

Investigation into X-STR haplotype frequencies for forensic human
identification in South Africa



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Abbreviations

| | |
|-------------|--|
| AMOVA | Analysis of molecular variance |
| AS-STRs | Autosomal short tandem repeats |
| bp | base pair |
| CA | California |
| CE | Capillary electrophoresis |
| ChrX | X chromosome |
| DNA | Deoxyribonucleic acid |
| EMPOP | European DNA Profiling Group (EDNAP) mtDNA Population Database |
| ENSI | European Network of Forensic Science Institute |
| Fig. | Figure |
| F_{ST} | Pairwise genetic distance |
| FTA | Flinders Technology Associates |
| GD | Gene diversity |
| GHEP-ISFG | Spanish and Portuguese Speaking Working Group of the International Society for Forensic Genetics |
| HD | Haplotypic diversity |
| HREC | Human Research Ethics Committee |
| HWE | Hardy-Weinberg equilibrium |
| <i>i.e.</i> | Latin – id est, meaning “namely” |
| IBD | Identity-by-descent |
| ISO | International Organisation of Standardisation |
| LD | Linkage disequilibrium |
| LG | Linkage group |
| MEC | Mean exclusion chance |
| MPS | Massively parallel sequencing |
| mtDNA | Mitochondrial DNA |
| NFDD | National Forensic DNA Database of South Africa |
| NTC | Non-template control |
| PAR | Pseudo-autosomal regions |
| PCR | Polymerase chain reaction |

| | |
|----------|--|
| PD | Power of discrimination |
| PIC | Polymorphism information content |
| PRISMA | Preferred reporting items for systematic reviews and meta-analyses |
| QS | Quality sensor |
| r^2 | Squared coefficient of correlation |
| RFU | Relative fluorescent units |
| RMP | Random match probability |
| SAPS | South African Police Service |
| SD | Standard deviation |
| STR | Short tandem repeat |
| SWGDM | Scientific Working Group on DNA Analysis Methods |
| UCT | University of Cape Town |
| URL | Uniform resource locators |
| USA | United States of America |
| WI | Wisconsin |
| X-INDELS | X-linked insertion and deletion polymorphisms |
| X-STRs | X-chromosome short tandem repeats |
| YHRD | Y-STR Haplotype Reference Database |
| Y-SNPs | Y chromosome single nucleotide polymorphisms |
| Y-STRs | Y-chromosome short tandem repeats |
| μ l | Microlitre |

Abstract

The utilisation of X-chromosome short tandem repeats (X-STRs) for DNA profiling has been demonstrated to be particularly useful in resolving distant familial relations and deficiency paternity testing. The implementation of X-STRs within a medico-legal context requires baseline frequency data for the general population to allow for appropriate statistical interpretations of results. This study aimed to generate the first X-STR data for the South African population and internally validate the Qiagen Investigator Argus X-12 QS kit. Biological samples from 781 South African individuals (517 males and 264 females) with either African, mixed, European, or Indian/Asian ancestry were processed. Statistical analyses were performed using StatsX and Arlequin. Herein, allele and haplotype frequencies and forensic parameters for the South African population are reported, as well as data related to the reproducibility, sensitivity, limit of detection, and concordance of the Investigator Argus X-12 QS kit. DXS10135 was the most informative locus, while DXS7423 was the least informative locus. The combined power of discrimination for both males and females was greater than 0.999999999. The haplotype diversity of all four linkage groups exceeded 0.993. Linkage group 1 was the most informative, with 421 unique haplotypes. Possible linkage disequilibrium was detected in five loci pairs in male samples and three loci pairs in female samples. However, it is expected that the effects of false linkage disequilibrium were present, and only loci pairs within the same linkage group may be in true linkage disequilibrium. All loci in female samples were in Hardy-Weinberg equilibrium, except DXS10148. Additionally, a total of 59 off-ladder alleles were identified. The discriminatory power of these results suggests X-STRs may be beneficial for forensic casework in South Africa. The availability of this data could allow this method to be used locally to assist with civil inheritance disputes and the identification of unknown individuals.

Keywords: X-STRs; haplotype frequencies; Investigator Argus X-12 QS; South Africa

Chapter 1: Systematic Literature Review

1.1. Introduction

The development and implementation of DNA profiling within the field of forensics has contributed greatly to the successful closure of many medico-legal investigations. This is because DNA profiling has enabled authorities to use DNA evidence to identify unknown missing persons and/or skeletal remains, identify victims and possible suspects of a crime, as well as provide exoneration to those who have previously been wrongfully convicted [1]. DNA profiling has also been utilised in civil disputes of paternity and kinship testing [2].

DNA profiles are typically produced using DNA of an unknown origin, and compared to a known reference profile [3] obtained from an individual's family or the national forensic DNA database. For this, autosomal short tandem repeats (STRs) are routinely targeted using polymerase chain reaction (PCR) and capillary electrophoresis-based methods. However, a match alone does not indicate positive identification. To provide statistical confidence to the match, the random match probability (RMP) must be calculated. This is done using allele frequencies previously generated for a representative subset of the general population [4].

Limitations regarding the discriminatory power and informativeness of autosomal STRs in resolving distant familial relations or cases involving complex pedigrees have been widely reported [2,5,6]. When using autosomal STR analysis, there is always a possibility that a true biological relationship will be excluded due to an insufficient number of shared alleles or certain discrepancies between individuals. These discrepancies may be attributed to several factors, including rare mutations or silenced alleles [7]. This highlights the need to utilise linkage markers, such as those on the sex chromosomes, to improve the efficiency of the identification and familial testing using DNA profiling [8].

The use of Y-chromosome STRs (Y-STRs) are particularly beneficial in sexual offence investigations, as they can be used to discern a male's identity, even in the presence of female DNA [9,10]. Additionally, Y-STRs can be utilised to resolve paternity disputes involving an alleged father and son. Similarly, X-chromosome STRs (X-STRs) can be implemented in complex kinship testing and deficiency paternity testing of female offspring [2], which refers specifically to situations where a DNA sample from the alleged father is unattainable.

This can be attributed to the differences observed in the recombination patterns of sex chromosomes when compared to that of autosomal chromosomes. As autosomal chromosomes occur in pairs, it is true that each chromosome will recombine with its homologous counterpart prior to being passed on to offspring. However, sex chromosomes differ between male (XY) and female (XX) individuals [9]. As such, only maternally inherited X-chromosomes undergo a recombination event prior to inheritance. Contrarily, the paternal sex chromosomes are transmitted to offspring almost entirely unaltered. This is because the X and Y chromosomes are mostly meiotically incompatible apart from small shared pseudo-autosomal regions (PAR1 and PAR2), wherein the occurrence of recombination between these two chromosomes is limited to [11]. Thus, most of the informative value of the X-chromosome lies within paternal sex chromosomes.

Additionally, the X-chromosome harbours unique inheritance patterns which can aid forensic applications. For example, STR markers on the X-chromosome are situated close to each other (*i.e.*, linked) and are therefore inherited as a haplotype. As males only inherit a maternal X-chromosome, their X-chromosome haplotype is easily detected. This X-chromosome haplotype is also passed on directly from father to female offspring, meaning it will be common amongst sisters who share a father [6,12]. It is also true that siblings who have the same mother will have a portion of their maternal X-chromosome haplotype in common [12]. Lastly, although STRs have a higher mutation rate than single nucleotide polymorphisms (SNPs), the X-chromosome has remained relatively stable overtime, making it possible to trace the lineage of X-STR haplotypes across previous familial generations [2,13].

Thus, the use of chromosomes that undergo separate meiotic processes provides a new perspective, which can be useful for certain cases that would otherwise remain inconclusive due to the insufficient discriminatory power possessed by autosomal STRs. However, the integration of X-STRs into forensic casework remains relatively new and as data from the general population is necessary for interpretation, the usefulness of X-STR profiling is dependent on the availability of appropriate haplotype frequency databases for local population groups. This type of data is yet to be collected in many regions of the world, which has led to an inconsistency in the success of identification between different populations. This has prompted researchers across the globe to produce X-STR population data for their relevant countries and population groups [9].

This literature review aims to provide an overview of the advancements of X-STRs in forensic and medico-legal casework. To address this aim appropriately, four objectives have been identified, namely (i) to briefly describe the applications of X-STRs in a medico-legal setting; (ii) to assess the evolution of commercial X-STR multiplex PCR kits; (iii) to evaluate the use of X-STR repositories/databases and (iv) to provide a global exhaustive summary of available X-STR population data, with a specific focus on data generated using the most popular commercial kit, the Investigator® Argus X-12 (QS) kit (Qiagen, Hilden).

1.2. Methods

1.2.1. Literature search strategy

This systematic literature review was performed using the “Preferred Reporting Items for Systematic reviews and Meta-Analyses” (PRISMA) guidelines. A literature search was conducted on major databases, namely Web of Science, Scopus and PubMed. Keywords were chosen to create a search string, separated by Boolean operators, to use across the aforementioned databases (Table 1.1). These keywords were systematically curated to ensure the search returned publications that encompassed the entirety of this literature review’s aim as well as the respective objectives.

Table 1.1 Keywords utilised in the systematic literature review.

| Keywords | |
|----------------------------|------------------------------|
| X-STR population data | X-STR database |
| Investigator Argus X-12 QS | Deficiency paternity testing |
| Investigator Argus X-12 | X-STR typing |
| X-STR | X-chromosomal STRs |

The search was limited to publications released between the years 2000 and 2022, as the introduction of X-STRs became more apparent approximately 20 years ago. An additional search of the available literature on the ChrX-STR.org 2.0 database was conducted.

Publications that were duplicated across databases or published under alternative titles by the same author(s) were removed from the results. The remaining publications were considered against all inclusion and exclusion criteria (Table 1.2) using the title and abstract, followed by the full text, to ascertain their relevance to the scope of this literature review. Hand-searching was performed on the reference lists of all included publications to ensure saturation of relevant literature.

Table 1.2 Search strategy inclusion and exclusion criteria

| Inclusion criteria | Exclusion criteria |
|---|--|
| Available in English | Non-human populations |
| X-STR multiplexes of four or more loci | Review articles |
| Investigation of X-STRs within a forensic genetic context | Investigation of X-STRs within a clinical context |
| X-STR population data produced using conventional DNA profiling | X-STR population data produced using massively parallel sequencing |

1.3. Results

The search produced a total of 1142 publications across all three databases. Additionally, 47 articles were identified on the ChrX-STR.org 2.0 database and a further 32 articles were identified through hand-searching. After the application of the eligibility criteria, a total of 293 unique publications were included (Fig. 1.1.).

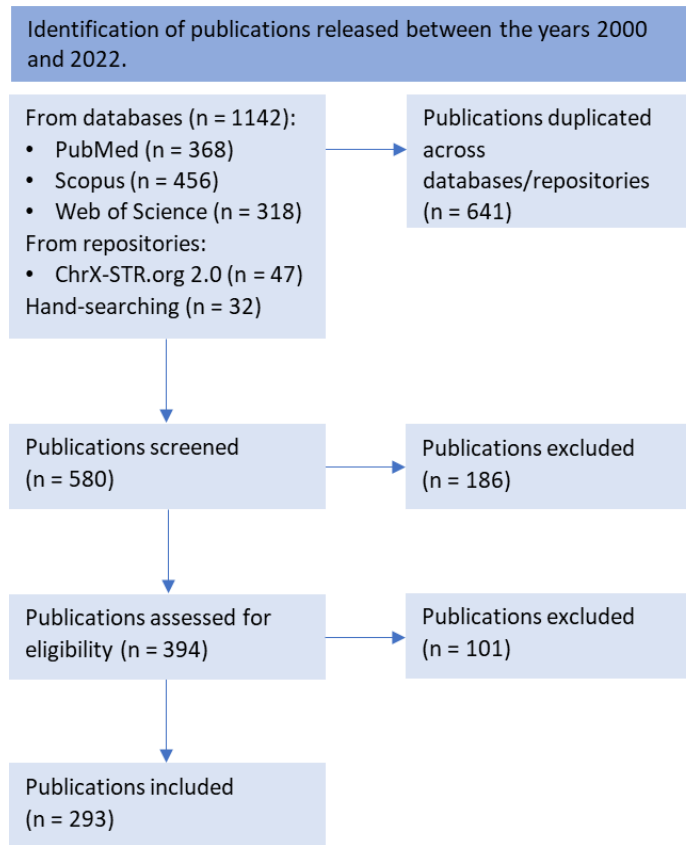


Fig. 1.1. Adapted PRISMA flow diagram representing the search strategy used and the implementation of the inclusion and exclusion criteria.

The results consisted of articles that were published across 35 different journals ([Appendix A](#)). The most prevalent journals are summarised in Fig. 1.2., with the leading journals in X-STR research being the International Journal of Legal Medicine ($n = 67$), Forensic Science International: Genetics Supplement Series ($n = 56$) and Forensic Science International: Genetics ($n = 56$).

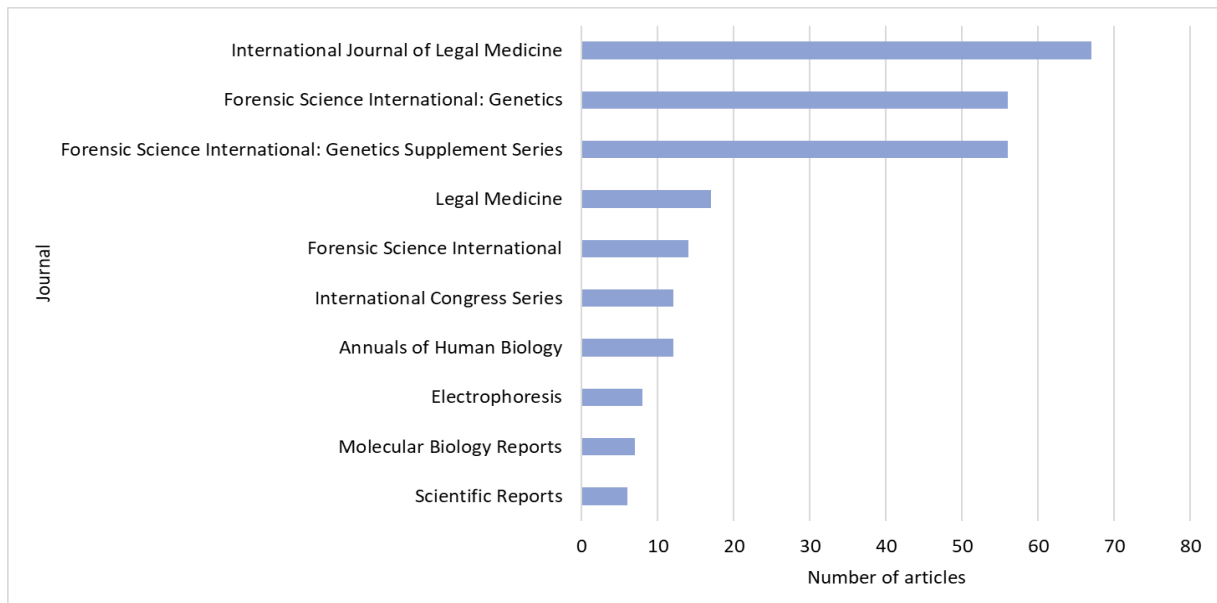


Fig. 1.2. The most prevalent journals identified during the search and their corresponding published articles.

The number of publications related to X-STR research per year fluctuated over time, with the year 2011 being the most popular. Since then, a slight decline in publications has been observed. However, the relevance of X-STRs remains, as seen by the surges in publications in recent years (*i.e.*, 2017, 2019, 2021) (Fig. 1.3.).

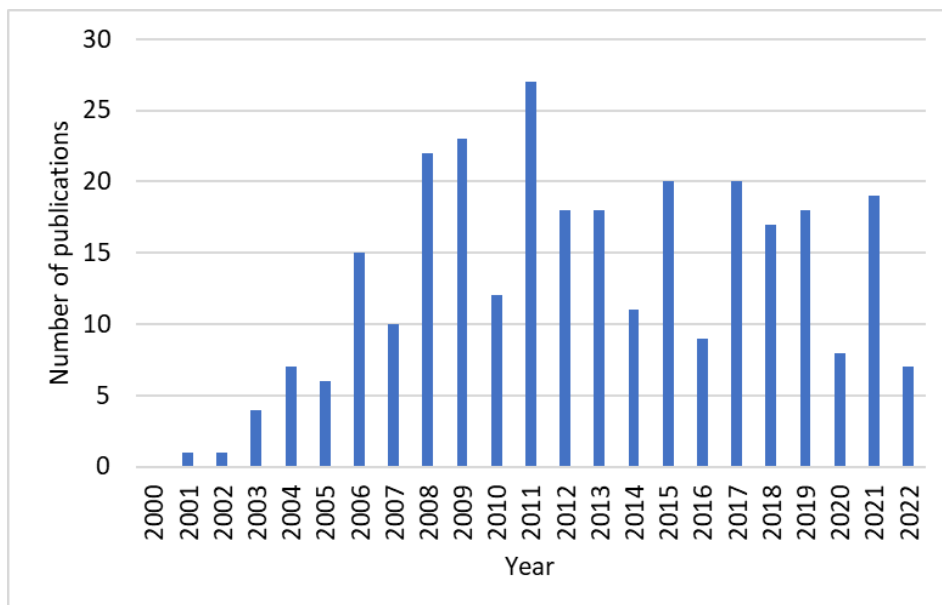


Fig. 1.3. The number of publications released during a 22-year period (2000 – 2022).

1.3.1. Applications of X-STRs

A total of 36 included articles focused on the use/applications of X-STRs within a medico-legal context ([Appendix A](#)). From these articles, three broad applications of X-STRs were identified. The first and most common is complex kinship testing, which includes deficiency paternity testing ($n = 29$, 80.56 %) [4,7,8,12,14–38], followed by human identification ($n = 5$, 13.89 %) [39–43] and lastly mixture interpretation ($n = 2$, 5.56 %) [44,45].

1.3.2. X-STR multiplex systems/kits

A variety of X-STR multiplex systems have been used for the various applications of X-STRs as well as the generation of X-STR population data. Many researchers have opted to develop laboratory-specific multiplex X-STR marker systems as a means of producing X-STR population data. Alternatively, companies have manufactured commercially available X-STR kits [46,47]. Table 1.3 provides a comprehensive summary of the most prevalent X-STR systems used throughout the literature. This summary shows that a multitude of advancements have been made to X-STR kits over time with a particular focus on the number of X-STR markers included and additional quality features. The most frequently used kit globally was the Investigator Argus X-12 (QS) (Qiagen, Hilden), followed by the GHEP-ISFG X-STR decaplex developed by the Spanish and Portuguese Speaking Working Group of ISFG [48] and the Mentype Argus X-8 (Biotype, Dresden).

Table 1.3 Evolution of X-STR multiplex kits/systems used in the reviewed literature.

| Kit Name | Number of X-STR loci | Additional features | Occurrence in population data |
|--|-----------------------------|---|--------------------------------------|
| Investigator Argus X-12 (QS) (Qiagen, Hilden) | 12 | Amelogenin marker, Quality Sensor, D21S11 | 66 |
| GHEP-ISFG X-STR decaplex | 10 | N/A | 26 |
| Mentype Argus X-8 (Biotype, Dresden) | 8 | Amelogenin marker | 26 |
| AGCU X19 (AGCU ScienTech, Jiangsu) | 19 | N/A | 23 |
| Goldeneye 17X (Peoplespot Incorporation, Beijing) | 16 | Amelogenin marker | 10 |

| Kit Name | Number of X-STR loci | Additional features | Occurrence in population data |
|---|----------------------|---------------------|-------------------------------|
| Mentype Argus X-UL (Biotype, Dresden) | 4 | Amelogenin marker | 7 |
| In-house [49]* | 17 | N/A | 5 |
| In-house (mini X-STRs) [50]* | 15 | N/A | 3 |
| ZJGA-X12 STR/AGCU X12 (AGCU ScienTech, Jiangsu) | 12 | N/A | 2 |
| Microreader 19X Direct ID System | 19 | Amelogenin marker | 2 |
| TYPER X19 | 18 | Amelogenin marker | 1 |

*Reference provided for in-house kits for better understanding.

1.3.3. X-STR databases/websites

The search returned three articles which described the creation of online X-STR databases (Table 1.4). This included two active databases which focus on the storage of X-STR population data and related calculations, namely the ChrX database and FamLinkX. Additionally, the Brazilian Genetic Database of X Chromosome was identified but the URL was unavailable.

Table 1.4 A list of available X-STR databases or websites from the reviewed literature.

| Database/Website | URL (date accessed) | Reference |
|--|--|-----------|
| ChrX database | https://www.chrx-str.org/xdb/ (Date Accessed: 27/12/2022) | [51] |
| Brazilian Genetic Database of X Chromosome | http://www.bgbx.com.br/ (Unavailable) | [52] |
| FamLinkX | https://www.famlink.se/fx_index.html (Date Accessed: 27/12/2022) | [53] |

1.3.4. Global X-STR population data

A multitude of publications pertaining to X-STR population data were identified ([Appendix B](#)). A comprehensive summary of the number of X-STR population studies that have been conducted on a global scale using any available X-STR multiplex system (Fig. 1.4.), revealed substantial variation between the X-STR population data available for each continent. Most

X-STR data was available for populations within Asia and Europe, while X-STR population data were limited for Africa and the Americas.

This variation is also present when focusing on X-STR population data that has been produced per country, with X-STR population data only available for a total of 72 countries worldwide, and most of these countries were only represented by the availability of limited X-STR data. A comparison of the number of population studies showed that China was the leading country in X-STR population data generation and thereafter, only five other countries, namely Japan, Germany, Italy, Spain and Brazil, have produced 10 or more publications related to X-STR population data (Fig. 1.4.).

The combined sample size of each country showed that within China, at least 47 different subpopulations and approximately 36 500 individuals, excluding families, were investigated. Other large countries, namely India ($n = 1132$), the United States of America ($n = 2525$), Brazil ($n = 6221$), Nigeria ($n = 218$) and Russia ($n = 803$), included much smaller sample sizes ([Appendix B](#)). In smaller countries such as Poland, Portugal and Croatia, the combined sample size for each country was greater than 1000 individuals. However, for many countries, only a single population study was available and often the sample sizes used were below 300 individuals ([Appendix B](#)). Some of these countries included East Timor ($n = 149$), Tunisia ($n = 139$), Bangladesh ($n = 209$), Saudi Arabia ($n = 200$), Greenland ($n = 198$) and Denmark ($n = 210$).

The X-STR population data generated with the most widely utilised commercial kit, the Investigator Argus X-12 (QS) kit (Qiagen, Hilden), is presented in Fig. 1.5. This figure illustrates that this kit has been utilised the most by researchers in China, which is consistent with China having produced the most X-STR population data publications. Following China, the use of this kit has also been frequently reported in smaller countries, namely Croatia and Portugal.

From Fig. 1.5., it is also clear that there is a paucity of the use of this kit in other largely populated countries such as India, the Russian Federation and the United States of America, with each of these countries only having one or two X-STR population data publications using this kit. Additionally, the use of this kit remains particularly scarce across countries on the African continent.

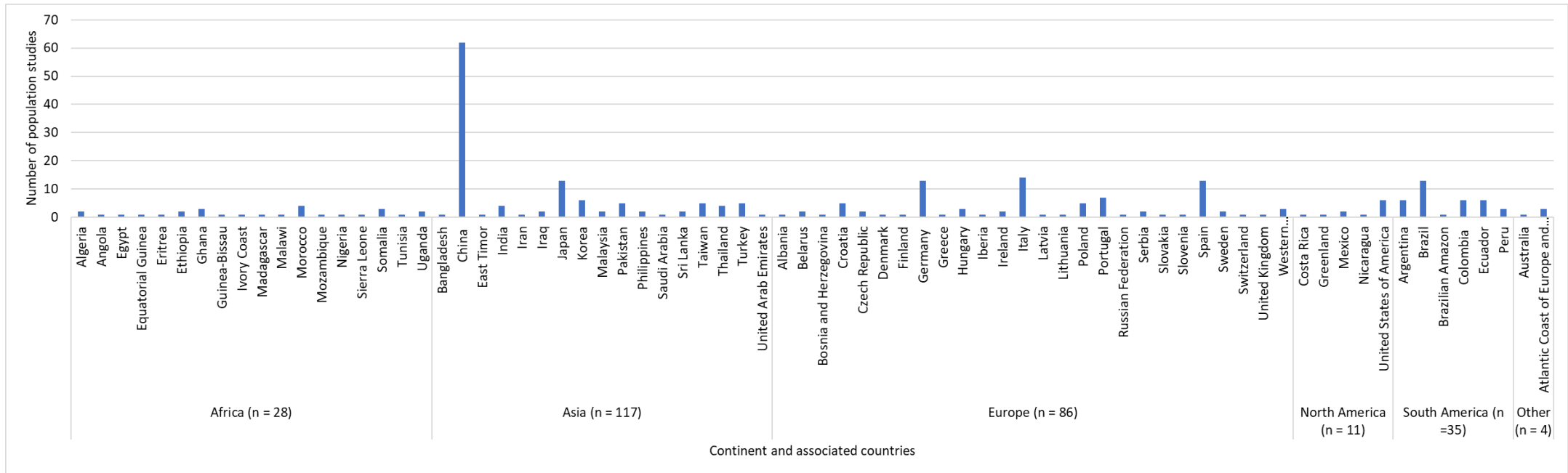


Fig. 1.4. A graphical illustration of the globally available X-STR population data based on the reviewed literature.

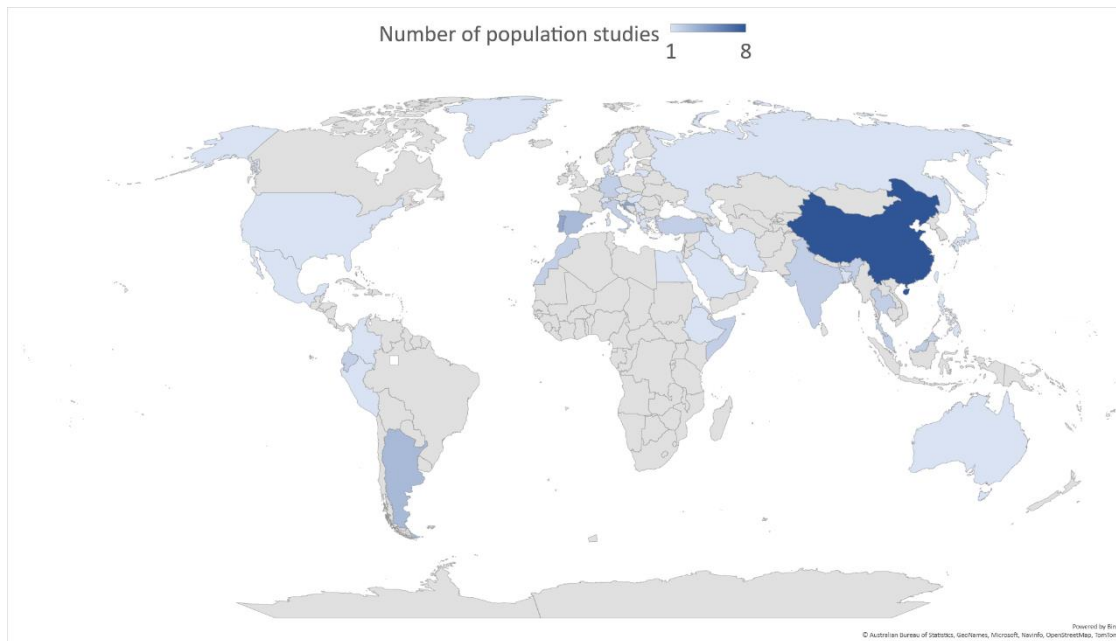


Fig. 1.5. A map illustrating the global use of the Investigator Argus X-12 (QS) kit (Qiagen, Hilden) across the reviewed literature.

1.4. Discussion

1.4.1. Applications of X-STRs

The incorporation of X-STRs within forensic laboratories, unlike autosomal STRs and Y-STRs, is not as well-established. This is because investigations into situations where X-STRs provide greater discriminatory value compared to other more extensively studied markers are more recent [2,6]. As explained in the introduction, the informativeness of autosomal STRs comes into question when there is a need to differentiate between related individuals or establish biological relationships while using an incomplete pedigree.

The most common application of X-STRs was identified as complex kinship testing and deficiency paternity testing. An example of complex kinship testing in which X-STRs would be useful is the analysis of possible incest cases involving a father, daughter and female offspring, as both the mother and female offspring would share the same paternal X-chromosome haplotype through identity-by-descent (IBD) [6].

