

Identification of Mother of Unidentified Infant

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Abstract

Application of DNA fingerprinting in crime investigation is a routine process across the globe. Next Generation Sequencing (NGS) is the recent advancement of the technology. Capillary electrophoresis is routinely used in most of the forensic laboratory globally. In this study, an infant is disposed of in a common toilet and postmortem showed that the infant was male of an intra-uterine period of about 30 ± 2 weeks. The birth of the infant was premature and he did not respire after birth.

Since no evidence was available to connect the mother with the unknown neonate. Scientific evidence like DNA test with autosomal STR markers was the last tool to investigate the situation. DNA profiling of infant and the suspected mother was prepared by using AmpF ℓ STR $\text{\textcircled{R}}$ Identifiler $\text{\textcircled{R}}$ kit having 16 autosomal markers. Half set of alleles of infant's DNA profile was accounted in the female DNA profile obtained from suspected mother. DNA profiling of both the infant and the suspected mother proved that she is the biological mother of the infant.

Key words: Maternity; DNA Fingerprinting; STRs; AmpF ℓ STR $\text{\textcircled{R}}$ Identifiler $\text{\textcircled{R}}$ kit.

Introduction

After the discovery of DNA fingerprinting, it became a very important tool in crime investigation. Since its discovery, many advancements have taken place. Initially, restriction fragment length polymorphism (RFLP) was used for the purpose but nowadays short tandem repeats (STRs) along with SNP (single nucleotide polymorphism) became a more convenient and discriminating tool in DNA analysis.

The urge of a male child is the main cause of female foeticide in India. The present investigation has its importance because several cases of infanticide are reported every year in the country which may be either due to act of commission or act of omission but the conviction rate is poor because of the absence of shreds of evidence in such cases.

There is a need for a technique which can identify an unknown person. DNA fingerprinting or profiling is the technique of identification by which we can identify a person or link a person with a particular crime. The blood samples of claimants are required for the identification of an unknown body in the absence of a national DNA database. The technology is based on the Mendelian principal of inheritance. This mainly shows that the genetic material of an infant is contributed equally by its biological father and mother.

Various Autosomal STR loci present on nuclear DNA are inherited by both the parents. In brief, the DNA fingerprinting technology in which we are working at the molecular level has ability to identify a person with great accuracy.¹ The possibility of matching of two samples is very remote (random match probability of about 7.2×10^{-10}).

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¹⁹⁾ with CODIS recommended STRs and a gender determining marker 'Amelogenin'. DNA analysis based on autosomal as well as sex chromosome STR is routinely used in forensic casework, population study, medico-legal examinations etc.²⁻⁶ In the past two decades, forensic science has revolutionized criminal investigations and prosecutions.⁷ Evidence material in sexual assault cases consists of stains of body fluids that may be contributed by both the victim and suspect. As a part of this, both autosomal STR as well as Y-STR study proved helpful in solving complex mixed samples in sexual assault cases.⁵

In this study, a neonate is disposed of in a common toilet of a town in Rajasthan state. Postmortem showed that the infant was a male child of an intra-uterine period of about 30 ± 2 weeks. The birth of the infant was pre-mature and he did not respire after birth. A case was registered in Police station under section 318 IPC. The police investigation was insufficient to correlate a suspected lady residing in the nearby area to be the mother of that infant. Since no evidence was available to connect the mother with the crime. DNA profiles of both the infant and the suspected mother proved that she is the biological mother of the infant. This technology provides a breakthrough in this type of criminal investigations.

Material and Methods

Liquid blood sample of the neonate collected during the postmortem by the medical officer and the reference blood sample of the suspected mother preserved in EDTA were received in the Forensic Science Laboratory for DNA analysis.

The blood samples immediately processed for DNA isolation by using the phenol-chloroform Isoamyl Alcohol (PCIA) organic extraction method. Extracted DNA was quantified by using Quantifiler duo kit (ThermoFisher scientific, USA-Thermo) on RT-PCR 7500 (Thermo) as per the recommended protocol of the manufacturer.

Before 2017, CODIS prescribed minimum 13 autosomal STR markers to decide a forensic case regarding paternity. The case was registered in the year 2013, therefore that time 13 CODIS core loci were used for the DNA examination to establish maternity of the child. These markers were D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, vWA, TPOX, D18S51, D5S818 and FGA. Quantified DNA template (1ng) was used for downstream processing by Amplification of 15 STR loci i.e., D8S1179, D21S11, D7S820,

CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, vWA, TPOX, D18S51, D5S818 and FGA along with one sex-determining locus Amelogenin, included in AmpF ℓ STR \circledR Identifiler \circledR kit (Thermo) as per the recommended protocol by the manufacturer except for the half-reaction volume. PCR amplicons were separated on Genetic Analyzer 3130 using POPTM-4 (Performance Optimized Polymer), 36cm capillary array. Each amplified samples mixed with 0.5 μ L LIZ 500 and 9.5 μ L Hi-Di Formamide. Samples were injected at 1.2 kV for 5s. Electrophoresis results were analyzed with GeneMapper ID-X v1.1 software (Thermo).

