Forensic Science Regulator
Overseeing Quality

DNA Analysis Specialist Group (DNASG)

Minutes of the twenty-third meeting held on 3 May 2016, at 5, St Philip's Place, Colmore Row, Birmingham

1. Welcome and apologies

1.1 The Chair welcomed all to the meeting. A full list of attendees is available at Annex A. Apologies had been received from Mark Bishop of the Crown Prosecution Service.

1.2 The Chair welcomed visitors Kevin Sullivan and Ian Evett from Principal Forensic Services (PFS).

2. Standards

DNA mixture interpretation software validation standard and guidance

2.1 PFS had been successful at tender to develop a DNA mixture interpretation software validation standard and guidance for the Forensic Science Regulator (FSR). The standard was required due to a high degree of variation between and within Forensic Science Providers (FSPs) in relation to the evaluation and reporting of results from mixed DNA samples and to ensure that DNA mixture interpretation software was properly validated. The existing ISO standards 17020 & 17025, only state that software should be suitably validated as being adequate for use. There were other standards from which guidance could be drawn including ISO 12207 and ISO 15288 which cover software life cycle processes and software engineering and system life cycle processes respectively. The existing Regulator's standards do not cover how software should be validated.

2.2 The tender for the standard and guidelines focused on DNA mixture interpretation software however it was questioned whether the guidelines should extend to software for the interpretation of all DNA profiles including DNA from a single source (i.e. non-mixed DNA profiles).

2.3 The group heard that PFS had sent out a questionnaire to all FSPs in the UK and to a few international FSPs seeking views on the draft standard and guidelines which had been developed. In particular, feedback had been sought on the overview of the validation requirements for mixture interpretation software including, the overall approach and the approach to breaking down the validation into software and the model.

2.4 A question was raised in relation to the scope of the document and whether it was intended that the principles would be applicable to simple binary mixture interpretation models as well as more complex models. It was noted that the focus of the document had been on the continuous model approach as it was anticipated these would primarily be used in the future however, the principles of validation should be sufficiently general to be applied to all models.

2.5 It was queried whether the standard should specify the performance parameters of a model and whether, as a minimum, it should be capable of interpreting four person mixtures. The Regulator expressed concerns about the variation in capacity between different FSPs in regards to complex mixture interpretations and put forward the view that instead of specifying that the software should have the capability to interpret 4 person mixtures it should be the FSPs who have the capability to interpret 4 person mixtures. The Regulator was aware of expert statements which indicated that software were available to interpret a mixed DNA profile however in the particular case in question, the software had not been used. If a statistical approach is available to interpret a profile then that approach should be used routinely.

2.6 The validation needed for off-the-shelf mixture interpretation packages was discussed as these were predicted to be commonly utilised amongst FSPs. Whilst this had been included in the standard it was agreed that clearer sign-posting to this was required. The models and coding used in off-the-shelf packages were likely to have been validated by the manufacturers, however it would also be necessary for FSPs to undertake end-user validation of the software when used with their own GeneMapper® settings and protocols. It would be the FSPs responsibility to ensure that there were no gaps in the end-to-end validation of the software. It was agreed that the FSR should engage with the manufacturers of software at an early stage in order to gain their feedback. A chart showing the requirements and responsibilities for each stage will be included in the guidelines.

2.7 Further comments were sought from members to ensure that FSPs were satisfied with the minimum standards of capability set for a piece of software and the timescale of when FSPs would be compliant with these requirements.

Action 1: Members to send their comments on the DNA mixture interpretation software validation standard and guidance to the Secretariat by 17 May 2016.

Formulation of propositions in the evaluation of DNA mixtures & discussion of issues relating to the presentation of qualitative opinions of evidential weight in relation to complex mixtures.

2.8 PFS had again been successful at tender to produce guidelines for mixture interpretations for the FSR. Members heard that during the development of the guidelines, PFS would consult with interested parties including the police, CPS, judiciary, FSPs and international bodies. Once the

guidance was completed the FSR would take forward a full public consultation on the guidelines.

2.9 Members were invited to comment on acceptable boundaries for scientists to formulate qualitative opinions and how it might be possible to establish the capabilities of scientists to formulate qualitative opinions.

2.10 When considering section 3 'Data & Observations' of the paper 'Formulation of propositions in the evaluation of DNA mixtures', it stated that the evidential value depends crucially on the scientists judgement regarding the data that can be included in the calculation but it should be made clear that it also depends on the validation and the analysis associated with that validation. When considering section 6 'Informativeness' of the same document it was suggested that the importance of the validation process be highlighted to determine the capabilities and limitations of the software.