For deficiency paternity testing, X-STR profiles obtained from paternal relatives or other female paternal offspring can be analysed in the alleged father's absence [27]. This may be necessary to resolve civil disputes, especially those regarding inheritance. A case example of

deficiency paternity testing was demonstrated by Serra *et al.*, (2008), whereby X-STRs were used to assist in a case involving a mother, a female child and alleged paternal grandparents. Initially, autosomal STRs were implemented, but the paternity results were inconclusive due to the presence of multiple mismatches between the DNA profiles. Thereafter, X-STR profiles were produced for the child and the alleged paternal grandmother. A comparison of these profiles, together with the data produced using autosomal STRs, provided sufficient evidence to exclude the alleged biological father from paternity [34]. This example especially illustrated that X-STRs can accompany routinely used autosomal-STRs to support findings and strengthen conclusions.

This same principle has been translated to forensic casework concerning the identification of missing individuals or human remains. Barbaro *et al.*, (2006) demonstrated the use of X-STRs to link a strand of hair found in a suspect's house, to a never-found missing female. An X-STR profile was generated for the hair, as well as the missing female's biological mother and sister. The sister's profile represented the paternal X-chromosome haplotype of the biological father in his absence. From this, a successful match was concluded [40].

Furthermore, the scope of X-STRs could be expanded into mortuary settings. Due to the low mutation rates of X-STRs, the use of X-STR profiling could also be utilised in an attempt to alleviate the burden of unidentified bodies. Many countries across the world face the harsh reality of an accumulation of unidentified bodies at their mortuaries year after year. Although an ongoing problem, this topic remains poorly documented, with minimal publications available [54–57]. It is possible that in some of these cases, the immediate family is unavailable for familial matching using autosomal STRs. If X-STRs were to be implemented as part of routine identification practice, alongside autosomal STRs, the analysis could be broadened to include distant familial members.

Although infrequently utilised, another application identified in the results was the use of X-STRs to support autosomal STRs and/or Y-STRs in mixture interpretation and criminal investigations, particularly those involving related suspects. An interesting case example by Lancia *et al.*, (2011) demonstrated the use of X-STRs in a rape and murder investigation involving a female victim and multiple male suspects, namely her husband and his brother. A mixed DNA sample was tested and X-STRs were able to differentiate between the victim's and

the suspect's DNA profiles, as well as used to discern the identity of the perpetrator by discriminating between the husband and his brother [41].

Therefore, the results show that the implementation of X-STRs may be useful in a variety of applications in both civil and forensic instances. However, the gap in which the use of X-STR markers fill lies particularly in deficiency paternity testing and complex kinship analyses involving at least one female (*i.e.*, mother/son, mother/daughter, father/daughter) [6].

1.4.2. Evolution of X-STR multiplex systems

There is a multitude of X-STRs spanning the X-chromosome. Over time, these STRs and the linkage properties thereof have been well-studied by many researchers [58–60]. STR loci that are inherited together as clusters are known as linkage groups. Four main linkage groups have been described at particular locations on the X-chromosome, namely Xq28, Xq26, Xq12 and Xp22 [2,61]. In addition to this, and similarly to the Y-chromosome, the X-chromosome houses the Amelogenin marker within its intronic region. This marker is traditionally used for biological sex determination, due to its 6 bp deletion on the X-chromosome, which allows for efficient differentiation between the sex chromosomes [62,63].

Many researchers have developed laboratory-specific multiplex X-STR systems as a means of simultaneously amplifying multiple X-STR loci for the generation of population data. For example, Sun *et al.* (2013) described the use of an internally developed 16 X-STR loci system (DXS9902, DXS6803, DXS10134, DXS6789, HPRTB, DXS6810, GATA31E08, DXS6795, DXS8378, GATA165B12, DXS6800, DXS10159, DXS7132, GATA172D05, DXS7424, and DXS6807) to perform X-STR typing on a Chinese Shanghai Han population [64]. However, the use of the PCR multiplex was limited to this publication.

The Spanish and Portuguese-speaking Working Group of the International Society for Forensic Genetics (GHEP-ISFG) have developed an X-STR decaplex, which includes 10 X-STR loci, namely GATA172D05, DXS6809, DXS9902, DXS7133, DXS8378, DXS6789, DXS9898, DXS7132, GATA31E08 and DXS7423 [48]. Although not commercially manufactured, this multiplex system has been widely used by many researchers across the globe and had the second highest prevalence in X-STR population studies (Table 1.3).

However, the increase in popularity in X-STR population research and the emergence of X-STRs in forensic and civil casework has prompted the need for the development of appropriate X-STR kits. X-STR loci located within the linkage groups, along with the Amelogenin marker have formed the basis for most of these commercially developed kits.

The Mentype® Argus X-UL kit (Biotype, Dresden) was the first commercially developed kit and amplified four X-STR loci (HPRTB, DXS7132, DXS7423 and DXS8378), along with the Amelogenin marker [46]. However, due to the limited number of loci included, this kit would have provided minimal discriminatory power within a forensic context. This prompted the development of the Mentype® Argus X-8 kit (Biotype, Dresden), which targeted a further four X-STR loci for amplification (DXS10135, DXS10101, DXS10134 and DXS10074) [65,66]. Thereafter, the Investigator® Argus X-12 kit (Qiagen, Hilden) was developed, which amplifies the Amelogenin marker and a total of twelve X-STR loci, consisting of the eight aforementioned loci, as well as four additional loci (DXS10148, DXS10079, DXS10103, DXS10146) [9]. Since then, advancements to this kit have been made and developmentally validated to create the current commercially available X-STR kit, known as the Investigator Argus X-12 QS kit (Qiagen, Hilden). A quality sensor (QS) was added to allow researchers to monitor PCR success, as well as an autosomal STR marker (D21S11) to facilitate an evaluation of concordance between X-STR and autosomal STR typing kits [47,67]. Furthermore, the Investigator Argus X-12 QS kit (Qiagen, Hilden) is ISO18385 certified, meaning that the appropriate measures were taken to reduce the risk of contaminating DNA during the manufacturing process.

The Investigator Argus X-12 (QS) kit (Qiagen, Hilden) was the most commonly used kit globally (Table 1.3, Fig. 1.5.), suggesting that this kit is widely accessible to many countries and may account for this kit becoming the most widely utilised for X-STR typing. However, it is also important to consider that the frequent use of this kit may be due to the limited number of manufacturers of commercial X-STR kits and none of these current competitors are as globally established within the field of forensics as Qiagen is.

Alternative X-STR multiplex kits that have been frequently described in the literature include the AGCU X19 X-STR amplification kit (AGCU ScienTech Inc., Wuxi, Jiangsu), which refers to a multiplex PCR system consisting of 19 X-STR loci (DXS10162, DXS6809, DXS10174, DXS10159, DXS10079, DXS8378, DXS101, DXS7132, DXS10103, DXS7424, DXS10135, DXS10148,

DXS10101, DXS10075, DXS10164, DXS6789, DXS7423, DXS10134, and HPRTB) [68] and the Goldeneye™ DNA identification system 17X kit (Peoplespot Inc., R&D, Beijing), which utilises the Amelogenin marker and 16 X-STR loci (DXS8378, DXS7424, DXS6789, GATA31E08, DXS6807, DXS10159, DXS9902, DXS7132, DXS6800, DXS6795, GATA165B12, DXS10134, HPRTB, DXS6810, DXS6803 and GATA172D05) [69]. However, the implementation of these two kits has been greatly limited to Chinese populations only ([Appendix B](#)). A reason for this may be that these kits are manufactured in China, thus providing ease of access to researchers in China, while remaining unavailable on a global scale.

1.4.3. X-STR databases/websites

A few researchers have attempted to develop a global X-STR database, which would allow for the storage of X-STR population data generated for different populations around the world (Table 1.4). However, these attempts have not yet been effective.

The Forensic ChrX Research database was created to serve as a repository for researchers to submit their published X-STR haplotype frequency data, provided the necessary quality requirements are met [51,70]. In theory, this would allow researchers to readily access this type of data for any population across the globe. Unfortunately, this database is currently underutilised and only provides frequency data for a few populations, namely Chinese Han, Ghanesen, German, and Japanese. This may be due to many articles not meeting the requirements stipulated on the database to qualify for publication to the database [70]. Albeit underutilised, the Forensic ChrX Research database provides researchers with information regarding the X-chromosome, as well as a mathematical model to calculate various forensic parameters related to X-STR population data. The latter aspect of the database has shown to be widely efficient and is often described by researchers as part of their data analysis in various publications [71–73].

Another database that stores X-STR population data and provides computational X-STR marker software for download is known as FamLinkX [53]. While FamLinkX provides a larger compilation of global X-STR population data, the last modification to the list of available population data occurred in 2019. This database is newer and lacks the quality standards

implemented by the Forensic ChrX Research database, possibly making it less well-known. Together, these reasons may explain its scarcity in the literature.

Lastly, a third database exists, known as the Brazilian Genetic Database of Chromosome X, which acted as a repository for Brazilian X-STR population data [52]. The authors of this database intended on expanding it to include various calculations related to X-STR markers which could be utilised by other researchers, as well as X-linked insertions and deletion polymorphisms (X-INDELS). However, it currently remains unavailable for use.

The underutilisation of these X-STR databases may also be due to the lack of consent from participants for their data to be stored on an openly accessible platform. Unlike other well-established databases, such as the Y-STR Haplotype Reference Database (YHRD) [74,75] and European DNA Profiling Group (EDNAP) mtDNA Population Database (EMPOP) [76], these X-STR databases do not have a privacy policy in place regarding the storage of the X-STR data. Furthermore, the data available on the YHRD and EMPOP databases are collated for each country and access to this population reference data must be requested, while population data available on the X-STR databases represent individual publications, which are readily downloadable. Although this is useful for population studies and comparisons, it could bring about ethical concerns around privacy. Additionally, the frequent contributions to the YHRD and EMPOP databases by researchers may be due to publication requirements. For example, the Forensic Science International: Genetics journal stipulates that all mtDNA and Y-STRs/Y-SNPs data must be submitted to the relevant database for quality assessment prior to submission for publication [77], while submissions of X-STR data do not hold this same requirement.

Therefore, the establishment of a single designated X-STR reference database with appropriate quality standards and management may increase the utilisation of such a database by researchers.

1.4.4. Global X-STR population data

There were a multitude of articles pertaining to X-STR population data (Fig. 1.4., [Appendix B](#)). As previously mentioned, the majority of the X-STR population data was within Asian and European populations, while African and American populations remained under-represented.

This may be because most STR multiplex kits are manufactured within these regions and are more readily accessible. However, this is highly disproportionate considering Africa has the second-largest continental population, after Asia. Furthermore, the African continent holds some of the world's most diverse populations [78], thus preventing the translation of X-STR datasets from one African population to another. This motivates for the generation of localised indigenous knowledge.

Fig. 1.4 also showed that as an individual country, China has the most extensive X-STR dataset. This is due to the availability of a large amount of X-STR data, which includes a large amount of separate Chinese populations from different regions across the country as well as a considerable sample size. However, evaluation of the sample size used and the number of different subpopulations/regions investigated for each country can provide a deeper understanding of how well a country is represented by the X-STR population data produced. An overview of the combined sample size utilised in each X-STR population study compared to the general population size of each country revealed that Croatia was the most well-represented country, as X-STR data is available for all major regions of the country and the density of Croatia's population is considerably smaller than China's. In contrast to this, the majority of the countries listed in Fig. 1.4 only have a limited number of X-STR population data publications available and these publications often lack the use of a sample size that appropriately mimics the relevant population. For example, publications for populations within East Timor [79], Tunisia [80], Bangladesh [81], Saudi Arabia [82], Greenland and Denmark [83] include less than 300 samples. In 2013, the Forensic Science International: Genetics journal released guidelines for the publication of X-STR population data, whereby a minimum of 500 individuals need to be included when performing X-STR typing [77]. Many publications fall short of adhering to these guidelines ([Appendix B](#)) and although the aforementioned studies were not published in this journal, researchers should consider this guideline when investigating X-STRs within different populations to ensure that the data they produce are informative.

When focusing on X-STR population data generated using the Investigator Argus X-12 (QS) kit as seen in Fig. 1.5., it showed that although this kit is the most popularly utilised globally, it has only been utilised for a maximum of eight publications within any particular country. Once again, China has produced the highest number of population studies, while other densely

populated countries such as India [84,85], the Russian Federation [86], the United States of America [87], have limited X-STR population data available. Therefore, although X-STR data is available for some populations, it is clear that unless these populations are homogenous throughout all regions, they will remain under-represented until additional X-STR data is collected. This further highlights the need for more extensive X-STR research within each population/region of a country. Fig. 1.5 also illustrates that Africa continues to have the least amount of available X-STR population data produced using the Investigator Argus X-12 QS kit (Qiagen, Hilden), as many countries, including South Africa, are not represented at all. This is unfortunate as African countries are considered to hold some of the most diverse populations globally [78], thus warranting exploration. However, the absence of this data may be due to the limited availability of resources for this type of research within Africa.

1.5. Conclusion

Sex-linked STRs (Y-STRs and X-STRs) have been shown to be critical in scenarios where autosomal STRs are not informative enough. This literature review focussed specifically on the use of X-STRs and the applications thereof. Published literature regarding X-STRs continuously highlights the importance of X-STRs in deficiency paternity testing and other kinship scenarios as a solution to dealing with incomplete and/or complex pedigrees. Although this supports the inclusion of X-STRs, it is clear that the scope thereof is limited to a certain niche. This may account for the lack of X-STR popularity in many countries.

There is a movement towards the incorporation of X-STRs as a supplementary analysis to support autosomal STRs. However, the literature suggests that there is a lack of uniformity within the kits that are used in forensic casework, as well as for population data generation. The Investigator® Argus X-12 (QS) kit (Qiagen, Hilden) is currently the most common commercially available multiplex PCR kit. Its wide availability and quality standards are currently unmatched. However, this is not to say that improvements are not necessary. In time, manufacturers may draw on literature to include a larger multiplex of X-STR markers to further increase the informativeness of the kit.

Furthermore, many researchers have undertaken the task of generating X-STR data for different populations. However, many countries remain underrepresented, limiting the use

of X-STRs in a forensic context, as the necessary statistical analyses cannot be adequately performed. X-STRs repositories are underutilised, meaning there is a lack of proper sorting and storing of population data for it to be made accessible to the rest of the world. Therefore, while the advancements of X-STRs in a medico-legal context over the years are appreciated, it is integral to acknowledge the gap in the field and the work that still lies ahead to ensure the inclusion of X-STRs during casework and research.

Chapter 2: Manuscript

2.1. Introduction

Human identification is primarily achieved through the use of DNA profiling [1]. During medico-legal investigations, DNA profiles are generated using autosomal short tandem repeats (AS-STRs) [8] and compared to a reference DNA profile of known origin. Random match probabilities (RMP) must be determined for each match, which requires allele frequency data from the general population [3]. However, limitations in the use of AS-STRs for resolving distant familial relationships have previously been identified [4,35]. This has led to research into the use of STR markers located on the sex chromosomes, which can be used to complement the results obtained from AS-STRs [8].

X-STRs have shown to be particularly useful in deficiency paternity testing involving daughters as well as complex kinship testing [6]. This is due to the unique way in which sex chromosomes recombine and are passed on, with only the mother's X-chromosomes undergoing recombination before being inherited by offspring, while the father's X-chromosome is passed on to female offspring virtually unchanged due to its hemizygous nature [2,9]. Consequently, females who share the same father have the same paternal X-chromosome haplotype. Additionally, X-STRs are located within linkage groups and inherited as a haplotype [12].

Many researchers have developed in-house laboratory X-STR multiplex polymerase chain reaction (PCR) systems which can be utilised to generate X-STR population data [88–91]. However, the use of developmentally validated and commercially available X-STR multiplex kits is often preferred within the framework of a forensic laboratory setting. The most popularly used commercially available kit for the investigation of X-STRs is the Investigator Argus X-12 QS (Qiagen, Hilden) [9]. This kit was developmentally validated according to the European Network of Forensic Science Institute (ENFSI) and the Scientific Working Group on DNA Analysis Methods (SWGDM) and allows for simultaneous amplification of 12 X-STR loci located within linkage groups 1 – 4 (LG1 – LG4) on the X-chromosome. Also included within this kit is a quality sensor (QS), the Amelogenin locus and an autosomal locus (D21S11) [92].

Currently, most X-STR data available has been generated for populations within Europe and Asia, while populations of African origin remain underrepresented. As such, the value of X-

STRs is often overlooked within many African countries, including South Africa, due to the lack of an appropriate reference database.

South Africa consists of nine provinces, namely Eastern Cape, Free State, Gauteng, KwaZulu-Natal, Limpopo, Mpumalanga, Northern Cape, North West and Western Cape, and has an overall national population exceeding 60 million individuals. These individuals have been classified into four major population groups according to the South African national census, namely Black African (81 %), Coloured (8.8 %), Indian/Asian (2.6 %) and White (7.7 %) [93]. The National Forensic DNA Database of South Africa (NFDD) utilised by the South African Police Service (SAPS) also makes use of these population groups. However, for this population study, population groups were rather reported as ancestry, whereby African ancestry, European ancestry, Mixed ancestry and Indian/Asian ancestry refers to the Black African, White, Coloured and Indian/Asian population groups, respectively. This was to ensure both the global and local applicability of this data.

The aim of this study is, therefore, to provide a 'starting point' and platform for expanding the application of forensic genetics in South Africa by generating X-STR data for the overall South African population.

2.2. Methods and materials

This population study was quantitative and prospective, with the purpose of generating X-STR frequency data for the South African population. Before this could be achieved, the workflow needed to be optimised and internally validated.

2.2.1. Sample selection and experimentation

This study was sub-linked to an umbrella population study (HREC 342/2016) and obtained ethical approval from the Faculty of Health Science Human Research Ethics Committee (HREC) at the University of Cape Town (UCT) (HREC 136/2022) ([Appendix C](#)).

The samples utilised in this study were collected from both living and deceased individuals. Participation in this study was voluntary and informed consent was provided by the participant themselves or if deceased, their next-of-kin, for their sample to be utilised in any

future research conducted within the Division of Forensic Medicine and Toxicology. Adult participants were at least 18 years of age at the time of sample collection. Any individual who had received a bone marrow transplant or blood transfusion was excluded from this study.

2.2.1.1. Internal validation

The DNA profiling workflow for the Investigator Argus X-12 QS kit (Qiagen, Hilden) was internally validated for lysate samples. These samples were created by processing buccal swabs with Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) or Promega SwabSolution™ (Promega Corporation, WI, USA), according to the respective manufacturer's handbook [94,95]. Internal validation experimentation included the establishment of the analytical threshold, stochastic threshold and stutter peak threshold as well as an evaluation of sensitivity, limit of detection, reproducibility and concordance.

For the analytical threshold, lysate blanks representing each lysate preparation methods were used (n = 6 (3x2)). These samples were subjected to the DNA profiling workflow in triplicate.

The stochastic threshold, stutter peak threshold, sensitivity and limit of detection were established using three female lysate samples, for which informed consent was obtained. These samples were serially diluted according to the manufacturer's developmental validation guidelines [96], and processed in triplicate.

To evaluate reproducibility using repeated measurements, control DNA 9947A and 2800M, as well as a subset of 20 lysate samples were subjected to the DNA profiling workflow on separate days.

For concordance testing, a subset of 483 samples was used to compare the genotype of each sample at the autosomal marker, D21S11, between the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the Investigator 24plex GO! kit (Qiagen, Hilden) [96]. Additionally, a subset of 340 samples was used to evaluate the concordance of 7 X-STR markers (DXS10135, DXS8378, DXS7132, DXS10074, DXS10103, HPRTB and DXS7423), which are common to both the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the massively parallel sequencing (MPS) ForenSeq™ DNA Signature Prep kit (Illumina, CA, USA).

2.2.1.2. Population study

For the population study, a total of 781 biological samples belonging to unrelated South African individuals (male = 517, female = 264) were utilised. These individuals were located across the nine provinces and fell within one of the four major population groups of South Africa, *i.e.*, African ancestry ($n = 178$; male = 131, female = 47), European ancestry ($n = 223$; male = 129, female = 94), mixed ancestry ($n = 240$; male = 161, female = 79) and Indian/Asian ancestry ($n = 140$; male = 96, female = 44). These samples included lysates ($n = 686$), which were previously collected under the umbrella study as buccal swabs and processed as described in section 2.2.1.1., as well as the DNA profiles of previously genotyped extracted DNA samples ($n = 95$).

For all experimentation, control DNA 9947A and a non-template control (NTC) were included in each batch as a quality control measure.

2.2.2. Workflow optimisation

The workflow was optimised for each lysate type, which resulted in certain adjustments to the manufacturer's protocol (Table 2.1) [67,94].

Table 2.1 Optimised DNA profiling workflow conditions for lysate samples.

| Optimised conditions | Qiagen STR GO! Lysis Buffer | Promega SwabSolution |
|---|------------------------------------|-----------------------------|
| Final reaction volume (μl) | 6.25 | 6.25 |
| Volume of 5X AmpSolution (μl) | - | 0.3 |
| Volume of lysate (μl) | 1 | 1 |
| Number of PCR cycles | 24 | 23 |
| CE injection time (seconds) | 15 | 20 |
| CE run time (seconds) | 1800 | 1800 |

2.2.3. X-chromosome STR typing

DNA amplification was performed using the Investigator Argus X-12 QS kit (Qiagen, Hilden) on the T100 thermal cycler (Bio-Rad, California). The resultant PCR products were separated using capillary electrophoresis (CE) on the Applied Biosystems 3500xl Genetic Analyser (Thermo Fisher Scientific, Massachusetts) with POP4 polymer and a 36 cm capillary array.

2.2.4. Data analysis

2.2.4.1. Internal validation

The data were analysed using GeneMapper® *ID-X* Software Version 1.5 (Thermo Fisher Scientific, Massachusetts) and subsequent calculations were performed using Microsoft® Excel 365. For the analytical threshold, the peak amplitude thresholds were set to 1 relative fluorescent unit (RFU) to identify the highest peak. The RFU of that peak was doubled and considered as the analytical threshold. For the stochastic threshold, the point at which allelic drop-out occurred for a known heterozygote was identified. The RFU of all false homozygotes were recorded to calculate the mean and standard deviation. The stochastic threshold was calculated using Equation 2.1.

$$\textit{Stochastic threshold} = \textit{mean} + 3 \times (\textit{standard deviation})$$

Equation 2.1 *Stochastic threshold calculation*

For the stutter peak threshold, peak amplitude thresholds were set to 25 RFU, to allow for the adequate calling of the stutter peaks. A stutter peak ratio for each marker was calculated using Equation 2.2 and an average was determined. The stutter peak threshold for each marker and overall was calculated using Equation 2.3.

$$\textit{Stutter peak ratio} = \frac{\textit{Height of stutter peak (RFU)}}{\textit{Height of true allelic peak (RFU)}}$$

Equation 2.2 *Stutter peak ratio calculation*

Stutter peak threshold = Mean stutter peak ratio + 3 x (standard deviation)

Equation 2.3 *Stutter peak threshold calculation*

The sensitivity of the kit was determined as the lowest amount of DNA at which a full, accurate and reliable DNA profile was obtained, whereas the limit of detection was considered to be the lowest amount of DNA that was able to be detected, even if the thresholds were not met or a full profile was not obtained. To determine reproducibility between samples, as well as concordance between workflows, the number of concordant alleles was summed and divided by the total number of alleles (Equation 2.4).

$$\text{Concordance} = \frac{\text{Number of concordant alleles}}{\text{Total number of alleles}}$$

Equation 2.4 *Frequency of concordance calculation*

2.2.4.2. Population study

The resultant DNA profiles were analysed using GeneMapper® *ID-X* Software Version 1.5 (Thermo Fisher Scientific, Massachusetts), with the internally validated thresholds applied. Off-ladder alleles were manually characterised according to allele size and verified using MPS data, where available.

StatsX v2.0 [97] was used to calculate allele and haplotype frequencies as well as relevant forensic parameters, namely gene diversity (GD)/haplotypic diversity (HD), polymorphism information content (PIC), power of discrimination (PD) for males and females and mean exclusion chance (MEC).

Arlequin software v3.5.2.2 was used to evaluate population differentiation for each locus between males and females, Hardy-Weinberg equilibrium (HWE) per locus using female samples, linkage disequilibrium (LD) between loci by exact test using a Markov chain for males and likelihood ratios test for females, a locus by locus analysis of molecular variance (AMOVA) using conventional F-statistics (only haplotype frequencies) and pairwise F_{ST} genetic distance matrix between the different population groups for male haplotypic data.

2.3. Results

2.3.1. Internal validation

2.3.1.1. Interpretation thresholds

Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) and Promega SwabSolution (Promega Corporation, WI, USA) lysate blanks were processed in triplicate, and the analytical threshold was calculated to be 60 RFU and 100 RFU, respectively.

Using three serially diluted female lysate samples, the stochastic threshold for Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) and Promega SwabSolution (Promega Corporation, WI, USA) lysate samples were established as 201 RFU and 264 RFU, respectively.

Similarly, the marker-specific and overall stutter peak thresholds were determined (Table 2.2). The overall stutter peak threshold for the Qiagen STR Go! Lysis Buffer (Qiagen. Hilden) and the Promega SwabSolution (Promega Corporation, WI, USA) lysates was 14 %.

Table 2.2 Marker-specific and overall stutter peak thresholds.

| Locus | Qiagen STR Go! Lysis Buffer | | | Promega SwabSolution | | |
|--------------------------------------|-----------------------------|--------|---------------------------------------|------------------------|--------|---------------------------------------|
| | Mean stutter ratio (%) | SD (%) | Marker-specific stutter threshold (%) | Mean stutter ratio (%) | SD (%) | Marker-specific stutter threshold (%) |
| DXS10103 | 6.80 | 1.93 | 12.59 | 6.96 | 2.15 | 13.41 |
| DXS8378 | 7.28 | 1.28 | 11.12 | 7.29 | 1.63 | 12.18 |
| DXS10101 | 7.48 | 2.96 | 16.36 | 6.74 | 1.50 | 11.24 |
| DXS10134 | 7.94 | 1.71 | 13.07 | 7.93 | 1.51 | 12.46 |
| DXS10074 | 5.13 | 1.18 | 8.67 | 5.26 | 1.33 | 9.25 |
| DXS7132 | 9.14 | 2.69 | 17.21 | 8.62 | 2.61 | 16.45 |
| DXS10135 | 11.09 | 1.80 | 16.49 | 11.01 | 1.69 | 16.08 |
| DXS7423 | 7.27 | 1.20 | 10.87 | 7.14 | 1.34 | 11.16 |
| DXS10146 | 7.70 | 1.77 | 13.01 | 7.86 | 1.71 | 12.99 |
| DXS10079 | 7.67 | 2.26 | 14.45 | 7.86 | 3.63 | 18.75 |
| HPRTB | 11.16 | 2.82 | 19.62 | 11.42 | 2.10 | 17.72 |
| DXS10148 | 7.83 | 2.85 | 16.38 | 8.56 | 3.31 | 18.49 |
| D21S11 | 8.93 | 2.20 | 15.53 | 9.06 | 2.80 | 17.46 |
| Average (%) | 8.11 | 2.05 | - | 8.13 | 2.10 | - |
| Overall stutter threshold (%) | 14 | | | 14 | | |

Abbreviation - SD: standard deviation.

2.3.1.2. Sensitivity and limit of detection

It was determined that the sensitivity was 1:32 for Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) lysates and 1:16 for Promega SwabSolution (Promega Corporation, WI, USA) lysates, while the limit of detection was determined to be 1:128 for Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) and Promega SwabSolution (Promega Corporation, WI, USA) lysates.

2.3.1.3. Reproducibility

A comparison between the DNA profiles produced for each of the 20 lysate samples, as well as the two control DNA samples (9947A and 2800M), revealed 100 % concordance between allele calls, indicating successful reproducibility.

2.3.1.4. Concordance

A comparison between the genotypes obtained at the D21S11 locus from the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the Investigator 24plex GO! kit (Qiagen, Hilden) revealed 99.59 % concordance. Two samples were found to be non-concordant, whereby the Investigator Argus X-12 QS kit (Qiagen, Hilden) produced the genotypes: 29, 32.2 and 28.3, 31.2 rather than 29, 29 and 29, 31.2 as seen with the Investigator 24plex GO! kit (Qiagen, Hilden), respectively. The first instance of non-concordance is likely due to allele drop-out in the Investigator 24plex GO! kit (Qiagen, Hilden), while the second instance may be due to variation in the migration of DNA fragments through the capillary.

