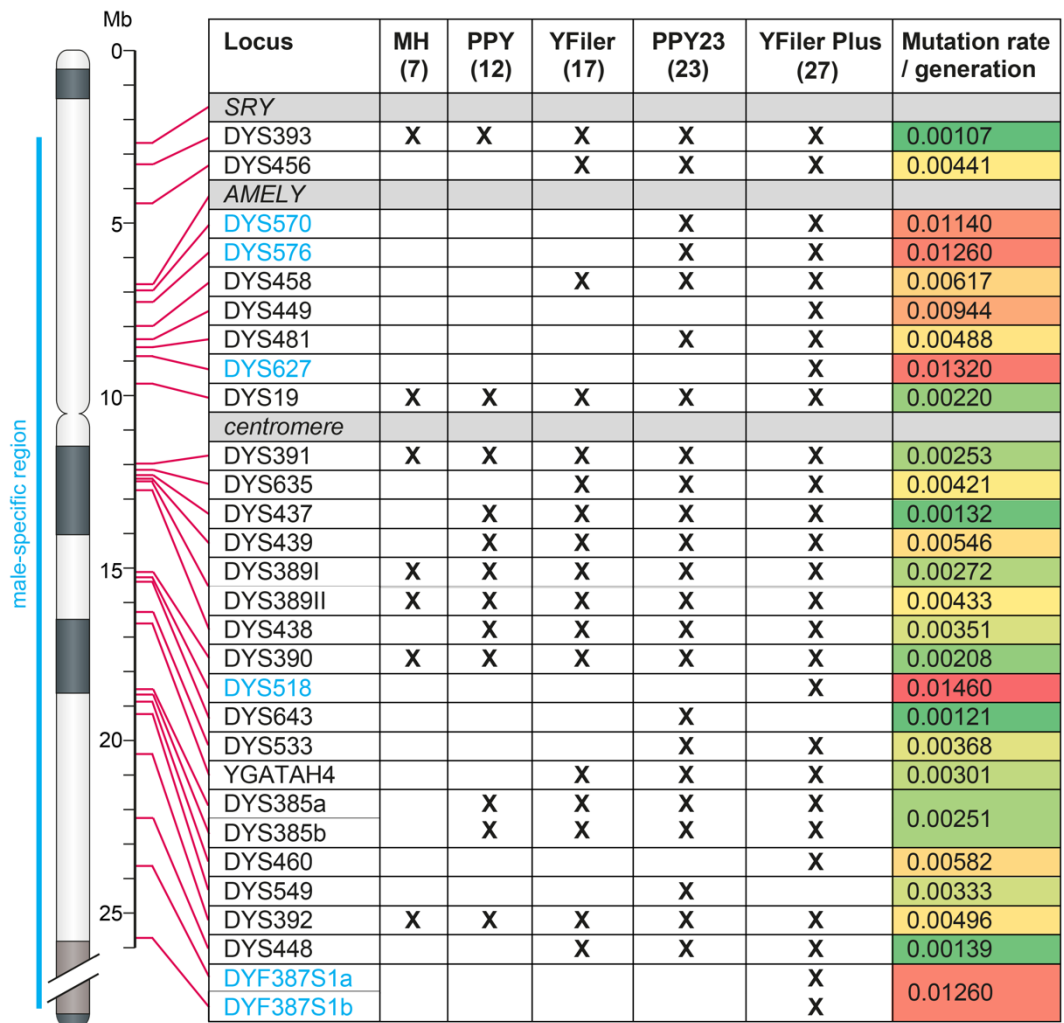


Figure 1: Physical positions of Y-STRs on the Y chromosome, inclusion in multiplexes, and mutation rates

Notes:

1. MH: minimal haplotype.
2. PPY: PowerPlex Y.

Physical positions of STRs are taken from Hanson and Ballantyne (2006) [4] and mutation rates are from the Y-STR Haplotype Reference Database (YHRD) Release 62; loci given in blue font (DYS570,576,627,518,387S1a and387S1b) are considered RM Y-STRs. The positions of the SRY and AMELY genes are also shown, as is that of the centromere.

1.2 Y-STR profile considerations

- 1.2.1 When the Y-STR profile from a crime stain does not match that from a suspect, this excludes the suspect. However, due to the Y chromosome's mode of inheritance, Y-STR profiling cannot distinguish between males who are paternally related unless an individual male displays a newly arisen mutation absent from his male-line relatives who would otherwise share the same profile. Even then there is a possibility that the same mutation may also have occurred in a more distant patrilineal relative, generating the same profile.
- 1.2.2 The complete absence of Y-STRs in females allows male-specific profiles to be clearly detected in mixtures even when the female DNA component is very much more abundant, for example, greater than 100,000-fold excess (Moore et al., 2016) [5]. Consequently, Y-STR profiling is invaluable in sexual assault casework where the male autosomal component cannot be isolated by alternative means such as preferential lysis (Gill et al., 1985) [6], for example, azoospermic or vasectomised males, non-ejaculation or digital penetration. Y-STR profiles may also be detectable for longer periods after intercourse, as DNA released from degraded sperm and epithelial cells could still be detected. In such cases the Y-STR profile can be compared with a reference Y-STR profile from a suspect or other persons of interest.
- 1.2.3 The UK currently lacks a database of either Y-STR crime stains or reference sample profiles from arrestees, and all comparisons are therefore made on a case-by-case basis. At the time of writing (February 2021) other countries (Austria, China, Italy and Singapore) have incorporated Y-STRs into their national databases, increasing the investigatory power of their DNA analysis.
- 1.2.4 Because Y-STR profiling tends to be used in challenging situations where recovery is low and the DNA may be degraded, it is not uncommon that the obtained profile is partial in nature. While the expectation of only a single allele at most STRs simplifies the interpretation of Y-STR profiles affected by drop-out, the much lower discriminating power of Y-STR profiles significantly increases the likelihood that partial profiles will result in adventitious matches with both close and distant male-line relatives.

