Forensic Science Regulator

Guidance: Autosomal DNA relationship testing

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1. Introduction

1.1 Background

1.1.1 The question of how individuals are related to each other arises in various contexts in criminal cases, as demonstrated by the following examples:

a. It may be necessary to establish the identity of a body using reference DNA samples from blood relatives of the deceased.

b. Following a rape allegation that has led to the conception of a child, the question of its paternity may be material evidence in establishing that an act of sexual intercourse took place between the parties. (This would not, however, address the issue of consent).

c. If an alleged rape occurs between parties who are themselves biologically related, then the child will have been 'incestuously' conceived. In such instances there may, therefore, be an important additional evidential element to establish: that the biological parents are themselves closely related in some way (as well as being the biological parents of the child).

d. A case may be brought against a couple who are in a consensual sexual relationship but who are believed to be biologically related in such a way that makes that relationship a criminal offence (incest).

e. In human trafficking or some kidnapping cases, individuals may assert that a child in their custody is biologically theirs.

1.1.2 In whatever context it arises, it is important that forensic practitioners providing DNA relationship testing evidence to the criminal courts adhere to the same standards and principles as the practitioners presenting more 'traditional' DNA evidence regarding identity.

1.1.3 However, it is important to recognise that DNA testing of a biological relationship is a different practice, with different emphases, from identity testing and so merits separate standards and guidance. This document is intended to fill that gap.

2. Other standards for DNA profile interpretation

2.1.1 National and international standards [1], [2] for testing and calibration in laboratories provide information on analytical methods used in forensic science and this document references those resources when applicable.

2.1.2 Any guidelines produced in this document are, wherever possible, based on published work. These publications are referenced where relevant.

2.1.3 The content of this document has also been the subject of widespread consultation with practitioner representatives of all of the major providers of forensic science services in the UK and Ireland via the DNA Analysis Specialist Group.

3. Scope

3.1.1 The purpose of this document is to provide guidance for those forensic practitioners delivering relationship testing services using autosomal DNA short tandem repeat (STR) profiling to the criminal justice system (CJS) but also, including a consideration of lineage markers (Y-STRs, X-STRs and mitochondrial DNA).

3.1.2 These guidelines apply to courts operating at any level within the CJS of England and Wales.

3.2 Exclusions

3.2.1 Although the ethos of the guidelines can be applied to other classes of autosomal markers such as insertion and deletion polymorphisms (indels) and single nucleotide polymorphisms (SNPs), these markers will not be addressed within this guidance.

3.2.2 For the avoidance of doubt, the Forensic Science Regulator's remit in requiring compliance to these standards and guidelines does not extend to:

a. Disaster victim identification (DVI), other than where there is a requirement to identify the remains of a suspected perpetrator.

b. Inquests held before HM Coroners in England and Wales, in which evidence is adduced regarding the identity of a deceased person based on an analysis of the DNA of his/her putative relatives.
c. Applications such as *in utero* paternity testing, non-invasive paternity testing (NIPT), and other forms of genetic testing for medical or research purposes.
d. Family law matters, such as child custody and maintenance arrangements.
e. Civil law matters, such as inheritance disputes.
f. Immigration matters, such as family repatriation.
g. Privately commissioned tests.

3.2.3 For those areas of relationship testing where these standards and guidelines do not officially apply, nevertheless, the principles of good practice espoused herein may still be applied to those spheres of work.

4. Implementation

4.1.1 This appendix is available for incorporation into a forensic unit's quality management system from the date of publication.

5. Terms and definitions

5.1.1 The word 'shall' has been used in this document where there is a corresponding requirement in ISO/IEC 17025:2017 [1], the Forensic Science Regulator's Code (the Code) [3] or there is a corresponding legal requirement.

5.1.2 The word 'should' has been used to indicate generally accepted practice, where the reason for not complying or deviating should be recorded.

5.1.3 The word 'may' or 'might' or 'can' or similar words indicates a voluntary component in the guidance.

5.1.4 The terms and definitions set out in the Code [3] and other related DNA guidance documents, apply to this document.

5.1.5 For the avoidance of doubt, the phrases ‘kinship testing’ and ‘pedigree testing’ are taken to be synonymous with ‘relationship testing’. For consistency, this document will use the phrase ‘relationship testing’ as a generic term to cover the testing of all potential biological relationships between individuals.

5.1.6 In this document, if the term ‘paternity testing’ is used, it is restricted to its specific meaning (for example, investigating an alleged father/child relationship).

6. Quality control

6.1 Laboratory processing

6.1.1 The laboratory processing the samples and preparing the DNA profiles shall be accredited to ISO17025 [1]. The requirements set out in the Code [3], section 92 shall be complied with.

6.2 Chain of custody

6.2.1 If it is anticipated that the evidence produced is to be used within the criminal justice system then the samples required for testing shall follow a chain of custody from sampling to presenting the evidence in court (code section 29.2.8). This includes unique labelling of exhibits, appropriate documentation and tracking\recording of the movement of items.

6.2.2 For any samples taken outside the police chain of custody, the handling, transportation and receipt of such samples should also be documented.

6.3 Formalities for taking DNA samples for relationship testing

6.3.1 The identity of an individual who has provided a DNA sample, should be established by the person taking that sample through:

a. a photograph of the person (or a copy of photographic identification they have supplied as proof of identity); or
b. a determination by an official authority on identity, e.g. during a prison visit.

6.3.2 Informed consent for the taking, use and retention of a voluntary sample (and the information derived from it) shall be obtained before the sample is taken [4]. The volunteer, or an appropriate adult, shall specifically consent to the use of the sample for relationship testing purposes in order to proceed (the Code, section 95.11). This consent shall be documented accordingly. A sampling kit containing a consent form[1] is specifically provided for this purpose (the Code, section 95.11).