An assessment of concordance was also conducted between the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the ForenSeq™ DNA Signature Prep kit (Illumina, CA, USA). Where MPS data were available and allele drop-out had not occurred, allelic calls were fully concordant between workflows across all eight loci, with six exceptions ([Appendix D](#)). Locus DXS10135 had the highest number of non-concordant alleles ($n = 3$), followed by locus DXS10074 ($n = 2$) and locus DXS8378 ($n = 1$).

2.3.2. Population study

2.3.2.1. Statistical analyses

Biological samples belonging to 264 female and 517 male South African individuals were processed using the Investigator Argus X-12 QS kit (Qiagen, Hilden). From these samples, full DNA profiles were obtained and a total of 120 unique alleles were identified. The Exact test performed per locus revealed no significant differentiation between males and females was present ($p > 0.05$) ([Appendix E](#)). This allowed for the allele frequencies of male and female individuals to be pooled ([Appendix F](#)) and subsequently used to calculate forensic parameters for the 12 loci (Table 2.3). Forensic parameters for the overall South African population were evaluated to determine each locus's usefulness in discerning the identity of different individuals. Locus DXS10135 was the most informative (PIC = 0.952), with 53 unique alleles, while locus DXS7423 was the least informative (PIC = 0.623), with only seven unique alleles. GD for all loci was greater than 0.6. DXS10135 had the highest GD (0.955) and DXS7423 had the lowest (0.681). The combined PD of males and females for the 12 loci were 0.999999999919563 and 1, respectively.

Table 2.3 Forensic parameters for the 781 South African individuals.

| Linkage group | Locus | Number of unique alleles | GD | PIC | PD _{Male} | PD _{Female} |
|-----------------|----------|--------------------------|-------|-------|--------------------|----------------------|
| 1 (Xp22) | DXS8378 | 7 | 0.699 | 0.643 | 0.698 | 0.853 |
| | DXS10135 | 53 | 0.955 | 0.952 | 0.954 | 0.996 |
| | DXS10148 | 56 | 0.937 | 0.933 | 0.936 | 0.993 |
| 2 (Xp11) | DXS7132 | 9 | 0.751 | 0.709 | 0.750 | 0.896 |
| | DXS10074 | 21 | 0.874 | 0.860 | 0.873 | 0.971 |
| | DXS10079 | 14 | 0.832 | 0.810 | 0.831 | 0.950 |
| 3 (Xp26) | HPRTB | 10 | 0.744 | 0.702 | 0.743 | 0.893 |
| | DXS10101 | 27 | 0.917 | 0.910 | 0.916 | 0.987 |
| | DXS10103 | 8 | 0.754 | 0.718 | 0.753 | 0.904 |
| 4 (Xp28) | DXS7423 | 7 | 0.681 | 0.623 | 0.681 | 0.841 |
| | DXS10134 | 38 | 0.882 | 0.870 | 0.881 | 0.975 |
| | DXS10146 | 41 | 0.917 | 0.910 | 0.916 | 0.987 |
| Combined | | - | - | - | 0.9999 | 1 |

Abbreviations - GD: gene diversity, PIC: polymorphism information content, PD: power of discrimination.

HWE was assessed in female samples, and after the application of a Bonferroni correction for multiple comparisons, all loci were found to be in HWE, except DXS10148 ($p < 0.0042$) ([Appendix G](#)).

Haplotype frequencies and haplotype forensic parameters for the 517 male individuals were determined ([Appendix H](#)). No shared haplotypes were observed for the full 12-loci haplotype definition: DXS8378-DXS10135-DXS10148-DXS7132-DXS10074-DXS10079-HPRTB-DXS10101-DXS10103-DXS7423-DXS10134-DXS10146. A total of 421, 228, 218 and 302 unique three loci haplotypes were observed in LG1, LG2, LG3 and LG4, respectively. LG1 was the most informative (PIC = 0.997), while LG2 and LG3 were the least informative (PIC = 0.992). Additionally, the HD values for all LGs were greater than 0.99. The most common haplotype in each LG was as follows, LG1: 11-24-18 ($n = 5$), LG2: 14-15-19 and 14-16-19 ($n = 13$), LG3: 12-30.2-19 ($n = 12$) and LG4: 14-35-28 ($n = 12$).

LD tests were performed separately for male and female samples to evaluate the presence of non-random associations between loci ([Appendix I](#)). After a Bonferroni correction was applied, significant LD ($p < 0.0008$) was detected in five loci pairs in males (DXS10148-DXS10101, HPRTB-DXS10101, DXS10101-DXS10103, DXS10101-DXS10134 and DXS10134-DXS10146) and three loci pairs in females (DXS10148-DXS10074, DXS10101-DXS10103 and DXS10134-DXS10146). The maximum squared coefficient of correlation (r^2) value for each unlinked loci-pair in LD for males, *i.e.*, DXS10148-DXS10101, HPRTB-DXS10101 and DXS10101-DXS10134, was 0.2485, 0.0574 and 0.1651, respectively.

Pairwise F_{ST} and an AMOVA were calculated using haplotype frequencies to assess the intra-population genetic structure within the South African population. Using pairwise F_{ST} values, significant differences were observed between the African population group and all three other population groups, namely European, Mixed ancestry and Indian/Asian, after a Bonferroni correction was applied ($p < 0.0083$) (Fig. 2.1). Additionally, the highest population differentiation was observed between the African and European groups (F_{ST} value = 0.02301). The AMOVA showed that molecular variation among the population groups was only 1.04 %, while molecular variation within the population groups was 98.96 % ($F_{ST} = 0.01042$; $p = 0.00000$).

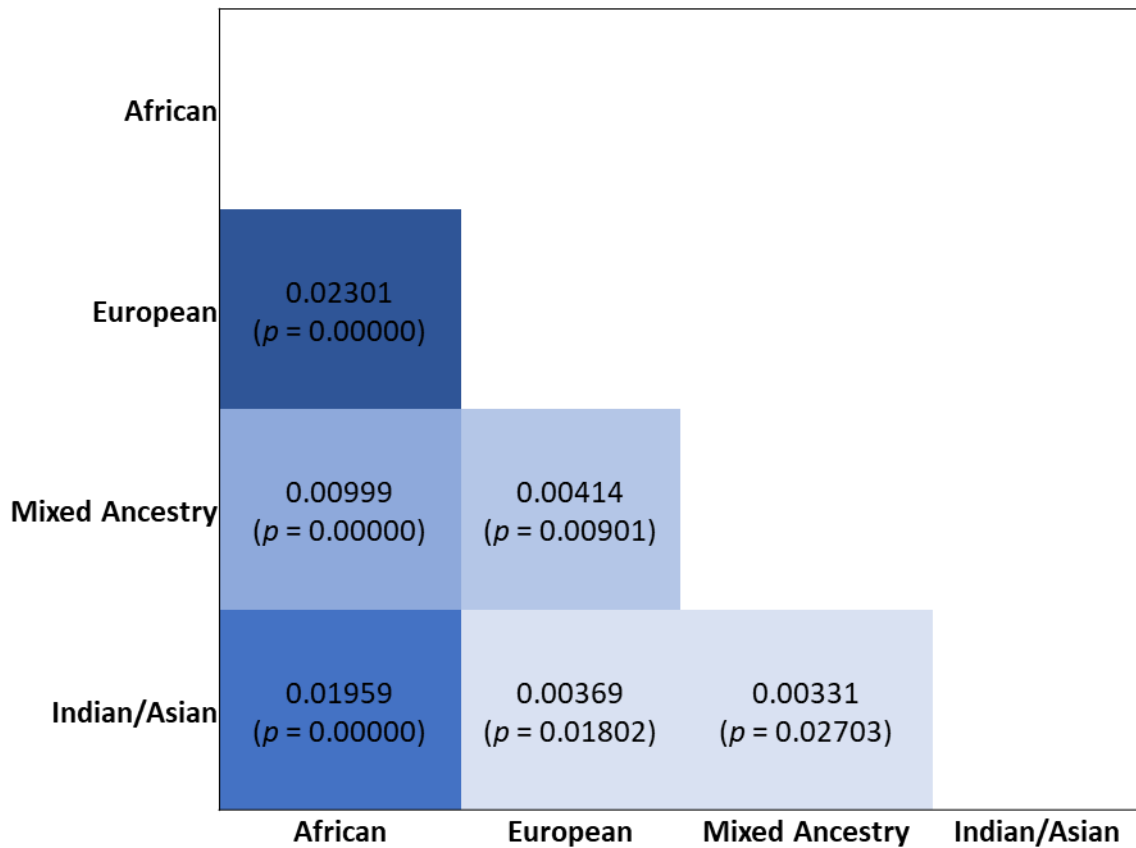


Fig. 2.1. Distance matrix of pairwise F_{ST} values (as indicated by the different shades of blue) for the four major South African population groups.

2.3.2.2. Off-ladder and null alleles

A total of 59 unique off-ladder peaks were identified across seven different loci included in the Investigator Argus X-12 QS kit (Qiagen, Hilden) ([Appendix J](#)). These peaks fell within regions of the loci where allelic representation is currently absent within the allelic ladder. These off-ladder alleles were detected in approximately 19 % ($n = 149$) of the total cohort utilised in this study, and a total of 166 off-ladder allele observations were made. From this, it was determined that 134 individuals had one off-ladder allele, 13 individuals had two off-ladder alleles and two individuals had three off-ladder alleles. Furthermore, the majority of the off-ladder alleles were observed within the African ($n = 96$) and Mixed ancestry ($n = 53$) population groups. Locus DXS10148 had the highest number of unique off-ladder alleles ($n = 23$), followed by locus DXS10134 ($n = 10$). However, the most prevalent off-ladder allele was 36.2 at locus DXS10146, with a total of 20 observations within the cohort (allele frequency = 0.0191). MPS data were able to verify alleles 26.3, 27.1, 28.3, 29.3, 30.1, 33.1 and 37.1 at

locus DXS10135, as well as alleles 13.2 and 16.3 at locus DXS10074. Null alleles were observed at locus DXS10146 in four male individuals and the Amelogenin locus in three male individuals.

2.4. Discussion

2.4.1. X-STR profiling in South Africa

The primary utilisation of X-STRs has been outlined within complex kinship testing, including deficiency paternity testing and testing distant familial relations [6]. These applications are relevant to the South African population and highlight the gap in which X-STRs can be implemented within medico-legal investigations. For example, South Africa is a country which legally permits polygynous marriages within indigenous communities [98]. Due to this, deficiency paternity inheritance disputes may arise. X-STRs can be used to resolve these complex family structures, particularly when involving daughters. Additionally, like many other countries [56,57], South Africa continues to be challenged by the large number of unidentified bodies remaining within the country's mortuary system [54,99]. These are individuals who were not able to be identified through routine processes [54] and lack a reference profile on the NFDD. Familial searching can be initiated in these instances, but this too is limited if only distant blood relatives are available as a reference. This is because DNA profiling in South Africa is routinely performed using AS-STRs, which lack sufficient discriminatory power to resolve these distant relationships. To overcome this, an alternative DNA profiling target, such as X-STRs, may be used.

However, the utilisation of X-STRs to assist in medico-legal investigations has not yet been implemented in South Africa. This is likely due to the absence of an appropriate X-STR population reference database, which is necessary to provide statistical confidence to X-STR profiling results [3]. Therefore, this study represents the first X-STR data generated for the South African population.

Previously, it was determined that allele frequency databases should contain at least 100 – 150 individuals per population group included [3,100]. As this study included 781 individuals from the four major population groups within South Africa, namely African ancestry ($n = 178$),

European ancestry ($n = 223$), Mixed ancestry ($n = 240$) and Indian/Asian ancestry ($n = 140$), it is believed that this data provides a reliable insight into the distribution of X-STRs within the South African population as a whole. Additionally, an overall sample size of greater than 500 individuals was utilised, which exceeds the Forensic Science International: Genetics journal guidelines [77].

2.4.2. The use of lysate samples

The conventional DNA profiling workflow requires a DNA extraction and quantification stage. Studies related to DNA profiling using the Investigator Argus X-12 QS kit (Qiagen, Hilden) typically include the use of extracted DNA from blood, buccal swabs or FTA cards [72,82,101–104]. Although effective, DNA extraction and quantification increase the processing time per sample and can result in sample loss. To address this, the direct PCR amplification approach has been introduced, whereby these stages within the DNA profiling workflow are eliminated. Shrivastava and Kumawat (2021) investigated the impact of direct PCR using FTA cards on the quality of DNA profiles produced for the Investigator Argus X-12 QS kit (Qiagen, Hilden) and concluded that this approach was indeed appropriate. This aligned with the developmental validation of the Investigator Argus X-12 QS kit (Qiagen, Hilden), which found the kit to be stable during direct amplification of FTA cards and lysates [105].

In this study, direct PCR amplification was performed on lysate samples prepared with two different lysis buffers, namely Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) and Promega SwabSolution (Promega Corporation, WI, USA). Following optimisation and internal validation, full DNA profiles were obtained for all included samples. This illustrates that lysate samples are compatible with the Investigator Argus X-12 QS kit (Qiagen, Hilden) and motivates the implementation of this workflow within medico-legal settings where a rapid turnaround time for identification is required.

2.4.3. Internal validation

This study presents the first internal validation data available on the Investigator Argus X-12 QS kit (Qiagen, Hilden) for direct amplification. As the developmental validation of the

Investigator Argus X-12 kit (Qiagen, Hilden) utilised extracted DNA samples, the Investigator validation guide [96] was implemented in this study instead.

Full DNA profiles were obtained for 98.24 % of individuals ($n = 781/795$). This indicates that the optimisation and the establishment of interpretation thresholds during the internal validation component of this study were successful. The analytical and stochastic thresholds were greater for Promega SwabSolution lysates compared to Qiagen STR GO! Lysis Buffer lysates. This was expected due to the difference in chemistry between the Promega SwabSolution lysates and the Investigator Argus X-12 QS kit (Qiagen, Hilden), thus requiring the addition of 5X AmpSolution (Promega Corporation, WI, USA). Subsequently, these DNA profiles often contained peaks of much higher RFU than the Qiagen STR GO! Lysis Buffer lysates.

The limit of detection for both lysate types was 1:128, while the sensitivity for the Qiagen STR GO! Lysis Buffer (Qiagen, Hilden) and the Promega SwabSolution (Promega Corporation, WI, USA) lysates were 1:32 and 1:16, respectively. The difference observed in sensitivity may be due to the use of different PCR and CE conditions for each lysate type (Table 2.1) and/or the differences in kit chemistry.

An evaluation of the reproducibility using repeated measurements for 20 lysate samples and two control DNA samples (9947A and 2800M) revealed 100 % concordance. This suggests that the DNA profiles obtained using the Investigator Argus X-12 QS kit (Qiagen, Hilden) are reliable.

Concordance testing between the autosomal locus, D21S11, included in the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the Investigator 24plex GO! kit (Qiagen, Hilden) showed 99.59 % concordance. This illustrates that the inclusion of this locus within the Investigator Argus X-12 QS kit (Qiagen, Hilden) to avoid sample mix-up, as suggested by the manufacturers [67], is valid. Furthermore, the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the ForenSeq™ DNA Signature Prep kit (Illumina, CA, USA) were fully concordant, except for six alleles ([Appendix D](#)). Similarly, Salvador *et al.* (2018) found 100 % concordance between these two workflows [106]. This indicates that the DNA profiles produced using the Investigator Argus X-12 QS kit (Qiagen, Hilden) are comparable to MPS workflows.

Overall, the internal validation of this study demonstrated that the Investigator Argus X-12 QS (Qiagen, Hilden) is sensitive and produces reliable results, making it appropriate for forensic use.

2.4.4. Population study

2.4.4.1. Shared haplotypes

The haplotypes of 517 males were evaluated. No shared haplotypes were observed when the full 12-loci haplotype definition was considered. This is interesting as the expectation of shared haplotypes increases when a large number of individuals are evaluated. Additionally, shared haplotypes have been observed in studies which utilised smaller sample sizes. For example, Curtis *et al.* (2022) investigated 298 Australian Aboriginal male individuals and found haplotype sharing between eight individuals [107].

Contrarily, the evaluation of three-loci haplotypes located at each linkage group showed haplotype sharing within the South African population ([Appendix H](#)). LG1 was identified as the most polymorphic (HD = 0.999, PIC = 0.997), while LG2 and LG3 were the least polymorphic (HD = 0.994, PIC = 0.992). A comparison to data generated for other African countries, namely Guinea-Bissau [108], Eritrea [109], Ethiopia [110] and Egypt [111], revealed that LG1 is consistently reported as the most diverse, while LG2 and LG3 were considered the least diverse in Eritrea and Ethiopia and Egypt, respectively. These trends were also observed in data generated globally for other populations [73,97,112,113]. Furthermore, 81.43 % ($n = 421/517$) of haplotypes identified at LG1 were unique for this South African cohort. This was the highest when compared to other reviewed literature [82,97,107,109,112]. Therefore, this indicates that LG1 is highly informative and of particular forensic interest, both locally and globally.

Together, these results illustrate the highly diverse nature of the South African population and suggest that X-STRs hold great discriminatory power within the population.

2.4.4.2. Forensic and population parameters

The individual population groups, namely African ancestry, European ancestry, Mixed ancestry and Indian/Asian ancestry were combined ($n = 781$) to evaluate the forensic

parameters of the 12 loci included in the Investigator Argus X-12 QS kit (Qiagen, Hilden) within a South African context. The results showed that locus DXS10135 was the most informative, while locus DXS7423 was the least informative (Table 2.3). These loci were also commonly reported as the most and least informative in many other population studies [97,101,106,107]. However, the number of unique alleles observed at locus DXS10135 ($n = 53$) was considerably higher in the South African population as opposed to the other populations. Loci DXS10148 and DXS10146 were also particularly informative within the South African population, with PIC values equivalent to 0.933 and 0.910, respectively. This may be due to the high number of novel alleles observed at these loci specifically. Lastly, the PIC value for all 12 loci was greater than 0.6, indicating a high level of polymorphism within this South African cohort.

Genetic comparisons between population groups revealed that all pairwise F_{ST} values were relatively low (Fig. 2.1). The greatest F_{ST} value (0.02301) was observed between the African ancestry and European ancestry population groups. According to Hartl and Clark (1997), an F_{ST} value between the range of 0.05 – 0.15 is considered to be “moderate” population differentiation [114]. Therefore, although there may be slight genetic variability between the population groups, the results suggest that sufficient gene flow between population groups has occurred to minimise the presence of any obvious intrapopulation substructure. This was supported by the AMOVA results, which showed that 98.96 % of the variability was within the population groups, while variability between population groups was only 1.04 % ($F_{ST} = 0.01042$; $p = 0.00000$). However, the significance indicated by the p -values should not be disregarded, suggesting these results should be interpreted with caution.

HWE was tested for the 12 loci using the genotypic data obtained from female individuals ($n = 264$). Deviations from HWE were identified at locus DXS10148, even after applying a Bonferroni correction ([Appendix G](#)). Deviations from HWE were also detected in other population studies. For example, Messaoudi *et al.*, (2021) reported HWE deviations at loci DXS10135, DXS10074, DXS10148 and DXS10101 in the Saudi population and Tomas *et al.* (2012) reported a deviation from HWE at locus DXS10148 in the Somali population [82,83]. These deviations from HWE could be due to the use of insufficient sample size within this study, resulting in sampling error [82,114]. Alternatively, null alleles have previously been reported at locus DXS10148 in African other populations [83,111]. Therefore, the significantly

low observed heterozygosity may be due to the presence of null alleles at locus DXS10148 [83] within the South African population. This would cause genotyping errors and ultimately lead to the false assignment of homozygosity to a female individual. As such, sequencing of alleles at this locus in female individuals may be more informative for HWE testing.

2.4.4.3. Linkage disequilibrium

Significant LD was found between several loci pairs after Bonferroni correction ([Appendix I](#)). In both male and female individuals, LD was detected between loci pairs DXS10101-DXS10103 and DXS10134-DXS10146, which belong to LG3 and LG4, respectively. This was expected as these markers are physically linked due to their proximity to each other, reducing the occurrence of recombination. Additionally, LD between these two loci pairs was also reported in other population studies [97,107,115]. LD was also detected between loci pairs DXS10148-DXS10101, HPRTB-DXS10101 and DXS10101-DXS10134 in males and DXS10148-DXS10074 in females. However, these loci pairs are not located within the same linkage groups. LD between unlinked loci (*i.e.*, from different linkage groups) has also previously been reported in the aforementioned studies [97,107,115]. The maximum r^2 value of these loci pairs was evaluated in males and showed that all values were relatively low ($r^2 < 0.25$). This may suggest that an unintentional association between these loci pairs may be present, rather than true LD.

2.4.4.4. Off-ladder and null alleles

A total of 59 unique off-ladder alleles were identified across seven loci, namely DXS10148, DXS10134, DXS10135, DXS10146, DXS10101, DXS10074, DXS10079 and HPRTB ([Appendix J](#)). The majority of these alleles were identified at locus DXS10148 ($n = 23$), of which 11 had previously been reported in other populations, particularly those of African origin. For example, Gomes *et al.* (2017) detected alleles 35.1, 37.1, 39.1, 40.1, 41.1, 42.1, 43.1 and 44.1 in the Guinea-Bissau population [116], while Elakkary *et al.* (2014) detected alleles 36.1, 37.1, 39.1, 42.1 and 43.1 in the Egyptian population [111]. Samejima *et al.* (2012) and Bottinelli *et al.* (2022) detected allele 40.1 at relatively low frequencies in the Malay ($f = 0.002$) and Swiss ($f = 0.0006$) populations, respectively [117,118]. Additionally, Samejima *et al.* (2012) reported

this as the largest allele to be detected within an eastern Asian population at that time. This may suggest that alleles with larger repeat core motifs (*i.e.*, > 35), at locus DXS10148, are more frequently observed within African populations compared to the rest of the world.

Further investigation into the off-ladder alleles revealed that allele 36.2, located at locus DXS10146, was the most prevalent within the South African cohort, with a total of 20 observations. A review of the literature revealed that this allele was also reported in a few other populations, namely Switzerland [118], Eritrea [109] and a Cabo Verde immigrant population residing in Lisbon [113]. Additionally, null alleles at locus DXS10146 were detected in four male individuals included in this study. However, this was not unexpected as null alleles at loci DXS10146 and DXS10148 have previously been described [110,111].

In this study, 134 individuals had one off-ladder allele, 13 individuals had two off-ladder alleles and two individuals had three off-ladder alleles identified from their DNA profile. This high frequency of off-ladder alleles is yet another demonstration of the diversity within the South African population. Most of these observations were in individuals with African ancestry, followed by individuals with Mixed ancestry, while only a few observations were in individuals with European and Indian/Asian ancestry ([Appendix J](#)). This suggests that there is an underrepresentation of alleles observed in African populations for approximately half of the loci included in the Investigator Argus X-12 QS kit (Qiagen, Hilden), and highlights the need for the development of an allelic ladder with improved allele coverage, to provide accurate allele calling in populations with different genetic compositions.

A total of 31 off-ladder alleles detected in this study had not previously been reported, which suggests that these alleles are likely to be novel. As such, these alleles must be verified, especially those which were only detected in a single individual ([Appendix J](#)). The use of MPS can be implemented for this purpose. For example, at locus DXS10135, only four of the nine off-ladder alleles identified in this study were reported in previous studies [108,109,113,118], while the use of available MPS data was able to verify the presence of seven of these alleles within the South African population. However, this approach is only appropriate for novel alleles located at loci included within the currently available MPS kits. Therefore, Sanger sequencing can be performed as an alternative method of novel allele verification.

Lastly, null alleles were also observed at the Amelogenin locus in two males from the Indian/Asian ancestry population group and one male from the Mixed Ancestry population group, whereby the Y-chromosome was absent from the DNA profile. This aligns with the literature, which suggests that this anomaly occurs more frequently in Indian populations compared to African and European populations [3]. Due to the hemizygous nature of the X-chromosome in male individuals, it was still possible to discern the sex of the individual using their DNA profile. However, this may become challenging when investigating mixed profiles or unknown individuals with X-chromosomal aberrations *e.g.*, Turner's syndrome (X0). Therefore, it may be beneficial for manufacturers of the Investigator Argus X-12 QS kit (Qiagen, Hilden) to include an additional locus for sex determination [63].

2.4.5. Limitations and future expansion

Although the cohort utilised in this study was appropriate [3,77], future studies could investigate a larger sample size due to the diversity within South Africa. Additionally, as this study served as a baseline for X-STRs within the South African population, it would be beneficial for future studies to investigate the composition of X-STRs within each of the four major population groups. This is because South Africa harbours multiple distinct population groups and is an admixed population [119]. As such, authorities have previously opted to generate autosomal STR allele frequency data per population group. Therefore, the separation of X-STR frequencies and other statistical calculations per population group would be necessary to ensure the casework applicability of this data within South Africa. Furthermore, as there are multiple cultural subgroups within the African ancestry population group itself [120], further investigation of X-STRs within and between each of these subgroups may be forensically informative. Lastly, verification of novel and null alleles using sequencing is required and a larger internal validation study should be performed for the Investigator Argus X-12 QS kit (Qiagen, Hilden), in which specificity, mixture, inhibition and degradation studies are included and tested.

2.5. Conclusion

In this study, X-STR data were successfully produced for lysate samples belonging to 781 South African individuals, using the Investigator Argus X-12 QS kit (Qiagen, Hilden). To this end, the data generated are highly polymorphic and informative. Internal validation of the Investigator Argus X-12 QS kit (Qiagen, Hilden) showed the kit to be reliable and comparable to other workflows, such as MPS. Multiple novel and null alleles were identified, which require confirmation in future studies. Comparisons to X-STR literature available for both other African and global populations illustrated the diverse nature of the South African population, as well as the paucity of African continental representation within the Investigator Argus X-12 QS kit (Qiagen, Hilden). Overall, the use of this kit for forensic purposes in the South African population is appropriate and motivates the generation of a national X-STR database for the South African population groups.

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- [335] J.F. Ferragut, K. Bentayebi, R. Pereira, J.A. Castro, A. Amorim, C. Ramon, A. Picornell, Genetic portrait of Jewish populations based on three sets of X-chromosome markers: Indels, Alu insertions and STRs, *Forensic Sci. Int. Genet.* 31 (2017) e5–e11. <https://doi.org/10.1016/j.fsigen.2017.09.008>.
- [336] E. Prieto-Fernández, A. Díaz-de Usera, M. Baeta, C. Núñez, F. Chbel, S. Nadifi, K. Rouault, C. Férec, O. Hardiman, F. Pinheiro, M.M. de Pancorbo, A genetic overview of Atlantic coastal populations from Europe and North-West Africa based on a 17 X-STR panel, *Forensic Sci. Int. Genet.* 27 (2017) 167–171. <https://doi.org/10.1016/j.fsigen.2016.11.011>.

Appendices:

Appendix A:

Table A1 List of all publications included within the systematic literature review.

