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DNA testing Falsehood: Conditions and Facts

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Abstract

DNA as an evidence plays a very important role in establishment of human identity through genetic profiling both in criminal and civil disputes. DNA has emerged as a powerful investigative tool as no two persons can have same DNA profile except for identical twin. The proper collection of any biological samples is must for effective utilization of DNA as medicolegal evidence. Single biological sample such as blood, semen, saliva, and hairs etc without any contamination can easily identify a single person. But, if there is mixing/contamination of the biological samples or there is insufficient sample collection from the object or surface for genetic profiling then the results can be ambiguous. In such situations, reports are inconclusive, also it could be difficult to exclude genetic sample of perpetrator from such mix profile results. Furthermore, the new age treatment therapies such as In-Vitro Fertilization (IVF), Blood transfusion, bone marrow transplantation and organ transplants can also hinder to present the actual identity or genetic profile of a person due to genetic mixing. Therefore, biological samples collected from crime scene can either match with suspect or acquit him/her from suspicion. Modern DNA forensic methods are powerful and sensitive, carelessness or ignorance in proper history and handling procedures for biological evidence can result in an unfit sample for analysis.

Keywords: DNA evidence; Genetic profiling; Reliability; Transplant therapies; Fallacies.

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Introduction

DNA is a powerful investigative tool because no two individuals have same DNA profile except identical twin. DNA evidence plays a very important role in identification of human identity through genetic profiling in civil dispute and criminal cases. The most reliable tests study as a part of the DNA testing is Short Tandem Repeats (STRs).[1] The method is called multiplex polymerase chain reaction (PCR). PCR is a method of amplifying and sequencing DNA. It is the method used to diagnose hereditary and infectious diseases for clinical aspects. DNA testing is considered to be the only completely accurate method of determining parentage in the medical community.Over the years, genetic parental testing technology has advanced substantially. During 1920's, paternity was determined through blood type. Blood types are not exclusive for genetic relationships; while they can be used to exclude certain people but cannot conclusively determine the parentage.[2,3] After more perceptive knowledge regarding DNA, an easier and correct techniques has been developed. Current methodology utilizes study of a particular section (short repeats) of DNA on chromosomes which determines genealogical heritage i.e. genetic characteristics passed from generation to generation. In forensic cases DNA testing, establishment of paternity or maternity of child were involved and revers parentage for identity of deceased.[4] The analysis of DNA testing worldwide is usually based on comparison of profiling of biological evidence with reference samples by using multiplex PCR technology. In this paper we are highlighting the possibilities of fallacies during DNA profiling.

Strengths of DNA testing in Forensic community

Although 99.9% of human DNA sequences are the same in every person in the world, there is still enough difference in order to distinguish one person from another. Using a method called DNA testing, also known as DNA profiling where experts or scientists analyses a long chain of DNA to identify specific "loci." These loci are very similar when they are comparing the loci of two closely related people, but the differences are much greater among unrelated people. Thus, in criminal prosecutions, DNA evidence is often offered to link the presence of accused with being at the scene of crime, as well as be often used by the defendant to prove his/her actual innocence.[5]

In contrast to this all DNA evidence is not equal. Sometimes its clear single biological sample such as blood or semen that can identifies a single person. If it's contains more than one person's biological samples or it is just a few skin cells left on an object as a genetic material, then it can be more ambiguous for experts. In such situations, report were not conclusive, or the defendant could not be excluded from the mix profile result.There is variation in statistical approaches used to evaluate the strength of the evidence where involvement of a particular person is decided than the approaches used must be supportable.[6,7]

A. Social and legal source of evidence

DNA analysis has brought a strong change in the world in medicine, especially forensic domain. So majority of forensic cases analysed by DNA testing involve disputes and crime via paternity or maternity of child or to identify unknown deceased. In this regards new methods have been developed, validated, and put into use to help in criminal investigations. New approaches for interpreting evidence via probabilistic modelling are being introduced. The validity and accuracy of older and current methods are even challenging. [7] The DNA analysis is based on comparison of result of biological evidence with reference samples such as blood /buccal swab. Sometimes stored or preserved biological and other intimated items of individuals like tooth brush, cloths, shaver, other object collected at crime scene are being send for DNA testing to establish the identity of deceased, victim, accuse; under such circumstances authentication of test is usually problematic.[8]

B. The technical reliability of DNA evidence depends on various aspects

1. Quantity and quality of the sample analysed
2. Laboratory equipment / technique in analysing the sample
3. Polymerase chain reaction (PCR) based testing is relatively insensitive to degradation
4. Analysis of poor quality DNA samples may lead to uncertain results requiring substantial interpretation by the forensic scientist
5. When a DNA sample contains a mixture of several persons' DNA, and the forensic scientist does not account for this, the resulting DNA profile may be incorrect.

C. Laboratory Accreditation

1. Accuracy of DNA analysis depends on the quality control and quality assurance procedures in the forensic laboratory.
2. Quality control refers to measures to help ensure that each DNA analysis result (and its interpretation) meets a required standard of quality.
3. Quality assurance refers to monitoring, verifying and documenting laboratory performance[9]

D. Human errors during sample handling

As it is very sensitive technique the DNA samples may be cross contaminate with other human DNA depending on nature of crime where crime scene samples may contain a mixture of fluids (biological and non-biological) or tissues from different persons. Moreover the crime scene sample may be contaminated during sample handling and transferring from the crime scene to the laboratory; or carry-over contamination can happen during PCR-based testing if the amplification products of one test are carried over into the mix for a subsequent PCR test. Sample mishandling, mislabelling or contamination is more likely to compromise a DNA analysis than an error in the analysis. Samples can be contaminated at any stage of collection, transportation and analysis of DNA samples. Laboratory staff unintentionally could make errors in conducting DNA analysis, in interpreting or reporting the results of the analysis, or in entering the DNA profile result after

analysing the allelic data. This might result from failure to comply with an established procedure, misjudgement by the expert, or some other mistake. While protocols and precautions can be introduced to minimise the opportunity for error during analysis or interpretation, the potential for human error cannot be fully eliminated. [10]