- 1.2.5 The unexpected occurrence of more than one allele at a Y-STR locus may indicate either a duplication event in the genome of the source individual, or DNA from multiple contributors. A check of whether the duplicated STRs are adjacent on the Y chromosome (Figure 1) can help to determine which explanation is more likely. The absence of a Y-STR allele in an otherwise good quality Y-STR profile indicates either a primer-binding site mutation or a deletion. When more than one Y-STR is absent, this is likely due to the same deletion event, and checking of the relative positions of the STRs on the Y chromosome can again help to determine this. However, it remains possible that alternative arrangements of the Y chromosome exist in some populations in which the relative positions of STRs as shown in Figure 1 do not apply.
- 1.2.6 The mode of inheritance of a Y-STR haplotype (profile) provides a useful means of establishing kinship through the male line. An identical profile is usually indicative of relatively recent shared ancestry and the more discriminating the multiplex (a combination of the number and mutation rates of the included STRs) the more recent that shared ancestry is likely to be. For example, the mean mutation rate of a haplotype defined by the PPY23 multiplex is 7.9% per generation (the sum of the per-STR mutation rates in Figure 1). Therefore two men who share a common male-line ancestor six generations ago (total, 12 generations) are extremely likely to carry different PPY23 profiles. In practice, most men observed to share a given male's PPY23 profile will be his close male-line relatives, such as a brother, uncle, nephew, or cousin. Y-STR profiling is therefore a useful tool for establishing the significance of autosomal profile similarity in familial screens where the relationship between the individual on the database and the source of the crime stain is patrilineal.
- 1.2.7 Because surnames, like Y chromosomes, are passed from father to son in most societies (King and Jobling, 2009) [7], men sharing surnames are more likely to share similar Y-STR profiles than men with different surnames. This relationship has been used in conjunction with genetic genealogy websites to identify very distant male-line relatives. On a longer timescale, men with ancestry tracing back to the same geographic region are more likely to share

similar profiles, and profiles can be grouped into clusters that are more abundant in particular continents and/or countries. However, because of recent population migration and admixture, association with geographic regions or ethnic groups is not wholly accurate. Nonetheless, the relative similarity of male profiles within such groups is significant and necessitates the use of a profile frequency reference database that reflects the likely origin of the person of interest.

- 1.2.8 The guidance in this document is based on published work. There has also been widespread consultation with all of the major providers of forensic science services in the UK and Ireland and academia via the Forensic Science Regulator DNA Specialist Group.

2. Scope

- 2.1.1 The purpose of this document is to provide guidance for Y-STR analysis delivered into the criminal justice system in England and Wales.
- 2.1.2 It applies to profile (haplotype) comparison and kinship testing and does not apply to ancestry testing.

3. Standards for Y-STR Profiling

- 3.1.1 National and international standards for testing and calibration in laboratories (British Standard BS EN ISO/IEC 17025; International Laboratory Accreditation Cooperation ILAC G19:08/2014) [8]; [9] provide guidance on analytical methods. However, there is much less detail for the type of interpretation of analytical results required for Y-STR analysis than for autosomal DNA analysis.

3.2 Existing Guidelines for Y-STR Profiling

- 3.2.1 Scientific and technical guidelines that are relevant to (but not mandatory for) the interpretation of Y-STR profiles have been published by the International Society for Forensic Genetics (ISFG) DNA Commission (Gill et al., 2001; Gusmão et al., 2006; Roewer et al., 2020) [10]; [11]; [12] and by the Scientific Working Group on DNA Analysis Methods (SWGDM, 2014) [13].

3.3 Guidelines Within the Forensic Science Regulator’s Codes

3.3.1 In addition to the Forensic Science Regulator’s (FSR’s) FSR-C-100 Codes of Practice and Conduct (the Codes) [14] the following are also relevant to this topic:

- a. FSR-C-108 DNA Analysis [15];
- b. FSR-G-201 Validation [16];
- c. FSR-G-208 The control and avoidance of contamination in laboratory activities, involving DNA evidence recovery and analysis [17];
- d. FSR-G-213 Allele frequency databases and reporting guidance for DNA (short tandem repeat) profiling [18];
- e. FSR-G-222 DNA mixture interpretation [19];
- f. FSR-G-223 Software validation for DNA mixture interpretation [20]; and
- g. FSR-P-302 DNA contamination detection: The management and use of staff elimination DNA databases [21].

4. Modification

4.1.1 This is the first issue of this document.

4.1.2 The Regulator uses an identification system for all documents. In the normal sequence of documents this identifier is of the form ‘FSR-#-###’ where (a) (the first ‘#’) indicates a letter to describe the type of document and (b) ‘###’ indicates a numerical, or alphanumerical code to identify the document. For example, this document is FSR-G-227, and the ‘G’ indicates that it is a guidance document. Combined with the issue number this ensures that each document is uniquely identified.

4.1.3 If it is necessary to publish a modified version of a document (for example, a version in a different language), then the modified version will have an additional letter at the end of the unique identifier. The identifier thus becoming FSR-#-####.

4.1.4 In all cases the normal document bearing the identifier FSR-#-### is to be taken as the definitive version. In the event of any discrepancy between the normal version and a modified version then the text of the normal version shall prevail.

5. Terms and Definitions

- 5.1.1 The terms and definitions set out in the Forensic Science Regulator's (FSR's) Codes of Practice and Conduct, FSR-C-100 (the Codes) [14], FSR-C-108 DNA Analysis [15], FSR-G-222 DNA mixture interpretation [19] (for ease of reference some definitions are reproduced) and the glossary (section 17) apply to this document.
- 5.1.2 The word 'shall' has been used in this document where there is a corresponding requirement in British Standard BS EN ISO/IEC 17025 [1], or the Codes [2]; the word 'should' has been used to indicate generally accepted practice where the reason for not complying or any deviation shall be recorded.

6. Implementation

- 6.1.1 This guidance is available for incorporation into a forensic unit's quality management system from the date of publication. The Regulator required that the Codes [14] were included in a provider's schedule of accreditation from October 2017 and the requirements in this guidance are effective from 01 October 2021.

7. Y-STR Profile Evaluation

7.1 Y-STR Profiles from Single Individuals ('Single-Source Profiles')

- 7.1.1 Y-STR profiles with no more than one component (allele) at each STR (other than the locus DYS385 and other constitutively duplicated STRs) can be considered as a single source.
- 7.1.2 If the alleles are well amplified, then the Y-STR profile can be confidently assigned as originating from a single individual (although the possibility of a mixture of male paternal-line relatives should also be considered). However, in less well amplified Y-STR profiles there may be reduced confidence in assigning the male DNA detected as being from a single source.

7.1.3 Where the male DNA in a crime sample appears to be from a single source, it can be compared directly with a reference Y-STR profile from a person of interest (POI) to determine whether they match or not.

7.1.4 If the profiles match at all loci for which a designation has been made then the crime sample may have originated from the individual who provided the reference sample, or from a member of the same paternal lineage. This finding should be evaluated as described below.

