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KINSHIP ANALYSIS

DNA-Based Kinship Analysis

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What is kinship analysis?

Relatedness between individuals and groups can be investigated using DNA markers. A child's DNA profile is a combination of alleles passed down from the father and mother. This means that relationships can be investigated between alleged family members.

DNA profiling is commonly used to test for potential paternity, parentage and sibship (whether people are related as brothers or sisters) relationships. In many forensic cases more complex relationships have to be considered.

What are common applications of kinship analysis?

In The Forensic Science Service's experience, common applications of kinship analysis are paternity, forensic casework and immigration disputes. More unusual circumstances can include identifying victims of mass disasters or recovered remains in missing person enquiries. The Forensic Science Service has designed the FSS-ibd application to analyse all of these types of relationship problems and to be particularly useful in more complex relationship scenarios, where the software application can complete the analysis in minutes. It is an effective tool that is cost-efficient, reduces processing times and creates unique reports according to the user's programmed specifications.

The FSS routinely uses kinship analysis in forensic casework and commercial applications for clients in the UK and internationally.

Case Example 1

Hyo Jung Jin and In Hea Song—The body in a suitcase mystery solved

The discovery of an almost-naked female body in a suitcase near York left police and scientists almost no clues as to her identity or who had killed her. But a stroke of luck helped detectives name the dead woman, and an investigation stretching across three continents began after her death was linked to the murder of a second Korean woman in London.

The gruesome discovery was made in November 2001 after a man walking along a lane in Askham Richard, North Yorkshire, spotted the suitcase and became suspicious when he was unable to lift it. The body inside appeared to be that of a young woman of Asian origin; her face was bound with distinctive tape.

Examination of the body by an anthropologist from Glasgow and ethnic testing carried out by The Forensic Science Service (FSS) using the red-hair analysis test as an ethnic marker, confirmed that the woman was South East Asian (Oriental) rather than Indo-Pakistani. Despite dental examination, DNA analysis and fingerprint recovery, investigators could not identify her, so a portfolio of this information was prepared for Interpol. Her details were matched to a Southeast Asian web site listing missing people.

The Korean government holds fingerprints of citizens for official purposes, and these were used to help identify the body of Hyo Jung Jin. The 21-year-old student at Lyon University had come from France to an address in west London on a

For kinship analysis, autosomal STR markers are the most discriminating and are used routinely in The Forensic Science Service. In addition, mitochondrial DNA can be used to examine the maternal line, and Y-STRs are used to study paternal-line inheritance patterns.

sightseeing trip but had disappeared almost immediately. Her death was quickly linked to the disappearance of a second Korean student, In Hea Song, whose body was later found in a concealed cupboard in March 2002 at an east London address in Poplar. The landlord of both properties was 31-year-old Kyo Soo Kim.

The death of Miss Jin and the disappearance but later the death of, Miss Song were investigated jointly following a meeting between the North Yorkshire Police, Metropolitan Police and The FSS. FSS scientists produced a range of evidence, which the prosecution used to link Kim with the discovery of the suitcase. The landlord had been living at the west London address when Miss Jin went missing.

DNA kinship analysis was used to identify the body of Miss Song, found in the concealed cupboard in Poplar, by comparing the dead woman's DNA profile with DNA profiles obtained from her parents.

Evidence from a property in Canada, where Kim had been staying, linked him to the body in the cupboard. Miss Song had been gagged and bound with tape. Orange paint found on a T-shirt taken from a house in Toronto matched paint found on the tape used to bind her wrists. DNA taken from the T-shirt matched Kim, linking all three elements. In addition, property in the cupboard with Miss Song was linked by DNA to Miss Jin.

Following a trial at the Old Bailey in 2003, Kim was convicted of both murders and sentenced to life imprisonment.

What types of markers are most useful for kinship analysis?

The circumstances of every case will be very different, and the scientist will select the technique to yield the most useful information according to these. Facility resources also may dictate which processes are deemed most suitable.

Autosomal STR markers are the most discriminating and are used routinely in The Forensic Science Service. Mitochondrial DNA can be used to examine the maternal line, and Y-STRs are used to study paternal-line inheritance patterns. Mitochondrial DNA and Y-STRs can provide useful information; however, the discriminating power is lower than that of autosomal STRs.

