Chimerism and Forensic DNA Profiling in Crime Cases

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ABSTRACT

An individual having two kinds of blood cell populations or two sets of deoxyribonucleic acid (DNA) types in different body tissues is called Chimeric and the situation is called as chimerism. This makes diagnosis and comparison of tissues for organ transplants and forensic investigations complicated and may lead to erroneous results.

A few such cases, highly publicised in the past, have been mentioned in this paper. How chimerism affects DNA profiling results in forensic science and can lead to situations where the accused may be exonerated, and a genuine offspring of one's parents may be denied parentage has been highlighted. How chimerism can be detected, and flawless reports can be given, the challenges faced in forensic cases have been mentioned in this paper.

Keywords: Chimerism, Tetra gametic chimerism, Cell population, DNA profiling, Forensics, Parentage, Maternity

INTRODUCTION

Chimerism is the situation when an individual or the tissues possess at least two types of different DNA, mostly coming from two different zygotes or fertilised eggs. The word *"chimera"* is believed to have been derived from Greek mythology, in which a chimera is said to be a fire-breathing individual with the head of a lion, body of a goat and the tail of a snake. [1]. The exact percentage of tetra gametic chimerism in the human population is unknown because most cases go undiagnosed, and there is no routine screening for it. In medical literature, only 100 cases of human chimerism have been documented in the world.

The condition is thought to be extremely rare, but some experts think the actual existence of natural human chimeric individuals may be up to 10% in human population. But, without authentic data it appears to be exaggerated. Natural human chimeras are divided into three groups. Micro chimerism which is formed by foetal-maternal cells mixing across the placenta. Fusion chimerism comes into being by the fusion of two zygotes, and twin chimerism, which also termed as blood chimerism or transfusion chimerism, occurs by the transfer of cells between dizygotic twins through fused placenta [2].

Appreciable signs of Chimerism

Human individuals with chimerism seldom show appreciable signs of their chimerism condition [3], while some may have physical symptoms as follows [4]:

- Both eyes in different colour.
- Separate kinds of skin colours due to hyper and hypopigmentation or differently coloured patches of skin.
- Differently coloured patches of hair.
- Due to disorderly sexual development, the genitalia may not be clear and may be infertile.

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Human individuals with chimeric situations may also depict the following, but rarely [5]:

- Initially, twins may start developing during early pregnancy but, due to vanishing twin syndrome, one of the twins may not survive.
- DNA test does not give clear maternity or paternity test results due to unknown reasons.
- There may be transgender identification due to involvement of X and Y chromosomes.
- The individuals may be ambidextrous.
- Individuals may exhibit two blood types.

Diagnosis, Detection and Effects

For the diagnosis and detection of chimerism, some laboratories use short tandem repeats (STRs). However, some other laboratories recommend more sensitive methods, such as Polymerase Chain Reaction, Next-Generation Sequencing, and Droplet Digital PCR.

If not diagnosed due to chimerism there may be false negative parental DNA paternity test [5]. There are several cases of paternity testing in which chimeric mothers or fathers lost paternal rights.

Highly Reported Cases in the Media

In one case of a woman named Karen, after a DNA test of family members for the donation of a kidney to her, she was informed that two of her sons out of three could not be her genetic offsprings [6]. The detailed analysis of the samples by multiple methods later revealed that they were chimeras.

In another case, a women Lydia, from Washington state was accused of fraud, when she claimed for welfare of her children as DNA test showed that the children were not her genetic offsprings. She was exonerated when her attorney's cited Karen case [7].

In the third case, a man living in California and his wife, both has blood group 'A', but during paternity test the child was found to be of blood group 'AB' failed to prove the paternity of the child [8]. The child was a chimera in fact.

All three cases turned out to be cases of chimerism, each made up of two individuals of the same sex. Thus, they are tetra-gametic chimeras. Further, investigations revealed that DNA profiles from some tissues of the parents did match with that of the disputed children.

It is not out of place to mention here that some persons also show chimerism with respect to blood groups, that is, having different cell populations, for example, A_1O. The secretor factor has a direct relationship with chimerism, particularly in resolving blood group discrepancies and confirming the presence of two different genetic cell populations in a chimeric person.

In blood group studies for practical applications, researchers have shown that:

- In twin chimeras, where twins were secretors and had different ABO groups, the true genetic group could be determined from their saliva.

- A study on male twin chimera found inconsistent genotypes while comparing his peripheral blood with his buccal cells, with the secretor status analysis of the buccal epithelium confirming the true, consistent genetic line of the tissue.

- Investigations of the interactions among the ABO, Lewis and secretor histo-blood group systems are standard procedures used to resolve complex cases of haematopoietic chimerism.

Thus, the secretor factor provides an independent genetic marker that can reveal the underlying consistent genetic identity in a person whose blood is a mixture of two different cell lines.

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Chimerism and Forensic Science

There are a large number of cases in forensic science, which come for DNA profiling. This takes place in the cases of:

- Babies are being swapped in hospitals and maternity homes, accidentally or deliberately.
- Rape and sexual assault cases, including gang rapes.
- Burglary and theft cases (Touch DNA from the scene of crime).
- Cases of bloodshed, that is, murders, terrorist activity, hurt and accident cases.
- Cases of human trafficking (Parentage testing for human identification)
- Cases of division of property among siblings (Parentage testing).
- Identification of the dead bodies and skeletons in events like earthquakes, disasters, and plane crashes.
- Identification of the parentage of long-lost children, who may claim to be the long-lost children of wealthy parents.