6.3.3 If the person is deceased, then a DNA sample shall only be taken under the lawful authority of an HM Coroner for England and Wales or pursuant to police powers of seizure in certain circumstances, as stated within section 39 of the Human Tissue Act [5].

6.3.4 A reference sample shall only be taken coercively from an arrested person using the powers granted to the police by virtue of the Police and Criminal Evidence Act 1984 (PACE) [6]. A sampling kit containing a controlled form[2] is specifically provided for this purpose.

6.3.5 Samples taken coercively pursuant to PACE [6] shall only be used for a limited set of purposes (which are set out in the Act) and which include the prosecution of crime and identification of the dead (i.e. for use in criminal and coronial cases). Such samples (which include the information derived from those samples) cannot therefore, by law, be used for a collateral purpose, such as in family proceedings or in a civil case, see FINDS Strategy Board policy [7].

6.3.6 Resampling and retesting would therefore be required if civil or family matters follow on from a criminal case.

[1] March 2021: England & Wales PACE DNA Sampling Kit – Elimination Product Code: G0010225
[2] March 2021: England & Wales PACE DNA Sampling Kit – Suspect Product Code: G00101-25

6.4 Guidance as to sample types suitable for relationship analysis

6.4.1 The type of sample available for relationship testing will depend on the circumstances of the individual case.

6.4.2 For living individuals, the preferred sample type will be a pair of buccal (mouth) swabs. However, if the individual is a young child or an adult undergoing medical treatment, then saliva, blood or freshly pulled hairs (with roots) could provide an alternative source of DNA. With neonates, blood from a heel stab preserved on a Guthrie card may be used, provided that the sample is traceable to the child via patient records.

6.4.3 For deceased individuals, any available post-mortem sample can be utilised. If the cadaver is in an advanced state of decay, testing of skeletal structures such as bones and teeth may be required.

6.4.4 For a child in utero, following either the planned termination of the pregnancy or, following a miscarriage, the police may use their powers of seizure to take possession of the products of conception so that DNA samples from the foetus may be secured: see Faculty of Forensic and Legal Medicine [8].

6.4.5 To associate human remains with missing individuals, or to identify a deceased, a ‘surrogate’ reference sample may be required. Such samples could be obtained from:

a. A traceable medical archive sample (such as wax embedded tissue, serum samples, cervical smears or other preparations on microscope slides); or
b. Personal effects attributable to the missing person or the deceased (preferably an item likely to have been used exclusively by that person) such as a toothbrush, dentures, razor or hairbrush; or
c. Clothing worn by the missing person.

6.4.6 When identifying a perpetrator’s remains in a disaster victim identification (DVI) incident, further guidance is provided in the INTERPOL guides [9], [10], the College of Policing authorised professional practice [11], and the International

Society for Forensic Genetics (ISFG) – see the recommendations in Prinz et al., 2007 [12].

6.5 Packaging

6.5.1 The packaging of collected biological material for forensic examination should be fit for purpose. Specifically, the packaging should:

a. Preserve the integrity of the material;
b. Minimise the risk of loss and/or degradation of the material;
c. Prevent adulteration or contamination of the material; and
d. Prevent the escape of any biohazardous material from the sample.

6.5.2 As a minimum this should include:

a. Separate packaging of items where the packaging of items together could compromise them;
b. Appropriate packaging for the size, condition and forensic analysis requirements of the material recovered;
c. Secure and tamper-evident seals; and
d. Where liquids are to be stored frozen, suitable containers that will neither rupture on freezing nor leak upon thawing.

6.6 Storage and preservation

6.6.1 Samples should be stored in such a way as to ensure that they are secure and suitably preserved for forensic examination.

6.6.2 Liquid samples and wet stains should generally be stored frozen (preferably at -20 degrees centigrade) unless they have otherwise been treated to render them stable for storage at an ambient temperature (for example, by drying or by use of a preservative or fixative).

6.6.3 It should be noted that the use of preservatives such as formalin and formaldehyde has a very deleterious effect on DNA and can negatively impact downstream DNA processes.

6.6.4 Unfrozen samples should be stored dry, at an ambient temperature and out of direct sunlight.

7. Case assessment and interpretation

7.1 Case assessment

7.1.1 The task-relevant (pedigree) information supplied by the authority submitting (or proposing to submit) the exhibits for relationship testing should be reviewed by the laboratory and propositions should be formulated accordingly.

7.1.2 In criminal cases, and when formulating the defence proposition or an alternative hypothesis (Hd), unless contrary information regarding a specific defence is received, the laboratory will be entitled to presume that the defendant will deny the alleged biological relationship. Any assumptions made should be stated explicitly (such as an assumption of maternity when considering paternity).

7.1.3 For body identification work, and when formulating Hd, the laboratory will be entitled to presume that the deceased is unrelated to the putative family of interest.

7.1.4 In addition, the laboratory should give due consideration as to whether the application of the autosomal STR tests at their disposal could adequately address the issues in the case. See Annex 1, which considers the power of autosomal STR tests for addressing certain biological relationships and contains additional guidance.

7.1.5 If there is doubt as to whether the proposed tests can satisfactorily address the propositions, the laboratory should inform the submitting authority of this. However, if the submitting authority nevertheless wishes to continue with testing, then this duty should be considered to have been discharged.

7.1.6 In disaster victim identification cases where there are multiple fatalities and some within the same family, the laboratory should alert the submitting authority

that the DNA tests employed may not be able to differentiate between close relatives involved in the same incident.