2.11 It was highlighted that in some cases there may be multiple defence propositions and also in some circumstances multiple prosecution propositions and this should be taken into account.

2.12 Section 10 on 'Number of contributors' should be re-phrased so that the role of the scientist in the evaluation was clear, emphasising that the scientist is not responsible for the prosecution's proposition.

2.13 It was queried whether the guidance covered activity level propositions and PFS clarified that the guidance would focus on sub-source and source level propositions.

2.14 The approach taken within the draft guidance in relation to qualitative opinions for mixture interpretations could be considered controversial as it had been written with the aim of discouraging scientists from providing qualitative opinions in courts for mixture interpretations. Feedback was requested in relation to this approach. The group heard that Cellmark Forensic Services had applied to UKAS seeking accreditation for qualitative opinions for mixture interpretations. Further details were sought as to the circumstances surrounding a situation when a scientist employed by Cellmark Forensic Services would provide a qualitative opinion for a mixture interpretation. The situation had arisen when the police had refused to pay for a statistical mixture interpretation using available software and had instead requested a qualitative opinion from the scientist. There were also instances when software was not available for the interpretation of a particular type of mixed profile. It was queried how it was possible to validate the opinion of a scientist and whether it was possible to validate a scientists opinion to interpret a mixture if it were not possible to validate the software to interpret the mixture. Huw Turk was invited to circulate to the group Cellmark Forensic Services' position on qualitative evaluation of mixtures.

Action 2: Huw Turk to circulate to the DNASG, Cellmark Forensic Services' position on qualitative evaluation of mixtures to the DNASG.

2.15 LGC Forensics reported that they had in the past provided a small number of qualitative assessments for mixture interpretations however they expected these numbers to decrease as new software was validated and they were therefore supportive of the direction of travel of the guidance. They did however believe the language to be overly robust in regards to the unsuitability of qualitative assessments in all circumstances. They also expected that there might be situations when even specialist software could not be used for mixture calculations, but it might be possible to undertake bespoke specific calculations rather than undertaking qualitative assessments. Only a limited number of individuals had the abilities to undertake bespoke calculations and if more individuals were trained, it would be possible to interpret more mixtures. This was discussed further under 2.18.

2.16 Key Forensics indicated that multiple police forces had asked them to provide qualitative opinions for mixture interpretations and that their refusal to provide these meant they were losing work.

2.17 The view held by the Regulator was that it was inappropriate for police forces to be involved in making decisions in regards to the processes used for mixture interpretation. Standards which prevented qualitative opinions would be able to defeat the commercial pressures being placed on FSPs to provide qualitative opinions.

2.18 It was queried whether the standard would allow for bespoke calculations to be used to interpret mixtures in situations where the accredited software was not available and whether simple mixtures using excel spreadsheets would be acceptable. Caution was exercised over the use of excel spreadsheets as they were considered to be error prone and questions were also raised about how the rarely used bespoke calculations could be validated.

Action 3: Members to review the documents 'Formulation of propositions in the evaluation of DNA mixtures' & 'Discussion of issues relating to the presentation of qualitative opinions of evidential weight in relation to complex mixtures' and feedback by 17 May 2016.

Anti-contamination standards

2.19 An update was provided from the FSR Unit on anti-contamination standards. The laboratory anti-contamination document had been published in December 2015 with an implementation date of April 2016. From October 2016, UKAS would be including in their inspections the incorporation of the anti-contamination guidance into laboratories' scope for 2017. The document would be updated with the version of ISO 18383 for 2016 and further reading material would be added to the reference section, however the substantive part of the document would not be changed.

2.20 The scene of crime anti-contamination document had undergone public consultation and had been updated and a conference would be held on 19 May 2016 in relation to the standard. There would be presentations from

various police forces who were already implementing the standard. The crime scene community have requested the implementation date of the anti-contamination document to be 2018 in order for crime scene examiners to be working to the standard when seeking accreditation to ISO 17020 in 2020. Interim guidance for Sexual Assault Referral Centres and Custody Suites would be published in July 2016.