7.1.5 If the allele designations mismatch at one or more loci (and non-concordance due to differing polymerase chain reaction chemistries can be ruled out) then the male DNA recovered from the crime sample is not from the individual who provided the reference sample and should be reported as such.

7.2 Y-STR Profiles from More Than One Individual ('Mixed Profiles')

7.2.1 Y-STR profiles with more than one component (allele) at a Y-STR locus (other than DYS385, which commonly has two components) should be considered as possible mixed profiles derived from DNA from more than one male.

7.2.2 In rare cases, genomic duplication events may result in single-source profiles presenting more than one component at one or more loci, but these are unusual and nearly always restricted to a small number of loci and the duplicated peaks are well balanced.

7.2.3 As with mixed autosomal DNA mixtures, where there is a clear major contributor to the mixed Y-STR profile obtained, this major Y-STR profile can be evaluated as if it is a single-source result. The rules for safe deconvolution should be defined based on laboratory validation data. This approach is only permissible if pursued with due regard for logic, taking into account all loci, and only where it is not based on the results of the comparison of the trace with that of the POI.

7.2.4 In some cases it may be possible to condition a mixed Y-STR profile on the assumed contribution of a known male other than the POI. Commonly, this may be a known sexual partner of a female victim of a sexual offence. In

such cases it may be possible to determine some or all of the Y-STR components in the mixture that originated from a male other than the known partner.

7.2.5 In both these circumstances the deduced profile can be compared directly with a reference Y-STR profile from a POI to determine whether they match or not, as described for single-source profiles.

7.2.6 However, mixed Y-STR profiles are often encountered where a clear major contributor cannot be identified. In such cases, a comparison of the mixed profile with the profile of a POI may be possible, but the findings cannot be evaluated using the currently available (February 2021) statistical tools described below.

8. Y-STR Statistical Evaluation

8.1 Criteria for Suitability for Statistical Evaluation

8.1.1 Only profiles interpreted as being from a single individual can currently be considered for statistical evaluation. Statistical evaluation is possible for complete or partial profiles meeting one of the following criteria. Profiles not meeting the following criteria cannot be considered for statistical evaluation.

- a. Single-source full profile.
- b. Single-source partial profile.
- c. Unambiguous major or minor contributor to a mixed profile. Full or partial designation of the major or minor contributor profile. Deduced profiles derived from mixed Y-STR profiles may be incomplete because of locus drop-out; where there is ambiguity in deconvolution, profiles should not be interpreted as if they were a single source.
- d. Contributor to a mixture, conditioned on a known contributor. Full or partial designation of the deduced contributor profile.

8.2 Haplotype (Profile) Frequency Databases

8.2.1 To estimate the weight of evidence in Y-STR cases where a suspect profile matches a crime-scene profile, an estimate of profile frequency needs to be determined. Because of the requirement for large population samples to

provide reasonable estimates of such frequencies it is common practice to use online databases to make such estimates. The most widely used example is the Y-STR Haplotype Reference Database (YHRD) established in 2000 as a web-based publicly-available reference database administered by Professor Lutz Roewer (Willuweit and Roewer, 2015) [22].

8.2.2 The YHRD (<https://yhrd.org/>) is currently recommended as the default database to be used to estimate the weight of evidence for cases involving Y-STRs.

8.3 Choice of Reference Population

8.3.1 By default, the YHRD returns the number of matches in its total dataset (a worldwide population sample) for the chosen multiplex. However, it is also possible to select a 'metapopulation' dataset (for example, western European metapopulation) or a national population (for example, the UK). Note, however, that the UK population here is not a subset of the western European metapopulation, because the UK population dataset also includes Black African and South Asian individuals resident and sampled in the UK, while the metapopulation refers to the bio-geographical origin (ancestry) of the population. There is also over-representation of Black, Asian, and minority ethnic groups within the UK population dataset as a result of attempts to provide similarly sized datasets for each ethnic group.

8.3.2 Guidance is required as to which population group it is appropriate to use. The current Forensic Science Regulator's guidelines for autosomal STRs recommend use of the population database corresponding to the group of the person of interest (POI) suspect ('suspect anchored') (Forensic Science Regulator FSR-G-213) [18]. Where there is no other information in the case to indicate a relevant population database, the database most applicable to the POI in the case may be used for Y-STR haplotype estimation where possible. If the population database is a point in issue in the case, the haplotype frequency estimation should be explored in any other relevant population groups.

- 8.3.3 Due to the general homogeneity of Y-chromosomal lineages within western Europe (Willuweit et. al. 2005) [23] it is appropriate to use the western Europe metapopulation to increase the database size for comparisons with White British individuals from the UK.
- a. For cases where the POI is White British or is unknown, the western European metapopulation should be selected.
 - b. For all other cases, where the POI is other than White British, use of alternative metapopulation groups should be considered. However, most or all other YHRD metapopulation datasets are much smaller than that for western Europe, and in addition it may not be clear which metapopulation may be applicable. If no appropriate population group is available then it may be appropriate to use the western European metapopulation, provided the result is clearly reported with disclosure of the limitations inherent in this approach.

8.4 Choice of Method for Probability Estimation

- 8.4.1 The primary result returned by the YHRD is the observed number of haplotypes (profiles) matching the queried haplotype. However, this count needs to be expressed as an estimated probability of observing the haplotype in the relevant population and there are a number of possible methods to do this.
- 8.4.2 The YHRD website currently offers three alternative estimates based on three different published methods.
- a. The $n+1/N+1$ pseudocounting or augmented count method (where N is the number of individual haplotypes in the relevant population dataset, and n is the number of those haplotypes matching that of the POI). This is equivalent to the frequency obtained when adding the haplotype in question to the dataset (Willuweit and Roewer, 2015) [22].
 - b. The 'kappa' correction (Brenner, 2010) [24] based on the observed number of singleton profiles in the database (and therefore applicable only to haplotypes not previously observed).
 - c. The 'discrete Laplace' method (Andersen *et al.*, 2013) [25].

- 8.4.3 A fourth possible alternative uses a modified version of the pseudocounting method, using $n+2/N+2$ proposed by Balding (2005) [26]. The Balding method is currently used by all UK forensic units and, in the absence of an international consensus on the matter, is currently considered the most appropriate method to use.
- 8.4.4 Further to these different methods to estimate the weight of evidence, alternative approaches that model haplotype distributions in living populations (Andersen and Balding, 2017) [27] offer a radically different solution. As these models develop, there may be a case for their adoption in casework as acceptance by the international forensic community grows.
- 8.4.5 It is recommended to use the $n+2/N+2$ approach until alternative methods emerge that have been demonstrated (through validation) to be a robust suitable replacement.