7.1.7 Where appropriate, the laboratory should recommend the application of additional testing (such as Y-STRs or mitochondrial DNA) or alternative techniques (such as SNPs) if these techniques offer a more suitable alternative to addressing the issue(s) in the case. This requirement exists whether the forensic science provider (FSP) is capable of providing these services or not.

7.2 Interpretation and evaluation of evidence

7.2.1 After testing is complete, the genetic findings (i.e. the resulting DNA profiles) should be evaluated in order to determine the degree of support for both the prosecution (Hp) and defence (Hd) propositions.

7.2.2 For those parent/child cases in which it is possible to formally exclude a biological relationship, the laboratory should establish and document their 'exclusion' criteria (see 9.10).

7.2.3 The statistical assessment of the strength of evidence in relationship cases is fundamentally different from 'standard' identification cases. The required formulae for some of the more common relationships are published in standard academic textbooks on the subject, see Buckleton et al., 2016 [13].

7.2.4 However, for more complicated situations, bespoke formulae would need to be derived individually based on the specific pedigree and so the assistance of software is advisable (see section 9.3).

7.2.5 The basic process of evidential evaluation follows essentially the same framework as 'standard' identification cases by:

a. Formulating both prosecution and defence propositions based on the case information;
b. Assigning probabilities to those propositions using the data; and
c. Computing a likelihood ratio as a measure of the evidential strength.

7.2.6 The same UK allele frequency data that is used for 'standard work' can be used for assignment of probabilities in relationship testing cases. Further information

regarding the allele frequency data can be found in the FSR's guidance document on the use of allele frequency databases to assign a likelihood ratio (LR) in human DNA cases, FSR-GUI-0012.

7.2.7 For the purposes of a calculation, a practitioner may elect to choose one allele frequency database (which is most similar to the biogeographic ancestry of the defendant) or perform multiple calculations using the different available frequency databases.

7.2.8 If multiple calculations are performed, the laboratory should document its policy and procedure for reporting those outcome(s).

7.2.9 Consideration should be given as to how to deal with 'rare' or 'zero frequency' alleles

7.2.10 An allowance for co-ancestry may be necessary in some cases.

7.2.11 An allowance for linked loci may be necessary in some cases and with some multiplexes.

7.2.12 An allowance for mutation may be necessary in some cases.

7.2.13 Further detailed guidance regarding the use of population databases and the employment of various statistical allowances is provided in section 9.11.

7.2.14 The policy for application/non-application of the above allowances by practitioners should be documented.

7.3 Use of lineage markers in questions of relationship

Lineage markers are those that are inherited directly from a parent to a child and can sometimes be useful when trying to infer more distant genetic relationships.

The Y chromosome is passed directly from a male to all of his male children. Males that share a paternal relationship may be expected to share the same Y-STR haplotype profile and that can be over many generations. While mutations in some Y-STRs are more common than others, it cannot be assumed that males sharing the same Y-haplotype are necessarily closely related.

There are two X chromosomes in females that recombine and female and male offspring of the mother will not inherit the same X-STR haplotype. However males will pass on their unrecombined X chromosome to their female children and so females that share the same biological father will be expected to share an allele at all X chromosome loci.

Mitochondrial DNA (MtDNA) is inherited from the mother and therefore all individuals sharing maternal inheritance, male or female, will be expected to share the identical MtDNA sequence that may be presented as a haplogroup. Only female children will pass this on to their descendants.

Y-STR frequencies are collated worldwide in YHRD [14] and MtDNA, similarly, in EMPOP [15]. Many of the X-STRs are also linked and therefore haplotype frequencies are often reported in linkage groups and require special statistical methodology to evaluate, such as available in the FamlinkX software application [16].

While each of these markers can be considered independent and therefore, statistically, could be combined with the genetic weight from autosomal genetic markers (as stated in the ISFG guidance [17]), it is recommended that, because of the limited patrilocal and matrilocal frequency knowledge and, in the case of X chromosome linkage haplogroups that have more limited coverage, information from these markers should be considered and reported separately from the autosomal LRs while potentially offering independent support in complex kinship.

7.4 General recommendations on the use of calculation software

7.4.1 Although manual calculations are possible for some of the simpler biological relationships, due to the large number of mathematical operations necessary and the error-prone nature of such calculations, it is strongly recommended that calculation software be employed to prevent mathematical errors.

7.4.2 All software applications require that genotypic information be entered and so it is recommended that, if possible, automatic import functionality be used to minimise the risk of transposition errors.

7.4.3 Where allele data are keyed in manually, additional transcription checks should be undertaken to ensure the accuracy of the inputted data.

7.4.4 Output files should be generated and stored (as either electronic copy or hard copy or both) so that these data can be inspected at a later date. The output should contain a record of the alternative prosecution and defence propositions considered and sufficient detail to enable checking, auditing and defence review.

7.4.5 Where such a record is not part of the software output, manual records shall be made and retained (code section 28.1).

7.4.6 It is recommended that the user interface should be intuitive and easy to use: (for example, using a graphical user interface for 'button and menu' clicking rather than requiring entry of command lines).

7.4.7 It is recommended that the source code and any embedded data files (such as allele frequency tables) be locked to prevent users from inadvertently altering it.

7.4.8 Prior to any release of updates and/or new versions of calculation software a risk assessment should be carried out and, if appropriate, the new versions should be re-validated.

7.5 Recommendations regarding functionality for calculation software

7.5.1 It is recommended that any software application possesses the following calculation functionality.

a. The ability to use different geographic ancestry (population) groups.
b. The ability to apply some form of sampling adjustment to allele frequencies.
c. The ability to allow for co-ancestry/population substructure.
d. The ability to make some form of statistical allowance for:

i. Germline mutation; and

ii. Linked loci.

