

# DNA profiling: Social, legal, or biological parentage

A. K. Sharma

Central Forensic Science Laboratory, Directorate of Forensic Science, Ministry of Home Affairs, Govt. of India,  
30 Gorachand Road, Park Circus, Kolkata - 700 014, India

DNA profiling in forensic casework is based on comparison of the results of biological evidence with direct reference samples of the individual concerned or with indirect references of his close blood relatives. The selection of reference samples for analysis is crucial to the success of a case; it not only depends on the authenticity of the reference samples, but also on the authenticity of the biological relation of the donors with the person in question. There are situations when the social or legal relationship is not the biological one and there is a need to educate investigating officers, forensic analysts, and the judiciary about the associated problems.

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The majority of the forensic cases analyzed by DNA testing involve establishment of paternity/maternity of the child or reverse parentage (for identity of the deceased). The analysis is based on comparison of the results of biological evidence with reference samples (blood or oral swab). Direct references like banked biological specimens preserved in medical or military repositories are a valuable source for identification purposes. Alternatively, intimate items of an individual, e.g., toothbrush, shaver, razor, etc. are good sources of DNA to establish the identity of deceased, but their authentication and exclusiveness is often problematic. Any doubt regarding the reliability of direct references will be detrimental for the case and may lead to false exclusions. Further, passengers often travel with their toothbrushes, razors, etc., and these items may not be available in an airline disaster.

Indirect references of close blood relatives of the person to be identified are usually desired for establishing identity. A DNA profile for a multiplex of 15 autosomal short tandem repeat (STR) markers is generated and obligatory alleles are compared with that of parents, siblings, or close relatives for kinship analysis. Indirect

reference samples of the following relatives are usually preferred in a kinship case:

- (a) Either or both biological parents of the victim.
- (b) Spouse (biological mate) of the victim and their child/children.
- (c) Biological full siblings, sharing the same parent as victim.<sup>[1]</sup>

An inconsistency at two or more loci (considering the mutation rate of STRs) generally leads to exclusion in a kinship case. Inclusion at all loci is statistically evaluated by calculating paternity, maternity, or sibship indices. In the absence of close relatives, mtDNA or Y-chromosomal markers are employed for establishing maternal or paternal lineage, respectively, by comparison with distant relatives. In mass disasters, identification with reference samples from multiple relatives is recommended to avoid false inclusions.<sup>[2]</sup>

The success of a DNA case not only depends on the authenticity of the reference samples but also on the authenticity of the biological relationship of the donors with the person in question, without which any comparison is futile. Often, the analysis in a DNA case is based on assumptions that:

- (a) Monozygotic twins or close blood relatives are not involved.
- (b) Reference samples belong to the persons they are attributed to.
- (c) Donors of samples have the same biological relationship with the person in question, as presumed.

However, such assumptions do not hold true in some exceptional situations and pitfalls should be clearly understood while collecting or analyzing reference samples.

## Monozygotic Twins and Close Relatives

Monozygotic twins are derived from the zygote of one sperm and an ovum which splits into two or more zygotes. Such twins cannot be differentiated with the conventional DNA typing methodologies and a case involving them can lead to false inclusion. A few cases of sexual assaults, involving monozygotic twins, in UK and USA have been solved based on other evidences like tattoo marks and fingerprints.

Similarly, closely related individuals like parent/child or full siblings often share more obligatory alleles than unrelated individuals and distinguishing them can be a difficult task in a parentage dispute. If there is a possibility of involvement of close relatives in a case, the DNA profiles of such relatives should be prepared, whenever feasible, for eliminating them from consideration.<sup>[3-4]</sup> However, if there is no information about the involvement of close relatives in a case, it may lead to complications. The following types of cases are sometimes difficult to examine with the present multiplexes of 15 autosomal STRs loci:

- (a) Motherless paternity or fatherless maternity-deficiency cases.
- (b) Incest cases.
- (c) Full siblings impersonating as parent/child.<sup>[5]</sup>
- (d) Cases involving mutations.