8.5 Choice of multiplex for evaluation purposes

- 8.5.1 Although all UK forensic units currently use a 23-locus Y-STR system (PPY23), it is possible to modify a YHRD search to truncate the existing query profile to look for the frequency of only 17 loci (Yfiler) or the 9 ‘minimal haplotype’ (MH) loci (Kayser et al., 1997) [3]. Because there are significantly more MH and Yfiler profiles in the YHRD than there are 23 loci (Y23) profiles, there will be cases where the estimated probability of observing the 17 Yfiler loci or the 9 MH loci of a given Y23 profile is smaller than that of observing all of the 23 loci. This will always be true for profiles that have not been seen previously in the YHRD dataset, which are frequently encountered.
- 8.5.2 Choosing to search databases with different groups of loci is referred to specifically in the Scientific Working Group on DNA Analysis Methods guidelines (SWAGDAM, 2014) [13], which support this approach, stating: “Population databases may contain profiles typed with different multiplexes, containing different numbers and/or sets of loci, such that only a subset of the database may have been typed at all of the loci present in the evidence profile. This can lead to a seemingly paradoxical situation: a more discriminating set of loci gives a less discriminating frequency estimate than a less discriminating set of loci, simply because there were fewer database

profiles typed with the more discriminating set. In such situations, it is acceptable to perform additional searches of the population database using reduced locus sets in an attempt to obtain the maximal discrimination potential for that combination of evidence and population database profiles.”

- 8.5.3 Given the inverse relationship between profile information content (number of loci) and number of records, there is no clear-cut position on which profile datasets are most appropriate. The following recommendation takes account of this by devolving this decision to a case-by-case assessment.
- 8.5.4 Profile probabilities should be calculated using both the Y23 (23-locus) and the Yfiler (17-locus) dataset within the YHRD.
- 8.5.5 The lowest frequency (i.e. the most discriminating) should be reported.

9. Reporting Outcomes of Statistical Evaluation

- 9.1.1 When a statistical evaluation has been performed and an estimated probability of observing the profile in the relevant population has been determined, the following information should be included in reports and statements.
 - a. That the Y- STR profile derived from the evidential sample matches that of the person of interest (POI).
 - b. That any male belonging to the same paternal lineage as the POI will also be likely to match the profile from the evidential sample.
 - c. That the YHRD was used to estimate the weight of evidence, citing the release number, the date the search was performed, and the (meta)population group selected.
 - d. The estimated probability of observing the profile in the population group selected. It is also be helpful to state the number of matching profiles found in the population searched, and the size of that population dataset.
 - e. An activity level interpretation, if at all possible.

9.2 Alternatives to Statistical Evaluation

9.2.1 In cases not reaching the requirements set out above for a statistical evaluation (generally mixed profiles), a comparison of a profile of a POI to the crime profile may, in some cases, still have been carried out. In doing so, the scientist may reach a conclusion that the POI cannot be excluded as a possible contributor to that mixture. In this situation, there are a number of possible approaches to reporting this outcome.

- a. If no statistical evaluation is possible, the forensic unit may report the profile as unsuitable for further evaluation and make no comment about the possibility of contribution, nor state the name of the POI.
- b. The forensic unit may only provide an expression of the possibility that the POI contributed to the mixture if it is presented in a manner that does not favour the prosecution; such an expression is likely to be uninformative. If an assessment of evidential weight is not possible, the scientist should make it clear that they can give no guidance to the court with regard to probative value. This is an analogous position to that recommended in the guidance document for autosomal STR mixture evaluation when no statistical evaluation can be conducted (Forensic Science Regulator FSR-G-222) [19].
- c. The forensic unit may provide a qualitative or subjective evaluation, if it is supported by scientific experimentation, such as non-contributor testing (whereby a large number of random profiles from individuals not associated with the investigation are compared to the evidential mixed profile to determine if they would have been considered as ‘possible’ contributors themselves). If such analysis is conducted it may be reasonable to conclude that the findings in the case are more likely if the POI (or a close paternal-line male relative of his) had contributed DNA to the mixed Y-STR profile obtained rather than if someone selected at random from the wider general population had contributed DNA to the mixed Y-STR profile. Andersen and Balding (2017) [27] demonstrate that the chance of a randomly selected man matching a POI is negligible. Andersen and Balding (2019) [29] show that a two-male mixture that includes the profile of a POI has almost exactly the same evidential value as a single contributor match to the POI.

9.3 Future Challenges

- 9.3.1 The statistical evaluation of Y-STR profiles differs significantly from that of autosomal profiles. In order to report and evaluate Y-STR results in a robust manner and to achieve the maximum evidential value from such results, a number of requirements can be identified.
- a. Provision of a larger reference population of UK-resident populations, preferably collected across different regions of the UK and with their geographical provenance recorded.
 - b. Further investigation and guidance from the international forensic community on appropriate methods to estimate profile probabilities and report weight of evidence for single-source profiles.
 - c. Further investigation and guidance from the international forensic community on appropriate methods to statistically evaluate mixed profiles where a clear single-source contributor cannot be deduced. This may include the development of probabilistic models applicable to Y-STR profiles.

10. Quality Assurance for Y-STR Profiling

10.1 Quality Assurance Checks

- 10.1.1 Quality assurance (QA) checks on Y-STR profiles should comply as much as possible with the requirements set out in the Forensic Science Regulator's FSR-C-108 DNA analysis [15]. Forensic units should take the same measures to minimise the risk of contamination in Y-STR profiling as when producing autosomal STR profiles.
- 10.1.2 Forensic units should take the same measures, where possible, to identify unknown Y-STR profiles in Y-STR profiling as when undertaking autosomal STR profiling. This includes the creation and maintenance of elimination databases. It is likely that only a local elimination database of Y-STR profiles can be maintained, including such profiles from male staff, visitors to laboratory areas (for example, engineers), and unsourced contaminants. It is less likely that Y-STR profiles will be obtained from police officers or people

involved in the manufacture of consumables. Given the risk of contamination, special consideration should be given to obtaining the Y-STR profiles of crime scene examiners and forensic medical examiners working in sexual assault referral centres, or similar.