7.6 Recommendations regarding the types of biological relationship that could be analysed by software

7.6.1 It is recommended that calculation software should be designed to evaluate biological relationships. These include the following biological relationships:

a. Parent/child;
b. Siblings/half siblings;
c. Aunt/uncle;
d. Grandparents;
e. First cousins; and
f. Any of the above relationships involving incestuous pedigrees.

7.7 General guidance on the validation of statistical software tools

7.7.1 It is a general requirement that all software tools deployed by FSPs shall have been validated to a standard acceptable to the key stakeholders (the Code section 24.3). Those designed for relationship testing applications are no different.

7.7.2 The International Society for Forensic Genetics (ISFG) has published guidelines for the validation of software performing bio-statistical calculations for forensic genetic calculations, see Coble et al., 2016 [18]. This sets out the minimum requirements for validation and covers both developmental and operational validation.

7.7.3 For the purposes of validation, the sections on ‘test methods and method validation’ and ‘estimation of uncertainty’ in the Code [3] shall apply.

7.7.4 For calculation tools, such as the use of Excel spreadsheets, the onus is on the party employing such a tool to validate it by demonstrating that the functions and calculations it performs are mathematically correct.

7.8 Dealing with apparent inconsistencies in the mendelian inheritance pattern

7.8.1 Where necessary, the laboratory should either take into account statistically, or address by sufficient additional testing, an apparent inconsistency in the Mendelian pattern of inheritance. It is not permissible, when estimating the strength of evidence, to ignore a locus exhibiting an incompatibility with the Mendelian inheritance pattern.

7.8.2 The laboratory should document its policy and procedure for dealing with apparent inconsistencies in the Mendelian inheritance pattern.

7.8.3 Inconsistencies in the Mendelian inheritance pattern can arise even though two individuals are biologically related as parent and offspring – via the mechanism of mutation. When these mutations occur in the germline cells (i.e. at meiosis) the change becomes a heritable trait.

7.8.4 There are three types of germline mutational change that can potentially affect the results of relationship tests performed using STRs.

a. Base mutations and/or indels affecting a primer binding site can abolish the priming of one allele at a locus leading to the presence of a silent (null) allele that is not observable via a polymerase chain reaction. Hereafter this is referred to as a primer binding site mutation.
b. Alterations (for example, due to strand slippage or unequal crossover) to the number of repeats at a STR locus. Mostly (but not exclusively) these produce ‘single-step’ changes (i.e. the number of repeats either expands or contracts by one full repeat unit).
c. Gross chromosomal rearrangements (such as microdeletions in the male Y chromosome, uniparental disomy [UPD] and aneuploidy).

7.8.5 Some of these can result in the biological offspring apparently having no allele in common with the biological mother (maternal mutation) and/or the biological father (paternal mutation), or the biological father not being recognised as male because of deletion of the AMELY locus that is generally used to determine sex

in a DNA test. These phenomena are therefore inconsistent with the expected pattern of Mendelian inheritance given that the biological relationship is true.

7.8.6 It is possible that some germline mutations can occur without affecting the observed Mendelian inheritance pattern (so-called 'covert' mutations).

7.8.7 It is recommended that primer binding site mutations be resolved by further analysis using alternative primer sets to render the null allele visible. If this is not possible, or the further analysis fails to demonstrate the presence of a null allele, then it is recommended that the locus be treated statistically.. Charles Brenner in his DNA View website [19] provides formulae that can be used when a null allele is suspected. Locus specific null allele frequencies and other discordances are available from the new National Institute of Standards and Technology (NIST) STRBase [20].

7.8.8 Suspected cases of UPD (based on the isolated observation of shared genotypes in the child with only one parent and nothing in common with the other parent) can be investigated by testing other markers located on the same section of chromosome: see Cavalheiro et al., 2020 [21]. If this is not possible, then it is recommended that the locus be treated statistically as though a slippage germline mutation had occurred.

7.8.9 Where it is necessary to allow statistically for the effects of an overt, repeat change mutation when computing the likelihood ratio (LR), implementation of a mutation model will be required.

7.8.10 Several mutation models are available but the one most commonly used in the forensic community is the 'stepwise' mutation model, see Ohta and Kimura, 1973 [22]. In this model, alleles are deemed to mutate in an iterative stepped fashion (with multistep changes being less probable than single-step changes).

7.8.11 Mutation rates from collections of data are published annually by the Association for the Advancement of Blood and Biotherapies (AABB) in a technical report [23] that can be used to derive or infer transition probabilities. Rates are known to be different for males and females and tables 1 and 2

within Annex 2 of this document provide a selected subset derived from the 2023 report.

7.8.12 In addition, in males, the mutation rate appears to increase with chronological age, see Qin et al., 2015 [24]. This has been ascribed to the fact that in females, the oocytes are laid down at birth, whereas spermatozoa are made continuously in men and so, over time, have the propensity to amass more mutations.

7.8.13 It is recommended that any calculation software contains the necessary functionality to include a statistical allowance for mutational events.

7.9 Dealing with linked loci

7.9.1 For any autosomal multiplex used by the laboratory to investigate biological relationships, the laboratory should establish which (if any) of the loci are syntenic and which of those syntenic loci will require a formal linkage allowance.

7.9.2 The laboratory should document this policy accordingly.

7.9.3 For those loci deemed to require it, the laboratory should incorporate an allowance for linkage into the reported LR.

7.9.4 The laboratory should document its policy and procedure for dealing with linked loci (see 9.8.9 to 9.8.10).

7.9.5 STR loci located on the same chromosome are said to be ‘syntenic’. With the increasing size of multiplex kits, it is more likely that syntenic loci will be present. The corollary of this is that because the loci are not on separate chromosomes, they will exhibit linkage, therefore a statistical allowance should be incorporated where necessary.