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|---------------------------|--|---|--|------------------|--------|-------|
| X-STR applications | Schmidt, D., Hummel, S., Herrmann, B | Brief communication: Multiplex X/Y-PCR improves sex identification in aDNA analysis | American Journal of Physical Anthropology | 2003 | 121 | 4 |
| | Szibor, R., Plate, I., Edelmann, J., Hering, S., Kuhlisch, E., Michael, M., Krause, D. | Chromosome X haplotyping in deficiency paternity testing principles and case report | International Congress Series | 2003 | 1239 | |
| | Edelmann, J., Lessig, R., Klintschar, M., Szibor, R. | Advantages of X-chromosomal microsatellites in deficiency paternity testing: Presentation of cases | International Congress Series | 2004 | 1261 | |
| | Szibor, R., Hering, S., Kuhlisch, E., Plate, I., Demberger, S., Krawczak, M., Edelmann, J. | Haplotyping of STR cluster DXS6801-DXS6809-DXS6789 on Xq21 provides a powerful tool for kinship testing | International Journal of Legal Medicine | 2005 | 199 | 6 |
| | Toni, C., Presciuttini, S., Spinetti, I., Rocchi, A., Domenici, R. | Usefulness of X-chromosome markers in resolving relationships: Report of a court case involving presumed half sisters | International Congress Series | 2006 | 1288 | |
| | Asamura, H., Sakai, H., Kobayashi, K., Ota, M., Fukushima, H. | MiniX-STR multiplex system population study in Japan and application to degraded DNA analysis | International Journal of Legal Medicine | 2006 | 120 | 3 |
| | Barbaro, A., Cormaci, P., Barbaro, A. | X-STR typing for an identification casework | International Congress Series | 2006 | 1288 | |
| | Hatsch, D., Keyser, C., Hienne, R., Bertrand, L. | Resolving paternity relationships using X-chromosome STRs and Bayesian networks | Journal of Forensic Sciences | 2007 | 52 | 4 |
| | Krawczak, M. | Kinship testing with X-chromosomal markers: Mathematical and statistical issues | Forensic Science International: Genetics | 2007 | 1 | |
| | Silveira, Debora., Silva, F., Jesus, P., Whittle, M. | Use of X-linked short tandem repeat loci in routine parentage casework | Transfusion | 2007 | 47 | 6 |
| | Branicki, W., Wolańska-Nowak, P., Parys-Proszek, A., Kupiec, T. | Application of the mentype ARGUS X-8 kit to forensic casework | Problems of Forensic Sciences | 2008 | 73 | |
| | Serra, A., Bento, A. M., Carvalho, M., Andrade, L., Batista, L., Oliveira, M. C., Lopes, V., Balsa, F., Corte-Real, F., Anjos, M. J. | X-chromosome STR typing in deficiency paternity cases | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Bobillo, C., Marino, M., Sala, A., Gusmão, L., Corach, D. | X-STRs: Relevance in complex kinship cases | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|---|--|---|--|------------------|--------|-------|
| X-STR applications | Builes, J. J., Manrique, A., Aguirre, D., Puerto, Y., Bravo, M. L., Gusmão, L. | Utility of Y- and X-STRs in the research of complex biological relationship | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Aquino, J., Peixe, C., Silva, D., Tavares, C., de Carvalho, E. | A X-chromosome STR hexaplex as a powerful tool in deficiency paternity cases | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Toscanini, U., Berardi, G., Gómez, A., Raimondi, E. | X-STRs analysis in paternity testing when the alleged father is related to the biological father | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Silva, D., Manta, F., Desidério, M., Tavares, C., de Carvalho, E. | Paternity testing involving human remains identification and putative half sister: Usefulness of an X-hexaplex STR markers | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Pinto, N., Gusmão, L., Amorim, A. | X-chromosome markers in kinship testing: A generalisation of the IBD approach identifying situations where their contribution is crucial | Forensic Science International: Genetics | 2011 | 5 | |
| | Lancia, M., Severini, S., Coletti, A., Margiotta, G., Dobosz, M., Carnevali, E. | Using X-chromosomal markers in rape investigation | Forensic Science International: Genetics Supplement Series | 2011 | 3 | |
| | Gomes, C., Magalhães, M., Amorim, A., Alves, C., Pinto, N., Gusmão, L. | How useful is your X in discerning pedigrees? | Forensic Science International: Genetics Supplement Series | 2011 | 3 | |
| | Gomes, C., Magalhaes, M., Alves, C., Amorim, A., Pinto, N., Gusmao, L. | Comparative evaluation of alternative batteries of genetic markers to complement autosomal STRs in kinship investigations: Autosomal indels vs. X-chromosome STRs | International Journal of Legal Medicine | 2012 | 126 | 6 |
| | Diegoli, T., Linacre, A., Coble, M. | A gonosomal marker multiplex to aid in mixture interpretation | Forensic Science International: Genetics Supplement Series | 2013 | 4 | |
| | Pinto, N., Gusmão, L., Egeland, T., Amorim, A. | Paternity exclusion power: Comparative behaviour of autosomal and X-chromosomal markers in standard and deficient cases with inbreeding | Forensic Science International: Genetics | 2013 | 7 | |
| | Trindade-Filho, A., Ferreira, S., Oliveira, S. | Impact of a chromosome X STR Decaplex in deficiency paternity cases | Genetics and Molecular Biology | 2013 | 36 | 4 |
| | Tavares, C., Loiola, S., Pontes, I., Silva, D. A., Carvalho, E. F. | A multiplex typing system composed of autosomal and X-chromosomal STR markers | Forensic Science International: Genetics Supplement Series | 2013 | 4 | |
| Shyla, A., Borovko, S., Tillmar, A., Kuzub, N., Kotova, S., Tsybovsky, I., Rębała, K. | Belarusian experience of the use of FamLinkX for solving complex kinship cases involving X-STR markers | Forensic Science International: Genetics Supplement Series | 2015 | 5 | | |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|---------------------------|---|---|--|------------------|--------|-------|
| X-STR applications | Hering, S., Edelmann, J., Haas, S., Graser, N. | Paternity testing of two female siblings with Investigator Argus X-12 kit: A case with several rare mutation and recombination events | Forensic Science International: Genetics Supplement Series | 2015 | 5 | |
| | Auler-Bittencourt, E., Iwamura, E., Lima, M., da Silva, I., dos Santos, S. | Exploring the applicability of analysing X chromosome STRs in Brazilian admixed population | Science and Justice | 2015 | 55 | 5 |
| | Liu, Q., Xue, L., Zhao, H., Lu, D. | A Case of Maternal Half-sisters Sharing Alleles at 18 X-chromosomal Short Tandem Repeat Loci | Journal of Forensic Science and Medicine | 2016 | 2 | 2 |
| | Votrubova, J., Saskova, L., Frolik, J., Vanek, D. | DNA identification of a 10th century female skeleton from the Prague Castle belonging to a member of the Przemyslids Dynasty | Forensic Science International: Genetics Supplement Series | 2017 | 6 | |
| | Tillmar, A., Kling, D., Butler, J., Parson, W., Prinz, M., Schneider, P., Egeland, T., Gusmão, L. | DNA Commission of the International Society for Forensic Genetics (ISFG): Guidelines on the use of X-STRs in kinship analysis | Forensic Science International: Genetics | 2017 | 29 | |
| | Palomo-Díez, S., Esparza Arroyo, Á., Tirado-Vizcaíno, M., Velasco Vázquez, J., López-Parra, A., Gomes, C., Baeza-Richer, C., Arroyo-Pardo, E. | Kinship analysis and allelic dropout: a forensic approach on an archaeological case | Annals of Human Biology | 2018 | 45 | 4 |
| | Kling, D. | Curiosities of X chromosomal markers and haplotypes | International Journal of Legal Medicine | 2018 | 132 | 2 |
| | Dumache, R., Puiu, M., Parvanescu, R., Rogobete, A., Enache, A. | Advantages of chromosome X-STRS markers in solving a father-daughter paternity case with one mismatch on SE33 locus | Clinical Laboratory | 2019 | 65 | 9 |
| | Spitzer, A., Sapir, L., Amiel, M. | What is she doing here? Klinefelter syndrome in forensic casework | Science and Justice | 2021 | 61 | 4 |
| | Zhang, J., Hao, S., Liu, Y., Yuan, L. | Identification of half-sisters from different mothers by autosomal and X chromosomal short tandem repeats: A case study | Journal of Forensic Science and Medicine | 2021 | 7 | 2 |
| X-STR kits | Gehrig, C., Teyssier, A. | Validation of the Mentype® Argus X-UL kit | International Congress Series | 2006 | 1288 | |
| | Gusmão, L., <i>et al.</i> | Results of the GEP-ISFG collaborative study on an X-STR Decaplex | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Scherer, M., König, M., Bussmann, M., Prochnow, A., Peist, R. | Development and validation of the new Investigator® Argus X-12 QS Kit | Forensic Science International: Genetics Supplement Series | 2015 | 5 | |
| X-STR databases | Szibor, R., Hering, S., Edelmann, J. | A new Web site compiling forensic chromosome X research is now online | International Journal of Legal Medicine | 2006 | 120 | 4 |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|---------------------------------|--|--|--|------------------|--------|-------|
| X-STR databases | Martins, J., Kawamura, B., Cardoso, A., Cicarelli, M., Regina B. | Brazilian genetic database of chromosome X | Molecular Biology Reports | 2014 | 41 | |
| | Kling, D., Dell'Amico, B., Tillmar, A. | FamLinkX - Implementation of a general model for likelihood computations for X-chromosomal marker data | Forensic Science International: Genetics | 2015 | 17 | |
| X-STR population studies | Edelman, J., Hering, S., Michael, M., Lessig, R., Deischel, D., Meier-Sundhausen, G., Roewer, L., Plate, I., Szibor, R. | 16 X-chromosome STR loci frequency data from a German population | Forensic Science International | 2001 | 124 | |
| | Zarrabeitia, M., Amigo, T., Sañudo, C., Zarrabeitia, A., González-Lamuño, D., Riancho, J. | A new pentaplex system to study short tandem repeat markers of forensic interest on X chromosome | Forensic Science International | 2002 | 129 | 2 |
| | Athanasiadou, D., Stradmann-Bellinghausen, B., Rittner, C., Alt, K. W., Schneider, P. M. | Development of a quadruplex PCR system for the genetic analysis of X-chromosomal STR loci | International Congress Series | 2003 | 1239 | |
| | Toni, C., Presciuttini, S., Spinetti, I., Domenici, R. | Population data of four x-chromosome markers in Tuscany and their use in a deficiency paternity case | Forensic Science International | 2003 | 137 | |
| | Shin, K., Kwon, B., Lee, S., Yoo, J., Park, M., Chung, U., Lee, H., Han, G., Choi, J., Kim, C. | Five highly informative X-chromosomal STRs in Koreans | International Journal of Legal Medicine | 2004 | 118 | |
| | Zarrabeitia, M. T., Alonso, A., Zarrabeitia, A., Castro, A., Fernández, I., Martínez De Pancorbo, M. | X-linked microsatellites in two Northern Spain populations | Forensic Science International | 2004 | 145 | |
| | Edelmann, J., Lessig, R., Hering, S., Brundirs, N., Kuhlisch, E., Szibor, R. | Allele frequencies for X-chromosomal microsatellites in different populations | International Congress Series | 2004 | 1261 | |
| | Chen, M., Pu, C | Population data on the X chromosome short tandem repeat loci DXS10011, DXS101, DXS6789, DXS7132, DXS8377, and DXS9895 in Taiwan | Forensic Science International | 2004 | 146 | |
| | Peloso, G., Grignani, P., Previderè, C. | Allele distribution of five X-chromosome STR loci in an Italian population sample | International Congress Series | 2004 | 1261 | |
| | Lee, H., Park, M., Jeong, C., Lee, S., Yoo, J., Chung, U., Choi, J., Kim, C., Shin, K. | Genetic characteristics and population study of 4 X-chromosomal STRs in Koreans: Evidence for a null allele at DXS9898 | International Journal of Legal Medicine | 2004 | 118 | 6 |
| | Bini, C., Ceccardi, S., Ferri, G., Pelotti, S., Alù, M., Roncaglia, E., Beduschi, G., Caenazzo, L., Ponzano, E., Tasinato, P., Turchi, C., Buscemi, L., Mazzanti, M., Tagliabracci, A., Toni, C., Spinetti, I., Domenici, R., Presciuttini, S. | Development of a heptaplex PCR system to analyse X-chromosome STR loci from five Italian population samples: A collaborative study | Forensic Science International | 2005 | 153 | |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|--|---|--|---|------------------|--------|-------|
| X-STR population studies | Poetsch, M., Petersmann, H., Repenning, A., Lignitz, E. | Development of two pentaplex systems with X-chromosomal STR loci and their allele frequencies in a northeast German population | Forensic Science International | 2005 | 155 | |
| | Pepinski, W., Skawronska, M., Niemcunowicz-Janica, A., Koc-Zorawska, E., Janica, J., Soltyszewski, I. | Polymorphism of four X-chromosomal STRs in a Polish population sample | Forensic Science International | 2005 | 151 | |
| | Shin, S., Yu, J., Park, S., Min, G., Chung, K. | Genetic analysis of 18 X-linked short tandem repeat markers in Korean population | Forensic Science International | 2005 | 147 | |
| | Tabbada, K., De Ungria, M., Faustino, L., Athanasiadou, D., Stradmann-Bellinghausen, B., Schneider, P. | Development of a pentaplex X-chromosomal short tandem repeat typing system and population genetic studies | Forensic Science International | 2005 | 154 | |
| | Zarrabeitia, M., Alonso, A., Martin, J., Gonzalez-Gay, M., Martin-Escudero, J. De Pancorbo, M., Sanz, P., Ruiz-Cabello, F., Riancho, J. | Study of six X-linked tetranucleotide microsatellites: Population data from five Spanish regions | International Journal of Legal Medicine | 2006 | 120 | |
| | Oguzturun, C., Thacker, C. R., Syndercombe Court, D. | Population study of four X-chromosomal STR loci in the UK and Irish population | International Congress Series | 2006 | 1288 | |
| | Pepinski, W., Niemcunowicz-Janica, A., Skawronska, M., Janica, J. R., Koc-Zorawska, E., Janica, J., Soltyszewski, I. | Polymorphism of four X-chromosomal STRs in a religious minority of Old Believers residing in northeastern Poland | International Congress Series | 2006 | 1288 | |
| | Cerri, N., Verzeletti, A., Gasparini, F., Bandera, B., De Ferrari, F. | Population data for four X-chromosomal STR loci in a population sample from Brescia (northern Italy) | International Congress Series | 2006 | 1288 | |
| | Edelmann, J., Lessig, R., Willenberg, A., Wildgrube, R., Hering, S., Szibor, R. | Forensic validation of the X-chromosomal STR-markers GATA165B12, GATA164A09, DXS9908 and DXS7127 in German population | International Congress Series | 2006 | 1288 | |
| | Tang, W. M., To, K. Y. | Four X-chromosomal STRs and their allele frequencies in a Chinese population | Forensic Science International | 2006 | 162 | |
| | Asamura, H., Sakai, H., Kobayashi, K., Ota, M., Fukushima, H. | MiniX-STR multiplex system population study in Japan and application to degraded DNA analysis | International Journal of Legal Medicine | 2006 | 120 | |
| | Poetsch, M., Sabule, A., Petersmann, H., Volkson, V., Lignitz, E. | Population data of 10 X-chromosomal loci in Latvia | Forensic Science International | 2006 | 157 | |
| | Robino, C., Giolitti, A., Gino, S., Torre, C. | Development of two multiplex PCR systems for the analysis of 12 X-chromosomal STR loci in a northwestern Italian population sample | International Journal of Legal Medicine | 2006 | 120 | |
| Asamura, H., Sakai, H., Ota, M., Fukushima, H. | Japanese population data for eight X-STR loci using two new quadruplex systems | International Journal of Legal Medicine | 2006 | 120 | | |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|--|--|---|--|------------------|--------|-------|
| X-STR population studies | Aler, M., Sánchez-Diz, P., Gomes, I., Gisbert, M., Carracedo, A., Amorim, A., Gusmão, L. | Genetic data of 10 X-STRs in a Spanish population sample | Forensic Science International | 2007 | 173 | |
| | Cainé, L., Pontes, L., Abrantes, D., Lima, G., Pinheiro, F. | Genetic data of four X-chromosomal STRs in a population sample of Santa Catarina, Brazil | Journal of Forensic Sciences | 2007 | 52 | 2 |
| | Gomes, I., Prinz, M., Pereira, R., Meyers, C., Mikulasovich, R., Amorim, A., Carracedo, A., Gusmão, L. | Genetic analysis of three US population groups using an X-chromosomal STR decaplex | International Journal of Legal Medicine | 2007 | 121 | |
| | Zalán, A., Völgyi, A., Jung, M., Peterman, O., Pamjav, H. | Hungarian population data of four X-linked markers: DXS8378, DXS7132, HPRTB, and DXS7423 | International Journal of Legal Medicine | 2007 | 121 | |
| | Gomes, I., Alves, C., Maxzud, K., Pereira, R., Prata, M., Sánchez-Diz, P., Carracedo, A., Amorim, A., Gusmão, L. | Analysis of 10 X-STRs in three African populations | Forensic Science International: Genetics | 2007 | 1 | |
| | Pereira, R., Gomes, I., Amorim, A., Gusmão, L. | Genetic diversity of 10 X chromosome STRs in northern Portugal | International Journal of Legal Medicine | 2007 | 121 | |
| | Turrina, S., Atzei, R., Filippini, G., De Leo, D. | Development and forensic validation of a new multiplex PCR assay with 12 X-chromosomal short tandem repeats | Forensic Science International: Genetics | 2007 | 1 | |
| | Cerri, N., Verzeletti, A., Gasparini, F., Poglio, A., Mazzeo, E., De Ferrari, F. | Population data for 8 X-chromosome STR loci in a population sample from Northern Italy and from the Sardinia island | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Masseti, S., Carnevali, E., Lancia, M., Coletti, A., Dobosz, M., Bacci, M., Argiolas, V., D'Aloja, E. | Analysis of 8 STR of the X-chromosome in two Italian regions (Umbria and Sardinia) | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Tavares, C., Gusmão, L., Domingues, C., Domingues, P., Silva, D., Aquino, J., Peixe, C., Amorim, A., de Carvalho, E. | Population data for six X-chromosome STR loci in a Rio de Janeiro (Brazil) sample: Usefulness in forensic casework | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Ribeiro Rodrigues, E., Leite, F., Hutz, M., Palha, T., Ribeiro dos Santos, Â., dos Santos, S. | A multiplex PCR for 11 X chromosome STR markers and population data from a Brazilian Amazon Region | Forensic Science International: Genetics | 2008 | 2 | |
| | Thiele, K., Löffler, S., Löffler, J., Günthner, F., Nitschke, K., Edelmann, J., Lessig, R. | Population data of eight X-chromosomal STR markers in Ewe individuals from Ghana | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Tariq, M., Ullah, O., Riazuddin, S., Riazuddin, S. | Allele frequency distribution of 13 X-chromosomal STR loci in Pakistani population | International Journal of Legal Medicine | 2008 | 122 | |
| Fracasso, T., Schürenkamp, M., Brinkmann, B., Hohoff, C. | An X-STR meiosis study in Kurds and Germans: Allele frequencies and mutation rates | International Journal of Legal Medicine | 2008 | 122 | | |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|------------------------------|--|---|---|------------------|--------|-------|
| X-STR population data | Ruivo, D., Ribeiro, T., Espinheira, R., Geada, H. | Use of eight X-chromosomal STRs in paternity investigation | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Tillmar, A., Mostad, P., Egeland, T., Lindblom, B., Holmlund, G., Montelius, K. | Analysis of linkage and linkage disequilibrium for eight X-STR markers | Forensic Science International: Genetics | 2008 | 3 | |
| | Branicki, W., Wolańska-Nowak, P., Parys-Proszek, A., Kupiec, T. | Application of the mentype ARGUS X-8 kit to forensic casework | Problems of Forensic Sciences | 2008 | 73 | |
| | Liu, Q., Lv, D., Wu, X., Sun, H., Wu, X., Lu, H. | Development of a five ChX STRs loci typing system | International Journal of Legal Medicine | 2008 | 122 | |
| | Zalán, A., Völgyi, A., Brabetz, W., Schleinitz, D., Pamjav, H. | Hungarian population data of eight X-linked markers in four linkage groups | Forensic Science International | 2008 | 175 | |
| | Hashiyada, M., Itakura, Y., Funayama, M. | Polymorphism of eight X-chromosomal STRs in a Japanese population | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Cybulska, L., Wysocka, J., Rebała, K., Kapińska, E., Mikulich, A., Tsybovsky, I., Siváková, D., Džupinková, Z., Szczerkowska, Z. | Polymorphism of four X-chromosomal STR loci in Belarusians and Slovaks | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Barbaro, A., Cormaci, P., Votano, S., Barbaro, A. | Population data of 8 X-STRs in South Italy (Calabria) using the Mentype 1 Argus X-8 PCR Amplification Kit (Biotype) | Forensic Science International: Genetics | 2008 | 1 | |
| | Martins, J. A., Silva, R. H.A., Freschi, A., Paneto, G. G., Oliveira, R. N., Cicarelli, R. M.B. | Population genetic data of five X-chromosomal loci in Bauru (São Paulo, Brazil) | Forensic Science International: Genetics Supplement Series | 2008 | 1 | |
| | Becker, D., Rodig, H., Augustin, C., Edelmann, J., Götz, F., Hering, S., Szibor, R., Brabetz, W. | Population genetic evaluation of eight X-chromosomal short tandem repeat loci using Mentype Argus X-8 PCR amplification kit | Forensic Science International: Genetics | 2008 | 2 | |
| | Pico, A., Castillo, A., Vargas, C., Amorim, A., Gusmão, L. | Genetic profile characterization and segregation analysis of 10 X-STRs in a sample from Santander, Colombia | International Journal of Legal Medicine | 2008 | 122 | |
| | Tamura, A., Tsutsumi, H., Hara, M., Takada, A., Saito, K., Suzuki, K., Komuro, T. | Genetic studies of eight X-STRs in a Japanese population | Legal Medicine | 2009 | 11 | |
| | Poetsch, M., El-Mostaqim, D., Tschentscher, F., Browne, E., Timmann, C., Horstmann, R., Von Wurmb-Schwark, N. | Allele frequencies of 11 X-chromosomal loci in a population sample from Ghana | International Journal of Legal Medicine | 2009 | 123 | |
| | Gusmão, L., Sánchez-Diz, P., Alves, C., Gomes, I., Zarrabeitia, M., Abovich, M. | A GEP-ISFG collaborative study on the optimization of an X-STR decaplex: Data on 15 Iberian and Latin American populations | International Journal of Legal Medicine | 2009 | 123 | |
| | Bekada, A., Benhamamouch, S., Boudjema, A., Fodil, M., Menegon, S., Torre, C., Robino, C | Analysis of 12 X-chromosomal STRs in an Algerian population sample | Forensic Science International : Genetics Supplement Series | 2009 | 2 | |

| Objective | Authors | Title | Journal | Publication Year | Volume | Issue |
|---|--|---|--|------------------|--------|-------|
| X-STR population data | Li, H., Tang, H., Zhang, Q., Jiao, Z., Bai, J., Chang, S. | A multiplex PCR for 4 X chromosome STR markers and population data from Beijing Han ethnic group | Legal Medicine | 2009 | 11 | |
| | Nadeem, A., Babar, M., Hussain, M., Tahir, M. | Development of pentaplex PCR and genetic analysis of X chromosomal STRs in Punjabi population of Pakistan | Molecular Biology Reports | 2009 | 36 | 7 |
| | Jêdrzejczyk, M., Jacewicz, R., Berent, J. | Distribution of chromosome X str markers DXS10135 , DXS10074 , DXS10101 and DXS10134 and their usefulness in forensic genetics | Problems of Forensic Sciences | 2009 | 77 | |
| | Martins, J. A., Costa, J. C., Paneto, G. G., Gusmão, L., Sánchez-Diz, P., Carracedo, A., Cicarelli, R. M.B. | Genetic data of 10 X-chromosomal loci in Vitória population (Espírito Santo State, Brazil) | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Toscanini, U., Gusmão, L., Berardi, G., Raimondi, E. | Genetic data of 10 X-STR in two Native American populations of Argentina | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Gomes, I., Amorim, A., Pereira, V., Carracedo, A., Gusmão, L. | Genetic patterns of 10 X chromosome short tandem repeats in an Asian population from Macau | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Lim, E., Lee, H., Sim, J., Yang, W., Shin, K. | Genetic polymorphism and haplotype analysis of 4 tightly linked X-STR duos in Koreans | Croatian Medical Journal | 2009 | 50 | |
| | Turrina, S., Filippini, G., De Leo, D. | Genetic studies of eight X-STRs in a Northeast Italian population | Forensic Science International: Genetics Supplement Series | 2009 | 2 | |
| | Tariq, M., Sabir, M., Riazuddin, S., Riazuddin, S. | Haplotype analysis of two X-chromosome STR clusters in the Pakistani population | International Journal of Legal Medicine | 2009 | 123 | |
| | Leite, F., Santos, S., Rodríguez, E., Callegari-Jacques, S., Demarchi, D., Tsuneto, L., Petzl-Erler, M., Salzano, F., Hutz, M. | Linkage disequilibrium patterns and genetic structure of Amerindian and non-Amerindian Brazilian populations revealed by long-range X-STR markers | American Journal of Physical Anthropology | 2009 | 139 | |
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Appendix B:

Table B1 Summary of the available global X-STR population data publications

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|---|--|---|----------------------------------|-----------|
| Asia | Bangladesh | 102 males, 107 females | Investigator Argus X-12 | [81] |
| | China | 80 males, 10 females | In-house (5 X-STRs) | [121] |
| | China (Aksu Uyghur) | 229 males, 271 females | AGCU X19 | [68] |
| | China (Beijing Han) | 200 males, 200 females | In-house (4 X-STRs) | [122] |
| | China (Beijing Han) | 592 males, 655 females, 770 families | Goldeneye 17X | [123] |
| | China (Beijing Han) | 494 males, 591 females | In-house (19 X-STRs) | [124] |
| | China (Chinese Han) | 500 males, 327 females | In-house (5 X-STRs) | [125] |
| | China (Chinese Han) | 203 males, 100 females | Mentype Argus X-8 | [126] |
| | China (Daur and Oroqen) | 141 males, 110 females | AGCU X19 | [127] |
| | China (Gelao) | 248 males, 265 females | AGCU X19 | [128] |
| | China (Guangdong Han, Xinjiang Uigur, Inner Mongolia Mongol) | 484 males, 406 females, 210 family trios, 170 family duos | In-house (9 X-STRs) | [129] |
| | China (Guangdong Han) | 144 males, 128 females | Investigator Argus X-12 | [130] |
| | China (Guangdong Han) | 191 males, 169 females | In-house (12 X-STRs) | [131] |
| | China (Guangdong Han) | 65 males, 135 females | Microreader 19X Direct ID System | [132] |
| | China (Guangdong Han and Xinjiang Kazakh) | 574 males, 431 females, 310 family trios, 170 family duos, 40 three-generation families | In-house (12 X-STRs) | [133] |
| | China (Guangdong Han, Uigur and Kazakh) | 670 males, 581 females | In-house (15 X-STRs) | [134] |
| | China (Guangdong Han, Xinjiang Uigur, Tacheng Kazakh, Mongolia Mongol) | 876 males, 646 females, 325 family trios, 286 family duos, 40 three-generation families | In-house (26 X-STRs) | [135] |
| | China (Guangdong Han, Tibetan, Mongolian, Korean, Uighur, Hui) | 1298 individuals | Investigator Argus X-12 | [136] |
| | China (Guangdong Han, Tibetan, Uighur, Hui) | 631 males, 301 females | AGCU X19 | [137] |
| | China (Guangzhong Han) | 252 males, 222 females | AGCU X19 | [138] |
| China (Guizhou Bouyei) | 188 males, 165 females | AGCU X19 | [139] | |
| China (Guizhou Chuanqing, Tujia and Yi) | 306 males, 208 females | AGCU X19 | [140] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|-----------|---|---|----------------------------|-----------|
| Asia | China (Guizhou Dong) | 272 males, 235 females | AGCU X19 | [141] |
| | China (Guizhou Han) | 819 males, 525 females | AGCU X19 | [142] |
| | China (Guizhou Miao) | 151 males, 117 females | AGCU X19 | [143] |
| | China (Guizhou Sui) | 195 males, 205 females | AGCU X19 | [144] |
| | China (Guizhou Tujia) | 258 males, 249 females | AGCU X19 | [145] |
| | China (Hainan Li, Hainan Han, Dujiangyan Tibetan and Wuzhong Hui) | 712 individuals | AGCU X19 | [146] |
| | China (Han) | 508 males | Goldeneye 17X | [147] |
| | China (Han, Hui, Uygur, Mongolian and Tibetab) | 250 males, 250 females | In-house (16 X-STRs) | [148] |
| | China (Hani) | 116 males, 335 females | Goldeneye 17X | [149] |
| | China (Hong Kong) | 250 males, 250 females | Mentype Argus X-UL | [150] |
| | China (Kazak) | 149 males, 151 females | AGCU X19 | [151] |
| | China (Liaoning Manchu) | 514 males, 258 females | Investigator Argus X-12 | [101] |
| | China (Macau) | 46 males, 22 females | GHEP-ISFG X-STR decaplex | [152] |
| | China (Mongolian and Eastern Han) | 316 males, 225 females | Investigator Argus X-12 | [153] |
| | China (Northern Han) | 268 males, 248 females | Investigator Argus X-12 QS | [97] |
| | China (Shaanxi Han) | 312 males, 206 females | AGCU X12 | [154] |
| | China (Shandong Han) | 97 males, 214 females | Goldeneye 17X | [155] |
| | China (Shanghai Han) | 106 males, 92 females | Mentype Argus X-8 | [156] |
| | China (Shanghai Han) | 200 males, 109 females | Investigator Argus X-12 | [157] |
| | China (Shanghai Han) | 293 males, 298 females, 400 two-generation families | In-house (16 X-STRs) | [64] |
| | China (Shanghai Han) | 127 males, 71 females, 43 families | In-house (9 X-STRs) | [158] |
| | China (Sichuan Han) | 93 males, 108 females | AGCU X19 | [159] |
| | China (Sichuan Tibetan) | 117 males, 118 females | AGCU X19 | [160] |
| | China (Southern Han) | 199 males, 199 females | TYPED X19 | [161] |
| | China (Tibetan and Northern Han) | 742 males, 863 females | In-house (11 X-STRs) | [162] |
| | China (Tibetan and Uygur) | 490 males | AGCU X19 | [163] |
| | China (Tibetan, Mongolian and Kazakh) | 259 males, 259 females | Goldeneye 17X | [164] |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|-----------|--------------------------------------|------------------------|----------------------------|-----------|
| Asia | China (Tongxin Hui and Wuzhong Hui) | 131 males, 100 females | ZJGA-X12 STR | [165] |
| | China (Uigur and Northern Han) | 470 males, 780 females | In-house (11 X-STRs) | [166] |
| | China (Xibe) | 87 males, 92 females | AGCU X19 | [167] |
| | China (Xinjiang Mongolian) | 156 males, 111 females | AGCU X19 | [168] |
| | China (Xinjiang Uygur) | 78 males, 231 females | In-house (11 X-STRs) | [169] |
| | China (Xinjiang Uygur) | 94 males, 139 females | AGCU X19 | [170] |
| | China (Yanbian, Korean) | 257 males, 316 females | Investigator Argus X-12 | [171] |
| | China (Yi) | 133 males, 198 females | AGCU X19 | [172] |
| | China (Yunnan Bai) | 202 males, 222 females | Goldeneye 17X | [173] |
| | China (Yunnan Han) | 247 males, 168 females | Goldeneye 17X | [174] |
| | China (Yunnan Miao) | 260 males, 180 females | Goldeneye 17X | [175] |
| | China (Zhejiang Han) | 121 males, 60 females | AGCU X19 | [176] |
| | China (Zhejiang She) | 129 males, 167 females | Goldeneye 17X | [177] |
| | China (Zhejiang) | 129 males, 167 females | Goldeneye 17X | [178] |
| | China (Zhuang and Mulao) | 388 males, 251 females | AGCU X19 | [179] |
| | East Timor | 101 males, 48 females | Investigator Argus X-12 | [79] |
| | India | 749 males | In-house (10 X-STRs) | [180] |
| | India | 749 males | In-house (11 X-STRs) | [181] |
| | India (Bhil, Madhya Pradesh) | 100 males, 83 females | Investigator Argus X-12 | [84] |
| | India (Jat Sikh, Punjab) | 100 males, 100 females | Investigator Argus X-12 QS | [85] |
| | Iran (Persians, Lurs, Kurds, Azeris) | 255 individuals | Investigator Argus X-12 | [182] |
| | Iraq | 105 males, 103 females | In-house (5 X-STRs) | [183] |
| | Iraq | 96 males | Investigator Argus X-12 | [184] |
| | Japan | 195 males, 138 females | In-house (8 mini X-STRs) | [39] |
| | Japan | 229 males, 172 females | In-house (8 X-STRs) | [185] |
| | Japan | 144 males, 114 females | Mentype Argus X-8 | [186] |
| | Japan | 313 males, 181 females | Mentype Argus X-8 | [187] |
| Japan | 95 males | In-house (5 X-STRs) | [121] | |
| Japan | 339 males, 173 females | In-house (16 X-STRs) | [188] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|-----------|---|--|----------------------------|-----------|
| Asia | Japan | 425 males, 323 females | In-house (27 X-STRs) | [189] |
| | Japan | 390 males, 179 females | Mentype Argus X-8 | [190] |
| | Japan | 37 males, 175 females | In-house (18 X-STRs) | [191] |
| | Japan | 290 males, 160 females, 34 family trios | In-house (12 X-STRs) | [192] |
| | Japan (Fukuoka) | 93 male | Mentype Argus X-8 | [65] |
| | Japan (Tokoyo) | 283 males, 209 females | Mentype Argus X-8 | [193] |
| | Japan (Tokoyo), China (Shenyang) | 438 males, 232 females (Japan), 263 males, 225 females (China) | Investigator Argus X-12 | [72] |
| | Korea | 150 males, 150 females | In-house (5 X-STRs) | [194] |
| | Korea | 150 males, 150 females | In-house (4 X-STRs) | [195] |
| | Korea | 220 males, 181 females, 95 family trios | In-house (18 X-STRs) | [90] |
| | Korea | 41 families (138 individuals) | Mentype Argus X-8 | [196] |
| | Korea (Seoul) | 300 males, 150 females | Mentype Argus X-8 | [197] |
| | Korea (Seoul) | 300 males, 150 females | In-house (4 X-STRs) | [198] |
| | Malaysia (Kedayan, Borneo) | 127 males, 72 females | Investigator Argus X-12 QS | [199] |
| | Malaysia (Kuala Lumpur) | 160 males, 123 females | Investigator Argus X-12 | [117] |
| | Pakistan | 285 males, 147 females | In-house (13 X-STRs) | [200] |
| | Pakistan | 302 males | In-house (5 X-STRs) | [201] |
| | Pakistan (Baluchi and Pakhtun) | 250 males, 250 females | In-house (9 mini X-STRs) | [202] |
| | Pakistan (Punjabi) | 94 males, 118 females, 84 family trios | In-house (5 X-STRs) | [203] |
| | Pakistan (Punjab, Sindh) | 712 individuals | In-house (11 mini X-STRs) | [204] |
| | Philippines | 57 males, 58 females | In-house (5 X-STRs) | [121] |
| | Philippines | 143 males | Investigator Argus X-12 | [106] |
| | Saudi Arabia | 105 males, 95 females | Investigator Argus X-12 | [82] |
| | Sri Lanka (Sinhalese) | 120 males, 80 females | In-house (16 X-STRs) | [205] |
| | Sri Lanka (Sinhalese, Sri Lankan Tamils, Indian Tamils and Moors) | 838 individuals | In-house (16 X-STRs) | [206] |
| | Taiwan | 101 males, 99 females | In-house (5 X-STRs) | [207] |
| Taiwan | 327 males, 187 females | Investigator Argus X-12 | [208] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|------------------------|-----------------------------|---|----------------------------|-------------------------|
| Asia | Taiwan | 208 males, 197 females | In-house (11 X-STRs) | [209] |
| | Taiwan (Chinese) | N/A | In-house (6 X-STRs) | [210] |
| | Taiwan (Han) | 113 males, 108 females | In-house (13 X-STRs) | [211] |
| | Thailand | 116 males, 41 females | In-house (5 X-STRs) | [121] |
| | Thailand | 50 individuals | In-house (17 X-STRs) | [49] |
| | Thailand | 282 males, 109 females | Investigator Argus X-12 | [212] |
| | Thailand | 61 males, 150 females | Investigator Argus X-12 | [213] |
| | Turkey | 135 males, 129 females | In-house (5 X-STRs) | [214] |
| | Turkey | 175 males, 52 females | In-house (11 X-STRs) | [215] |
| | Turkey | 56 males, 73 females | Mentype Argus X-8 | [216] |
| | Turkey | 100 males, 91 females | Investigator Argus X-12 | [217] |
| | Turkey | 61 males | Investigator Argus X-12 | [184] |
| | United Arab Emirates | 501 males | Investigator Argus X-12 QS | [218] |
| | Europe | Albania | 180 males, 77 females | Investigator Argus X-12 |
| Belarus | | 360 males, 423 females | In-house (19 X-STRs) | [219] |
| Belarus, Slovakia | | 180 males (Belarus), 116 males (Slovakia) | In-house (4 X-STRs) | [220] |
| Bosnia and Herzegovina | | 86 males, 86 females | In-house (15 mini X-STRs) | [221] |
| Croatia | | 78 males, 99 females | Mentype Argus X-8 | [222] |
| Croatia | | 100 males, 100 females | Investigator Argus X-12 | [223] |
| Croatia | | 102 males, 100 females | Investigator Argus X-12 | [103] |
| Croatia | | 99 males, 98 females | Investigator Argus X-12 | [104] |
| Croatia | | 249 males, 148 females | Investigator Argus X-12 | [224] |
| Czech Republic | | 307 males, 216 females | Investigator Argus X-12 | [225] |
| Czech Republic | | 234 males, 197 females | GHEP-ISFG X-STR decaplex | [226] |
| Denmark | | 210 individuals | Investigator Argus X-12 | [83] |
| Finland | | 200 males, 100 females | Mentype Argus X-8 | [227] |
| Germany | | 60 family trios | In-house (4 X-STRs) | [228] |
| Germany | | N/A | In-house (5 X-STRs) | [229] |
| Germany | | 105 males, 100 females | In-house (10 X-STRs) | [230] |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|-----------|--|---|-------------------------|-----------|
| Europe | Germany | 50 males, 55 females | In-house (5 X-STRs) | [121] |
| | Germany | 574 individuals | In-house (4 X-STRs) | [91] |
| | Germany | N/A | In-house (16 X-STRs) | [231] |
| | Germany | 110 males, 107 females | In-house (5 X-STRs) | [183] |
| | Germany | 354 males, 184 females | In-house (6 X-STRs) | [232] |
| | Germany | 159 males, 159 females | Mentype Argus X-8 | [233] |
| | Germany | 1037 males | Investigator Argus X-12 | [234] |
| | Germany | 700 males | Investigator Argus X-12 | [235] |
| | Germany (Dresden) | 80 males, 52 females | In-house (7 X-STRs) | [236] |
| | Germany (Dresden, Leipzig, Magdeburg, Hamburg) | 439 male, 556 female | Mentype Argus X-8 | [65] |
| | Greece | 223 individuals | Investigator Argus X-12 | [237] |
| | Hungary | 219 males, 165 females, 96 family trios | Mentype Argus X-UL | [238] |
| | Hungary | 219 males, 165 females | Mentype Argus X-8 | [239] |
| | Hungary | 219 males, 188 females | Investigator Argus X-12 | [240] |
| | Iberia (western) | 244 males, 83 females | Mentype Argus X-8 | [241] |
| | Ireland | N/A | In-house (5 X-STRs) | [229] |
| | Italy (Tuscany) | 80 males, 80 females | In-house (4 X-STRs) | [242] |
| | Italy (Pavia) | 60 males, 60 females, 40 family trios | In-house (5 X-STRs) | [243] |
| | Italy (Bologna, Modena, Padova, Ancona, Pisa) | 288 males, 268 females | In-house (7 X-STRs) | [244] |
| | Italy (Brescia) | 30 males, 90 females | Mentype Argus X-UL | [245] |
| | Italy (Piedmont) | 80 males, 80 females | In-house (12 X-STRs) | [246] |
| | Italy | 100 males, 100 females | In-house (12 X-STRs) | [247] |
| | Italy (Brescia), Sardinia island | 131 individuals | Mentype Argus X-8 | [248] |
| | Italy (Umbria and Sardinia island) | 200 individuals | In-house (5 X-STRs) | [249] |
| | Italy (Calabria) | 25 males, 75 females | Mentype Argus X-8 | [250] |
| | Italy | 29 males, 147 females | Mentype Argus X-8 | [251] |
| Italy | 80 families | Mentype Argus X-8, In-house (12 X-STRs) | [252] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|---|---|---|--|-----------|
| Europe | Italy | 118 males, 89 females | Investigator Argus X-12 | [253] |
| | Italy | 200 males | Investigator Argus X-12 | [254] |
| | Sardinia | 318 males, 198 females | Investigator Argus X-12 | [255] |
| | Lativa (Riga, Liepaja, Ogre, Jelgava, Aizkraukle, Rezenkne, Preili, Balvi, Kuldiga, Madona) | 78 males, 45 females | In-house (10 X-STRs) | [256] |
| | Lithuania | 33 males, 77 females | Investigator Argus X-12 | [184] |
| | Poland | 120 males, 120 females | Mentype Argus X-UL | [257] |
| | Poland | 103 males, 101 females | Mentype Argus X-8 | [33] |
| | Poland | 60 males, 60 females | Mentype Argus X-8 | [258] |
| | Poland (Kuyavua-Pomerania) | 152 males, 159 females | Mentype Argus X-8, In-house (7 X-STRs) | [259] |
| | Poland (Old Believers) | 140 males, 70 females | Mentype Argus X-UL | [260] |
| | Portugal | 53 males, 48 females | Mentype Argus X-8 | [261] |
| | Portugal | 150 males | Investigator Argus X-12 | [262] |
| | Portugal | 150 males, 73 females | Investigator Argus X-12 | [263] |
| | Portugal (Cabo Verde, Lisboa) | 95 males, 54 females | Investigator Argus X-12 | [113] |
| | Portugal (Miranda do Douro) and Spain (Zamora) | 121 individuals (Portugal), 202 individuals (Spain) | Investigator Argus X-12 | [264] |
| | Portugal (Porto, Braga, Viana do Castelo, Vila Real, Bragança) | 347 males | In-house (10 X-STRs) | [265] |
| | Portugal, Spain | 1492 males, 1467 females (total) | GHEP-ISFG X-STR decaplex | [266] |
| | Russian Federation | 803 females | Investigator Argus X-12 | [86] |
| | Serbia | 604 males | In-house (10 X-STRs) | [267] |
| | Serbia (Vojvodina) | 220 males, 105 females | Investigator Argus X-12 | [268] |
| | Slovenia | 136 males, 21 females | Investigator Argus X-12 | [184] |
| | Spain | 63 males, 68 females | In-house (5 X-STRs) | [269] |
| | Spain (Cantabria and Basque Country) | 244 individuals | In-house (5 X-STRs) | [270] |
| Spain (Cantabria, Basque Country, Galicia, Castilla, Andalucia) | 324 males, 298 females | In-house (6 X-STRs) | [271] | |
| Spain (Castellón, Valencia, Alicante) | 145 females | In-house (10 X-STRs) | [272] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|-------------------------------|---|--|----------------------------|-----------|
| Europe | Spain (Cantabria, Pas Valley, Basque Country), Portugal | 252 males, 273 females | GHEP-ISFG X-STR decaplex | [273] |
| | Spain (Murcia) | 311 sets of female twins, 4 sets of triplets | GHEP-ISFG X-STR decaplex | [274] |
| | Spain | 192 males | In-house (10 X-STRs) | [275] |
| | Spanish Levant | 172 individuals | GHEP-ISFG X-STR decaplex | [276] |
| | Spain (Basque, Navarre) | 91 males, 94 females | GHEP-ISFG X-STR decaplex | [277] |
| | Spain | 50 individuals | In-house (17 X-STRs) | [49] |
| | Spain (Alicante, Basque Country, Andalusia, Galicia, Madrud, Barcelona) | 391 males, 202 females | In-house (17 X-STRs) | [278] |
| | Spain (Alicante) | 62 males, 59 females | Investigator Argus X-12 | [279] |
| | Sweden | 718 males, 106 females | Mentype Argus X-8 | [280] |
| | Sweden | 652 males | Investigator Argus X-12 | [112] |
| | Switzerland | 606 males, 592 females | Investigator Argus X-12 QS | [118] |
| | UK and Irish | 600 individuals | Mentype Argus X-UL | [281] |
| | Mediterranean | 672 individuals | In-house (15 X-STRs) | [282] |
| | Western Mediterranean (Valencia, Majorca, Minorca, Ibiza) | 160 males, 95 females | Investigator Argus X-12 | [283] |
| | Western Mediterranean | 716 individuals | Investigator Argus X-12 | [284] |
| South America | Argentina | 457 father-daughter duos | Investigator Argus X-12 | [285] |
| | Argentina | 419 individuals | GHEP-ISFG X-STR decaplex | [286] |
| | Argentina (Entre Rios) | 110 males | Investigator Argus X-12 | [287] |
| | Argentina (Salta) | 78 males, 100 females | Investigator Argus X-12 | [288] |
| | Argentina (Toba and Colla) | 72 males | GHEP-ISFG X-STR decaplex | [289] |
| | Argentina, Brazil, Colombia | 1492 males, 1467 females (total) | GHEP-ISFG X-STR decaplex | [266] |
| | Brazil | 1355 males, 879 females | In-house (12 X-STRs) | [290] |
| | Brazil (Santa Catarina) | 114 males, 70 females | Mentype Argus X-UL | [291] |
| | Brazil (Rio de Janeiro) | 129 males, 134 females | In-house (6 X-STRs) | [292] |
| | Brazilian Amazon (Pará City) | 182 males, 142 females | In-house (11 X-STRs) | [293] |
| | Brazil (Bauru, São Paulo) | 90 males and females | In-house (5 X-STRs) | [294] |
| Brazil (Espírito Santo State) | 77 males, 147 females | GHEP-ISFG X-STR decaplex | [295] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|----------------------|--|----------------------------------|--------------------------|-----------|
| South America | Brazil (Rio Grande do Sul) | 200 males | In-house (11 X-STRs) | [296] |
| | Brazil (Alagoas, Rio de Janeiro) | 423 males, 323 females | In-house (5 X-STRs) | [297] |
| | Brazil (Japanese) | 102 males, 130 females | In-house (12 X-STRs) | [298] |
| | Brazil (Santa Catarina) | 112 males, 72 females | GHEP-ISFG X-STR decaplex | [299] |
| | Brazil (São Paulo, Rio de Janeiro, Vitória and Belo Horizonte) | 424 males, 577 females | GHEP-ISFG X-STR decaplex | [300] |
| | Brazil (Rio Grande do Sul) | 141 males, 124 females | In-house (14 X-STRs) | [301] |
| | Brazil (Mato Grosso) | 174 males, 100 females | GHEP-ISFG X-STR decaplex | [302] |
| | Colombia (Antioquia, Cauca, Santander, Cundiboyacense) | 806 males | Investigator Argus X-12 | [303] |
| | Colombia | 50 individuals | In-house (17 X-STRs) | [49] |
| | Colombia (Chocó) | 285 individuals | GHEP-ISFG X-STR decaplex | [304] |
| | Colombia, Bolivar | 101 males, 89 females | GHEP-ISFG X-STR decaplex | [305] |
| | Colombia (Santander) | 108 males, 110 females | In-house (10 X-STRs) | [306] |
| | Ecuador | 72 males, 67 females | GHEP-ISFG X-STR decaplex | [307] |
| | Ecuador | 200 males | GHEP-ISFG X-STR decaplex | [308] |
| | Ecuador | 100 males, 100 females | Investigator Argus X-12 | [309] |
| | Ecuador (Imbabura) | 51 males, 49 females | GHEP-ISFG X-STR decaplex | [310] |
| | Ecuador (Mestizos) | 377 males, 209 females | Investigator Argus X-12 | [311] |
| | Ecuador (Pichincha) | 52 males, 48 females | GHEP-ISFG X-STR decaplex | [312] |
| | Peru | N/A | In-house (5 X-STRs) | [229] |
| | Peru (Ayacucho) | 203 individuals | Investigator Argus X-12 | [313] |
| Peru (Lima) | 141 males, 141 females | GHEP-ISFG X-STR decaplex | [314] | |
| North America | Costa Rica | 1492 males, 1467 females (total) | GHEP-ISFG X-STR decaplex | [266] |
| | Greenland | 198 individuals | Investigator Argus X-12 | [83] |
| | Mexico | 529 females | GHEP-ISFG X-STR decaplex | [315] |
| | Mexico (Mestizos and Amerindian) | 641 females | Investigator Argus X-12 | [316] |
| | Nicaragua | 164 males | GHEP-ISFG X-STR decaplex | [317] |
| | America (Native American and Mestizo) | 404 males, 237 females | In-house (17 X-STRs) | [318] |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|------------------------|---|---|---|------------------|
| North America | United States of America (African Americans, Asians, Hispanics) | 377 males | In-house (10 X-STRs) | [89] |
| | United States of America (African American, Asian, Caucasian, Hispanic) | 621 males, 742 females | In-house (15 mini X-STRs) | [50] |
| | United States of America (African American, Asian, Caucasian, Hispanic) | 663 males, 38 females | Investigator Argus X-12 | [87] |
| | United States of America (Basque Diaspora) | 40 males, 44 females (United States), 60 males, 112 females (Argentina) | GHEP-ISFG X-STR decaplex | [319] |
| | United States of America (African American, Caucasian, Hispanic) | 423 males, 723 females | In-house (15 mini X-STRs) | [320] |
| Africa | Algeria | 104 males, 106 females | In-house (12 X-STRs) | [321] |
| | Algeria | 104 males, 106 females | In-house (21 X-STRs) | [322] |
| | Angola, Mozambique, Uganda | 74 males (Angola), 112 males (Mozambique), 51 males (Uganda) | In-house (10 X-STRs) | [88] |
| | Eritrea | 255 males | Investigator Argus X-12 | [109] |
| | Ethiopia | N/A | In-house (5 X-STRs) | [229] |
| | Ethiopia (Tigray) | 248 individuals | Investigator Argus X-12 | [110] |
| | Egypt (Alexandria) | 250 males | Investigator Argus X-12 | [111] |
| | Ghana | 59 male | Mentype Argus X-8 | [65] |
| | Ghana (Ashanti) | 129 males, 114 females | In-house (11 X-STRs) | [323] |
| | Ghana (Ewe) | 74 males, 108 females | Mentype Argus X-8 | [324] |
| | Guinea-Bissau | 332 males | Investigator Argus X-12 | [108] |
| | Ivory Coast (Ouangolodougou) | 51 males, 74 females | Mentype Argus X-8, In-house (13 X-STRs) | [325] |
| | Malawi and Equatorial Guinea | 50 individuals | In-house (17 X-STRs) | [49] |
| | Morocco | 97 males, 48 females | Investigator Argus X-12 | [326] |
| | Morocco (Arabs, Berbers, Sahrawi) | 97 males | Investigator Argus X-12 | [327] |
| | Morocco (Casablanca) | 246 males | GHEP-ISFG X-STR decaplex | [328] |
| Morocco and Madagascar | 50 males, 50 females (Morocco), 107 males, 62 females (Madagascar) | In-house (11 X-STRs) | [329] | |

| Continent | Country (Region/Population) | Sample size used | Multiplex system used | Reference |
|------------------|---|--|----------------------------------|------------------|
| Africa | Nigeria (Igbo, Hausa and Yoruba) | 218 males | GHEP-ISFG X-STR decaplex | [330] |
| | Sierra Leone (Freetown) | 265 males, 285 females | Microreader 19X Direct ID System | [331] |
| | Somalia | 200 males, 100 females + 100 additional Somali individuals | Mentype Argus X-8 | [227] |
| | Somalia | 692 males | Investigator Argus X-12 | [332] |
| | Somalia | 441 individuals | Investigator Argus X-12 | [83] |
| | Tunisia (Nabeul) | 26 males, 113 females | In-house (17 X-STRs) | [80] |
| | Uganda (Karamoja) | 117 males, 138 females | GHEP-ISFG X-STR decaplex | [333] |
| Oceania | Australia (Aboriginal) | 298 males | Investigator Argus X-12 QS | [107] |
| Other | Jewish (Middle Eastern, Ashkenazi, Sephardic, North Africa) and Chuetas | 152 males, 161 females | Investigator Argus X-12 | [334] |
| | Jewish (Middle Eastern, Ashkenazi, Sephardic, North Africa) and Chuetas | 276 males, 224 females | Investigator Argus X-12 | [335] |
| | Atlantic Coast of Europe and North-West Africa | 500 males, 13 females | In-house (17 X-STRs) | [336] |

Appendix C:

Ethical approval letter



UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee



Room 45 E-52-E-Floor- Old Main Building
Groota Schuur Hospital
Observatory 7925
Telephone [021] 406 6492
Email: hrec-submissions@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

01 March 2022

HREC REF: 136/2022

Dr L Heathfield

Division of Forensic Medicine & Toxicology
Entrance 3 Level 1, Falmouth Building-FHS
Email: Laura.heathfield@uct.ac.za
Student: WHTAMY003@myuct.ac.za

Dear Dr Heathfield

PROJECT TITLE: INVESTIGATION INTO X-STR HAPLOTYPE FREQUENCIES FOR FORENSIC HUMAN IDENTIFICATION IN SOUTH AFRICA-MPHIL CANDIDATE-MISS AMY-LEIGH WHITTAKER-SUB-STUCY LINKED TO 342/2016

Thank you for submitting your study to the Faculty of Health Sciences Human Research Ethics Committee (HREC) for review.

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study.

This approval is subject to strict adherence to the HREC recommendations regarding research involving human participants during COVID -19, our letter dated 02 February 2022 provides guidance found on our website:
<http://www.health.uct.ac.za/fhs/research/humanethics/forms>

Approval is granted for one year until the 30 March 2023.

Please submit a progress form, using the standardised Annual Report Form if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed within the approval period.

(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

The HREC acknowledges that the student: Miss Amy-Leigh Whittaker will also be involved in this study.

Please quote the HREC REF 136/2022 in all your correspondence.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate institutional approval, where necessary, before the research may occur.

Yours sincerely



PROFESSOR M BLOCKMAN

CHAIRPERSON, FACULTY OF HEALTH SCIENCES HUMAN RESEARCH ETHICS COMMITTEE

Federal Wide Assurance Number: FWA00001637. Institutional Review Board (IRB) number:

IRB00001938 NHREC-registration number: REC-210208-007

This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Council for Harmonisation of Technical Requirements for Pharmaceuticals for Human Use: Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2020), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI), and Declaration of Helsinki (2013) guidelines. The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.