10.2 Co-processing of Quality Assurance Controls

- 10.2.1 Where possible, extraction negative controls should be processed alongside Y-STR samples. These will provide assurance for the Y-STR process. Negative controls can also provide information on the rate of drop-in seen within the Y-STR profiling method.
- 10.2.2 In some instances, the DNA extract from controls processed with samples during autosomal STR profiling may be consumed to such an extent (for example, in the investigation of a suspected contamination event) that insufficient DNA extract remains for co-processing of the controls with the samples using Y-STRs. In such instances, the reason why QA controls from autosomal DNA profiling were not reworked with Y-STRs alongside the co-processed samples must be recorded.
- 10.2.3 Validation of Y-STR profiling may provide assurance that, where no autosomal STR profile is obtained from a negative control using the kit used for regular processing of crime stains, then the expectation is that no Y-STR profile will be obtained either. Where such a demonstration has been made during validation, or derived from processing a sufficient number of controls with both autosomal and Y-STR kits, consideration may be given to not co-processing extraction negative controls with Y-STR samples.
- 10.2.4 Where an unsourced contaminant sufficient for retention on a local elimination database is identified in a QA control sample in autosomal STR profiling, and the profile is not certain to have derived from a female source, a Y-STR profile should be produced from the control where possible. This should be done even if no samples co-processed with that control have been subject to Y-STR profiling, as samples from other batches may be processed in the same laboratory or with the same consumables.

11. Investigation of Potential Contamination in Y-STR Profiles

- 11.1.1 It is likely that not every sample generated from crime stains and associated reference samples will automatically undergo Y-STR profiling. A Y-STR profile may be generated from a crime stain sample and not found to match a nominal from whom a reference sample has been submitted. Considered to be of unknown origin, such a sample may originally have been processed for the purpose of generating an autosomal STR profile alongside samples for which no Y-STR profile was requested or produced. If the unknown Y-STR profile is the result of cross-contamination from another sample co-processed during autosomal STR profiling, it is possible that the contamination event will go undetected if the sample from which the contaminant derived has not also been Y-STR profiled.
- 11.1.2 This restricts the investigation of potential cross-contamination events leading to the generation of a contaminant Y-STR profile. Forensic units should not produce Y-STR profiles for any crime stain or reference samples other than those for which a request is submitted. Generating Y-STR profiles from samples in other cases, where no such profiling was requested, will require the disclosure of any Y-STR profiles and may lead to further Y-STR profiling of samples from nominals and other crime stains that were also not requested. Aside from the legal complexity of Y-STR profiling that was not requested or required, such investigative Y-STR profiling would consume DNA recovered from a sample that might be required for re-work with the regular testing method.
- 11.1.3 In some instances, for Y-STR profiles for which the origin is unknown, comparison between the autosomal STR profile obtained for that sample with other autosomal STR profiles obtained from co-processed samples can be considered. This may be carried out with the understanding that, due to differences in the amount of template DNA input, or to the presence of large amounts of female DNA in the autosomal DNA reaction, no evidence of sample-to-sample contamination may be found by comparing autosomal STR

profiles even where sample-to-sample contamination has occurred and resulted in a contaminant Y-STR profile.

- 11.1.4 Forensic units should investigate the origin of any unknown Y-STR profile obtained as much as possible. The limitations of the investigation should be noted along with any findings. Given the above limitations, contamination can almost never be ruled out as the source of an unknown Y-STR profile; this does not justify failure to investigate the origin of unknown Y-STR profiles.

11.2 Sharing Unsourced Contaminant Y-STR Profiles

- 11.2.1 Currently (February 2021), the National DNA Database provides a mechanism for retaining and sharing information on unsourced contaminant profiles generated with approved autosomal STR kits. No such mechanism is available for the sharing of Y-STR unsourced contaminant profiles. Forensic units should circulate a list of points of contact for the purposes of sharing such profiles.

11.3 Y-STR Elimination Databases

- 11.3.1 As for autosomal DNA profiling, an elimination database containing the Y-STR profiles of members of staff should be created and maintained for comparison with unknown Y-STR profiles prior to their being reported or loaded to a DNA database.
- 11.3.2 Use of a Y-STR staff elimination database should comply as much as possible with the requirements set out in the Forensic Science Regulator's FSR-P-302 Protocol: DNA contamination detection – The management and use of staff elimination databases [21].

11.4 Creation of a Y-STR Elimination Database

- 11.4.1 The nature of Y-STR profiles, and the likely approach taken to routinely generating such profiles in the laboratory, creates some instances where the approaches taken to using a Y-STR elimination database (YED) must differ from using an elimination database containing autosomal STRs. These issues include shared ancestry, fertility, sex-reversal syndromes and gender.

- 11.4.2 Y-STR profiles from male employees may reveal facts about them that should not be made known to colleagues and about which, in some cases, they themselves may be unaware.
- a. Different Y-STR profiles obtained from males believing themselves to be from the same paternal line (for example, brothers) may reveal a difference in their paternity.
 - b. The absence of several loci from a Y-STR profile may be indicative of a deletion if the loci are adjacent to each other on the Y-chromosome. Genes linked to fertility may also have been deleted, perhaps preventing the individual from fathering children.
- 11.4.3 In rare sex-reversal syndromes chromosomal sex is discordant with phenotypic sex. For example, an individual with androgen insensitivity syndrome carries a Y-chromosome, but is phenotypically female. In autosomal DNA profiling such an individual will show an Amelogenin Y (AMELY) result, and would provide a full Y-STR profile if tested.
- 11.4.4 Obtaining Y-STR elimination profiles from transgender staff members must be considered carefully. Those who have transitioned from male to female will still be a potential source of Y-STR contamination. Those who have transitioned from female to male will not. Employers should be mindful of any legal protections for such individuals under the Equality Act 2010 [30].
- 11.4.5 When new recruits are first offered a job within a forensic unit, employers should inform them of the need to produce autosomal profiles from employees, and to retain those profiles for comparison in order to detect cross-contamination. Employees should also be told that, where AMELY is observed in the autosomal profile, or in those presenting themselves as male, Y-STR profiling will be progressed and that both tests will reveal the chromosomal status of the donor.
- 11.4.6 In anticipation of these scenarios, forensic units must carefully consider how to use and manage Y-STR profile information obtained from staff and visitors for the purposes of elimination, and especially how best to restrict access to any database holding Y-STR profiles so as to avoid revealing personal information to other employees. It is therefore likely that the YED must be

more closely protected than the regular autosomal STR elimination database. Creation of a separate local Y-STR elimination database by each forensic unit will make it easier to enact protective measures. Communicating matches without naming the individual on the YED to whom the profile matched will ensure that anonymity is maintained.