7.9.6 The closer together the loci are on the chromosome, the more tightly they will be linked. Some syntenic loci are so far apart on the chromosome that the effects of linkage will be negligible. For some more closely linked loci, the effects can be more significant.

7.9.7 Of the loci currently (2021) used for the UK National DNA Database®, only vWA and D12S391 are considered close enough (12.9cM) to warrant an allowance to be made.

7.9.8 Other syntenic loci that are present in DNA24 (TPOX-D2S441-D2S1338 and D5S818- CSF1PO) are more distant from each other – at least 87.7cM on chromosome 2 and at least 25.9cM distance on chromosome 5.

7.9.9 Although genetic linkage has no impact on LR calculations in standard parentage cases, LRs can be substantially impact other relationships where the effects of linkage are not taken into account, see Tillmar and Phillips, 2017 [25]. Furthermore, linkage effects can result in an overestimation of the LR in scenarios involving incest, see O'Connor and Tillmar, 2012 [26].

7.9.10 A formal statistical adjustment can be made using recombination frequencies, see Bright et al., 2014 [27].

7.9.11 Alternatively, a simpler adjustment can be achieved by dropping one locus from each syntenic pair post-calculation. It is most conservative (i.e. most favourable to the defendant) to drop the locus with the highest calculated LR.

7.10 Making an allowance for co-ancestry (fixation index)

7.10.1 An allowance for co-ancestry should be applied to all calculations for criminal cases.

7.10.2 An allowance for co-ancestry is not required for body identifications or for those cases where the defendant is positively asserting (as opposed to denying) a biological relationship.

7.10.3 An allowance for population substructure can be made using a fixation index (Fst) adjustment (θ). This is in line with the policy for criminal cases involving DNA identification. This has been set at 0.03 (or 3%) and ensures that a calculation is not unfavourable towards a defendant who is taken to be disputing the biological relationship, see Steele and Balding, 2014 [28] and Steele et al., 2014 [29].

7.10.4 However, if a person on trial is positively asserting a biological relationship then θ may be set to zero (or at a much lower value than 0.03) at the discretion of the practitioner. This is because in such cases, any over reduction in evidential strength caused by applying a larger than required co-ancestry allowance operates against the interests of the defendant.

7.10.5 In body identification cases, there is no prima facie reason to skew the calculation in the favour of any particular party and so θ may be set at a more realistic value than 0.03 at the discretion of the practitioner.

7.11 Guidance on declaring an exclusion

7.11.1 In theory a LR can be calculated for any given pedigree (irrespective of how many mutational events might be required to explain the evidence under the Hp). If the LR was less than 1, the evidential strength could then be expressed in terms of the degree of support for the Hd. For each locus for which a mutation must be invoked under the Hp, the corresponding probability will be very low (often in the order of 0.001). When several loci are involved the LR will become very small very quickly.

7.11.2 In reality, therefore, and for pragmatic reasons, rather than report a large LR supporting the Hd a simple ‘exclusion’ may be declared.

7.11.3 It follows logically from this analysis that if the pedigree is fully consistent with the Hp, a LR calculation should be undertaken. However, if there are inconsistencies under the Hp at ‘several’ loci, then an ‘exclusion’ may be declared instead of undertaking a calculation.

7.11.4 A laboratory policy should be set and guidance issued to practitioners as to how many loci must exhibit inconsistencies under the Hp in order to trigger a declaration of an exclusion. That number is generally set at between at least 2 or 3 inconsistencies, dependent on the number of loci examined..

7.11.5 The number of steps required for the proposed mutation(s) will have a bearing on this decision because, under the stepwise mutation model, multistep

changes to the repeat units are inherently less probable than single-step repeat unit changes.

7.12 Guidance on using allele frequency (population) databases

7.12.1 Allelic frequency (population) databases are required for statistical evaluations to be performed in all DNA cases. A population database is usually defined according to ethnic appearance (biogeographic ancestry group) and/or geographical descriptors.

7.12.2 In identification work, forensic providers in the UK make use of four different data collections, which represent most of the population of the UK. These same data collections can be used for relationship testing purposes. Further guidance can be found in FSR-GUI-0012, the use of allele frequency databases to assign a likelihood ratio (LR) in human DNA cases.

8. Reporting Considerations

8.1 Reporting format and contents

8.1.1 The specific content of a report is only covered in this section insofar as it concerns the additional elements that are required for reporting the outcomes of investigations into biological relationships.

8.1.2 The principles of determining and reporting a biological relationship stem from family law when blood tests were used to determine paternity, the statutory instrument being subsequently amended to allow for DNA tests from mouth swabs [32].

8.1.3 In the above legislation, the report requires a conclusion, the reason for the conclusion, a comment on the value of the test in answering the question, and the test results. These regulations are fulfilled by the following proposed content of a written report when dealing with the investigation of biological relationships in criminal investigations.

8.1.4 The written report should include:

a. A summary of the pedigree information supplied and on which the analysis has been based;
b. A statement of the specific propositions that were used to compute the likelihood ratio (LR); and
c. A declaration of any assumptions that have been included within the analysis (for example, that a child's maternity has been assumed to be true).

8.1.5 The LR should always be reported but may be rounded to 2 significant figures (2SF).

8.1.6 The calculation of a posterior probability (or any other value that requires an assessment of the priors of a biological relationship) should not be reported.

8.1.7 In line with other policies, a LR of 'one billion' should be adopted as the upper reporting boundary for all relationship tests.

8.1.8 Forensic science providers may unilaterally decide to self-impose a lower testing boundary for certain types of relationship (for example, an internal laboratory policy to try to obtain a minimum LR). However, the test results obtained should still be disclosed to the submitting authority even if the required threshold has not been reached.