3 Minutes of the last meeting

3.1 Clarity was requested in relation to item 12.2 of the previous minutes which related to a grant which had been awarded to Dundee University, in conjunction with the Royal Society, to establish a research centre for forensic science. It had been agreed that the work to write all the primers for the judiciary, including the DNA primer, would be subsumed under this grant. A group had been set up to develop the primer which would be chaired by Lord Hughes (Justice of the Supreme Court). The Regulator had spoken with Lord Hughes and had agreed to contribute to the primer. Consequently, the DNASG no longer needed to develop the DNA primer. The Chair noted that a primer covering DNA 17, primer binding site mutations and adventitious matches had been written and agreed by the judiciary and would be published in Science and Justice shortly.

3.2 The minutes were agreed as an accurate reflection of the discussions held and could be published on the FSR website.

4 Actions and matters arising

4.1 Members considered outstanding actions and matters arising from the previous meeting.

Action 3: June Guiness to redraft the papers on Syntenic Loci recommendations as an FSR Guidance note for publication

4.2 No progress had been made in relation to this action and consideration was being given to whether this could be tagged onto another document or published as a separate guidance document.

Action 6: The approach to using n-2 DNA matches to be pursued and both the policy and scientific aspects to be considered.

4.3 The group heard that the National DNA Database Delivery Unit (NDU) were considering a tender for a piece of research to develop the statistical assessment model and the outcomes of the research was likely to feed into the Home Office forensics and biometrics work.

4.4 A match of 31 out of the 32 alleles had been reported (with the only difference occurring at loci SE33) between a PACE sample and a crime scene sample recovered from the handle of a suitcase. The FSP were in the process of obtaining and profiling hand swabs from the person whom the PACE

sample originated. The NDU were currently classifying the two samples as not matching.

Action 4: Huw Turk to provide Adam Shariff with details of the apparent first adventitious match that had resulted with DNA-17 technology and feedback to the DNASG.

5 Work plan

5.1 Members considered the work plan and indicated that they were supportive of the timescales which had been provided. It was suggested that Next Generation Sequencing (NGS) should be added to the list of emerging technologies and it was noted that the International Society of Forensic Genetics (IFSG) had published a framework for NGS and that the group should maintain a watching brief in this area at this stage.

6 Emerging technologies

Y-STRs

6.1 The minutes from the Y-STR meeting held on 11 March 2016 were provided as well as a draft of the Quality Assurance section of the Y-STR standard which had been drafted by Stephen Ferguson[1]. Members were invited to feedback on these documents.

6.2 The Quality Assurance section of the standard as drafted expected that FSPs would create and maintain Y-STR elimination databases populated by staff who worked in the FSPs. It was suggested that staff who worked in Sexual Assault Referral Centres (SARCs) should also be included in the Y-STR elimination databases. The document also provided guidance on processing of extraction negatives and positives and guidance for the investigation of contamination incidents.

6.3 A number of ethical issues were raised in the Quality Assurance section of the Y-STR standard including how to deal with Y-STR profiling of transgender staff, the possibility that Y-STR profiling might unintentional reveal information about genetic relationships and the possibility that it might reveal information about male fertility.

6.4 A representative from one FSP requested that the document be re-worded so that FSPs would be able to create a single elimination database containing both autosomal and Y-STR profiles.

6.5 It was suggested that the ethical issues associated with Y-STR elimination databases should be put to the Home Office National DNA

[1] The minutes of the Y-STR sub-group meeting held on 11 March 2016 are available at Annex B.

Database Ethics Group for consideration and to feedback their advice to the group and the FSR.

Action 5: Secretariat to request advice from the National DNA Database Ethics Group in relation to the creation of Y-STR elimination databases.

6.6 The Quality Assurance section of the standard would be combined with a section on interpretation however this section of the standard could be published earlier. Members were asked to feedback on this section of the standard.

Action 6: Members to feedback on the Quality Assurance section of the Y-STR standard.

6.7 An update was provided on the discussion held at the Y-STR sub-group meeting on population data, including concerns about using Y-STR databases which are populated from individuals from other countries due to differences in Y-STR profile distributions between countries. The UK Y-STR profile database is small and only holds 800 profiles and a project was being undertaken at King's College London in collaboration with the Association of Forensic Science Providers (AFSP) to attempt to add a further 3000 profiles to the UK Y-STR database. Further concerns had been expressed about the use of the YHRD database and software due to the lack of control UK providers would have as the implementation of the model by the developers of the YHRD software had not been published and therefore had not been externally validated. A proposal had been put forward by the Y-STR subgroup for UK forensic providers to become self sufficient and develop their own statistical tool for evaluating the weight of evidence of Y haplotypes. The software tool would house the UK and Irish haplotype frequency datasets and be able to carry out statistical evaluations.