Appendix D:

Table D1 Non-concordant alleles identified between the Investigator Argus X-12 QS kit (Qiagen, Hilden) and the ForenSeq™ DNA Signature Prep kit (Illumina, CA, USA).

| Locus | Investigator Argus X-12 QS kit allele called | Forenseq DNA Signature Prep kit allele called |
|-----------------|--|---|
| DXS10135 | 23.1 | 23 |
| | 34.1 | 34 |
| | 29 | 29.1 |
| DXS10074 | 14.2 | 14 |
| | 17 | 16 |
| DXS8378 | 11 | 12 |

Appendix E:

Table E1 Exact test for population differentiation per locus between allele frequencies for male and female individuals using 100000 steps in the Markov chain. Significance level = 0.05.

| Locus | p-value |
|-----------------|------------------|
| DXS8378 | 0.71555 ± 0.0235 |
| DXS10135 | 0.88583 ± 0.0114 |
| DXS10148 | 0.93992 ± 0.0091 |
| DXS7132 | 0.65209 ± 0.0149 |
| DXS10074 | 0.93204 ± 0.0083 |
| DXS10079 | 0.65388 ± 0.0191 |
| DXS10101 | 0.89960 ± 0.0095 |
| DXS10103 | 0.54168 ± 0.0149 |
| HPRTB | 0.72595 ± 0.0155 |
| DXS7423 | 0.86183 ± 0.0081 |
| DXS10134 | 0.50997 ± 0.0253 |
| DXS10146 | 0.41480 ± 0.0226 |

Appendix F:

Table F1 Pooled allele frequencies and forensic parameters for 781 South African individuals.

| Allele | DXS8378 | DXS10135 | DXS10148 | DXS7132 | DXS10074 | DXS10079 | HPRTB | DXS10101 | DXS10103 | DXS7423 | DXS10134 | DXS10146 | Combined |
|-------------|---------|----------|----------|---------|----------|----------|--------|----------|----------|---------|----------|----------|----------|
| 7 | | | | | 0.045 | | | | | | | | |
| 8 | 0.0029 | | | | 0.0603 | | | | | 0.0019 | | | |
| 9 | 0.0278 | | | | 0.0067 | | 0.0029 | | | | | | |
| 10 | 0.2096 | | | 0.001 | 0.0029 | | 0.0067 | | | | | | |
| 11 | 0.3828 | | | 0.0153 | 0.0096 | | 0.0938 | | | | | | |
| 11.2 | | | | | | | 0.001 | | | | | | |
| 12 | 0.3292 | | | 0.0871 | 0.0364 | | 0.3502 | | | 0.0048 | | | |
| 13 | 0.045 | | | 0.2517 | 0.0402 | 0.0105 | 0.3033 | | | 0.0775 | | | |
| 13.2 | | | | | 0.0019 | | | | | | | | |
| 13.3 | | | 0.0354 | | | | | | | | | | |
| 14 | 0.0029 | 0.001 | 0.001 | 0.3378 | 0.0565 | 0.0019 | 0.1732 | | | 0.3971 | | | |
| 14.2 | | | | | 0.001 | | 0.001 | | | | | | |
| 15 | | | | 0.2498 | 0.1397 | 0.0278 | 0.0555 | | 0.0077 | 0.3732 | | | |
| 15.3 | | | | | 0.001 | | | | | | | | |
| 16 | | 0.0096 | 0.0019 | 0.044 | 0.199 | 0.0201 | 0.0124 | | 0.1483 | 0.1263 | | | |
| 16.1 | | 0.0048 | | | | | | | | | | | |
| 16.3 | | | 0.001 | | 0.0048 | | | | | | | | |
| 17 | | 0.0153 | 0.0124 | 0.0115 | 0.1828 | 0.0699 | | | 0.0842 | 0.0191 | | | |
| 17.1 | | 0.0096 | | | | | | | | | | | |
| 17.3 | | | | | 0.0019 | | | | | | | | |
| 18 | | 0.0517 | 0.1282 | 0.0019 | 0.1388 | 0.1407 | | | 0.2182 | | | | |
| 18.1 | | 0.0287 | | | | | | | | | | | |
| 18.2 | | | 0.001 | | | | | | | | | | |
| 18.3 | | | | | 0.0019 | | | | | | | | |
| 19 | | 0.0498 | 0.0383 | | 0.0545 | 0.2364 | | | 0.3914 | | | | |

| Allele | DXS8378 | DXS10135 | DXS10148 | DXS7132 | DXS10074 | DXS10079 | HPRTB | DXS10101 | DXS10103 | DXS7423 | DXS10134 | DXS10146 | Combined |
|--------|---------|----------|----------|---------|----------|----------|-------|----------|----------|---------|----------|----------|----------|
| 19.1 | | 0.0201 | | | | | | | | | | | |
| 19.3 | | | 0.001 | | | | | | | | | | |
| 20 | | 0.0421 | 0.0163 | | 0.0144 | 0.2239 | | | 0.1292 | | | | |
| 20.1 | | 0.0201 | 0.0096 | | | | | | | | | | |
| 20.3 | | 0.001 | 0.001 | | | | | | | | | | |
| 21 | | 0.0775 | 0.0077 | | 0.001 | 0.1799 | | | 0.0201 | | | 0.001 | |
| 21.1 | | 0.0325 | 0.001 | | | | | | | | | | |
| 21.2 | | | | | | 0.0029 | | | | | | | |
| 22 | | 0.0689 | 0.0019 | | | 0.0641 | | | 0.001 | | | | |
| 22.1 | | 0.0182 | 0.0144 | | | | | | | | | 0.0019 | |
| 22.2 | | | | | | | | 0.001 | | | | | |
| 22.3 | | | 0.001 | | | | | | | | | | |
| 23 | | 0.0651 | 0.0249 | | | 0.0144 | | | | | | 0.001 | |
| 23.1 | | 0.0134 | 0.0622 | | | | | | | | | | |
| 24 | | 0.0699 | 0.023 | | | 0.0057 | | 0.001 | | | | 0.0182 | |
| 24.1 | | 0.0057 | 0.0842 | | | | | | | | | | |
| 24.2 | | | | | | | | 0.0124 | | | | | |
| 24.3 | | | 0.001 | | | | | | | | | | |
| 25 | | 0.0584 | 0.0124 | | | 0.0019 | | 0.001 | | | | 0.0488 | |
| 25.1 | | 0.0019 | 0.1254 | | | | | | | | | | |
| 25.2 | | | | | | | | 0.001 | | | | | |
| 26 | | 0.0555 | 0.0153 | | | | | 0.0038 | | | | 0.0852 | |
| 26.1 | | | 0.0861 | | | | | | | | 0.001 | | |
| 26.2 | | | | | | | | 0.0153 | | | | 0.001 | |
| 26.3 | | 0.0029 | 0.001 | | | | | | | | | | |
| 27 | | 0.0603 | 0.0268 | | | | | 0.0048 | | | | 0.1033 | |
| 27.1 | | 0.0019 | 0.0632 | | | | | | | | 0.001 | | |
| 27.2 | | | | | | | | 0.0373 | | | | | |
| 27.3 | | 0.001 | | | | | | | | | | 0.001 | |

| Allele | DXS8378 | DXS10135 | DXS10148 | DXS7132 | DXS10074 | DXS10079 | HPRTB | DXS10101 | DXS10103 | DXS7423 | DXS10134 | DXS10146 | Combined |
|--------|---------|----------|----------|---------|----------|----------|-------|----------|----------|---------|----------|----------|----------|
| 28 | | 0.0459 | 0.0191 | | | | | 0.0526 | | | 0.0029 | 0.155 | |
| 28.1 | | 0.0019 | 0.0383 | | | | | 0.001 | | | | | |
| 28.2 | | | 0.0019 | | | | | 0.0785 | | | 0.001 | | |
| 28.3 | | 0.0048 | 0.001 | | | | | | | | | | |
| 29 | | 0.0383 | 0.0211 | | | | | 0.0536 | | | 0.001 | 0.1407 | |
| 29.1 | | | 0.0211 | | | | | | | | 0.001 | | |
| 29.2 | | | 0.0019 | | | | | 0.0746 | | | 0.001 | 0.0019 | |
| 29.3 | | 0.0019 | | | | | | | | | | | |
| 30 | | 0.0344 | 0.0105 | | | | | 0.0679 | | | 0.0086 | 0.0909 | |
| 30.1 | | 0.001 | 0.0105 | | | | | | | | 0.0019 | 0.001 | |
| 30.2 | | | | | | | | 0.1072 | | | | | |
| 31 | | 0.0191 | 0.0077 | | | | | 0.1321 | | | 0.0048 | 0.0785 | |
| 31.1 | | | 0.0048 | | | | | | | | | | |
| 31.2 | | 0.0019 | | | | | | 0.0689 | | | | 0.0038 | |
| 31.3 | | | 0.001 | | | | | 0.0019 | | | | 0.001 | |
| 32 | | 0.0086 | 0.0067 | | | | | 0.134 | | | 0.0364 | 0.0411 | |
| 32.1 | | 0.0057 | 0.0096 | | | | | | | | 0.001 | | |
| 32.2 | | | | | | | | 0.0325 | | | | 0.0019 | |
| 32.3 | | | 0.001 | | | | | | | | | | |
| 33 | | 0.0086 | 0.0029 | | | | | 0.0833 | | | 0.0632 | 0.0172 | |
| 33.1 | | 0.0019 | | | | | | 0.001 | | | | | |
| 33.2 | | | | | | | | 0.0105 | | | 0.001 | 0.0086 | |
| 33.3 | | | | | | | | | | | 0.0038 | | |
| 34 | | 0.0077 | | | | | | 0.0172 | | | 0.1215 | 0.0057 | |
| 34.1 | | 0.0019 | | | | | | | | | | | |
| 34.2 | | | | | | | | 0.001 | | | 0.0038 | 0.0258 | |
| 34.3 | | | | | | | | | | | 0.0067 | | |
| 35 | | 0.0057 | | | | | | 0.0048 | | | 0.1646 | 0.0019 | |
| 35.1 | | 0.0019 | 0.0038 | | | | | | | | 0.001 | | |

| Allele | DXS8378 | DXS10135 | DXS10148 | DXS7132 | DXS10074 | DXS10079 | HPRTB | DXS10101 | DXS10103 | DXS7423 | DXS10134 | DXS10146 | Combined |
|--------|---------|----------|----------|---------|----------|----------|-------|----------|----------|---------|----------|----------|----------|
| 35.2 | | 0.001 | | | | | | | | | | 0.0067 | |
| 35.3 | | | | | | | | | | | 0.001 | | |
| 36 | | 0.0038 | | | | | | | | | 0.1904 | 0.0019 | |
| 36.1 | | | 0.001 | | | | | | | | | | |
| 36.2 | | 0.0019 | | | | | | | | | 0.0019 | 0.0191 | |
| 36.3 | | | | | | | | | | | 0.0048 | | |
| 37 | | 0.0048 | | | | | | | | | 0.1579 | | |
| 37.1 | | 0.0019 | 0.001 | | | | | | | | | | |
| 37.2 | | 0.0038 | | | | | | | | | 0.0067 | 0.0067 | |
| 37.3 | | | | | | | | | | | 0.0077 | | |
| 38 | | | | | | | | | | | 0.0861 | 0.001 | |
| 38.1 | | | 0.001 | | | | | | | | | | |
| 38.2 | | 0.001 | | | | | | | | | 0.0029 | 0.0153 | |
| 38.3 | | | | | | | | | | | 0.0077 | | |
| 39 | | | | | | | | | | | 0.0392 | | |
| 39.1 | | | 0.0086 | | | | | | | | | | |
| 39.2 | | 0.0029 | | | | | | | | | | 0.0153 | |
| 39.3 | | | | | | | | | | | 0.0096 | | |
| 40 | | | 0.001 | | | | | | | | 0.0096 | | |
| 40.1 | | | 0.0067 | | | | | | | | | | |
| 40.2 | | 0.001 | | | | | | | | | | 0.0258 | |
| 40.3 | | | | | | | | | | | 0.022 | 0.001 | |
| 41 | | | | | | | | | | | 0.001 | | |
| 41.1 | | | 0.0115 | | | | | | | | | | |
| 41.2 | | | | | | | | | | | | 0.0086 | |
| 41.3 | | | | | | | | | | | 0.0124 | | |
| 42.1 | | | 0.0067 | | | | | | | | | | |
| 42.2 | | | | | | | | | | | | 0.0153 | |
| 42.3 | | | | | | | | | | | 0.0057 | | |

| Allele | DXS8378 | DXS10135 | DXS10148 | DXS7132 | DXS10074 | DXS10079 | HPRTB | DXS10101 | DXS10103 | DXS7423 | DXS10134 | DXS10146 | Combined |
|-------------------|---------|----------|----------|---------|----------|----------|-------|----------|----------|---------|----------|----------|----------|
| 43.1 | | | 0.001 | | | | | | | | | | |
| 43.2 | | | | | | | | | | | | 0.0115 | |
| 43.3 | | | | | | | | | | | 0.0067 | | |
| 44.1 | | | 0.001 | | | | | | | | | | |
| 44.2 | | | | | | | | | | | | 0.0144 | |
| 45.2 | | | | | | | | | | | | 0.0105 | |
| 46.2 | | | | | | | | | | | | 0.0057 | |
| 47.2 | | | | | | | | | | | | 0.001 | |
| Null allele | | | | | | | | | | | | 0.0038 | |
| MEC_Kruger | 0.440 | 0.907 | 0.874 | 0.523 | 0.747 | 0.666 | 0.516 | 0.831 | 0.541 | 0.424 | 0.764 | 0.832 | 1 |
| MEC_Kishida | 0.643 | 0.952 | 0.934 | 0.709 | 0.861 | 0.810 | 0.702 | 0.910 | 0.719 | 0.623 | 0.871 | 0.910 | 1 |
| MEC_Desmarais | 0.643 | 0.952 | 0.933 | 0.709 | 0.860 | 0.810 | 0.702 | 0.910 | 0.718 | 0.623 | 0.870 | 0.910 | 1 |
| MEC_Desmarais_Duo | 0.498 | 0.910 | 0.878 | 0.571 | 0.766 | 0.696 | 0.564 | 0.841 | 0.582 | 0.478 | 0.781 | 0.842 | 1 |

Abbreviations – MEC: mean exclusion chance.

Appendix G:

Table G1 Hardy-Weinberg equilibrium test for female samples. Significance level = 0.0042.

| Locus | Observed Heterozygosity | Expected Heterozygosity | p-value |
|-----------------|-------------------------|-------------------------|----------------|
| DXS8378 | 0.73485 | 0.69691 | 0.30851 |
| DXS10135 | 0.95455 | 0.95395 | 0.00878 |
| DXS10148 | 0.88636 | 0.9284 | 0.00009 |
| DXS7132 | 0.72727 | 0.75265 | 0.62951 |
| DXS10074 | 0.89015 | 0.87666 | 0.34980 |
| DXS10079 | 0.84848 | 0.83867 | 0.53975 |
| HPRTB | 0.73485 | 0.73787 | 0.38804 |
| DXS10101 | 0.93561 | 0.91888 | 0.18347 |
| DXS10103 | 0.67803 | 0.74371 | 0.06018 |
| DXS7423 | 0.67803 | 0.68759 | 0.13571 |
| DXS10134 | 0.89773 | 0.8798 | 0.30600 |
| DXS10146 | 0.91288 | 0.90864 | 0.03020 |

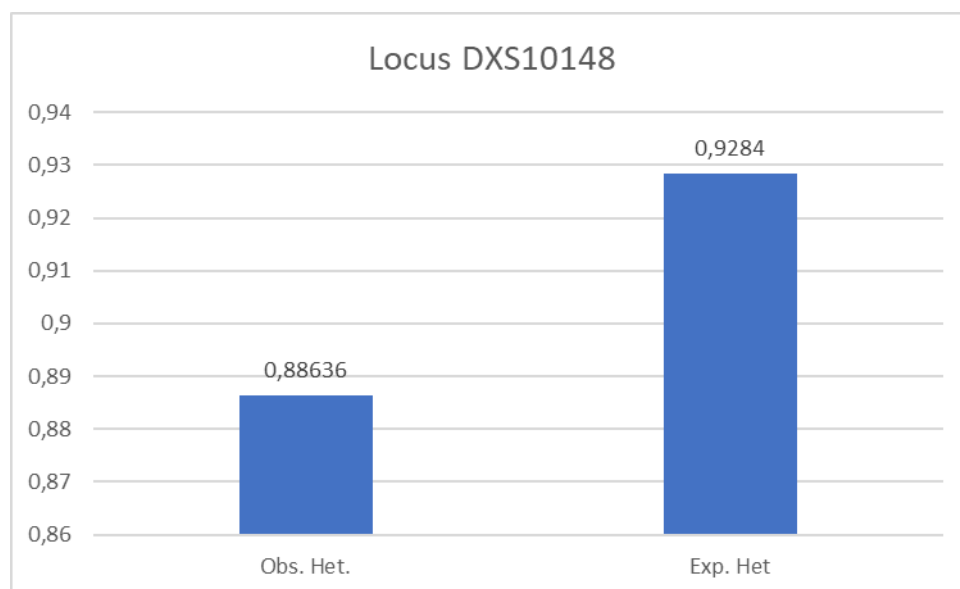


Fig. G1. Bar graph illustrating the significant difference between observed and expected heterozygosity at locus DXS10148 ($p < 0.0042$).

Appendix H:

Table H1 Haplotype frequencies for 517 South African males.