8.1.9 Unless a specific defence is advanced, or the submitting authority otherwise requests it, there is no duty placed on the practitioner to unilaterally consider, explore and report on, other scenarios involving individuals related to the defendant.

8.2 Ethical dilemmas and The Equality Act 2010

8.2.1 During DNA profiling tests information as to biological sex is obtained, which could contradict the stated gender of a person in the pedigree. On 16 April 2025 the UK Supreme Court ruled that, legally, 'sex' will refer to the biological sex of an individual, while not removing any existing protections of transgender individuals under the Equality Act 2010 [33]. This ruling ensures that DNA tests will be considered based on biological sex of the individual.

8.2.2 Data collection should, however, respect both gender identity and legal sex with appropriate privacy protections in place.

8.2.3 As contradictory sex information may also reveal a sample mix-up prior to receipt at the laboratory, it is good practice to request a repeat test from the instructing authority without revealing any contradiction to third parties, including directly to the donor(s) themselves.

8.2.4 Under normal circumstances it is not necessary to make reference to observed genetic anomalies in DNA testing. If considered necessary a clinical geneticist should be approached for guidance and consideration of providing a joint report after discussion with the submitting authority.

8.2.5 Contradictory sex information can arise in the following different ways:

a. A genetic anomaly at the Amelogenin locus (AMELY) can result in the non-priming of the Y homolog, see Steinlechner et al., 2002 [34]. This produces an apparently female DNA profile even though the person is genetically male. Newer multiplexes often incorporate additional markers from the Y chromosome to assist recognition.

b. In rare conditions, such as androgen insensitivity syndrome (AIS), 46XY males do not respond to the hormones needed for the development of male characteristics due to androgen receptor mutations. These individuals appear phenotypically female at a prevalence of about 6.4 in 100,000 live-born apparent females.

8.2.6 When testing a foetus, the biological mother may not know the sex of the child and might wish not to receive this information. The foetal sex can be reported, and the relevant authority should consider whether or not they should disclose this information.

8.2.7 For deceased individuals, and for products of conception, it is recommended that any tissue samples be held by the DNA testing laboratory for the minimum amount of time possible (for example, only until a DNA profile has been prepared) and thereafter should be returned promptly to the seizing authority.

8.2.8 Human tissue should always be handled in accordance with the provisions of the Human Tissue Act 2004 [5].

9. Dealing with unusual features and genetic anomalies in DNA profiles

9.1.1 The majority of reference samples processed for relationship testing are expected to be single source with each locus having one or two alleles (depending on its heterozygosity). Where two alleles are present, and provided sufficient DNA is present, the allelic peaks should be balanced. Using neoplastic tissue as a surrogate sample can reveal gross chromosomal rearrangements and loss of whole chromosomes, see Budimlija et al., 2009 [35].

9.1.2 Occasionally, however, samples exhibit reproducible aberrant di-allelic or triallelic patterns, see Clayton et al., 2004 [36] and/or the sample appears to be reproducibly mixed despite being expected to be single source, see Rettner, 2016 [37].

9.1.3 Aberrant allelic patterns are the results of genetic rearrangements (somatic mutations, copy number variation, trisomy and so on). These will only matter if the rearrangement affects the gametes and then causes an inconsistency the Mendelian inheritance pattern.

9.1.4 Additionally, they may present problems for software in subsequent calculations when a genotype has to be input. In such instances, and in order to use a software calculator, it is permissible to exclude such a locus from a calculation provided that the results at the locus are still compatible with the asserted biological relationship.

9.1.5 Unexpected mixtures obtained from the testing of a reference sample can be the result of a number of biological reasons (other than sample contamination).

9.1.6 If a personal effect (such as a toothbrush) has been tested as surrogate reference sample, then this could simply mean that another person has used the article concerned.

9.1.7 Samples taken from products of conception often produce mixed DNA profiles due to the presence of mixed maternal and foetal tissues. If a sample from the mother has been provided it may be possible to condition the mixture on the mother's DNA profile, thereby determining the paternally inherited components in the DNA profile of the foetus. It is possible to compare these paternal DNA components to the DNA profile of any alleged father.

9.1.8 Occasionally, there can be medical or biological reasons for the individual being tested to exhibit a mixed DNA profile. Such reasons include being the recipient of an allogenic bone marrow transplant, see Pope et al., 2006 [38] or rare instances of human chimerism, Yunis et al., 2007 [39].

9.1.9 All samples giving unexpected results should be duplicated prior to initiating further investigation. If multiple additional peaks are observed reproducibly within a DNA profile, it may be necessary to first consider and investigate whether the original donor sample could have been contaminated with exogenous DNA at the laboratory (or during the original sampling).

9.1.10 The next step is to request from the submitting authority a replacement sample for the individual in question. If the phenomenon persists then it is likely that the person is exhibiting an *in vivo* mixed DNA profile for one of the reasons outlined above.

9.1.11 Sometimes, targeting a different type of sample (for example, hair follicles rather than blood) can resolve the issue, see Chaudhary et al., 2015 [40].

10. Acknowledgements

10.1.1 This guidance has been adapted from the previous non-statutory guidance (FSR-G-228, DNA Relationship Testing using Autosomal Short Tandem Repeats, Issue 1) and reviewed by the Regulator's Human DNA Sub-Specialist Group.

11. Modification

11.1.1 This is the first issue of this document under section 9 of the Forensic Science Regulator Act 2021.

11.1.2 The PDF is the primary version of this document.

11.1.3 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 three '#') indicate letters to describe the type of document and (b) (the second four '#') indicates a numerical code to identify the document. For example, this document is FSR-GUI-0014, and the 'GUI' indicates that it is a guidance document. Combined with the issue number this ensures that each document is uniquely identified.