6.8 The group considered possible sources of funding for the development of the UK and Ireland Y-STR population database and statistical tool. It was noted that due to the UK and Irish focus, this project was unlikely to gain European Network of Forensic Science Institutes (ENFSI) funding. It was suggested that if the developer were able to find an academic partner, then research funding might be a possibility or alternatively if a police force were willing to collaborate then it might be possible to gain funding through the Police Innovation Fund. Further suggestions for sources of funding included the Home Office Biometrics Programme and the Home Office Centre for Applied Science & Technology (CAST).

6.9 As a way forward, it was suggested that the Y-STR subgroup would define the user specifications for the work at an early stage in order for an estimate of the costs to be drawn-up. Following this, the Regulator would explore the possibility of funding with the National Police Chief's Council Science and Innovation Board and Susan Hale would explore the possibility of funding through the Police Innovation Fund.

Action 7: Y-STR subgroup to be asked to define the user specifications of the Y-STR software.

6.10 Members consider the action from the Y-STR minutes to seek clarification of the interpretation of the Protection of Freedoms Act (2012) legislation and whether a Y-STR profile could be obtained after an autosomal profile had been obtained. It was agreed that this issue should be raised with the Biometrics Commissioner.

Action 8: Raise with the Biometrics Commissioner the issue of requiring clarification of the interpretation of the Protection of Freedoms Act (2012) legislation and whether a Y-STR profile can be obtained after an autosomal profile has been obtained.

6.11 Members considered the statistical methods currently available for interpreting Y-STR profiles, including the Pseudocount method (N+1/N+1), Brenner's Kappa methods and Andersen's Discrete Laplace method. Currently, there was no guidance or standards for FSPs as to which statistical method should be used and FSPs must make a decision themselves which method they believed to be acceptable. It was suggested that the International Society for Forensic Genetics (IFSG) establish a DNA commission to develop advice in this area including Y-STR single source and mixed profiles and that someone from the FSR DNA Analysis Specialist group could be co-opted onto the working group. Denise Syndercombe-Court agreed to put this suggestion forward.

Action 9: Denise Syndercombe-Court to put forward the suggestion to the IFSG that they establish a DNA commission to develop advice as to which statistical methods to be used for Y-STR single source and mixed profiles.

6.12 In relation to the minutes of the Y-STR meeting held on 11 March 2016 it was suggested that in para 3.7 the following line should be deleted 'It was agreed that Roberto Puch-Solis would develop a Laplace model, with the confidence intervals and FST agreed amongst the providers with the UK'.

Rapid DNA

6.13 The Rapid DNA project board had been established by the Home Office to evaluate the economics and efficiencies of on-site DNA testing at crime scenes. The board consisted of representatives from police forces, CAST, the NDU, the DNA Database Ethics Group and a representative from the FSR DNA Analysis Specialist Group. It was reported that there were currently issues with the cost of consumables and the impact of these costs on police budgets as well as issues with the requirements for validation and control samples. The project board work was on-going and was not currently at a stage where it could make a recommendation to police forces. It was highlighted that when considering the impact on budgets for rapid DNA work it would be necessary to consider the impact on the costs of the end-to-end

process within the Criminal Justice System and not solely the impact on forensic budgets.

Allele Frequencies for 20+ STR kits

6.14 Kings College London had undertaken work looking at allele frequency data in kits with additional loci to those in the DNA17 kit and had carried a direct comparison with DNA 17 data. Organisations using kits with additional loci would be able to use the allele frequency data that Kings College London had obtained rather than undertaking this work themselves. Kings College London were still in the process of evaluating the data and additional resources might be required in order to make the data available to FSPs.

Standard/requirements for provision of supporting data and documents for new technology

6.15 The Regulator noted that a high priority in her annual report was the requirement for the provision of supporting data and documents for new technology. Manufacturers were often focused on the technology however it was important that the FSPs focus on the data which was required to underpin these new methods and technologies so that they can be relied upon in court.

7 Professional and Scientific updates

Body Fluid Forum

7.1 The Body Fluid Forum (BFF) had coordinated work looking at the suitability of lubricants in forensic sampling and the effect of the lubricants on both presumptive testing for body fluids and DNA analysis. Testing of a small number of samples at each FSP was close to completion and the initial results had indicated that Aquagel has no adverse effects on either presumptive testing or DNA analysis.