Following an extensive kinship analysis using autosomal STR and mitochondrial markers, The FSS concluded that the bones are those of the last Tsar, Nicholas II, and his family.

Case Example 2

The Romanovs—The mystery of the Russian family solved

Skeletons found by construction workers in a shallow grave in Yekaterinburg, Russia, in July 1991 were tentatively identified as the remains of the last Tsar, Tsarina and three of their five children—the Romanov family.

Tsar Nicholas II, his wife Tsarina Alexandra and their five children Olga, Tatiana, Maria, Anastasia and Alexei were executed by Bolshevik troops following months of captivity. They were shot in the house of a local merchant on 17 July 1918.

Their bodies were to have been transported to a mine shaft for disposal. However, the transport truck developed a mechanical fault along the way. A shallow grave was hastily dug on the roadside, then the bodies were burned and sprayed with acid to make them unidentifiable. The bodies of two of the children never were found.

Despite the collection of extensive forensic evidence, this version of events never had been positively verified, and in 1992 The Forensic Science Service (FSS) was approached by the Russian authorities to initiate an Anglo-Russian investigation to authenticate the remains using DNA analysis.

Using samples taken from the surviving bones, The FSS performed DNA-based sex testing and STR analysis, the results of which confirmed that a family group was present in the grave. In addition, mitochondrial DNA analysis was employed.

Mitochondrial DNA is a tiny amount of the total DNA present and can be used when samples are too small, old or degraded for analysis by normal means. Where there is no body fluid or tissue available mitochondrial DNA can be taken from bone. Mitochondrial DNA is more likely to survive for prolonged periods than chromosomal DNA and is particularly suited to tracing maternal inheritance and testing relatedness if there are several generations between ancestor and living descendant.

Following this extensive analysis, The FSS concluded that the bones are those of the last Tsar and his family.

KINSHIP ANALYSIS

What are some limitations of kinship analysis?

The Forensic Science Service understands that there are limitations to kinship analysis. The 'strength of evidence' can depend on a number of factors such as the availability of close relatives for testing. The potential evidential strength of a kinship analysis decreases as the genetic 'distance' between the individuals increases. The Forensic Science Service would expect kinship analysis to provide a higher level of evidence if the case involved two parents and a child rather than two half siblings.

In some instances scientists may suspect that a genetic mutation has occurred. A mutation is a genetic event where an allele changes identity when transmitted from parent to child. In relationship analysis this is often perceived as an apparent inconsistency in the Mendelian pattern of inheritance, such that at one locus a child apparently inherits an allele that is not represented in either parent. An example is shown in Figure 1.

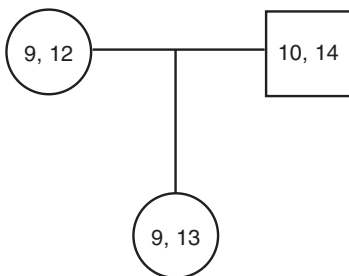


Figure 1. An inconsistency in the Mendelian pattern of inheritance. The 'mother' has apparently passed the '9' allele to the child. If the 'putative father' is the natural father of the child, he has passed either the '10' or '14' allele, and there has been a mutation event with the transition '10' to '13' or '14' to '13'.

How often does The Forensic Science Service perform kinship analysis?

The FSS undertakes kinship analysis on a daily basis when cases involve body identification, familial searching, incest and relationship testing for questioned paternity or immigration casework.

What tools are available to labs interested in using kinship analysis?

The Forensic Science Service has introduced a product called FSS-ibd, which automates relationship analysis. It can improve the speed, efficiency and cost-effectiveness of a laboratory process dramatically.

FSS-ibd accepts input files from the FSS-i³™ Expert Systems Software and other commercially available analysis systems, and data can be added manually. The relationship 'problem' is defined by drawing pedigree trees using a graphic user interface (Figure 2). DNA profiles generated for individuals involved in a case or investigation are assigned to the pedigree. FSS-ibd takes care of the mathematics—even for complex analyses.

FSS-ibd can analyse data using a variety of user-defined parameters, including: mutation, theta corrections for population data and size-bias corrections for rare alleles, and can use any of the commercially available STR systems. In addition, FSS-ibd can be programmed to produce user-specific reports.

Who would benefit from using kinship analysis software?