11.1.4 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 - ### - #### - #.

11.1.5 In the event of any discrepancy between the primary version and a modified version then the text of the primary version should prevail.

12. Review

12.1.1 This document is subject to review by the Forensic Science Regulator at regular intervals.

12.1.2 The Forensic Science Regulator welcomes views on this guidance. Please send any comments to the address as set out at the following web page: www.gov.uk/government/organisations/forensic-science-regulator or send them to the following email address: FSREnquiries@forensicscienceregulator.gov.uk.

13. Abbreviations and acronyms

Abbreviation	Meaning
cM	CentiMorgans
DNA	Deoxyribonucleic acid
FSI	Forensic Science International
FSP	Forensic science providers
FSR	Forensic Science Regulator
F_{ST}	Fixation index
Hd	The defence proposition or hypothesis
Hp	The prosecution proposition or hypothesis
ISFG	International Society for Forensic Genetics
LR	Likelihood ratio
NDNAD	National DNA database®
NIST	National Institute of Standards and Technology
PACE	Police and Criminal Evidence Act 1984
SNP	Single nucleotide polymorphism
STR	Short tandem repeat

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Annex 1

15. Additional considerations for addressing certain biological relationships

15.1 The 'power' of STR kits for investigating specific biological relationships

15.1.1 In general, the power of any particular multiplex (or combination of multiplexes) in addressing a specific relationship will depend on the number and the heterozygosity of the loci employed. The more loci that are present and the more discriminating those loci are, the greater will be the power of the tests.

15.1.2 However, of much greater impact is the type of biological relationship being investigated and the number of other 'known' samples within the pedigree. So for instance, in a paternity test, the test will generally have more power if the non-questioned mother is also included in the testing rather than being absent (i.e. if a 'trio' is tested rather than a 'duo').

15.1.3 The greater the genetic distance between the parties, the less powerful the tests will be at addressing the disputed relationship. So, tests of first-degree relationships (such as maternity, paternity and siblingship) are more powerful than tests of second-degree relationships (such as grandpaternity tests or avuncular – uncle/nephew or uncle/niece – tests).

15.1.4 For example, using the 16 loci in DNA17 and a fixation index (Fst) of 0.03, about 87% of true full siblings will provide a LR greater than 100 with a 0.05% false positive rate.

15.1.5 Making use of additional loci, such the additional loci provided in Globalfiler™, or PowerPlex® Fusion, can improve the separation further. In this case 93% of true full siblings will provide a LR greater than 100 with a 0.05% false positive rate.

15.1.6 Figure 1 illustrates the separation between full siblings and unrelated pairs when using the 16 National DNA Database® (NDNAD) loci [20] and applying an Fst of 0.03 for Caucasians.

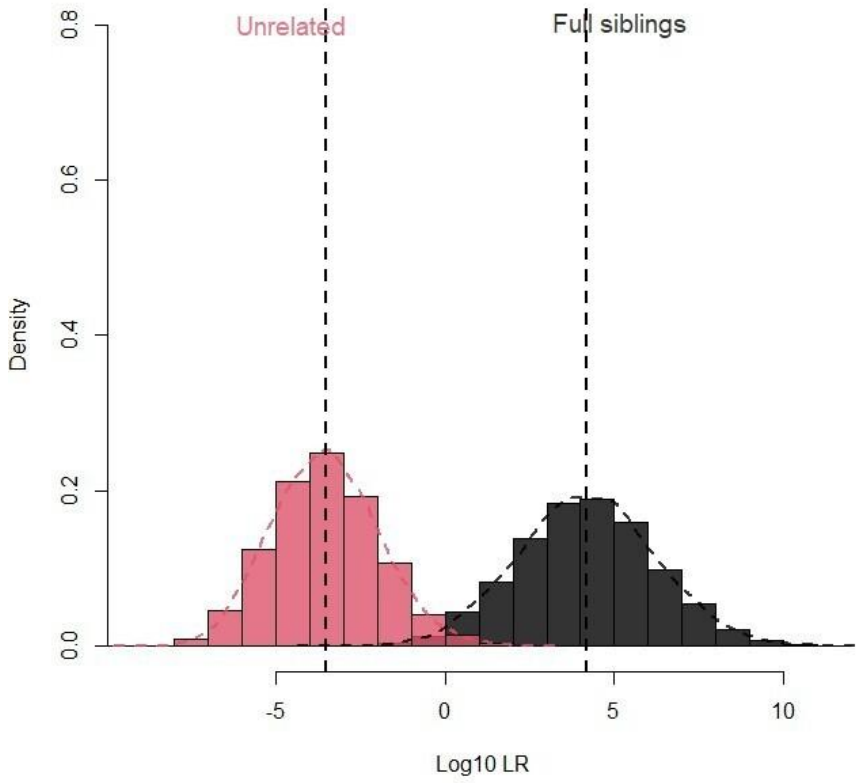

Figure 1: Separation between full siblings and unrelated pairs for 16 loci with Fst 0.03 for Caucasians

15.1.7 Additional loci could be used to improve confidence of a second-degree relationship if a LR greater than 100 is not achieved.

15.2 Testing of second and third-degree pedigrees

15.2.1 The relationship testing of second-degree pedigrees will often lead to low or modest LRs. For example, Figure 2 illustrates the poor separation between half siblings and unrelated pairs when using when using the 16 NDNAD loci and applying an Fst of 0.03 for Caucasians.