7.2 Four papers had been written and would be submitted for publication. These were ‘Effect of Fabric Colour on Subsequent DNA Extraction and Quantification’, ‘Degradation of DNA and Enzymatic Activity’, ‘Drying Times of Semen, Saliva and Vaginal Material’ and ‘The Detection of Semen, Saliva and Vaginal Material following Secondary Transfer’.

7.3 The BFF hoped to be joining with the Association of Forensic Science Providers (AFSP) DNA working group (DNAWG) for its next conference, which was likely to be in 2018. The Chair of the AFSP DNAWG had been invited to the next BFF meeting where this would be discussed.

7.4 The Regulator raised an issue that some forensic scientists were not making reference to the considerable amount of published material when considering transfer and persistence in witness statements. The statements provided by some forensics scientists were not providing a clear link as to how opinions had been developed and it was expressed that using ‘In my opinion’ alone was insufficient and the literature should be referenced. It was

suggested that there was considerable published data on transfer and persistence and the AFSP were expecting to publish soon their work which evaluates the relevant literature in this area.

Association of Forensic Science Providers DNA working group

7.5 Members heard that the AFSP DNAWG were collating and reviewing literature on secondary transfer and persistence and were also heavily engaged in developing a programme for the joint conference with the BFF. They were also collating views from suppliers as to their issues in relation to the National DNA Database. The main AFSP group had been asked to represent the views of the DNAWG at the DNA Operations Group meeting in relation to best practice for obtaining victim elimination samples.

Other – ISFG/EuroForGen/ENFSI

7.6 A set of tools had been developed to use with mitochondrial DNA which would allow screening of mitochondria in situations when a large volume of mitochondria needed to be analysed.

7.7 Members heard about an ISFG project involving NGS of mitochondrial DNA to predict the age of the provider of an unknown sample. Fourteen laboratories had agreed to take part in an initial exercise and samples had been sent out to allow the laboratories to create a standard curve. The technique was currently only validated for blood samples and work in future would focus on validation of other tissues and searches for new markers.

7.8 STRbASE which was an ENFSI population database was being converted to a database named STRIDA which would be freely available and would be centrally curated in Austria. The database would have quality control and plausibility checks built in and would be able to accept NGS data.

7.9 The IFSG were also working on a system for nomenclature of STRs due to the increase in alleles which were being detected with NGS to prevent redundancy in the future.

7.10 Members heard that the development of two programs for mixture interpretation had been sponsored through EuroForGen; these were LRmix and EuroForMix. Discussion had been held at a EuroForGen meeting about the issue of different validated mixture interpretation software platforms producing different likelihood ratios (LR) and how this might be confusing to the courts. The view had been put forward that a verbal scale to explain the LR might be beneficial.

8 AOB

8.1 The Regulator informed the group that she meets with the NDU on a quarterly basis to review performance data from suppliers in order to investigate where errors might be occurring in the process. Errors as a result of manual transcriptions were highlighted as an area where there was a

continuing high volume and FSPs were asked to review the processes they have in place for manual transcriptions and to introduce effective checks to reduce the number of errors.

8.2 The Regulator noted that the use of Streamlined Forensic Reports and partial mixtures would be discussed at the next DNA Database Strategy Board due to the dissatisfaction from police forces with the decision not to include statistical interpretations in these reports.

8.3 June Guiness provided feedback from a recent ENFSI working group biology session. The importance for manufacturers to inform FSPs if the composition of their swabs changed was highlighted due to recent changes to the Flock swab having detrimental impact on one of the presumptive blood tests – the Opti test. ENFSI had undertaken two recent surveys to find out which presumptive and confirmatory tests were used by FSPs and a further survey to find out about case management and resources. A presentation by Tacha Champod which examined the prosecution and defence hypotheses, demonstrated that there was evidence to suggest that a neutral statement could support the prosecution hypothesis when qualitatively it should have supported the defence hypothesis, even if an individual could not be excluded as a contributor to a mixture. This would support the view for witness statements not to make qualitative statements.