- 11.4.7 Forensic units may use staff volunteers as donors with consent for validation, research, and blind testing of the processes and systems in place. The use of staff for repeated quality assurance batch controls should be avoided. Care should be taken when requesting donor samples of any sort for producing Y-STR profiles where a potential donor has a Y-STR profile that may involve a genetic privacy issue. Repeat use of that donor will draw attention to the affected profile. Where the issue is with fertility, any impact on the quality of sperm donated for use as control or experimental material for non-DNA processes should also be considered.

11.5 Searching Y-STR Elimination Databases

- 11.5.1 Careful consideration should be given to the minimum number of alleles used to search the YED. Searching supposed contaminant profiles that are very partial might result in adventitious matches and unnecessary investigations. Searching only nearly complete profiles may prevent detection of genuine contamination. Presumptive alleles not labelled or included in the search should also be compared against any matches obtained.
- 11.5.2 Unlike complete autosomal STR profiles commonly generated from crime stains, many individuals in a population may share the same Y-STR profile. Also, the size of a local YED may not be very different, perhaps only an order of magnitude, from the size of any database, or subset thereof, used to determine the statistical significance of a Y-STR profile. Given the similarity between Y-STR profiles, partial profiles may produce several matches when searched on a YED. As with autosomal DNA elimination databases, the larger a YED is, the more likely a matching profile will be found. However, a match between an unknown Y-STR profile and a record on a local YED may not be as significant as a match between autosomal STR profiles.

11.5.3 As patrilineal male relatives are expected to share the same Y-STR profile, search of unknown profiles against a local YED may be more likely to produce matches where a crime has been committed in the same community in which those sampled, or their relatives, have lived. There is also some evidence for a correlation between Y-STR haplotypes and surnames for which a single origin is likely. Forensic units should understand that a local YED may operate as a 'de facto' mini-database, highlighting possible familial connections.

12. Requirements for Forensic Unit Guidelines for Interpreting the Presence and Designation of Peaks

12.1.1 As with autosomal short tandem repeat (STR) typing, forensic units conducting Y-STR analyses should characterise the performance of their systems and the profiles produced as part of their validation process. These data should be used to develop appropriate guidelines for the interpretation of such profiles.

12.1.2 Forensic units should consider the following thresholds and parameters.

12.1.3 Analytical threshold: The peak height below which alleles cannot safely be designated. This threshold may be dependent on the multiplex kits used, the amplification and detection systems in place, and other factors that may impact on the baseline noise and the signal strength. An appropriate threshold should be determined by each forensic unit.

12.1.4 Stutter: Polymerase chain reaction amplification of Y-STRs is likely to generate stutter artefacts similar to those seen with autosomal STRs. These will most commonly be one repeat unit shorter than the primary allele, but additional stutter products (for example, two repeats smaller or one repeat larger) may also be observed. Interpretation guidelines to help to identify stutter may be developed at the multiplex level, the locus level or at the level of individual alleles.

12.1.5 Stochastic threshold: For an autosomal locus, this is defined as the peak height below which a single allele peak at a locus cannot safely be designated as a homozygote at an autosomal locus (because of possible

allele drop-out of a second allele). Most Y-STRs commonly used for forensic analyses are single-copy loci present only once in the male genome. A stochastic threshold is not applicable to single copy loci. However, for duplicated Y-STR loci (such as locus DYS385a,b) it is appropriate that a stochastic threshold is determined.

12.1.6 Peak height ratio / heterozygote balance: Thresholds based on the ratio of two allele peaks presumed to be heterozygous alleles from the same individual provide useful guidance in the interpretation of autosomal STRs. Peak height ratios are not applicable to single copy loci. However, for duplicated Y-STR loci (such as DYS385a,b) it is appropriate that the characteristics of peak height ratio of the two alleles are determined and appropriate thresholds developed.

12.1.7 Non-specific artefacts: Y-STR multiplexes designed for forensic analysis are highly specific for the intended Y-chromosome loci. However, it has been demonstrated that in the presence of very large excesses of human genomic DNA (usually female) low levels of non-specific amplification products may be observed (Moore et al., 2016) [4]. These appear in characteristic positions likely to be dependent on the exact multiplex kit used. Forensic units should have appropriate guidelines for identifying and reporting such artefacts.

12.1.8 Forensic units should also have guidance to identify other non-allelic artefact peaks that may occur in Y-STR profiles.

12.1.9 Deletions and duplications: The Y chromosome is prone to copy number variation including deletion and duplication of stretches of the genomic sequence, which may impact on STR loci (see 1.1.3 and 1.2.5). Forensic units should have appropriate guidelines for identifying and reporting such occurrences.

13. Acknowledgements

13.1.1 This guidance was produced by the Forensic Science Regulator's DNA Analysis Specialist Group. The FSR would like to thank Cellmark Forensic Services, Eurofins Forensic Services, Forensic Science Ireland, Forensic Science Northern Ireland, Key Forensic Services Ltd, King's College London,

Scottish Police Authority Forensic Services, University of Leicester and the Forensic Science Regulation Unit (FSRU).

14. Review

- 14.1.1 This published guidance will form part of the review cycle as determined by the Forensic Science Regulator.
- 14.1.2 The Forensic Science Regulator welcomes comments. Please send them to the address as set out at: www.gov.uk/government/organisations/forensic-science-regulator, or email: FSREnquiries@homeoffice.gov.uk

15. References

- [1] Jobling, M. A. (2008) 'Copy number variation on the human Y chromosome', *Cytogenet. Genome Res.*, vol. 123 (1–4), pp 253–62.
- [2] Jobling, M. A., Lo, I. C., Turner, D. J., Bowden, G. R., Lee, A. C., Xue, Y., Carvalho-Silva, D., Hurles, M. E., Adams, S. M., Chang, Y. M., Kraaijenbrink, T., Henke, J., Guanti, G., McKeown, B., van Oorschot, R. A., Mitchell, R. J., de Knijff, P. and Tyler-Smith, C. (2006) 'Structural variation on the short arm of the human Y chromosome: recurrent multigene deletions encompassing Amelogenin', *Human Molecular Genetics*, vol. 16 (3), pp 307–316, Dec 22.
- [3] Ballantyne, K. N., Goedbloed, M., Fang, R., Schaap, O., Lao, O., Wollstein, A., Choi, Y., van Duijn, K., Vermeulen, M., Brauer, S., Decorte, R., Poetsch, M., von Wurmb-Schwark, N., de Knijff, P., Labuda, D., Vézina, H., Knoblauch, H., Lessig, R. and Roewer, L. (2010) 'Mutability of Y-chromosomal microsatellites: rates, characteristics, molecular bases, and forensic implications', *American Journal of Human Genetics*, vol. 87 (3), pp 341–353.
- [4] Hanson, E. K. and Ballantyne, J. (2006) 'Comprehensive annotated STR physical map of the human Y chromosome: Forensic implications', *Legal Medicine*, vol. 8, pp 110–120.
- [5] Moore, D., Clayton, T. and Thomson, J. (2016) 'Description of artefacts in the PowerPlex Y23((R)) system associated with excessive quantities of background female DNA', *Forensic Science International: Genetics* 2016, vol. 24, pp 44–50.