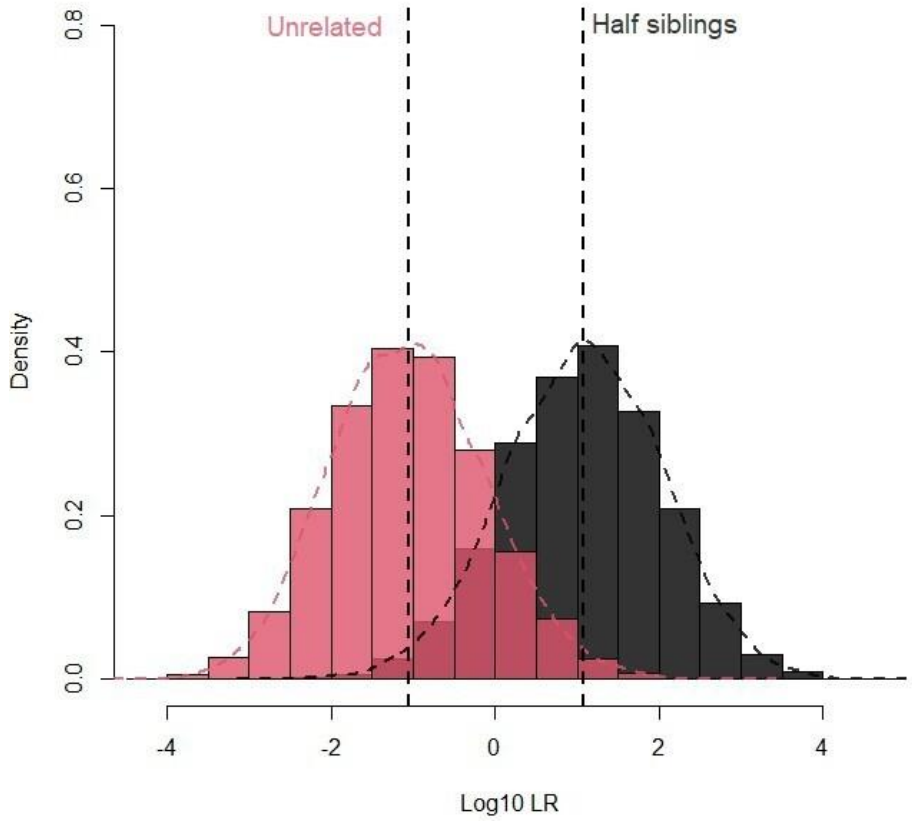

Figure 2: Separation between half siblings and unrelated pairs for 16 loci with Fst 0.03 for Caucasians

15.2.2 Adding ‘known’ relatives into such a pedigree is likely to improve the power of the test and may produce more meaningful results.

15.2.3 Testing third-degree pedigrees (without any other individuals in the pedigree) is unlikely to be satisfactorily addressed using standard autosomal STR testing and LRs obtained are unlikely to be meaningful even where multiplex kits of 20+ loci are employed.

15.2.4 For second- and third-degree relationships, consideration should therefore be given to applying other supplementary DNA profiling techniques to investigate the relationship or to supplement the STR analyses.

15.2.5 Tests using lineage markers and/or large single nucleotide polymorphism (SNPs) panels and/or large panels of sequenced STRs will have much greater investigative capacity for more distant relationships.

15.2.6 Given the information contained within this annex, it is not generally recommended to use a verbal scale, such as that published by the Association of Forensic Science Providers [41], but a qualifying comment on level of support may be provided.

Annex 2 – Germline mutation rates

15.2.7 Data regarding Germline mutation rates is available via the AABB [23], which is updated annually. Trends have shown that there are minimal changes within the data, over a number of years. It is recommended that this table is used to define the mutation rates for such calculations, it is not necessary to update this on a yearly basis.

Locus	ObsMut	Meioses	Rate		
CSF1PO	43	500469	0.0000859		
D10S1248	58	498010	0.0001165		>400,000
D12S391	66	117069	0.0005638		<400,000
D13S317	78	511637	0.0001525		<200,000
D16S539	70	510506	0.0001371		<20,000
D18S51	128	509382	0.0002513		
D19S433	107	508980	0.0002102		
D1S1656	54	499646	0.0001081		
D21S11	247	508527	0.0004857		
D22S1045	20	505247	0.0000396		
D2S1338	41	505391	0.0000811		
D2S441	41	500458	0.0000819		
D3S1358	37	511973	0.0000723		
D5S818	51	511757	0.0000997		
D7S820	44	501980	0.0000877		
D8S1179	62	510511	0.0001214		
FGA	122	508661	0.0002398		
Penta D	6	14734	0.0004072		
Penta E	7	14707	0.0004760		
SE33	216	374599	0.0005766		
TH01	11	510659	0.0000215		
TPOX	19	505110	0.0000376		
vWA	80	507618	0.0001576		

Table 1: maternal mutations 2023 data, loci sorted alphanumerically

Locus	ObsMut	Meioses	Rate
CSF1PO	424	500469	0.0008472
D10S1248	224	498010	0.0004498
D12S391	512	117069	0.0043735
D13S317	400	511637	0.0007818
D16S539	284	510506	0.0005563
D18S51	611	509382	0.0011995
D19S433	209	508980	0.0004106
D1S1656	14	499646	0.0000280
D21S11	371	508527	0.0007296
D22S1045	59	505247	0.0001168
D2S1338	398	505391	0.0007875
D2S441	125	500458	0.0002498
D3S1358	377	511973	0.0007364
D5S818	301	511757	0.0005882
D7S820	282	501980	0.0005618
D8S1179	464	510511	0.0009089
FGA	896	508661	0.0017615
Penta D	20	14734	0.0013574
Penta E	50	14707	0.0033997
SE33	937	374599	0.0025013
TH01	29	510659	0.0000568
TPOX	54	505110	0.0001069
vWA	692	507618	0.0013632

>400,000
<400,000
<200,000
<20,000

Table 2: paternal mutations 2023 data, loci sorted alphanumerically.

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