9 Date of the next meeting

9.1 The date of the next meeting was confirmed as Monday 28 November 2016 from 11am to 3pm.

Annex A

Members:

Sue Pope (Chair)	Principal Forensic Services
Lesley Ann Beck	Forensic Service of Northern Ireland
Maggie Boyce	Acting-chair, Body Fluid Forum
Susan Hales	Metropolitan Police
Des Van Hinsbergh	Key Forensic Services
Lorraine Hall-Ramsay	National DNA Database Delivery Unit
Ben Mallinder	Scottish Police Authority
Shirley Marshall	Chartered Society of Forensic Sciences
Roberto Puch-Solis	Royal Statistical Society
Dorothy Ramsbottom	Forensic Science Ireland
Denise Syndercombe-Court	International Society for Forensic Genetics
Jim Thomson	LGC Forensics
Huw Turk	Orchid Cellmark
Andy Ward	UKAS

In attendance:

Emma Burton-Graham	Home Office, Science Secretariat
Gill Tully	Forensic Science Regulator
June Guiness	Forensic Science Regulation Unit
Ian Evett	Principal Forensic Services
Kevin Sullivan	Principal Forensic Services

Apologies:

Mark Bishop	Crown Prosecution Service

Annex B – Minutes of the Y-STR working group on 11 March 2016

Forensic Science Regulator
Overseeing Quality

YSTR Working Group

Minutes of the meeting held on 11 March 2016
at Room 5C, 5 St.Philip's Place, Colmore Row, Birmingham, B3 2PW

1.0 Welcome, Introduction and Apologies

1.1 June Guiness welcomed all to the meeting and invited everyone to introduce themselves. A full list of attendees is at Annex A.

1.2 The aim of the meeting was discussed which was to consider the areas to be covered by the YSTR standard and determine who would draft the various sections of the standard. In general the standard document could be split into technical and interpretative issues.

1.3 The group consider technical issues that would apply to YSTRs which would not be covered by the broader autosomal DNA standard. The potential for YSTR profiling to diagnose health problems was raised; specifically, there is the possibility of diagnosing male infertility when examining YSTR profiles. Whilst the mutations were rare it would be important for this area to be considered including whether the consent form would disclose that this information might be revealed and whether the individuals would be informed about the results of mutations which could result in male infertility if they were identified. A further ethical issue which could arise with YSTR profiling is the possibility of revealing information regarding biological relatedness if male members of the same family were employed within the same organisation and were requested to submit samples for elimination purposes.

2.0 Quality Assurance

2.1 Contamination issues were discussed and it was reported that with the Y23 kit, when microgram levels of female DNA were present in a sample, the resulting profile contained non-specific artifacts. The peaks did not appear allelic and were spread across the profile but one peak fell within an allelic bin. Similar artifacts had been observed with the Yfiler plus kit. It was not clear which primers were binding to the autosomal DNA to create the peaks and the areas of the genome where they were binding.

2.2 The group expressed caution about how the standard dealt with the requirement to include an extraction negative with crime scene samples and concerns were raised that if a crime scene sample had been extracted and tested using autosomal STR processes there may be insufficient extraction negative left to run through the YSTR process. It was recognised that ideally the extraction negative should be run through both the autosomal and the YSTR systems but there would be occasions when this were not feasible. To prepare for these situations, consideration should be given to whether an extraction negative which had been through an autosomal analysis system could be assumed to be free of contamination for the YSTR system. To test this, it would be necessary for individual laboratories to prove that their autosomal system was as sensitive as their YSTR system and the sensitivity of the two systems would need to be validated using a dilution series that took into account a range of factors including the impact of DNA degradation and the impact of mixtures on the sensitivity of the two systems. Mixtures containing excess amounts of female DNA should be included in the validation.

2.3 The issues which might arise in relation to a single extraction batch containing samples which would be sent for both YSTR analysis and autosomal analysis were considered. The wording needed to be reviewed in relation to checking extraction batches of samples for contamination and it would be necessary for the standard to recognised that an extraction batch of samples would consist of samples that would be sent for both autosomal and YSTR analysis. In addition, when checking YSTR profiles for contamination it would only be possible to check those samples within the batch which were submitted for YSTR analysis and there would be a subset of samples which were submitted for autosomal analysis which could not be checked. From a reporting perspective with YSTR profiles, there may be a requirement to state that contamination had been checked for as much as possible but it would not be possible to rule out that the YSTR profile had not come from contamination from another case. It was important for these challenges to be accounted for within the standard.

2.4 Members discussed the importance of determining the threshold for the number of alleles a profile must contain for the profile to be used in criminal cases. If the threshold was set too low, too many adventitious matches would be obtained and if it were set too high, some profiles will be rejected.