- [6] Gill, P., Jeffreys, A. J. and Werrett, D. J. (1985) 'Forensic application of DNA 'fingerprints', *Nature*, vol. 318, pp 577–579.
- [7] King, T. E. and Jobling, M. A. (2009) 'What's in a name? Y chromosomes, surnames and the genetic genealogy revolution', *Trends in Genetics*, vol. 25, pp 351–360.
- [8] British Standard, BS EN ISO/IEC 17025, General requirements for the competence of testing and calibration laboratories.
- [9] International Laboratory Accreditation Cooperation, ILAC G19:08/2014 Modules in a Forensic Science Process, [Online]. Available at: <http://ilac.org/news/ilac-g19082014-published/>. [Accessed 10 03 2021].
- [10] Gill, P., Brenner, C., Brinkmann, B., Budowle, B., Carracedo, A., Jobling, M. A., de Knijff, P., Kayser, M., Krawczak, M., Mayr, W. R., Morling, N., Olaisen, B., Pascali, V., Prinz, M., Roewer, L., Schneider, P. M., Sajantila, A. and Tyler-Smith, C. (2001) 'DNA Commission of the International Society of Forensic Genetics: Recommendations on forensic analysis using Y-chromosome STRs', *Forensic Science International*, vol. 124, pp 5–10.
- [11] Gusmão, L., Butler, J. M., Carracedo, A., Gill, P., Kayser, M., Mayr, W. R., Morling, N., Prinz, M., Roewer, L., Tyler-Smith, C. and Schneider, P. M. (2006) 'DNA Commission of the International Society of Forensic Genetics (ISFG): An update of the recommendations on the use of Y-STRs in forensic analysis', *Forensic Science International*, vol. 157, pp 187–197.
- [12] Roewer, L., Andersen, M. M., Ballantyne, J., Butler, J. M., Caliebe, A., Corach, D., D'Amato, M. E., Gusmão, L., Hou, Y., de Knijff, P., Parson, W., Prinz, M., Schneider, P. M., Taylor, D., Vennemann, M., Willuweit, S. and Corach, D. (2020) 'DNA commission of the International Society of Forensic Genetics (ISFG): Recommendations on the interpretation of Y-STR results in forensic analysis', *Forensic Science International: Genetics*, vol. 48, p 10230.
- [13] SWAGDAM (2014) 'Interpretation Guidelines for Y-Chromosome STR Typing 2014', Scientific Working Group on DNA Analysis Methods [Online]. Available at: www.swgdam.org/publications. [Accessed 10 03 2021].

- [14] Forensic Science Regulator, Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System,” FSR-C-100, [Online]. Available at: www.gov.uk/government/collections/forensic-science-providers-codes-of-practice-and-conduct. [Accessed 10 03 2021].
- [15] Forensic Science Regulator, DNA Analysis,” FSR-C-108, [Online]. Available at: www.gov.uk/government/collections/forensic-science-providers-codes-of-practice-and-conduct#appendices [Accessed 10 03 2021].
- [16] Forensic Science Regulator, Validation,” FSR-G-201, [Online]. Available at: www.gov.uk/government/publications/forensic-science-providers-validation. [Accessed 10 03 2021].
- [17] Forensic Science Regulator, The control and avoidance of contamination in laboratory activities, involving DNA evidence recovery and analysis,” FSR-G-208, [Online]. Available at: www.gov.uk/government/publications/laboratory-dna-anti-contamination-guidance. [Accessed 10 03 2021].
- [18] Forensic Science Regulator, Allele frequency databases and reporting guidance for the DNA (short tandem repeat) profiling,” FSR-G-213, [Online]. Available at: www.gov.uk/government/publications/allele-frequency-databases-and-reporting-guidance-for-the-dna-17-profiling. [Accessed 10 03 2021].
- [19] Forensic Science Regulator, DNA mixture interpretation,” FSR-G-222, [Online]. Available at: www.gov.uk/government/publications/dna-mixture-interpretation-fsr-g-222. [Accessed 10 03 2021].
- [20] Forensic Science Regulator, Software validation for DNA mixture interpretation FSR-G-223, [Online]. Available at: www.gov.uk/government/publications/software-validation-for-dna-mixture-interpretation-fsr-g-223. [Accessed 10 03 2021].
- [21] Forensic Science Regulator, DNA contamination detection: The management and use of staff elimination DNA databases [Online]. Available at: www.gov.uk/government/publications/dna-contamination-detection. [Accessed 10 03 2021].

- [22] Willuweit, S. and Roewer, L. (2015) 'The new Y Chromosome Haplotype Reference Database', *Forensic Science International: Genetics*, vol. 15, pp 43–48.
- [23] Roewer, L., Croucher, P. J. P., Willuweit, S., Lu, T. T., Kayser, M., Lessig, R., de Knijff, P., Jobling, M. A., Tyler-Smith, C. and Krawczak, M. (2005) 'Signature of recent historical events in the European Y-chromosomal STR haplotype distribution', *Hum. Genet.*, vol. 116, pp 279–291.
- [24] Brenner, C. H. (2010) 'Fundamental problem of forensic mathematics — The evidential value of a rare haplotype', *Forensic Science International: Genetics*, vol. 4, pp 281–291.
- [25] Andersen, M., Svante, P. and Morling, N. (2013) 'The discrete Laplace exponential family and estimation of Y-STR haplotype frequencies', *Journal of Theoretical Biology*, vol. 329, pp 39–51.
- [26] Balding, D. (2005) *Weight-of-evidence for Forensic DNA*. John Wiley & Sons Ltd, ISBN: 9780470867648.
- [27] Andersen, M. M. and Balding, D.J. (2017) 'How convincing is a matching Y-chromosome profile?', *PLoS Genet.* 2017, vol. 13, p. e1007028.
- [28] Kayser, M., Caglia, A., Corach, D., Fretwell, N. and Gehrig, C. (1997) 'Evaluation of Y-chromosomal STRs: a multicenter study', *International Journal Legal Medicine*, vol. 110, pp 125–133.
- [29] Andersen, M. M. and Balding, D. J. (2019) 'Y-profile evidence: Close paternal relatives and mixtures', *Forensic Science International: Genetics*, vol. 38, pp 48–53.
- [30] The Equality Act 2010 [Online]. Available at: www.legislation.gov.uk/ukpga/2010/15/contents. [Accessed 10 03 2021].