E. Tempering or indirect transfer with the samples at crime site

A suspect's DNA profile might match the profile found at a crime scene as a result of tampering with the crime scene, or subsequent substitution of DNA samples. This might occur where the actual offender, a police investigator, or another person deliberately leaves a suspect's genetic sample at the crime scene. [11] Alternatively, it is possible that a suspect's sample might later be substituted for the actual crime scene sample to falsely implicate the suspect in the offence. There may be possibilities of such instances during practice and procedure for the collection of crime scene samples and handling of these samples for transfer to the laboratory, and at the laboratory itself, the steps to minimise the opportunity for tampering must be assured. In contrast, complexity and different possibilities of DNA transfer make the identification of contamination incidents quite difficult as biological material can not only be transferred by means of direct but also by means of indirect transfer during sample collection and transportation.

A match between the crime scene profile and a defendant's profile does not prove that the defendant committed the particular offence. There may be several alternative explanations for a match, because there is possibility that sample was deliberately left at the crime scene during or immediately after the offence and could be 'planted' at the crime scene. False positive result may be possible due to error during DNA handling.[12]

F. Samples of close relatives

Close genetic relatives have more common genes than unrelated persons. Therefore, it is possible that an innocent person's DNA profile could match with the profile obtained from a crime scene, where the offender was in fact that person's sibling or other close relative. However, the chance of such a coincidence will decrease inversely as the number of loci examined along the DNA molecule increases [13]. In this condition sample originated from a close relative of the suspect; or it originated from an unrelated person who by coincidence has the same DNA profile as the suspect.[14]

G. Cases of Mix DNA profiling

The evaluation and interpretation of forensic DNA mixture evidence has greater interpretational challenges due to increasing complex mixture evidence. Such type of challenges are occurs when low quantity or degraded DNA evidence shows dropouts of locus and allele, allele staking and stutter artefacts instead of true alleles. There are common concerns that methods utilized for interpretation of complex forensic DNA mixtures may not be implemented properly in some casework. Similar questions are being raised which lead to some confusion about mixture interpretation for current and previous case works.[15]

H. Disputed Parentage test

Parentage testing mainly relies on autosomal STRs DNA profiling, the cases of monozygotic twin, motherless paternity or fatherless maternity (where child and one parent are available), in endogamous and consanguineous generally showed inconclusive DNA profile because of common alleles where as in cases of mutation or degradation showed incomplete DNA profile. The conventional DNA test methodology by 15 STR loci can lead to false inclusion in very closely related parent and child or siblings as they share more obligatory alleles than the unrelated.As parentage testing determines an alleged individual is biological parent of disputed child are based on likelihood ratio, if alleged parent are sharing common alleles it is difficult to eliminate.[16,17] Similarly for monozygotic twins involved in sexual assault cases DNA testing is challenging and even the sample of product of conception (POC) can be contaminated or produced mixed DNA profile.In cases of child swapping in hospital or other places and adopted child may give mismatched DNA results.

I. Cases of rape survivor

Rape is the great social problem of society and it is heinous crime in many develop and underdeveloped countries. It has been reported that rape cases are often committed by persons who are known to the victim and 5%-15% of assaults are perpetrated by a stranger or unknown to victim. The biological evidences play important role in sexual assault cases. In such crime for DNA profiling, biological samples of victim and accused required which includes epithelia cell and spermatozoa

(semen). In rape cases improper or delayed collection (after 24 to 72 hours) of samples and low quantity of DNA can give inconclusive DNA profiling results. On other side if involvement of more than one person (gang rape), generates a mix DNA profile.The technique used for differentiation of male and female DNA is not always successful. [18]

J. Methods of treatment: Bone marrow/Organ transplant and IVF

Advancements in treatment procedures and new methods of treatment have evolves to save lives by transplant therapies. An organism having two or more genetically distinct cells is called chimera. In cases of bone marrow or organ transplantation therapy, chimeras testing can be performed for acceptance or rejection of patients who have received a hematopoietic stem cell transplant. The test involves identifying the genetic profiles of the recipient and of the donor and then evaluating the extent of mixture in the recipient's blood or bone marrow. There are cases when man has transplant bone marrow, his DNA changed to that donor who has live far away from the recipient.[19–21] In in-vitro fertilization (IVF) biological father can be donor for the children or any other complication during such procedure can be misinterpreted DNA profile. Other hematological disease condition where blood transfusion is the only treatment procedure like thalassemia, anemia, and also some form of chemotherapy may also cause some misinterpretation of results.[22–24] It can be inferred that in criminal offence cases, some specific medical treatments can hinder the actual identity of person due to mix profile.

Conclusion

In the DNA profiling procedures to choose a suspect, investigators try to match the sample for loci and the suspect's loci as closely as possible. But often, crime scene samples are imperfect and the DNA breaks down, so the loci are weak and unable to generate good profile. That makes finding of 100 percent match very challenging. Modern DNA forensic methods are powerful and sensitive, but great care must be taken to prevent unfairness of justice. The methodology for DNA test opted by the expert is also a crucial, good practice of laboratory protocol is also important to make the uniformity in DNA profile. Carelessness or ignorance in handling procedures as well as case false history for biological evidence can result in an unfit sample for analysis.

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