Development of software solutions in The FSS is driven by the need to improve efficiency and quality in forensic processes and/or drive down analysis costs. By making the application commercially available we can bring these benefits to other international forensic laboratories.

The Forensic Science Service has introduced the FSS-ibd software, which automates relationship analysis, to improve speed, efficiency and cost-effectiveness.

The FSS-ibd has application in a wide range of laboratory casework situations where relationship analysis plays a part:

- Forensic relationship analyses, including incest
- Paternity or parentage issues
- Immigration and nationality testing
- Identification of missing persons
- Identification of victims of mass fatality incidents

FSS-ibd even has the potential for use in animal pedigree testing. As long as the laboratory uses an STR marker system for the pedigree analysis, data can be added to the FSS-ibd application. The user can input new multiplexes and allele frequency tables.

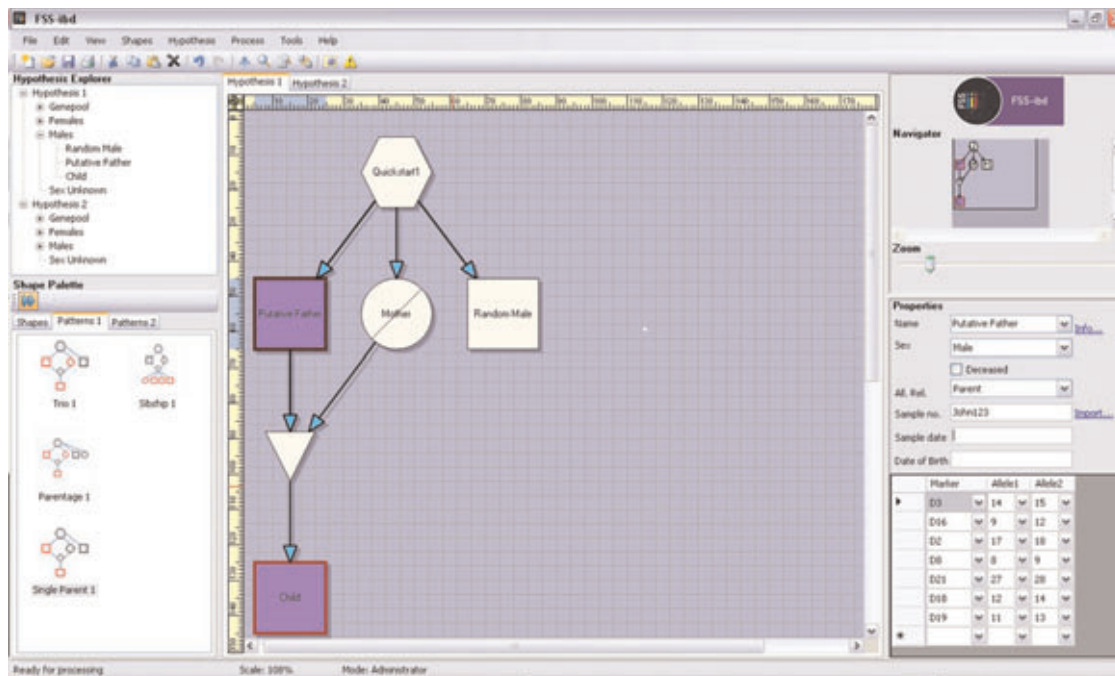


Figure 2. The interface for the FSS-ibd software for relationship analysis.

What are some unique features of the FSS-ibd software?

Like all of our DNA expert systems, FSS-ibd is built to the highest scientific and technical standards. It has a clear graphical interface in which pedigrees can be built rapidly (by a 'drag and drop' technique) and adapted to represent the specific case under investigation.

User-definable STR marker systems and allele frequency databases can be added to the FSS-ibd application. In this way the user defines the parameters used to calculate probabilities and likelihood ratios from the hypotheses under test. The analysis parameters can be chosen to reflect laboratory-specific requirements to take account of rare alleles (minimum default frequencies or size-bias corrections), mutations and Theta values (coefficient of inbreeding of a population).

FSS-ibd also creates a user-defined technical audit file, which contains case details, process settings, DNA profiles, allele frequencies, probabilities for each hypothesis by locus, likelihood ratio by locus and overall likelihood ratio. The application also has the functionality to produce user-defined statements or reports.