Additional DNA markers help to differentiate closely related individuals or situations involving mutations. Goodwin *et al.*<sup>[6]</sup> reported a case where 21 autosomal STR loci were required to identify the true father between two brothers while determining paternity of a child. Advances in epigenetics may help in differentiating monozygotic twins in future by exploiting the differences that accumulate over a period of time.<sup>[7]</sup> Similarly, use of parent-of-origin specific DNA methylation markers to identify the parental origin of alleles can help in the deficiency cases as well as in cases involving close blood relatives.<sup>[8]</sup>

## Chimeras

A kidney transplant patient underwent HLA typing test to identify a suitable donor from among her children. To her surprise, the results from her blood concluded that

two out of her three sons did not belong to her. While all three sons shared haplotype with the father, only one shared it with the patient. Further investigations into her DNA from buccal mucosa, hair follicle, skin, thyroid, and bladder, along with samples of close relatives, led to the discovery that she was a tetragametic chimera - a mixture of two individuals, formed by the fusion of two zygotes in the womb which grows as a single individual.<sup>[9]</sup>

A chimera is an organism having two or more different populations of genetically distinct cells, originating from different zygotes. If different cells emerge from the same zygote, it is called mosaicism. A chimera may be due to transfusion, transplantation, or inheritance. In the non-spontaneous form of human chimeras, the transplant or transfusion recipient has a mixture of different organs or different blood, respectively, which is intentional. A person can be a spontaneous chimera through inheritance, either by the free passage of blood between mother and child or between child and child in the uterus. This free passage of blood may result in a condition known as microchimerism. Forensic geneticists are aware of the complications that can arise to transfusion or transplantation. However, the occurrence of a rare spontaneous tetragametic chimera, formed by the merging of embryos in the uterus, can lead to a false interpretation in the analysis of stain material in crime cases and in paternity testing because a mixed pattern of different genotypes can be obtained in one individual.<sup>[10]</sup> Since in most of the cases maternity is not in doubt, any discrepancy in the results should be checked by collecting samples of different tissues from the mother. But if the father in a paternity case is a chimera, with different genetic make up in sperm and blood cells, he can be falsely excluded by DNA profiling. Though tetragametic chimerism is a rare event, its incidence is likely to increase with the increase in accessibility to *in vitro* fertilization, as the embryos are in close contact.

## Authentic Reference Samples

History of sample swapping is as old as the use of DNA profiling in criminal cases. In the first criminal case analyzed by DNA testing, the blood sample of the rapist and murderer, Colin Pitchfork, was swapped. Any change in the reference samples or fudging will lead to erroneous results. If investigated properly, sample swapping can be identified. Unintentional swapping of samples at the time

of collection by the authorities in a paternity case has been observed in this laboratory; the error was identified from the labels present on the samples and was later proved by DNA test.

### **Biological Parentage vs. Social or Legal Parentage**

The authenticity of the relationship of the person in question with the donors of reference samples should be clearly understood at the time of collection of samples, evaluation of results, as well as at the time of evidence deposition in the court. There are situations when the social or legal relationship is not the biological one. For an investigating officer collecting the reference samples and the DNA analyst, the donor contributing to the biological (genetic) formation of the child is more important rather than social or legal close relative.

The donor of the reference sample in a forensic DNA case:

- (a) may know the true biological relation he has with the person in question and disclose it (like adoption, step relationships, assisted reproductive technologies (ARTs)).
- (b) may know the true biological relation of other donor has with the person in question and disclose it.
- (c) may know the true biological relation of other donor has with the person in question and may not disclose it (like false paternity).
- (d) may not know the true biological relation he has with the person in question (child swapping, ARTs).

Following are the scenarios where we may find conflict among biological parentage with the social or legal parentage:

#### *Adoption*

Adoption of children is usually well documented and known. It is expected that such facts will be disclosed at the time of collection of blood samples in a forensic case, should the need arise. Similarly, details of step relationships are usually known and must be disclosed.