In all such types of cases, blood group typing and DNA profiling are the methods that are used for solving these cases of crime. Under chimeric genotypes and different cell populations in blood group situations, there is a likelihood of miscarriage of justice if the typing is done from blood or other single tissue and by a single method. Especially under tetra gametic chimeric situations, we can face situations of getting wrong results, as in the cases of Karen [6] and Lydia [7] and the case in California, USA [8].

Scientific Examination of Crime Cases and Chimerism

Genetic chimerism can lead to misleading results. These misleading results can further lead to false accusations and wrongful exoneration of the guilty individuals. It is because of the reason that a chimera possesses two or more genetically separate cell populations, that is, different tissue samples from the same individual may give different DNA profiles.

Challenges in Forensic Examination

1. If the DNA profile of a blood or semen sample collected from the scene of crime belongs to a Chimera's minor cell line and the reference sample in a cheek swab taken from the same individual. These may not match together and will lead to the false exclusion of the suspect.
2. Chimera's that have XY as well as XX, that is, both male and female line cells, will give confusing test results in DNA-based sex determination. A sample may show markers of both sexes or the opposite sex of physical appearance. Thus, it makes the sex determination discordant. The DNA of chimeras in forensic analysis may be interpreted as a mixture of two individuals' DNA, complicating the investigation and potentially misleading.
3. Paternity and maternity disputes may be wrongly decided.
4. A forensic scientist by mistake can interpret a single person's chimeric DNA as a mixture of two individuals.
5. There is tissue-specific variability of DNA when chimeric situations are due to blood or bone marrow transplantation. The donor's DNA may be present in the bone marrow, saliva, skin cells, etc., but may not be present in the hair roots.

The challenges can be overcome by multiple tissue sampling, DNA analysis by advanced techniques like STR typing, Y-chromosome STR typing and SNP pyro-sequencing. These techniques are more discriminating and can differentiate different cell lines in a chimeric individual. In the cases when DNA profiling and blood group typing results are inconclusive, confirmatory tests may be done to determine whether the person is a chimeric individual or not. As in Indian courts benefit of doubt goes to the accused, and all evidences should be beyond

a reasonable doubt to convict a person; the slightest doubt in DNA and blood typing reports can lead to the acquittal of the criminals based on the slightest doubt.

DISCUSSION

Right from the beginning of the 20th century, when ABO blood group system was discovered by Karl Landsteiner in 1901, there have been tremendous development in the field of blood group serology, enzyme typing, serum protein typing and ultimately in 1985, the technology of DNA profiling in forensic science was introduced by Professor Alec Jeffreys while working in the University of Leicester in England. Initially, blood and body fluid-stained forensic samples were examined for match or differentiation of the biological fluid on the basis of blood groups, enzyme and serum protein typing. From 1985 onwards, more emphasis was laid on typing biological fluid-stained exhibits through DNA profiling using the method Restriction Fragment Length Polymorphism (RFLP). Later, Short Tandem Repeats (STR), Single Nucleotide Polymorphism (SNP) have been used for examination and comparison of the biological fluids and their stains in paternity dispute cases, homicide cases, hurt cases, sexual assault cases, shootouts and explosion cases. Not only has DNA profiling been carried out in almost all kinds of crime cases in which touch DNA was recovered from the scene of the crime. DNA profiling has also been carried out for the identification of persons/ dead bodies in cases of mass disasters, plane crashes and railway accidents for human identification.

Man was always in search of a technique that could match and differentiate the biological source of the stain to 100% extent. Discovery of DNA profiling provided the probability of a near 100% match; however, there have been generic processes like genetic drifts, mutation and chimerism which created doubt in DNA technology being a foolproof technology in forensic analysis. The cases of paternity and maternity reported herein pertain to Karen, Lydia and the case of California, USA, where DNA results were misleading due to chimerism.

As in forensic case sample analysis, the samples received in the laboratories are old, putrefied, and sometimes mixtures of biological fluids of two or more persons. Chimerism can lead to highly misleading results, as a chimeric individual contains two sets of DNAs or can be from tetra gametic individuals. In such situations, forensic experts should analyse samples where negative, confusing or inconclusive results are obtained by advanced methods like PCR, NGS, ddPCR, etc. Moreover, where the accused or the victim has taken a blood transfusion or has undergone a bone marrow transplant, the investigators should inform FSLs of these facts to avoid chimeric complications. One of the authors had personal discussions with a few DNA scientists who examine forensic case exhibits/samples in forensic science laboratories (FSLs) in India. They stated that they have an excessive workload in forensic laboratories and the pendency of cases. They aren't carrying out DNA profiling of crime case exhibits with multiple and advanced methods. They have also not come across any specific case of chimerism, and the forensic laboratories are also not examining samples for diagnostic purposes.

CONCLUSIONS

DNA profiling in forensic science is a powerful tool in crime investigation and the administration of justice, but it isn't infallible under situations like chimerism, gene mutations, point mutations and genetic drift or when the population is not obeying the principles of Hardy-Weinberg's law of random mating. DNA typing in forensic science should be done with utmost care and attention, keeping in view the rare genetic anomalies as well.

A declaration regarding the use of AI tools in the writing process:

I declare that no generative AI tools were used to develop the article I am submitting.

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