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 10 16 29.1 | 1 | 0.0019 | 10 18 20 | 1 | 0.0019 | 10 24.2 16 | 1 | 0.0019 | 12 35 38.2 | 2 | 0.0039 |
| 10 16 40.1 | 1 | 0.0019 | 11 12 17 | 1 | 0.0019 | 10 28.2 20 | 1 | 0.0019 | 13 32 30.1 | 1 | 0.0019 |
| 10 16 42.1 | 1 | 0.0019 | 11 13 20 | 1 | 0.0019 | 10 32 18 | 1 | 0.0019 | 13 34 27 | 1 | 0.0019 |
| 10 18 22.1 | 1 | 0.0019 | 11 14 18 | 2 | 0.0039 | 10 32 20 | 1 | 0.0019 | 13 34 28 | 1 | 0.0019 |
| 10 18 26.1 | 3 | 0.0058 | 11 14 19 | 1 | 0.0019 | 11 26.2 18 | 1 | 0.0019 | 13 34 29 | 1 | 0.0019 |
| 10 18 30.1 | 1 | 0.0019 | 11 15 19 | 1 | 0.0019 | 11 27.2 19 | 1 | 0.0019 | 13 34 30 | 1 | 0.0019 |
| 10 18.1 13.3 | 1 | 0.0019 | 11 17 20 | 1 | 0.0019 | 11 28.1 19 | 1 | 0.0019 | 13 35 27 | 3 | 0.0058 |
| 10 18.1 25.1 | 1 | 0.0019 | 11 17 21 | 1 | 0.0019 | 11 28.2 16 | 1 | 0.0019 | 13 35 29 | 2 | 0.0039 |
| 10 18.1 29.1 | 2 | 0.0039 | 11 18 20 | 1 | 0.0019 | 11 28.2 18 | 3 | 0.0058 | 13 35 30 | 2 | 0.0039 |
| 10 18.1 30.1 | 1 | 0.0019 | 12 12 19 | 1 | 0.0019 | 11 28.2 19 | 2 | 0.0039 | 13 36 25 | 3 | 0.0058 |
| 10 18.1 31.1 | 1 | 0.0019 | 12 12 22 | 1 | 0.0019 | 11 28.2 21 | 3 | 0.0058 | 13 36 26 | 1 | 0.0019 |
| 10 19 22.1 | 1 | 0.0019 | 12 14 19 | 4 | 0.0077 | 11 29.2 19 | 9 | 0.0174 | 13 36 27 | 2 | 0.0039 |
| 10 19 23.1 | 1 | 0.0019 | 12 14 20 | 1 | 0.0019 | 11 29.2 20 | 1 | 0.0019 | 13 36 28 | 1 | 0.0019 |
| 10 19.1 28.1 | 1 | 0.0019 | 12 14 22 | 1 | 0.0019 | 11 30 17 | 1 | 0.0019 | 13 36 39.2 | 1 | 0.0019 |
| 10 19.1 28.2 | 1 | 0.0019 | 12 15 19 | 1 | 0.0019 | 11 30 18 | 2 | 0.0039 | 13 37 25 | 1 | 0.0019 |
| 10 19.1 44.1 | 1 | 0.0019 | 12 15 20 | 1 | 0.0019 | 11 30 19 | 1 | 0.0019 | 13 37 26 | 3 | 0.0058 |
| 10 20 28 | 1 | 0.0019 | 12 15 22 | 1 | 0.0019 | 11 30.2 18 | 1 | 0.0019 | 13 37 29 | 1 | 0.0019 |
| 10 20.1 32.1 | 1 | 0.0019 | 12 16 17 | 2 | 0.0039 | 11 30.2 19 | 2 | 0.0039 | 13 37 34.2 | 1 | 0.0019 |
| 10 21 18 | 4 | 0.0077 | 12 16 19 | 2 | 0.0039 | 11 31 17 | 1 | 0.0019 | 13 37 35.2 | 1 | 0.0019 |
| 10 21 24.1 | 1 | 0.0019 | 12 16 20 | 4 | 0.0077 | 11 31 18 | 2 | 0.0039 | 13 38 25 | 1 | 0.0019 |
| 10 21 25.1 | 1 | 0.0019 | 12 16 21 | 3 | 0.0058 | 11 31 19 | 2 | 0.0039 | 13 38 29 | 1 | 0.0019 |
| 10 21 26 | 2 | 0.0039 | 12 16 23 | 1 | 0.0019 | 11 31 20 | 2 | 0.0039 | 13 38 30 | 2 | 0.0039 |
| 10 21 26.1 | 1 | 0.0019 | 12 16.3 22 | 1 | 0.0019 | 11 31.2 18 | 1 | 0.0019 | 13 38 31 | 1 | 0.0019 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 10 21 27 | 1 | 0.0019 | 12 17 18 | 1 | 0.0019 | 11 31.2 19 | 3 | 0.0058 | 13 38 36.2 | 1 | 0.0019 |
| 10 21 27.1 | 1 | 0.0019 | 12 17 19 | 1 | 0.0019 | 11 31.2 20 | 1 | 0.0019 | 13 38.3 44.2 | 1 | 0.0019 |
| 10 21 28.1 | 1 | 0.0019 | 12 17 20 | 2 | 0.0039 | 11 32 17 | 1 | 0.0019 | 13 39 28 | 1 | 0.0019 |
| 10 21.1 13.3 | 1 | 0.0019 | 12 17 21 | 2 | 0.0039 | 11 32 18 | 2 | 0.0039 | 13 39 40.2 | 1 | 0.0019 |
| 10 21.1 18 | 1 | 0.0019 | 12 17.3 20 | 1 | 0.0019 | 11 32 19 | 1 | 0.0019 | 13 39 46.2 | 1 | 0.0019 |
| 10 21.1 23.1 | 1 | 0.0019 | 12 18 19 | 4 | 0.0077 | 11 32 20 | 2 | 0.0039 | 13 40.3 27 | 1 | 0.0019 |
| 10 21.1 24.1 | 1 | 0.0019 | 12 18 21 | 1 | 0.0019 | 11 33 16 | 1 | 0.0019 | 13 41.3 40.2 | 1 | 0.0019 |
| 10 22 18 | 1 | 0.0019 | 12 19 15 | 1 | 0.0019 | 11 33 18 | 2 | 0.0039 | 14 29.1 27 | 1 | 0.0019 |
| 10 22 23 | 1 | 0.0019 | 12 19 17 | 1 | 0.0019 | 11 33 19 | 1 | 0.0019 | 14 30 31 | 1 | 0.0019 |
| 10 22 23.1 | 1 | 0.0019 | 12 19 18 | 2 | 0.0039 | 11 34 16 | 1 | 0.0019 | 14 30 32 | 1 | 0.0019 |
| 10 22 24.1 | 1 | 0.0019 | 12 19 20 | 2 | 0.0039 | 11 34 17 | 1 | 0.0019 | 14 32 29 | 2 | 0.0039 |
| 10 22 25 | 1 | 0.0019 | 12 7 19 | 2 | 0.0039 | 11 34 18 | 1 | 0.0019 | 14 32 29.2 | 1 | 0.0019 |
| 10 22 25.1 | 2 | 0.0039 | 12 8 18 | 1 | 0.0019 | 11.2 33.2 18 | 1 | 0.0019 | 14 32 30 | 1 | 0.0019 |
| 10 22 26.1 | 1 | 0.0019 | 13 10 19 | 1 | 0.0019 | 12 22.2 19 | 1 | 0.0019 | 14 32 31.2 | 1 | 0.0019 |
| 10 22 27.1 | 1 | 0.0019 | 13 11 21 | 1 | 0.0019 | 12 24.2 18 | 2 | 0.0039 | 14 32 32 | 2 | 0.0039 |
| 10 22 29 | 1 | 0.0019 | 13 12 19 | 3 | 0.0058 | 12 24.2 19 | 2 | 0.0039 | 14 32 36.2 | 1 | 0.0019 |
| 10 22 29.1 | 1 | 0.0019 | 13 12 20 | 5 | 0.0097 | 12 25 18 | 1 | 0.0019 | 14 32 43.2 | 1 | 0.0019 |
| 10 22.1 28.1 | 1 | 0.0019 | 13 13 16 | 1 | 0.0019 | 12 25.2 18 | 1 | 0.0019 | 14 32.1 27 | 1 | 0.0019 |
| 10 22.1 32.1 | 1 | 0.0019 | 13 13 19 | 1 | 0.0019 | 12 26 18 | 1 | 0.0019 | 14 33 25 | 1 | 0.0019 |
| 10 22.1 39.1 | 1 | 0.0019 | 13 13 20 | 1 | 0.0019 | 12 26.2 16 | 4 | 0.0077 | 14 33 26 | 1 | 0.0019 |
| 10 23 18 | 1 | 0.0019 | 13 13 21 | 1 | 0.0019 | 12 26.2 18 | 1 | 0.0019 | 14 33 27 | 3 | 0.0058 |
| 10 23 20 | 1 | 0.0019 | 13 13 22 | 1 | 0.0019 | 12 26.2 20 | 1 | 0.0019 | 14 33 28 | 2 | 0.0039 |
| 10 23 23 | 1 | 0.0019 | 13 14 19 | 2 | 0.0039 | 12 27 20 | 1 | 0.0019 | 14 33 29 | 2 | 0.0039 |
| 10 23 24.1 | 1 | 0.0019 | 13 14 20 | 1 | 0.0019 | 12 27.2 19 | 5 | 0.0097 | 14 33 30 | 2 | 0.0039 |
| 10 23 25.1 | 2 | 0.0039 | 13 14 21 | 1 | 0.0019 | 12 27.2 20 | 3 | 0.0058 | 14 33 31 | 2 | 0.0039 |
| 10 23 27 | 1 | 0.0019 | 13 14 21.2 | 1 | 0.0019 | 12 28 17 | 1 | 0.0019 | 14 33 32 | 1 | 0.0019 |
| 10 23 28.1 | 1 | 0.0019 | 13 14 22 | 1 | 0.0019 | 12 28 18 | 6 | 0.0116 | 14 33 38.2 | 2 | 0.0039 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 10 23 29.1 | 1 | 0.0019 | 13 15 13 | 2 | 0.0039 | 12 28 19 | 11 | 0.0213 | 14 34 25 | 4 | 0.0077 |
| 10 23 32.1 | 1 | 0.0019 | 13 15 17 | 1 | 0.0019 | 12 28 20 | 2 | 0.0039 | 14 34 26 | 8 | 0.0155 |
| 10 23.1 25.1 | 1 | 0.0019 | 13 15 18 | 2 | 0.0039 | 12 28.2 16 | 3 | 0.0058 | 14 34 27 | 1 | 0.0019 |
| 10 24 18 | 1 | 0.0019 | 13 15 19 | 5 | 0.0097 | 12 28.2 17 | 1 | 0.0019 | 14 34 29 | 5 | 0.0097 |
| 10 24 19 | 1 | 0.0019 | 13 15 20 | 6 | 0.0116 | 12 28.2 18 | 1 | 0.0019 | 14 34 30 | 3 | 0.0058 |
| 10 24 23 | 2 | 0.0039 | 13 15 21 | 2 | 0.0039 | 12 28.2 19 | 8 | 0.0155 | 14 34 31 | 2 | 0.0039 |
| 10 24 23.1 | 1 | 0.0019 | 13 15 22 | 1 | 0.0019 | 12 28.2 20 | 4 | 0.0077 | 14 34 34.2 | 1 | 0.0019 |
| 10 24 24.1 | 1 | 0.0019 | 13 16 13 | 1 | 0.0019 | 12 29 18 | 1 | 0.0019 | 14 34 40.2 | 1 | 0.0019 |
| 10 24 25 | 1 | 0.0019 | 13 16 16 | 1 | 0.0019 | 12 29 20 | 1 | 0.0019 | 14 34 44.2 | 1 | 0.0019 |
| 10 24 25.1 | 2 | 0.0039 | 13 16 18 | 3 | 0.0058 | 12 29.2 16 | 1 | 0.0019 | 14 34.3 31 | 1 | 0.0019 |
| 10 24 28 | 1 | 0.0019 | 13 16 19 | 3 | 0.0058 | 12 29.2 19 | 5 | 0.0097 | 14 34.3 44.2 | 1 | 0.0019 |
| 10 24 28.1 | 1 | 0.0019 | 13 16 20 | 8 | 0.0155 | 12 29.2 21 | 1 | 0.0019 | 14 35 24 | 2 | 0.0039 |
| 10 24 29.1 | 1 | 0.0019 | 13 16 21 | 7 | 0.0135 | 12 30 17 | 1 | 0.0019 | 14 35 25 | 1 | 0.0019 |
| 10 24 42.1 | 1 | 0.0019 | 13 16 21.2 | 1 | 0.0019 | 12 30 18 | 8 | 0.0155 | 14 35 26 | 2 | 0.0039 |
| 10 24.1 18 | 1 | 0.0019 | 13 16 22 | 1 | 0.0019 | 12 30 19 | 2 | 0.0039 | 14 35 27 | 1 | 0.0019 |
| 10 25 23.1 | 1 | 0.0019 | 13 16 23 | 2 | 0.0039 | 12 30 20 | 2 | 0.0039 | 14 35 28 | 12 | 0.0232 |
| 10 25 24.1 | 1 | 0.0019 | 13 16.3 20 | 1 | 0.0019 | 12 30.2 16 | 1 | 0.0019 | 14 35 29 | 2 | 0.0039 |
| 10 25 25.1 | 2 | 0.0039 | 13 17 15 | 1 | 0.0019 | 12 30.2 18 | 3 | 0.0058 | 14 35 30 | 2 | 0.0039 |
| 10 25 26.1 | 1 | 0.0019 | 13 17 17 | 2 | 0.0039 | 12 30.2 19 | 12 | 0.0232 | 14 35 31 | 6 | 0.0116 |
| 10 25 28 | 1 | 0.0019 | 13 17 18 | 3 | 0.0058 | 12 30.2 20 | 2 | 0.0039 | 14 35 32 | 1 | 0.0019 |
| 10 26 23.1 | 1 | 0.0019 | 13 17 19 | 5 | 0.0097 | 12 30.2 21 | 1 | 0.0019 | 14 35 34.2 | 2 | 0.0039 |
| 10 26 24.1 | 1 | 0.0019 | 13 17 20 | 7 | 0.0135 | 12 31 16 | 3 | 0.0058 | 14 35 35.2 | 2 | 0.0039 |
| 10 26 26.1 | 1 | 0.0019 | 13 17 21 | 5 | 0.0097 | 12 31 18 | 10 | 0.0193 | 14 35 40.2 | 2 | 0.0039 |
| 10 27 17 | 1 | 0.0019 | 13 18 15 | 2 | 0.0039 | 12 31 19 | 8 | 0.0155 | 14 36 24 | 1 | 0.0019 |
| 10 27 18 | 1 | 0.0019 | 13 18 18 | 2 | 0.0039 | 12 31 20 | 1 | 0.0019 | 14 36 25 | 3 | 0.0058 |
| 10 27 19 | 1 | 0.0019 | 13 18 19 | 4 | 0.0077 | 12 31.2 16 | 1 | 0.0019 | 14 36 26 | 1 | 0.0019 |
| 10 27 24.1 | 1 | 0.0019 | 13 18 20 | 1 | 0.0019 | 12 31.2 18 | 2 | 0.0039 | 14 36 27 | 6 | 0.0116 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 10 27 26.1 | 3 | 0.0058 | 13 18 21 | 3 | 0.0058 | 12 31.2 19 | 8 | 0.0155 | 14 36 28 | 10 | 0.0193 |
| 10 27 28.1 | 1 | 0.0019 | 13 18 22 | 2 | 0.0039 | 12 31.2 20 | 4 | 0.0077 | 14 36 29 | 7 | 0.0135 |
| 10 27 43.1 | 1 | 0.0019 | 13 18 23 | 1 | 0.0019 | 12 31.3 20 | 1 | 0.0019 | 14 36 30 | 1 | 0.0019 |
| 10 28 23.1 | 1 | 0.0019 | 13 18.3 18 | 1 | 0.0019 | 12 32 16 | 3 | 0.0058 | 14 36 31 | 5 | 0.0097 |
| 10 28 26.1 | 1 | 0.0019 | 13 19 17 | 1 | 0.0019 | 12 32 17 | 3 | 0.0058 | 14 36 32 | 1 | 0.0019 |
| 10 29 26 | 1 | 0.0019 | 13 19 20 | 2 | 0.0039 | 12 32 18 | 3 | 0.0058 | 14 36 32.2 | 1 | 0.0019 |
| 10 29 27.1 | 1 | 0.0019 | 13 20 19 | 1 | 0.0019 | 12 32 19 | 4 | 0.0077 | 14 36 33 | 2 | 0.0039 |
| 10 29 28 | 1 | 0.0019 | 13 20 20 | 2 | 0.0039 | 12 32.2 16 | 1 | 0.0019 | 14 36 33.2 | 1 | 0.0019 |
| 10 30 18 | 1 | 0.0019 | 13 20 22 | 1 | 0.0019 | 12 32.2 17 | 1 | 0.0019 | 14 36 34.2 | 1 | 0.0019 |
| 10 30 24.1 | 1 | 0.0019 | 13 7 18 | 1 | 0.0019 | 12 32.2 18 | 2 | 0.0039 | 14 36 36.2 | 1 | 0.0019 |
| 10 31 28.1 | 1 | 0.0019 | 13 7 19 | 4 | 0.0077 | 12 32.2 19 | 3 | 0.0058 | 14 36 40.2 | 1 | 0.0019 |
| 10 31 41.1 | 1 | 0.0019 | 13 7 20 | 2 | 0.0039 | 12 33 16 | 1 | 0.0019 | 14 36 42.2 | 1 | 0.0019 |
| 10 33 23 | 1 | 0.0019 | 13 7 23 | 1 | 0.0019 | 12 33 18 | 2 | 0.0039 | 14 36 44.2 | 1 | 0.0019 |
| 10 34 18 | 1 | 0.0019 | 13 7 24 | 1 | 0.0019 | 12 33 19 | 1 | 0.0019 | 14 36.3 31 | 1 | 0.0019 |
| 11 16 19 | 1 | 0.0019 | 13 8 17 | 2 | 0.0039 | 12 33.2 18 | 1 | 0.0019 | 14 36.3 33 | 2 | 0.0039 |
| 11 17 18 | 1 | 0.0019 | 13 8 18 | 1 | 0.0019 | 12 34 18 | 1 | 0.0019 | 14 37 0 | 1 | 0.0019 |
| 11 17 25.1 | 1 | 0.0019 | 13 8 20 | 1 | 0.0019 | 12 34 19 | 2 | 0.0039 | 14 37 26 | 3 | 0.0058 |
| 11 17.1 23.1 | 1 | 0.0019 | 14 12 19 | 1 | 0.0019 | 12 35 16 | 1 | 0.0019 | 14 37 27 | 5 | 0.0097 |
| 11 17.1 31 | 1 | 0.0019 | 14 12 21 | 4 | 0.0077 | 13 24 19 | 1 | 0.0019 | 14 37 28 | 4 | 0.0077 |
| 11 18 18 | 3 | 0.0058 | 14 12 22 | 1 | 0.0019 | 13 26.2 16 | 2 | 0.0039 | 14 37 29 | 3 | 0.0058 |
| 11 18 20 | 1 | 0.0019 | 14 13 18 | 3 | 0.0058 | 13 27.2 16 | 1 | 0.0019 | 14 37 30 | 3 | 0.0058 |
| 11 18 23 | 1 | 0.0019 | 14 13 20 | 2 | 0.0039 | 13 27.2 18 | 1 | 0.0019 | 14 37 31 | 5 | 0.0097 |
| 11 18 24.1 | 1 | 0.0019 | 14 13 23 | 1 | 0.0019 | 13 27.2 21 | 1 | 0.0019 | 14 37 31.3 | 1 | 0.0019 |
| 11 18 26.1 | 1 | 0.0019 | 14 13.2 19 | 1 | 0.0019 | 13 28 16 | 1 | 0.0019 | 14 37 32 | 1 | 0.0019 |
| 11 18 27.1 | 1 | 0.0019 | 14 13.2 20 | 1 | 0.0019 | 13 28 19 | 3 | 0.0058 | 14 37 33 | 2 | 0.0039 |
| 11 18.1 13.3 | 1 | 0.0019 | 14 14 18 | 3 | 0.0058 | 13 28.2 16 | 3 | 0.0058 | 14 37 34 | 1 | 0.0019 |
| 11 18.1 19 | 1 | 0.0019 | 14 14 19 | 2 | 0.0039 | 13 28.2 18 | 2 | 0.0039 | 14 37 34.2 | 1 | 0.0019 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 11 18.1 24.1 | 1 | 0.0019 | 14 14 20 | 2 | 0.0039 | 13 28.2 19 | 3 | 0.0058 | 14 37 43.2 | 1 | 0.0019 |
| 11 18.1 27 | 1 | 0.0019 | 14 14 21 | 1 | 0.0019 | 13 28.2 20 | 2 | 0.0039 | 14 37.2 27 | 1 | 0.0019 |
| 11 18.1 29.2 | 1 | 0.0019 | 14 15 13 | 1 | 0.0019 | 13 29 17 | 1 | 0.0019 | 14 37.2 28 | 1 | 0.0019 |
| 11 18.1 30.1 | 1 | 0.0019 | 14 15 15 | 1 | 0.0019 | 13 29 18 | 2 | 0.0039 | 14 37.3 43.2 | 1 | 0.0019 |
| 11 19 18 | 1 | 0.0019 | 14 15 17 | 1 | 0.0019 | 13 29 19 | 8 | 0.0155 | 14 38 26 | 1 | 0.0019 |
| 11 19 23.1 | 1 | 0.0019 | 14 15 18 | 2 | 0.0039 | 13 29 20 | 1 | 0.0019 | 14 38 27 | 1 | 0.0019 |
| 11 19 25.1 | 1 | 0.0019 | 14 15 19 | 13 | 0.0251 | 13 29.2 16 | 4 | 0.0077 | 14 38 28 | 1 | 0.0019 |
| 11 19 28 | 1 | 0.0019 | 14 15 20 | 7 | 0.0135 | 13 29.2 18 | 2 | 0.0039 | 14 38 29 | 3 | 0.0058 |
| 11 19 32.1 | 1 | 0.0019 | 14 15 21 | 6 | 0.0116 | 13 29.2 19 | 3 | 0.0058 | 14 38 30 | 1 | 0.0019 |
| 11 19 40 | 1 | 0.0019 | 14 16 15 | 2 | 0.0039 | 13 29.2 22 | 1 | 0.0019 | 14 38 32 | 3 | 0.0058 |
| 11 19.1 16.3 | 1 | 0.0019 | 14 16 16 | 1 | 0.0019 | 13 30 16 | 3 | 0.0058 | 14 38 33 | 1 | 0.0019 |
| 11 19.1 17 | 3 | 0.0058 | 14 16 17 | 2 | 0.0039 | 13 30 17 | 2 | 0.0039 | 14 38 36.2 | 5 | 0.0097 |
| 11 19.1 18 | 2 | 0.0039 | 14 16 18 | 6 | 0.0116 | 13 30 18 | 3 | 0.0058 | 14 38 38 | 1 | 0.0019 |
| 11 20 13.3 | 1 | 0.0019 | 14 16 19 | 13 | 0.0251 | 13 30 19 | 5 | 0.0097 | 14 38 40.3 | 1 | 0.0019 |
| 11 20 24 | 1 | 0.0019 | 14 16 20 | 8 | 0.0155 | 13 30.2 17 | 2 | 0.0039 | 14 38 45.2 | 1 | 0.0019 |
| 11 20 24.1 | 2 | 0.0039 | 14 16 21 | 4 | 0.0077 | 13 30.2 18 | 5 | 0.0097 | 14 38.3 33 | 1 | 0.0019 |
| 11 20 25.1 | 1 | 0.0019 | 14 16 22 | 1 | 0.0019 | 13 30.2 19 | 6 | 0.0116 | 14 38.3 42.2 | 1 | 0.0019 |
| 11 20 27.1 | 1 | 0.0019 | 14 16 23 | 1 | 0.0019 | 13 30.2 20 | 2 | 0.0039 | 14 38.3 44.2 | 1 | 0.0019 |
| 11 20 28.1 | 2 | 0.0039 | 14 16 24 | 1 | 0.0019 | 13 30.2 21 | 1 | 0.0019 | 14 39 26 | 3 | 0.0058 |
| 11 20 29.1 | 2 | 0.0039 | 14 17 17 | 3 | 0.0058 | 13 31 16 | 8 | 0.0155 | 14 39 28 | 1 | 0.0019 |
| 11 20 30 | 1 | 0.0019 | 14 17 18 | 5 | 0.0097 | 13 31 17 | 5 | 0.0097 | 14 39 29 | 1 | 0.0019 |
| 11 20 32 | 1 | 0.0019 | 14 17 19 | 10 | 0.0193 | 13 31 18 | 4 | 0.0077 | 14 39 31 | 1 | 0.0019 |
| 11 20.1 16 | 1 | 0.0019 | 14 17 20 | 10 | 0.0193 | 13 31 19 | 5 | 0.0097 | 14 39 32 | 1 | 0.0019 |
| 11 20.1 19 | 1 | 0.0019 | 14 17 21 | 6 | 0.0116 | 13 31 20 | 4 | 0.0077 | 14 39 34 | 1 | 0.0019 |
| 11 20.1 25 | 1 | 0.0019 | 14 17 22 | 1 | 0.0019 | 13 31.2 16 | 1 | 0.0019 | 14 39 36.2 | 3 | 0.0058 |
| 11 20.1 29.2 | 1 | 0.0019 | 14 17 23 | 1 | 0.0019 | 13 31.2 17 | 1 | 0.0019 | 14 40 26 | 1 | 0.0019 |
| 11 20.1 32 | 1 | 0.0019 | 14 18 15 | 2 | 0.0039 | 13 31.2 18 | 4 | 0.0077 | 14 40 27 | 1 | 0.0019 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 11 21 18 | 1 | 0.0019 | 14 18 17 | 3 | 0.0058 | 13 31.2 19 | 4 | 0.0077 | 14 40 31 | 1 | 0.0019 |
| 11 21 23 | 1 | 0.0019 | 14 18 18 | 3 | 0.0058 | 13 31.2 20 | 1 | 0.0019 | 14 40 38.2 | 1 | 0.0019 |
| 11 21 25.1 | 1 | 0.0019 | 14 18 19 | 5 | 0.0097 | 13 31.2 21 | 1 | 0.0019 | 14 40.3 30 | 1 | 0.0019 |
| 11 21 26.1 | 2 | 0.0039 | 14 18 20 | 3 | 0.0058 | 13 31.3 16 | 1 | 0.0019 | 14 40.3 31 | 1 | 0.0019 |
| 11 21 28.1 | 1 | 0.0019 | 14 18 21 | 5 | 0.0097 | 13 32 15 | 1 | 0.0019 | 14 41.3 27 | 1 | 0.0019 |
| 11 21 31 | 1 | 0.0019 | 14 18 22 | 1 | 0.0019 | 13 32 16 | 7 | 0.0135 | 14 41.3 31 | 1 | 0.0019 |
| 11 21 31.1 | 1 | 0.0019 | 14 19 18 | 1 | 0.0019 | 13 32 17 | 4 | 0.0077 | 14 42.3 30 | 1 | 0.0019 |
| 11 21.1 20.1 | 2 | 0.0039 | 14 19 19 | 3 | 0.0058 | 13 32 18 | 6 | 0.0116 | 15 26.1 24 | 1 | 0.0019 |
| 11 21.1 29 | 1 | 0.0019 | 14 19 20 | 3 | 0.0058 | 13 32 19 | 3 | 0.0058 | 15 30 43.2 | 1 | 0.0019 |
| 11 21.1 31 | 1 | 0.0019 | 14 19 21 | 1 | 0.0019 | 13 32 20 | 4 | 0.0077 | 15 30.1 30 | 1 | 0.0019 |
| 11 21.1 41.1 | 1 | 0.0019 | 14 19 22 | 1 | 0.0019 | 13 32.2 19 | 3 | 0.0058 | 15 31 42.2 | 1 | 0.0019 |
| 11 21.1 42.1 | 1 | 0.0019 | 14 7 18 | 3 | 0.0058 | 13 32.2 20 | 1 | 0.0019 | 15 32 29 | 1 | 0.0019 |
| 11 22 18 | 1 | 0.0019 | 14 7 20 | 4 | 0.0077 | 13 33 16 | 2 | 0.0039 | 15 32 30 | 1 | 0.0019 |
| 11 22 19 | 2 | 0.0039 | 14 8 16 | 1 | 0.0019 | 13 33 17 | 7 | 0.0135 | 15 32 33.2 | 1 | 0.0019 |
| 11 22 24.1 | 2 | 0.0039 | 14 8 17 | 4 | 0.0077 | 13 33 18 | 3 | 0.0058 | 15 33 24 | 1 | 0.0019 |
| 11 22 25.1 | 4 | 0.0077 | 14 8 18 | 1 | 0.0019 | 13 33 19 | 1 | 0.0019 | 15 33 26 | 1 | 0.0019 |
| 11 22 26.1 | 2 | 0.0039 | 14 8 19 | 2 | 0.0039 | 13 33 20 | 3 | 0.0058 | 15 33 26.2 | 1 | 0.0019 |
| 11 22 29 | 1 | 0.0019 | 14 8 20 | 3 | 0.0058 | 13 33 21 | 1 | 0.0019 | 15 33 27 | 1 | 0.0019 |
| 11 22 29.1 | 1 | 0.0019 | 14 8 21 | 1 | 0.0019 | 13 33.1 20 | 1 | 0.0019 | 15 33 29 | 1 | 0.0019 |
| 11 22 41.1 | 1 | 0.0019 | 14 9 16 | 1 | 0.0019 | 13 33.2 18 | 2 | 0.0039 | 15 33 30 | 3 | 0.0058 |
| 11 22.1 18 | 2 | 0.0039 | 14 9 19 | 1 | 0.0019 | 13 33.2 19 | 1 | 0.0019 | 15 33 31 | 2 | 0.0039 |
| 11 22.1 19 | 1 | 0.0019 | 15 11 19 | 2 | 0.0039 | 13 34 15 | 1 | 0.0019 | 15 33 32 | 1 | 0.0019 |
| 11 22.1 22.1 | 1 | 0.0019 | 15 12 21 | 1 | 0.0019 | 13 34 19 | 1 | 0.0019 | 15 33 36.2 | 1 | 0.0019 |
| 11 22.1 24.1 | 3 | 0.0058 | 15 12 24 | 1 | 0.0019 | 14 24.2 19 | 1 | 0.0019 | 15 33 44.2 | 2 | 0.0039 |
| 11 23 18 | 1 | 0.0019 | 15 13 19 | 1 | 0.0019 | 14 27 17 | 1 | 0.0019 | 15 33.3 31 | 1 | 0.0019 |
| 11 23 18.2 | 1 | 0.0019 | 15 13 20 | 3 | 0.0058 | 14 27 18 | 1 | 0.0019 | 15 34 21 | 1 | 0.0019 |
| 11 23 19 | 2 | 0.0039 | 15 13 21 | 1 | 0.0019 | 14 27.2 19 | 1 | 0.0019 | 15 34 26 | 2 | 0.0039 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 11 23 21 | 1 | 0.0019 | 15 13 22 | 1 | 0.0019 | 14 28.2 19 | 1 | 0.0019 | 15 34 27 | 3 | 0.0058 |
| 11 23 24 | 1 | 0.0019 | 15 14 17 | 1 | 0.0019 | 14 29 19 | 5 | 0.0097 | 15 34 28 | 1 | 0.0019 |
| 11 23 25.1 | 1 | 0.0019 | 15 14 18 | 1 | 0.0019 | 14 29 20 | 1 | 0.0019 | 15 34 29 | 2 | 0.0039 |
| 11 23 28.2 | 1 | 0.0019 | 15 14 19 | 1 | 0.0019 | 14 29.2 16 | 1 | 0.0019 | 15 34 31 | 4 | 0.0077 |
| 11 23 30 | 1 | 0.0019 | 15 14 20 | 1 | 0.0019 | 14 29.2 17 | 1 | 0.0019 | 15 34 32 | 3 | 0.0058 |
| 11 23 31 | 1 | 0.0019 | 15 14 21 | 2 | 0.0039 | 14 29.2 18 | 2 | 0.0039 | 15 34 33 | 1 | 0.0019 |
| 11 23 32 | 1 | 0.0019 | 15 14 22 | 1 | 0.0019 | 14 29.2 19 | 1 | 0.0019 | 15 34 42.2 | 3 | 0.0058 |
| 11 23.1 24.1 | 1 | 0.0019 | 15 14.2 19 | 1 | 0.0019 | 14 30 17 | 1 | 0.0019 | 15 34.2 24 | 1 | 0.0019 |
| 11 23.1 41.1 | 1 | 0.0019 | 15 15 15 | 2 | 0.0039 | 14 30 18 | 1 | 0.0019 | 15 34.2 26 | 1 | 0.0019 |
| 11 24 18 | 5 | 0.0097 | 15 15 17 | 1 | 0.0019 | 14 30 19 | 2 | 0.0039 | 15 34.2 28 | 1 | 0.0019 |
| 11 24 20 | 1 | 0.0019 | 15 15 18 | 4 | 0.0077 | 14 30.2 16 | 1 | 0.0019 | 15 34.2 30 | 1 | 0.0019 |
| 11 24 21 | 1 | 0.0019 | 15 15 19 | 1 | 0.0019 | 14 30.2 18 | 5 | 0.0097 | 15 34.3 32 | 1 | 0.0019 |
| 11 24 24 | 1 | 0.0019 | 15 15 20 | 3 | 0.0058 | 14 30.2 19 | 6 | 0.0116 | 15 34.3 33 | 2 | 0.0039 |
| 11 24 25.1 | 1 | 0.0019 | 15 15 21 | 10 | 0.0193 | 14 30.2 20 | 2 | 0.0039 | 1534.3362 | 1 | 0.0019 |
| 11 24 27.1 | 2 | 0.0039 | 15 15 22 | 2 | 0.0039 | 14 31 16 | 3 | 0.0058 | 15 35 25 | 3 | 0.0058 |
| 11 24 28.1 | 2 | 0.0039 | 15 16 17 | 2 | 0.0039 | 14 31 17 | 5 | 0.0097 | 15 35 26 | 2 | 0.0039 |
| 11 24 32 | 1 | 0.0019 | 15 16 18 | 4 | 0.0077 | 14 31 18 | 5 | 0.0097 | 15 35 27 | 4 | 0.0077 |
| 11 25 18 | 3 | 0.0058 | 15 16 19 | 5 | 0.0097 | 14 31 19 | 7 | 0.0135 | 15 35 28 | 5 | 0.0097 |
| 11 25 21 | 1 | 0.0019 | 15 16 20 | 6 | 0.0116 | 14 31 20 | 3 | 0.0058 | 15 35 29 | 4 | 0.0077 |
| 11 25 24.1 | 3 | 0.0058 | 15 16 21 | 5 | 0.0097 | 14 31.2 18 | 2 | 0.0039 | 15 35 30 | 3 | 0.0058 |
| 11 25 25 | 1 | 0.0019 | 15 16 22 | 1 | 0.0019 | 14 31.2 19 | 4 | 0.0077 | 15 35 31 | 1 | 0.0019 |
| 11 25 25.1 | 1 | 0.0019 | 15 17 14 | 1 | 0.0019 | 14 32 16 | 5 | 0.0097 | 15 35 32 | 2 | 0.0039 |
| 11 25 26.1 | 1 | 0.0019 | 15 17 15 | 3 | 0.0058 | 14 32 17 | 1 | 0.0019 | 15 35 34.2 | 1 | 0.0019 |
| 11 25 27.1 | 2 | 0.0039 | 15 17 17 | 1 | 0.0019 | 14 32 18 | 2 | 0.0039 | 15 35 38.2 | 1 | 0.0019 |
| 11 25 30.1 | 1 | 0.0019 | 15 17 18 | 2 | 0.0039 | 14 32 19 | 5 | 0.0097 | 15 35 45.2 | 1 | 0.0019 |
| 11 25.1 25.1 | 1 | 0.0019 | 15 17 19 | 3 | 0.0058 | 14 32 20 | 2 | 0.0039 | 15 35.1 27 | 1 | 0.0019 |
| 11 26 18 | 1 | 0.0019 | 15 17 20 | 7 | 0.0135 | 14 32.2 18 | 3 | 0.0058 | 15 36 25 | 4 | 0.0077 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 11 26 23.1 | 2 | 0.0039 | 15 17 21 | 3 | 0.0058 | 14 32.2 19 | 1 | 0.0019 | 15 36 26 | 2 | 0.0039 |
| 11 26 24.1 | 2 | 0.0039 | 15 17 21.2 | 1 | 0.0019 | 14 33 16 | 6 | 0.0116 | 15 36 27 | 3 | 0.0058 |
| 11 26 25.1 | 2 | 0.0039 | 15 17 22 | 1 | 0.0019 | 14 33 17 | 1 | 0.0019 | 15 36 28 | 6 | 0.0116 |
| 11 26 26.1 | 1 | 0.0019 | 15 17 24 | 1 | 0.0019 | 14 33 18 | 2 | 0.0039 | 15 36 29 | 4 | 0.0077 |
| 11 26.3 13.3 | 1 | 0.0019 | 15 18 15 | 2 | 0.0039 | 14 33 19 | 3 | 0.0058 | 15 36 30 | 4 | 0.0077 |
| 11 26.3 28 | 1 | 0.0019 | 15 18 18 | 1 | 0.0019 | 14 34 16 | 1 | 0.0019 | 15 36 31 | 6 | 0.0116 |
| 11 27 18 | 3 | 0.0058 | 15 18 19 | 2 | 0.0039 | 14 34 20 | 2 | 0.0039 | 15 36 32 | 1 | 0.0019 |
| 11 27 23.1 | 1 | 0.0019 | 15 18 20 | 6 | 0.0116 | 14.2 35 15 | 1 | 0.0019 | 15 36 32.2 | 1 | 0.0019 |
| 11 27 24 | 2 | 0.0039 | 15 18 21 | 4 | 0.0077 | 15 27 18 | 1 | 0.0019 | 15 36 39.2 | 1 | 0.0019 |
| 11 27 25.1 | 1 | 0.0019 | 15 18 22 | 2 | 0.0039 | 15 28.2 16 | 1 | 0.0019 | 15 36 40.2 | 3 | 0.0058 |
| 11 27 26.1 | 2 | 0.0039 | 15 19 18 | 2 | 0.0039 | 15 29 18 | 2 | 0.0039 | 15 36 41.2 | 1 | 0.0019 |
| 11 27 27 | 1 | 0.0019 | 15 19 19 | 1 | 0.0019 | 15 29 20 | 1 | 0.0019 | 15 36 44.2 | 1 | 0.0019 |
| 11 27 27.1 | 2 | 0.0039 | 15 19 21 | 5 | 0.0097 | 15 29 21 | 1 | 0.0019 | 15 36 45.2 | 1 | 0.0019 |
| 11 27 29 | 1 | 0.0019 | 15 20 18 | 1 | 0.0019 | 15 29.2 19 | 1 | 0.0019 | 15 36.3 26 | 1 | 0.0019 |
| 11 27 32.1 | 1 | 0.0019 | 15 7 22 | 1 | 0.0019 | 15 30 16 | 1 | 0.0019 | 15 36.3 27 | 1 | 0.0019 |
| 11 27.1 13.3 | 2 | 0.0039 | 15 8 17 | 1 | 0.0019 | 15 30 18 | 1 | 0.0019 | 15 37 0 | 1 | 0.0019 |
| 11 27.3 27 | 1 | 0.0019 | 15 8 18 | 3 | 0.0058 | 15 30.2 16 | 2 | 0.0039 | 15 37 23 | 1 | 0.0019 |
| 11 28 21 | 1 | 0.0019 | 15 8 19 | 3 | 0.0058 | 15 31 16 | 1 | 0.0019 | 15 37 24 | 2 | 0.0039 |
| 11 28 23 | 1 | 0.0019 | 15 8 20 | 4 | 0.0077 | 15 31 17 | 1 | 0.0019 | 15 37 25 | 1 | 0.0019 |
| 11 28 24.1 | 4 | 0.0077 | 15 9 20 | 1 | 0.0019 | 15 31 19 | 1 | 0.0019 | 15 37 26 | 1 | 0.0019 |
| 11 28 25.1 | 2 | 0.0039 | 16 12 22 | 1 | 0.0019 | 15 31.2 20 | 1 | 0.0019 | 15 37 27 | 1 | 0.0019 |
| 11 28 26 | 1 | 0.0019 | 16 13 18 | 1 | 0.0019 | 15 32 16 | 1 | 0.0019 | 15 37 28 | 2 | 0.0039 |
| 11 28 26.1 | 1 | 0.0019 | 16 13 20 | 1 | 0.0019 | 15 32 18 | 1 | 0.0019 | 15 37 29 | 4 | 0.0077 |
| 11 28 27.1 | 1 | 0.0019 | 16 15 16 | 2 | 0.0039 | 15 32 19 | 4 | 0.0077 | 15 37 30 | 3 | 0.0058 |
| 11 28 28.1 | 2 | 0.0039 | 16 15 18 | 1 | 0.0019 | 15 32.2 20 | 1 | 0.0019 | 15 37 31 | 5 | 0.0097 |
| 11 28.1 26.1 | 1 | 0.0019 | 16 15 22 | 1 | 0.0019 | 15 33 16 | 1 | 0.0019 | 15 37 34.2 | 1 | 0.0019 |
| 11 28.3 19 | 1 | 0.0019 | 16 16 18 | 1 | 0.0019 | 15 33 17 | 1 | 0.0019 | 15 37 38.2 | 1 | 0.0019 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 11 28.3 24 | 1 | 0.0019 | 16 16 19 | 1 | 0.0019 | 16 29.2 19 | 1 | 0.0019 | 15 37 40.2 | 1 | 0.0019 |
| 11 29 18 | 1 | 0.0019 | 16 16 20 | 2 | 0.0039 | 16 32 19 | 1 | 0.0019 | 15 37 43.2 | 1 | 0.0019 |
| 11 29 23 | 1 | 0.0019 | 16 17 21 | 2 | 0.0039 | 16 33 19 | 1 | 0.0019 | 15 37 45.2 | 1 | 0.0019 |
| 11 29 23.1 | 1 | 0.0019 | 16 17 22 | 1 | 0.0019 | 16 34 16 | 1 | 0.0019 | 15 37.2 27 | 1 | 0.0019 |
| 11 29 24 | 1 | 0.0019 | 16 18 17 | 1 | 0.0019 | 9 30.2 18 | 1 | 0.0019 | 15 37.3 34.2 | 1 | 0.0019 |
| 11 29 24.1 | 1 | 0.0019 | 16 18 21 | 2 | 0.0039 | 9 31.2 18 | 1 | 0.0019 | 15 38 27 | 2 | 0.0039 |
| 11 29 25 | 1 | 0.0019 | 16 18 22 | 1 | 0.0019 | | | | 15 38 27.3 | 1 | 0.0019 |
| 11 29 25.1 | 2 | 0.0039 | 16 19 18 | 2 | 0.0039 | | | | 15 38 28 | 3 | 0.0058 |
| 11 29 28 | 1 | 0.0019 | 16 19 19 | 1 | 0.0019 | | | | 15 38 29 | 2 | 0.0039 |
| 11 29 29 | 1 | 0.0019 | 16 20 18 | 1 | 0.0019 | | | | 15 38 30 | 3 | 0.0058 |
| 11 29.3 27 | 1 | 0.0019 | 16 7 19 | 1 | 0.0019 | | | | 15 38 31 | 1 | 0.0019 |
| 11 30 18 | 2 | 0.0039 | 16 8 18 | 2 | 0.0039 | | | | 15 38 34.2 | 2 | 0.0039 |
| 11 30 23 | 1 | 0.0019 | 17 15 21 | 1 | 0.0019 | | | | 15 38 35.2 | 1 | 0.0019 |
| 11 30 24.1 | 1 | 0.0019 | 17 16 18 | 1 | 0.0019 | | | | 15 38 36.2 | 1 | 0.0019 |
| 11 30 26.1 | 3 | 0.0058 | 18 20 21 | 1 | 0.0019 | | | | 15 38 44.2 | 1 | 0.0019 |
| 11 30 27.1 | 1 | 0.0019 | 18 7 21 | 1 | 0.0019 | | | | 15 38.2 40.2 | 2 | 0.0039 |
| 11 30 28.1 | 1 | 0.0019 | | | | | | | 15 38.3 28 | 1 | 0.0019 |
| 11 30 29 | 1 | 0.0019 | | | | | | | 15 39 27 | 1 | 0.0019 |
| 11 30.1 24.1 | 1 | 0.0019 | | | | | | | 15 39 28 | 3 | 0.0058 |
| 11 31 18 | 1 | 0.0019 | | | | | | | 15 39 30 | 1 | 0.0019 |
| 11 31 25.1 | 1 | 0.0019 | | | | | | | 15 39 31 | 1 | 0.0019 |
| 11 31 40.1 | 1 | 0.0019 | | | | | | | 15 39.3 28 | 1 | 0.0019 |
| 11 31.2 27.1 | 1 | 0.0019 | | | | | | | 15 39.3 30 | 1 | 0.0019 |
| 11 32 24.1 | 1 | 0.0019 | | | | | | | 15 39.3 31 | 1 | 0.0019 |
| 11 32 25.1 | 1 | 0.0019 | | | | | | | 15 39.3 39.2 | 1 | 0.0019 |
| 11 32.1 14 | 1 | 0.0019 | | | | | | | 15 39.3 43.2 | 1 | 0.0019 |
| 11 32.1 18 | 1 | 0.0019 | | | | | | | 15 40 0 | 1 | 0.0019 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 11 32.1 35.1 | 1 | 0.0019 | | | | | | | 15 40 26 | 1 | 0.0019 |
| 11 33 26.1 | 1 | 0.0019 | | | | | | | 15 40.3 27 | 1 | 0.0019 |
| 11 34 24 | 1 | 0.0019 | | | | | | | 15 40.3 28 | 1 | 0.0019 |
| 11 34.1 28 | 1 | 0.0019 | | | | | | | 15 40.3 39.2 | 1 | 0.0019 |
| 12 16 18 | 1 | 0.0019 | | | | | | | 15 40.3 43.2 | 1 | 0.0019 |
| 12 17 26.1 | 2 | 0.0039 | | | | | | | 15 41 32 | 1 | 0.0019 |
| 12 17.1 25 | 1 | 0.0019 | | | | | | | 15 41.3 27 | 1 | 0.0019 |
| 12 18 18 | 1 | 0.0019 | | | | | | | 15 41.3 44.2 | 1 | 0.0019 |
| 12 18 24.1 | 1 | 0.0019 | | | | | | | 15 42.3 28 | 1 | 0.0019 |
| 12 18 25.1 | 1 | 0.0019 | | | | | | | 15 42.3 29 | 1 | 0.0019 |
| 12 18 26 | 1 | 0.0019 | | | | | | | 15 43.3 43.2 | 1 | 0.0019 |
| 12 18 26.1 | 1 | 0.0019 | | | | | | | 16 32 27 | 1 | 0.0019 |
| 12 18 27 | 1 | 0.0019 | | | | | | | 16 32 29 | 3 | 0.0058 |
| 12 18 27.1 | 2 | 0.0039 | | | | | | | 16 32 32 | 1 | 0.0019 |
| 12 18.1 13.3 | 2 | 0.0039 | | | | | | | 16 32 37.2 | 1 | 0.0019 |
| 12 18.1 19 | 1 | 0.0019 | | | | | | | 16 34 26 | 1 | 0.0019 |
| 12 18.1 24 | 1 | 0.0019 | | | | | | | 16 34 27 | 1 | 0.0019 |
| 12 18.1 25.1 | 1 | 0.0019 | | | | | | | 16 34 29 | 6 | 0.0116 |
| 12 18.1 28.1 | 1 | 0.0019 | | | | | | | 16 34 31 | 1 | 0.0019 |
| 12 18.1 31 | 1 | 0.0019 | | | | | | | 16 34.3 32 | 1 | 0.0019 |
| 12 19 19 | 1 | 0.0019 | | | | | | | 16 35 0 | 1 | 0.0019 |
| 12 19 20 | 2 | 0.0039 | | | | | | | 16 35 26 | 1 | 0.0019 |
| 12 19 23.1 | 1 | 0.0019 | | | | | | | 16 35 27 | 2 | 0.0039 |
| 12 19 24 | 1 | 0.0019 | | | | | | | 16 35 29 | 1 | 0.0019 |
| 12 19 25.1 | 1 | 0.0019 | | | | | | | 16 35 30 | 1 | 0.0019 |
| 12 19 27.1 | 1 | 0.0019 | | | | | | | 16 35 32 | 1 | 0.0019 |
| 12 19 29 | 1 | 0.0019 | | | | | | | 16 35 35.2 | 1 | 0.0019 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 12 19.1 13.3 | 1 | 0.0019 | | | | | | | 16 35 40.2 | 1 | 0.0019 |
| 12 19.1 19 | 2 | 0.0039 | | | | | | | 16 35 41.2 | 1 | 0.0019 |
| 12 19.1 24.1 | 1 | 0.0019 | | | | | | | 16 35 45.2 | 1 | 0.0019 |
| 12 20 13.3 | 2 | 0.0039 | | | | | | | 16 36 22.1 | 1 | 0.0019 |
| 12 20 18 | 3 | 0.0058 | | | | | | | 16 36 25 | 1 | 0.0019 |
| 12 20 23 | 2 | 0.0039 | | | | | | | 16 36 28 | 4 | 0.0077 |
| 12 20 24.1 | 1 | 0.0019 | | | | | | | 16 36 29 | 1 | 0.0019 |
| 12 20 39.1 | 1 | 0.0019 | | | | | | | 16 36 30 | 1 | 0.0019 |
| 12 20.1 17 | 1 | 0.0019 | | | | | | | 16 36 32 | 2 | 0.0039 |
| 12 20.1 25 | 1 | 0.0019 | | | | | | | 16 37 26 | 1 | 0.0019 |
| 12 20.3 40.1 | 1 | 0.0019 | | | | | | | 16 37 28 | 3 | 0.0058 |
| 12 21 17 | 1 | 0.0019 | | | | | | | 16 37 30 | 2 | 0.0039 |
| 12 21 22 | 1 | 0.0019 | | | | | | | 16 37 32 | 1 | 0.0019 |
| 12 21 24.1 | 2 | 0.0039 | | | | | | | 16 37 43.2 | 1 | 0.0019 |
| 12 21 25.1 | 1 | 0.0019 | | | | | | | 16 37 46.2 | 1 | 0.0019 |
| 12 21 26 | 1 | 0.0019 | | | | | | | 16 37.3 30 | 1 | 0.0019 |
| 12 21 26.1 | 2 | 0.0039 | | | | | | | 16 38 26 | 1 | 0.0019 |
| 12 21 27 | 1 | 0.0019 | | | | | | | 16 38 27 | 1 | 0.0019 |
| 12 21 28.1 | 1 | 0.0019 | | | | | | | 16 38 28 | 2 | 0.0039 |
| 12 21 32.1 | 1 | 0.0019 | | | | | | | 16 38 39.2 | 1 | 0.0019 |
| 12 21.1 13.3 | 1 | 0.0019 | | | | | | | 16 39 35 | 1 | 0.0019 |
| 12 21.1 27.1 | 2 | 0.0039 | | | | | | | 16 39.3 28 | 1 | 0.0019 |
| 12 21.1 29 | 1 | 0.0019 | | | | | | | 16 40.3 39.2 | 2 | 0.0039 |
| 12 21.1 41.1 | 1 | 0.0019 | | | | | | | 16 40.3 40.2 | 2 | 0.0039 |
| 12 21.1 42.1 | 1 | 0.0019 | | | | | | | 16 41.3 40.2 | 1 | 0.0019 |
| 12 22 18 | 1 | 0.0019 | | | | | | | 16 43.3 28 | 1 | 0.0019 |
| 12 22 28.1 | 1 | 0.0019 | | | | | | | 17 33 28 | 2 | 0.0039 |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 12 22 32.3 | 1 | 0.0019 | | | | | | | 17 33 31 | 1 | 0.0019 |
| 12 22.1 27 | 1 | 0.0019 | | | | | | | 17 34 40.2 | 1 | 0.0019 |
| 12 22.1 27.1 | 1 | 0.0019 | | | | | | | 17 36 31 | 1 | 0.0019 |
| 12 23 13.3 | 1 | 0.0019 | | | | | | | 17 37 27 | 2 | 0.0039 |
| 12 23 19 | 3 | 0.0058 | | | | | | | 17 37 29 | 1 | 0.0019 |
| 12 23 23.1 | 1 | 0.0019 | | | | | | | 17 39.3 30 | 1 | 0.0019 |
| 12 23 24 | 1 | 0.0019 | | | | | | | 17 42.3 38.2 | 1 | 0.0019 |
| 12 23 24.1 | 3 | 0.0058 | | | | | | | 8 35 37.2 | 1 | 0.0019 |
| 12 23 25.1 | 1 | 0.0019 | | | | | | | 8 39 24 | 1 | 0.0019 |
| 12 23 29.1 | 1 | 0.0019 | | | | | | | | | |
| 12 23 31.3 | 1 | 0.0019 | | | | | | | | | |
| 12 23.1 18 | 1 | 0.0019 | | | | | | | | | |
| 12 23.1 19 | 1 | 0.0019 | | | | | | | | | |
| 12 24 18 | 1 | 0.0019 | | | | | | | | | |
| 12 24 20.3 | 1 | 0.0019 | | | | | | | | | |
| 12 24 21 | 1 | 0.0019 | | | | | | | | | |
| 12 24 23.1 | 1 | 0.0019 | | | | | | | | | |
| 12 24 24 | 1 | 0.0019 | | | | | | | | | |
| 12 24 24.1 | 1 | 0.0019 | | | | | | | | | |
| 12 24 25.1 | 1 | 0.0019 | | | | | | | | | |
| 12 24 26 | 1 | 0.0019 | | | | | | | | | |
| 12 24 26.1 | 1 | 0.0019 | | | | | | | | | |
| 12 24 39.1 | 1 | 0.0019 | | | | | | | | | |
| 12 25 18 | 3 | 0.0058 | | | | | | | | | |
| 12 25 22.1 | 2 | 0.0039 | | | | | | | | | |
| 12 25 23.1 | 1 | 0.0019 | | | | | | | | | |
| 12 25 24.1 | 1 | 0.0019 | | | | | | | | | |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 12 25 25.1 | 1 | 0.0019 | | | | | | | | | |
| 12 25 30.1 | 1 | 0.0019 | | | | | | | | | |
| 12 26 17 | 2 | 0.0039 | | | | | | | | | |
| 12 26 18 | 1 | 0.0019 | | | | | | | | | |
| 12 26 20.1 | 1 | 0.0019 | | | | | | | | | |
| 12 26 24.1 | 1 | 0.0019 | | | | | | | | | |
| 12 26 25.1 | 2 | 0.0039 | | | | | | | | | |
| 12 26 26.1 | 1 | 0.0019 | | | | | | | | | |
| 12 26 28.1 | 1 | 0.0019 | | | | | | | | | |
| 12 26 31 | 1 | 0.0019 | | | | | | | | | |
| 12 26 32.1 | 1 | 0.0019 | | | | | | | | | |
| 12 27 19 | 1 | 0.0019 | | | | | | | | | |
| 12 27 23.1 | 1 | 0.0019 | | | | | | | | | |
| 12 27 24 | 1 | 0.0019 | | | | | | | | | |
| 12 27 24.1 | 3 | 0.0058 | | | | | | | | | |
| 12 27 25 | 1 | 0.0019 | | | | | | | | | |
| 12 27 25.1 | 1 | 0.0019 | | | | | | | | | |
| 12 27 26.3 | 1 | 0.0019 | | | | | | | | | |
| 12 27 27.1 | 1 | 0.0019 | | | | | | | | | |
| 12 27 30 | 1 | 0.0019 | | | | | | | | | |
| 12 28 18 | 2 | 0.0039 | | | | | | | | | |
| 12 28 24.1 | 1 | 0.0019 | | | | | | | | | |
| 12 28 26.1 | 2 | 0.0039 | | | | | | | | | |
| 12 28 27.1 | 1 | 0.0019 | | | | | | | | | |
| 12 28 28.1 | 1 | 0.0019 | | | | | | | | | |
| 12 28.1 22.3 | 1 | 0.0019 | | | | | | | | | |
| 12 29 25.1 | 1 | 0.0019 | | | | | | | | | |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 12 29 26 | 1 | 0.0019 | | | | | | | | | |
| 12 29 26.1 | 1 | 0.0019 | | | | | | | | | |
| 12 30 22.1 | 1 | 0.0019 | | | | | | | | | |
| 12 30 24 | 1 | 0.0019 | | | | | | | | | |
| 12 30 25.1 | 1 | 0.0019 | | | | | | | | | |
| 12 30 27.1 | 1 | 0.0019 | | | | | | | | | |
| 12 31 18 | 1 | 0.0019 | | | | | | | | | |
| 12 31 23 | 1 | 0.0019 | | | | | | | | | |
| 12 31 25.1 | 2 | 0.0039 | | | | | | | | | |
| 12 32 25.1 | 1 | 0.0019 | | | | | | | | | |
| 12 32 26 | 1 | 0.0019 | | | | | | | | | |
| 12 32 27 | 1 | 0.0019 | | | | | | | | | |
| 12 32 28.1 | 1 | 0.0019 | | | | | | | | | |
| 12 32 30.1 | 1 | 0.0019 | | | | | | | | | |
| 12 33 18 | 2 | 0.0039 | | | | | | | | | |
| 12 33 27 | 1 | 0.0019 | | | | | | | | | |
| 12 33.1 13.3 | 1 | 0.0019 | | | | | | | | | |
| 12 34 19 | 1 | 0.0019 | | | | | | | | | |
| 12 34 251 | 1 | 0.0019 | | | | | | | | | |
| 12 34 27 | 1 | 0.0019 | | | | | | | | | |
| 12 34 32 | 1 | 0.0019 | | | | | | | | | |
| 12 34.1 30 | 1 | 0.0019 | | | | | | | | | |
| 12 35 28 | 2 | 0.0039 | | | | | | | | | |
| 12 35.1 23.1 | 1 | 0.0019 | | | | | | | | | |
| 12 36 26 | 1 | 0.0019 | | | | | | | | | |
| 12 36 32 | 1 | 0.0019 | | | | | | | | | |
| 12 36.2 13.3 | 1 | 0.0019 | | | | | | | | | |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 12 36.2 23 | 1 | 0.0019 | | | | | | | | | |
| 12 37 25 | 1 | 0.0019 | | | | | | | | | |
| 12 37.1 23.1 | 1 | 0.0019 | | | | | | | | | |
| 12 37.2 13.3 | 2 | 0.0039 | | | | | | | | | |
| 12 38.2 13.3 | 1 | 0.0019 | | | | | | | | | |
| 12 39.2 40.1 | 1 | 0.0019 | | | | | | | | | |
| 13 17 20 | 1 | 0.0019 | | | | | | | | | |
| 13 17 25.1 | 1 | 0.0019 | | | | | | | | | |
| 13 18 26 | 1 | 0.0019 | | | | | | | | | |
| 13 18 27.1 | 2 | 0.0039 | | | | | | | | | |
| 13 18.1 19 | 1 | 0.0019 | | | | | | | | | |
| 13 19 27 | 1 | 0.0019 | | | | | | | | | |
| 13 19 27.1 | 1 | 0.0019 | | | | | | | | | |
| 13 20 13.3 | 1 | 0.0019 | | | | | | | | | |
| 13 21 22.1 | 1 | 0.0019 | | | | | | | | | |
| 13 21 23 | 1 | 0.0019 | | | | | | | | | |
| 13 21 24.1 | 1 | 0.0019 | | | | | | | | | |
| 13 21 25.1 | 3 | 0.0058 | | | | | | | | | |
| 13 24 24 | 1 | 0.0019 | | | | | | | | | |
| 13 24 42.1 | 1 | 0.0019 | | | | | | | | | |
| 13 25 18 | 1 | 0.0019 | | | | | | | | | |
| 13 25 24 | 1 | 0.0019 | | | | | | | | | |
| 13 25 25 | 1 | 0.0019 | | | | | | | | | |
| 13 25 25.1 | 1 | 0.0019 | | | | | | | | | |
| 13 25 40.1 | 1 | 0.0019 | | | | | | | | | |
| 13 25 41.1 | 1 | 0.0019 | | | | | | | | | |
| 13 26 18 | 1 | 0.0019 | | | | | | | | | |