2.5 The importance of Sexual Assault Referral Centres working to appropriate standards was raised as having a significant impact on YSTR profiling as a considerable portion of samples to be tested through the YSTR profiling system were taken in sexual assault cases with victims examined in the centres.

2.6 Steven Ferguson agreed to draft the Quality Assurance section of the YSTR standard.

Action 1: Steven Ferguson to draft the Quality Assurance section of the YSTR standard.

3.0 Population Data

3.1 Members agreed that the distribution of YSTR profiles differed between countries and within different regions of the UK and therefore using databases populated with individuals from other countries was not ideal. However, the UK database was small and only holds 800 profiles. To produce accurate statistical estimations of haplotype frequencies, the size of the database needed to be increased. The committee heard that a project being undertaken at King's College, London in collaboration with the Association of Forensic Science Providers (AFSP) attempted to add a further 3000 profiles to the UK YSTR database from individuals from Caucasian, Asian, Afro-Caribbean and Irish Caucasian backgrounds. The AFSP would also coordinate the addition of YSTR profiles to the database from staff working for Forensic Providers. In addition, Leicester University were undertaking a project which would add a further 1000 YSTR profiles to the database in a years time. It would be important to ensure that individuals were not represented on the database twice as this would impact on certain haplotype frequencies and it may be necessary for the consent form to request that individuals do not donate their DNA for the YSTR database more than once. Whilst this increase to around 5000 samples was recognised as an improvement it may still be insufficient and it would be necessary to assess the impact on the weight of evidence if the YSTR database was various sizes.

3.2 The group heard about on-going work at Leicester University to compare statistically UK sub-populations to global populations to determine whether it would be feasible to use global populations to estimate UK sub-population haplotype frequencies. However, there were a number of challenges including obtaining sufficiently large global datasets and discrepancies between how individuals describe themselves and their Y-chromosome lineage.

3.3 The group discussed the methods which could be used to estimate the frequency of the YSTR haplotypes in the population. The methods were the Pseudocount method (n+1/N+1), Brenner's Kappa method which uses the frequencies of singletons within a population sample and Andersen's Discrete Laplace method which takes allelic distribution into account. The Pseudocount method was being used by experts and whilst its advantages include that it has been previously used and is simple, members noted concerns with the Pseudocount method in that the weight of evidence was governed by the size of the dataset and probably underestimates the true weight of the evidence. Concerns were also raised in relation to the use of confidence intervals

as these would be a departure from usual practice and whether the jury would understand the meaning of a confidence interval.

3.4 The following choices needed to be made: which counting method to use, which choice of interval to use (either the binomial confidence interval or the beta distribution) and the percentages for the distribution.

3.5 At previous international YSTR conferences, preferences had moved towards the adoption of the Laplace method, and the committee noted that it would determine international views at the next YSTR conference in May in Berlin.

3.6 Validation of the YHRD software was discussed and LGC noted that they had initiated work with the developers of the software in order to attempt to validate the software. Whilst members were supportive of the Laplace approach to estimate the frequency of YSTR haplotypes in the population there were concerns about using the YHRD database and software, around which the UK providers had very little control. The implementation of the models by the developers of the YHRD software has not been published and therefore had not been externally validated. Concerns were raised that UK providers would not be able to demonstrate sufficient validation of the implementation of the software and would be reliant on programming and data which they were unable to access to validate.

3.7 An alternative option was proposed whereby the UK forensic providers become self sufficient and developed their own statistical tool for evaluating the weight of evidence for Y haplotypes. The software tool would house the UK and Irish haplotype frequency datasets, carry out statistical evaluations for both single source and mixed profiles and be validated centrally. It was agreed that a Laplace model should be developed, with the confidence intervals and FST agreed amongst the providers within the UK. This model could be used with the YHRD database until the UK database had been sufficiently developed. In the meantime, laboratories would continue with the method they currently had in place.

3.8 Developing the statistical tool and building up the UK YSTR database would require external funding and consideration was given to where the funding may be sought. As the tool would be used by all Forensic Providers it wasn’t considered appropriate for a single provider to pay the costs associated with the work. One possibility would be to seek funding from the European Network of Forensic Science Institute (ENFSI) monopoly money as the platform developed could also be adopted by other countries and populated with their YSTR data. In addition, discussions would be held with the Forensic Science Regulator and the Home Office Biometrics Strategy to determine whether there was an appropriate department to commission this work.

Action 2: Jim Thomson and Tim Clayton to draft a paper which outlines the work which needs to be undertaken for the UK to develop their own statistical tool for evaluating the weight of evidence of Y haplotypes.