16. Abbreviations and Acronyms

Abbreviation	Meaning
AMELY	Amelogenin Y
BS EN	British Standard European Norm

DNA	Deoxyribonucleic acid
DNA17	17 STR loci system including the gender marker Amelogenin
FSR	Forensic Science Regulator
IEC	International Electrotechnical Commission
ILAC	International Laboratory Accreditation Cooperation
ISFG	International Society for Forensic Genetics
ISO	International Organization for Standardization
MH	Minimal Haplotype (9 Y-STR loci)
POI	Person of interest
PPY23	PowerPlex® Y23 System (23 Y-STR loci)
QA	Quality assurance
STR	Short tandem repeat
SWGDM	Scientific Working Group on DNA Analysis Methods
YED	Y-STR elimination database
YHRD	Y-STR Haplotype Reference Database

17. Glossary

Allele:

A genetic variant at a particular location within an individual's DNA. DNA profiling tests examine a range of alleles that are known to vary widely between individuals. Alleles are represented by peaks in a DNA profile.

Allelic Drop-In:

Additional random alleles present in a profile originating from random fragmented sources and regarded as independent events.

Allelic Drop-Out:

Alleles missing from a DNA profile, so that it is partially represented.

Amelogenin:

A single-copy gene located on the X chromosome (AMELX) and on the male-specific region of the Y chromosome (AMELY). A 6-bp deletion in AMELX enables each to be visualised after electrophoretic separation, informing the determination of sex.

Artefact:

Artefacts are ‘nuisance’ peaks in a profile; often associated with the amplification and detection processes, such as spikes, dye blobs, spectral pull-up. They do not represent genuine alleles; they are screened out by the scientist or the software.

Autosomal DNA:

DNA from the 22 pairs of non-sex chromosomes.

Complex DNA Profile:

A crime-sample profile exhibiting allele drop-out, drop-in or which originated from two or more individuals.

Duplication Events:

Genetic duplication of stretches of the genomic sequence. Most duplication events result in two alleles at a single-copy locus that differ by a single repeat unit. These may result in single source profiles having more than one component at one or more loci.

Elimination Database:

Collection of DNA profiles held in a searchable format from staff whose access/role/activities are deemed to be a potential DNA contamination risk. This may include not just the staff working within a specific facility, but also profiles from visitors to the facility, staff of manufacturers supplying consumables for DNA processing, and unsourced contamination profiles. The profiles are used to identify instances of inadvertent contamination.

Forensic Unit:

A forensic unit is a legal entity or a defined part of a legal entity that performs any part of the forensic science process. [SOURCE: ILAC-G19:08/2014 Modules in a Forensic Science Process.] It is interchangeable with ‘provider’, and is also used for small teams or sole practitioners for accreditation purposes.

Haplotype:

A group of linked alleles that are inherited together from one parent. Alleles (for example, short tandem repeats) on the Y-chromosome represent a single haplotype inherited from father to son.

Locus (plural Loci):

The specific genetic location of an allele on a chromosome. Short tandem repeats are examples of loci that are used in forensic science because they are polymorphic and are therefore highly discriminatory when several are analysed in combination to generate a DNA profile.

Mixture:

A DNA profile that contains more designated alleles than would be expected if there were only one contributor to the sample.

Negative Control (Blank):

Contains the analyte at a concentration below a specified limit. The intention is that no DNA is present and no profile or alleles above the drop-in rate are expected.

Partial Profile:

A DNA profile that is missing one or more alleles from the donor. This can be because the DNA has been degraded, or because DNA is present at such low levels that accurate marker information cannot be obtained.

Patrilineal:

Relating to descent through the male line. Grandfather, father and son share a patrilineal relationship.

Quantitative Evaluation:

A statistical evaluation of evidence based on empirical and mathematical data. The calculation of a numerical likelihood ratio in relation to a pair of propositions or hypotheses. This will almost always be achieved by means of validated software and will incorporate reference to one or more databases of allele proportions.

Short Tandem Repeat (STR):

A microsatellite consisting of one to six or more nucleotides that is repeated adjacent to each other along the DNA strand.

Stutter:

An artefact of the amplification process that leads to smaller peaks close to the main allelic peak. The most common stutter peak is one repeat unit smaller than the allelic peak (for a tetranucleotide short tandem repeat, - 4). Stutters with other numbers of repeats are also possible, but less common. Over-stutters are one repeat unit larger than the allelic peak (+4).

Validation:

The process of providing objective evidence that a method, process or device is fit for the specific purpose intended.

Y-STR Profile:

A short tandem repeat (STR) profile derived from the combination of several STRs on the Y-chromosome, also known as a Y-STR haplotype.

18. Further Reading

Forensic Science Regulator, Validation – Use of casework material validation, FSR-P-300 [Online]. Available at:

www.gov.uk/government/publications/protocol-using-casework-material-for-validation-purposes [Accessed 10 03 2021].

Peach, C. (2006) 'South Asian migration and settlement in Great Britain, 1951–2001', *Contemporary South Asia*, 15:2, pp 133-146, DOI: 10.1080/09584930600955234.

The Royal Society and the Royal Society of Edinburgh (2017) *Forensic DNA Analysis – A primer for courts*. [Online]. Available at:

www.royalsociety.org/~media/about-us/programmes/science-and-law/royal-society-forensic-dna-analysis-primer-for-courts.pdf [Accessed 10 03 2021].

UKAS® (2016) *UKAS Policy on Participation in Proficiency Testing, TPS 47*, edition 3, issued November 2016. United Kingdom Accreditation Service. [Online]. Available at:

www.ukas.com/download/publications/Technical%20Policy%20Statements/TPS-47-Participation-in-PT-Edition-4-February-2020.pdf [Accessed 10 03 2021].

Published by:

The Forensic Science Regulator

5 St Philip's Place

Colmore Row

Birmingham

B3 2PW

www.gov.uk/government/organisations/forensic-science-regulator