| LG1 | | | LG2 | | | LG3 | | | LG4 | | |
|---------------------------|-------|-----------|---------------------------|-------|-----------|-------------------------|-------|-----------|---------------------------|-------|-----------|
| DXS8378-DXS10135-DXS10148 | | | DXS7132-DXS10074-DXS10079 | | | HPRTB-DXS10101-DXS10103 | | | DXS7423-DXS10134-DXS10146 | | |
| Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency | Haplotype | Count | Frequency |
| 13 26 25.1 | 1 | 0.0019 | | | | | | | | | |
| 13 26 27.1 | 1 | 0.0019 | | | | | | | | | |
| 13 27 27 | 1 | 0.0019 | | | | | | | | | |
| 13 30 17 | 1 | 0.0019 | | | | | | | | | |
| 13 33 27.1 | 1 | 0.0019 | | | | | | | | | |
| 13 37 33 | 1 | 0.0019 | | | | | | | | | |
| 14 19 18 | 1 | 0.0019 | | | | | | | | | |
| 14 31 26.1 | 1 | 0.0019 | | | | | | | | | |
| 8 28 26.1 | 1 | 0.0019 | | | | | | | | | |
| 9 17.1 24.3 | 1 | 0.0019 | | | | | | | | | |
| 9 18 18 | 1 | 0.0019 | | | | | | | | | |
| 9 18 27.1 | 1 | 0.0019 | | | | | | | | | |
| 9 19 35.1 | 1 | 0.0019 | | | | | | | | | |
| 9 20 23.1 | 1 | 0.0019 | | | | | | | | | |
| 9 21 23.1 | 1 | 0.0019 | | | | | | | | | |
| 9 22 18 | 2 | 0.0039 | | | | | | | | | |
| 9 23 19.3 | 1 | 0.0019 | | | | | | | | | |
| 9 24 23.1 | 1 | 0.0019 | | | | | | | | | |
| 9 28 27.1 | 1 | 0.0019 | | | | | | | | | |
| 9 31 20 | 1 | 0.0019 | | | | | | | | | |

Table H2 Haplotype forensic parameters for 517 South African males.

| | LG1 | LG2 | LG3 | LG4 | Combined |
|------------------------------------|-------------------------------|-------------------------------|-----------------------------|-------------------------------|-------------------|
| | DXS8378-DXS10135- DXS10148 | DXS7132-DXS10074- DXS10079 | HPRTB-DXS10101- DXS10103 | DXS7423-DXS10134- DXS10146 | |
| Number of unique haplotypes | 421 | 228 | 218 | 302 | - |
| HD | 0.999 | 0.994 | 0.994 | 0.996 | - |
| PIC | 0.997 | 0.992 | 0.992 | 0.994 | - |
| PD_Male | 0.997 | 0.992 | 0.992 | 0.994 | 0.99999998975966 |
| PD_Female | 1.000 | 1.000 | 1.000 | 1.000 | 1 |
| MEC_Kruger | 0.984 | 0.981 | 0.981 | 0.983 | 0.999999898770688 |
| MEC_Kishida | 0.987 | 0.989 | 0.989 | 0.988 | 0.99999980879369 |
| MEC_Desmarais | 0.997 | 0.992 | 0.992 | 0.994 | 0.9999999895124 |
| MEC_Desmarais_duo | 0.994 | 0.984 | 0.984 | 0.989 | 0.99999983927333 |

Abbreviations - LG: linkage group. HD: haplotype diversity. PD: power of discrimination. MEC: mean exclusion chance.

Appendix I:

Table I1 Linkage disequilibrium test p-values for all pairs of loci. For male samples (upper triangle) the Exact test using a Markov chain (Chain length = 10000) was used. For female samples (lower triangle) likelihood ratio test for linkage disequilibrium was used. Significant p-values are bolded. i.e., $p < 0.0008$.

| Linkage group (LG) | | 1 | | | 2 | | | 3 | | | 4 | | |
|--------------------|----------|---------|----------|----------------|---------|----------|----------|---------|----------------|--------------|---------|----------------|--------------|
| | Locus | DXS8378 | DXS10135 | DXS10148 | DXS7132 | DXS10074 | DXS10079 | HPRTB | DXS10101 | DXS10103 | DXS7423 | DXS10134 | DXS10146 |
| 1 | DXS8378 | - | 0.015 | 0.146 | 0.695 | 0.823 | 0.738 | 0.092 | 0.265 | 0.343 | 0.644 | 0.228 | 0.840 |
| | DXS10135 | 0.44703 | - | 0.289 | 0.719 | 0.229 | 0.905 | 0.004 | 0.923 | 0.413 | 0.370 | 0.001 | 0.552 |
| | DXS10148 | 0.09327 | 0.00267 | - | 0.857 | 0.480 | 0.421 | 0.630 | 0.000 | 0.549 | 0.127 | 0.416 | 0.379 |
| 2 | DXS7132 | 0.80990 | 0.95772 | 0.73050 | - | 0.045 | 0.222 | 0.070 | 0.364 | 0.692 | 0.191 | 0.618 | 0.959 |
| | DXS10074 | 0.58139 | 0.54713 | 0.00020 | 0.22218 | - | 0.147 | 0.040 | 0.380 | 0.226 | 0.120 | 0.160 | 0.080 |
| | DXS10079 | 0.93881 | 0.96000 | 0.28475 | 0.51604 | 0.14614 | - | 0.384 | 0.214 | 0.583 | 0.513 | 0.362 | 0.782 |
| 3 | HPRTB | 0.37010 | 0.63327 | 0.47396 | 0.33554 | 0.20713 | 0.17040 | - | 0.000 | 0.026 | 0.810 | 0.946 | 0.013 |
| | DXS10101 | 0.06396 | 0.69614 | 0.13802 | 0.99673 | 0.00733 | 0.51554 | 0.05327 | - | 0.000 | 0.048 | 0.000 | 0.655 |
| | DXS10103 | 0.41347 | 0.52713 | 0.03366 | 0.93347 | 0.20050 | 0.44663 | 0.00366 | 0.00000 | - | 0.141 | 0.161 | 0.339 |
| 4 | DXS7423 | 0.70020 | 0.96455 | 0.72604 | 0.63505 | 0.36178 | 0.01782 | 0.11891 | 0.09416 | 0.18891 | - | 0.426 | 0.236 |
| | DXS10134 | 0.74267 | 0.43634 | 0.15139 | 0.04446 | 0.19416 | 0.38030 | 0.74723 | 0.53287 | 0.08416 | 0.15158 | - | 0.000 |
| | DXS10146 | 0.57228 | 0.29960 | 0.03059 | 0.26228 | 0.18515 | 0.45802 | 0.56129 | 0.08158 | 0.46594 | 0.01396 | 0.00010 | - |

Appendix J:

Table J1 List of off-ladder alleles observed within the South African population. Bolded alleles have previously been reported [71,97,103,108,109,113,117,118,157].

| Marker | Allele | Total number of observations | Number of observations | | | |
|-----------------|-------------|------------------------------|------------------------|-------------------|----------------|-----------------------|
| | | | African ancestry | European ancestry | Mixed ancestry | Indian/Asian ancestry |
| DXS10148 | 16.3 | 1 | | | 1 | |
| | 19.3 | 1 | | | 1 | |
| | 20.3 | 1 | | | 1 | |
| | 22.3 | 1 | 1 | | | |
| | 24.3 | 1 | | | 1 | |
| | 26.3 | 1 | | | 1 | |
| | 28.3 | 1 | | | 1 | |
| | 29.2 | 2 | 2 | | | |
| | 31.3 | 1 | 1 | | | |
| | 32 | 7 | 3 | 1 | 3 | |
| | 32.1 | 10 | 8 | 1 | 1 | |
| | 32.3 | 1 | 1 | | | |
| | 33 | 3 | 2 | | 1 | |
| | 35.1 | 4 | 3 | | 1 | |
| | 36.1 | 1 | 1 | | | |
| | 37.1 | 1 | | | 1 | |
| | 39.1 | 9 | 3 | 2 | 4 | |
| | 40 | 1 | 1 | | | |
| | 40.1 | 7 | 6 | | 1 | |
| | 41.1 | 12 | 8 | 1 | 3 | |
| 42.1 | 7 | 4 | 1 | 2 | | |
| 43.1 | 1 | 1 | | | | |
| 44.1 | 1 | 1 | | | | |
| DXS10134 | 26.1 | 1 | 1 | | | |
| | 27.1 | 1 | | | 1 | |
| | 28.2 | 1 | 1 | | | |
| | 29.1 | 1 | | | 1 | |
| | 29.2 | 1 | | | 1 | |
| | 30.1 | 2 | 1 | | 1 | |
| | 33.2 | 1 | 1 | | | |
| | 33.3 | 4 | 3 | | 1 | |
| | 35.1 | 1 | | 1 | | |
| | 36.2 | 2 | | | 1 | 1 |
| DXS10135 | 26.3 | 3 | 1 | | 2 | |
| | 27.1 | 2 | | | 2 | |
| | 28.3 | 5 | | | 5 | |
| | 29.3 | 2 | 1 | | 1 | |
| | 30.1 | 1 | 1 | | | |
| | 33.1 | 2 | 1 | | | 1 |
| | 37.1 | 2 | | | 2 | |

| Marker | Allele | Total number of observations | Number of observations | | | |
|--------------|--------|------------------------------|------------------------|-------------------|----------------|-----------------------|
| | | | African ancestry | European ancestry | Mixed ancestry | Indian/Asian ancestry |
| DXS10135 | 38.2 | 1 | 1 | | | |
| | 40.2 | 1 | 1 | | | |
| DXS10146 | 22.1 | 2 | | | 1 | 1 |
| | 26.2 | 1 | 1 | | | |
| | 30.1 | 1 | | | | 1 |
| | 31.2 | 4 | 3 | | 1 | |
| | 36 | 2 | 2 | | | |
| | 36.2 | 20 | 15 | | 5 | |
| | 38 | 1 | | | 1 | |
| | 40.3 | 1 | 1 | | | |
| DXS10101 | 22.2 | 1 | 1 | | | |
| | 28.1 | 1 | | | 1 | |
| | 31.3 | 2 | 1 | | | 1 |
| | 33.1 | 1 | | | | 1 |
| DXS10074 | 13.2 | 2 | | 2 | | |
| | 14.2 | 1 | 1 | | | |
| | 16.3 | 5 | 3 | | 2 | |
| DXS10079 | 13 | 11 | 9 | | 1 | 1 |
| HPRTB | 14.2 | 1 | | | | 1 |
| Total | | 166 | 96 | 9 | 53 | 8 |