Result and Discussion

Main applications of DNA fingerprinting in forensic are paternity testing and criminal investigation which are routinely used by forensic laboratories across the globe^{8,5}. The analysis of electropherogram obtained from GeneMapper ID-X software reveals that all the 16 loci including 15 STRs Viz: D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, vWA, TPOX, D18S51, D5S818, FGA and Amelogenin are amplified in both the challenging samples.

These results are depicted in table 1. The alleles of the male DNA profile of the infant are accounted in the DNA profile of suspected mother in accordance with Mendelian inheritance.⁹

Table 1: DNA profile of neonate and suspected mother at 16 genetic markers.

Locus	DNA Profile of Neonate	DNA Profile of suspected Mother
D8S1179	15,16	10,16
D21S11	30,32.2	32.2,35.2
D7S820	11,11	8,11
CSF1PO	11,13	11,11
D3S1358	15,16	15,16
TH01	6,9	9,9.3
D13S317	10,11	10,11
D16S539	8,11	8,11
D2S1338	19,22	19,26
D19S433	12,13	13,14.2
vWA	16,17	17,18
TPOX	8,11	8,11
D18S51	14,14	13,14
D5S818	10,11	10,12
FGA	20,21	21,21
AMELOGENIN	X, Y	X, X

Table 2: Calculation of complete maternity examination using genetic data.

Locus	DNA profile of suspected Mother of neonate	DNA profile of neonate	obligate Allele		AF		PI
D8S1179	10,16	15,16	16		0.081		3.08641975
D21S11	32.2,35.2	30,32.2	32.2		0.191		1.30890052
D7S820	8,11	11,11	11		0.241		2.07468880
CSF1PO	11,11	11,13	11		0.304		1.64473684
D3S1358	15,16	15,16	15	16	0.289	0.341	1.59818973
TH01	9,9.3	6,9	9		0.254		0.98425197
D13S317	10,11	10,11	10	11	0.092	0.274	3.62980006
D16S539	8,11	8,11	8	11	0.076	0.296	4.13406828
D2S1338	19,26	19,22	19		0.158		1.58227848
D19S433	13,14.2	12,13	13		0.278		0.89928058
vWA	17,18	16,17	17		0.308		0.81168831
TPOX	8,11	8,11	8	11	0.377	0.3376	1.32802359
D18S51	13,14	14,14	14		0.258		1.93798450
D5S818	10,12	10,11	10		0.120		2.08333333
FGA	21,21	20,21	21		0.133		3.75939850
AMELOGENIN	X,X	X,Y					
PI= Paternity Index, CPI-Combined Paternity index, POP= Probability of Paternity, AF-Allele Frequency					CPI		7575.41852219
					POP		99.986801151%

These results were statistically analyzed for the probable son by comparing the locus wise contribution of alleles. The parentage was established based on an exclusion that no other woman could be the mother of this neonate. Paternity Index (PI)¹⁰ which is a comparison of the relative chance of transmitting the obligate allele from probable mother, son, and any other random individual of the population, was calculated using likelihood ratio (LR).¹⁰ Allele frequencies used in the calculation of Paternity Index (PI) for the studied population (Table 2) was previously reported on 21 markers.² Table 2 shows that the Probability of Paternity (POP) between neonate and probable mother is 99.986801151%. Probability of paternity is calculated by using the formula -Probability of paternity = $1/1+$ (1/the value of combined paternity index).

The conviction rate of maternity/maternity based criminal/conflict cases before DNA test came in existence in forensic was very low because of limitations of blood group analysis. When it

is applied in forensic and acknowledged by the court of law, significant rise has been observed in conviction rate.

Conclusion

On the basis of Mendelian inheritance, it is concluded beyond the doubt that the source of blood sample of the neonate is the biological son of the source of blood sample of the suspected mother. This DNA profiling report helps the court of law to decide maternity of the infant, which was not possible before the availability of DNA fingerprinting.

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Author's contribution:

AK and GK designed the study, the manuscript drafted by AK which was reviewed by GK. All authors reviewed and approved the final manuscript.

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