Action 3: June Guiness to hold discussion with the Forensic Science Regulator, officials within the Home Office and the European Network of Forensic Science Institute in order to seek funding for the UK to develop its own statistical tool for evaluating the weight of evidence of Y haplotypes.

4.0 Legislation

4.1 The committee understood the current legislation to be as follows. The Police and Criminal Evidence Act (PACE), as amended by the Protection of Freedoms Act 2012 (PoFA), required DNA samples, taken from an arrested person or volunteers, to be destroyed as soon as a DNA profile had been derived from it and in any event within six months of the date it was taken. Police forces could apply under the Criminal Procedures and Investigations Act (1996) to have the samples, including PACE samples retained, if they might be needed as evidence in court. The samples retained under CPIA may not be used for purposes other than for the purposes of any proceedings for the offence in connection with which the PACE sample was taken[2]. This rules outs the possibility of further analysis being carried out to derive genetic information for other cases. Under the current legislation, if a PACE sample had been retained under CPIA, it would be possible to use that PACE sample to obtained a YSTR profile, for the case in which the PACE sample was taken. However, if the individual became a person of interest in another case it would not be possible to use the PACE sample to obtain a YSTR profile for the other case. The police would need to obtain a new PACE sample which would have time and resource implications. This does not apply to samples which are held under PACE (and not the CPIA exemption) as these can be used for the investigation of any crimes. The legislation does not state restrictions on the type of technology or profiling apart from it should be non-coding regions. There was disagreement amongst members as to the interpretation of the PoFA legislation with some believing that once an autosomal profile had been obtained no further analysis was possible with that sample, unless it was made CPIA exempt. Others thought that the legislation was not that restrictive and allowed for all appropriate tests to be undertaken within the six month timeframe, prior to the sample being destroyed. Members suggested that the situation required clarification and the police forces required clear direction to ensure a unified approach.

[2] https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/251330/22_Factsheet_-_samples.pdf

Action 4: June Guiness to seek clarification of the interpretation of the PoFA legislation and whether a YSTR profile can be obtained after an autosomal profile has been obtained.

5.0 Interpretation & statistical analysis

5.1 The interpretation of mixtures was considered and it was noted that a mixture interpretation tool existed which was based on a paper published by Wolf *et al* (2005) but was not being used by any of the forensic providers. In some respects interpreting mixed YSTR profiles was considered more straight forward than interpreting autosomal profiles as it would be relatively straight forward to determine if the profile was mixed and there would be no concerns about heterozygous balance. The forensic providers were currently interpreting mixtures using the following methods: once interpretation parameters had been set based on validations, the mixtures were treated as having major or minor profiles and profiles were conditioned on a known profile if it was likely that the conditioned profile contributed to the mixture. Further consideration was given to whether subjective evaluations (but not statistical evaluations) should be undertaken with partial mixed YSTR profiles where the expert believed that an individual was present in the mixture. Caution was expressed about this approach as un-weighted evidence which is admitted into court can falsely give the impression to the jury of having greater evidential weight. It was agreed that mixture interpretation needed to be built into the statistical tool for evaluating the weight of evidence.

Action 5: Jim Thomson and Tim Clayton to include mixture interpretation in their paper which outlines the work which needs to be undertaken for the UK to develop their own statistical tool for evaluating the weight of evidence of Y haplotypes.

6.0 Designation & Nomenclature

6.1 It was agreed that the recommendations on the use of YSTRs in forensic analysis outlined in the International Society of Forensic Genetics paper[3] should be adopted.

7.0 Going forward

7.1 A standard for YSTRs would be developed and published as a regulators guidance document with specific sections annexed to the DNA standard. LGC agreed for sections of their paper to be used when drafting the document.

[3] Gusmao L *et al* (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 Sci Int*; 157(2-3): 187-97.

Annex A

Present:

Tim Clayton	LGC
Steven Ferguson	Forensic Services, Scottish Police Authority
June Guiness	Home Office, Forensic Science Regulator
John Lowe	Key Forensic Services
Andrew McDonald	Cellmark Forensics
Dave Mallett	Cellmark Forensics
Charlotte Murphy	Forensic Science Ireland
Sue Pope	Principal Forensic Services
Roberto Puch-Solis	RSS
Jim Thomson	LGC
Jon Wetton	Leicester University
Emma Burton-Graham, Secretariat	Home Office Science Secretariat