#### *Assisted reproductive technologies (ARTs)*

Children conceived through new reproductive technologies employing donor sperm, egg, or gestational

surrogates lead to different opinions regarding their social or legal parentage. A child conceived by ART can have as many as five parents: a genetic father who donates sperm, a genetic mother who provides the egg, a surrogate mother, and two parents who have no biological connection to the child but who commission the other parties to start a family. ART has introduced new definitions of parenthood: the genetic, the gestational (which is also biological, but is different from genetic), and the social and nurturing. Most legal frameworks place the rights of the gestational mother and/ or social parents above the rights of biological donors, particularly if the donation has been anonymous. Anonymous donation may also lead to the existence of several half-siblings who have no knowledge of one another.<sup>[11]</sup>

There are valid fears that fertility clinics, after having failed to help the woman conceive with her husband's sperm, may inseminate her with frozen sperm without divulging the information to appear successful. Dr. Cecil Jacobson, nicknamed the "Babymaker" or "Sperminator," during artificial insemination procedures at his fertility clinic, fathered a number of children by fraudulently injecting his own sperm into his patients, instead of the sperm of the husband or of an anonymous donor.

The Indian Council of Medical Research guidelines makes it voluntary on the part of ART clinics (with the approval of the couple) to keep on record DNA fingerprints of the donor, the child, the couple, and the surrogate mother. The information about the donors can be released when specified and required for the legal purposes.<sup>[12]</sup> Instead of having a DNA profile for a limited number of markers, the storage of biological samples of all persons contributing to the biological formation of the child would be more meaningful from the medical as well as forensic point of view in case of any eventuality in the future. A legal act is required to enforce mandatory maintenance of such repositories of biological samples by ART clinics and for regulation of the sector.

#### *False paternity*

In the situation of false paternity, the mother usually fails to disclose the information, fearing social stigma or the embarrassment it can lead to. Establishing the identity of a child (missing or deceased) born in this situation not only complicates the analysis of results but also raises

ethical questions regarding the reporting of the results. Similarly, establishing the identity of a (deceased) father from, for example, the skeleton, employing indirect reference blood samples of the wife and child will lead to false exclusion if the social father did not sire the child.

### *Child swapping*

There may be situations where the donors do not know the truth about their biological relationship with the child. This may happen when the child has been swapped at an early age without the knowledge of the parents. Several cases of baby swapping in hospitals, inadvertently or fraudulently, have been reported. Identification of the swapped child later by DNA using indirect reference samples will give misleading results.

In an unusual case of paternity determination received in our laboratory, the blood samples of the three were examined. The alleged father in the case did not share an allele with the child at six autosomal STR loci. Surprisingly, the woman also did not share an allele with the child at seven autosomal STR loci, showing maternity exclusion. Mitochondrial DNA analysis excluded her having maternal lineage with the child and also excluded the possibility of a tetragametic chimera. Due to the unexpected results, the court ordered a re-collection of the blood samples; reexamination yielded identical results. If the possibility of fudging of samples is excluded, this situation may be either due to:

- (a) Concealment of facts about the maternal origin of the child by the putative mother.
- (b) Child swapping.

In the latter scenario, though the complaint of the putative mother may be genuine, DNA technology can not help her. Instead, DNA profiling results may lead to strained relations between her and the child, adding to misery.

### **Legal Preference to Social Parentage**

Indian courts have given more importance to social parentage than the biological one. Echoing the maxim *Pater est quem nuptiae demonstrant* (the father is he whom the nuptials indicate), Section 112 of the Indian Evidence Act, 1872, is based on the rule that the child born in wedlock should be treated as the child of the

man who was then the husband of his mother. The only exception is when the husband proves that he had no access to his wife at the time of conception of that child. The legislative concern is against illegitimizing a child as he should not suffer social disability because of the lapses of parents. The Supreme Court emphasized that Section 112 of the Evidence Act was enacted when DNA tests were not even in contemplation of the legislature. The result of a genuine DNA test is said to be scientifically accurate, but even that is not enough to escape from the conclusiveness of Section 112 of the Act, e.g., if a husband and wife were living together during the time of conception but the DNA test reveals that the child was not born to the husband, the conclusiveness in law would remain un rebuttable. This may seem to be hard on the husband who would be compelled to bear the fatherhood of a child of which he may be innocent, but even in such a case, the law leans in favor of the innocent child from being bastardized, if his mother and her spouse was living together during the time of conception.<sup>[13-15]</sup>

DNA profiling is the most effective tool for justice in criminal and civil cases. The above-mentioned exceptional situations are rare, but it is important that investigating officers, forensic analysts, and members of the judiciary be aware of the necessity of obtaining authentic biological (genetic) samples and of the problems that may be encountered.

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