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Deception in DNA Profiling: A Case Report

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Abstract

The product of conception and fetus of initial development stage, more often than not, become a challenging task to address human identity issues due to number of reasons. A case of identity of abandoned male fetus was required to be established by an investigation agency from small quantity of tissue material preserved by the autopsy surgeon. The DNA analysis revealed that material was of female origin and not in consonance with the gender mentioned in the postmortem report. Rare Y-deletion in Amelogenin marker reported in the Indian population was ruled out through Y-STR analysis. The remaining fetal tissue was analyzed and yielded a complete male DNA profile. The application of genetic genealogy helped in resolving the issue as relationship between the two DNA profiles was established.

Keywords: Fetal Tissue; Relationship Analysis

Introduction

The unintended pregnancy in any society is a social taboo especially in case of unwed mother. The social stigma is often associated with termination of pregnancy, as a consequence thereof, many may resort to terminate pregnancy without intervention of approved public/private facility. The incidence of abortion is a worldwide phenomenon and is prevalent in all societies by adopting different means to terminate the pregnancy. There are globally 73.3 million abortions every year as per WHO report [1]. The data from American population showed that a total of 625,346 abortions were reported. The rate of abortion was 11.4 abortions per 1000 women aged 15-44 years and 79% of abortions were performed at ≤ 9 weeks gestation and 92.7% were performed ≤ 13 weeks gestation [2]. The total numbers of abortions in India were 15.6 million, out of which 3.4 million (22%) took place in health facilities, 11.5million (73%) were done with medical methods outside facilities and 5% are expected to have been done through other methods. The rate of incidence of abortion was 47 per 1000 women of reproductive age (15-49 years) which was close to the figure of neighboring countries like Pakistan (50), Nepal (42), and Bangladesh (39) [3]. In this case report, an aborted fetus (approx.12 weeks gestation period) was found abandoned in an open drain of the town which was noticed by the police and sent to the autopsy surgeon for preservation of samples for DNA testing in order to generate investigating lead to provide insight about the perpetrator of crime.

Materials and Methods

Approximately 5-8 gms tissue preserved from finger of an aborted fetus was used for DNA analysis. Prior to DNA isolation, the tissue sample weighing about 3-4 gms was taken and washed thoroughly with nuclease free water (Ambion, Life Technologies Corporation, Austin). The sample was subjected to DNA isolation through automated DNA extraction (using Magnetic bead-based method using Qiagen EZ1 Advanced XL). The isolated DNA was checked for quality and quantity through 0.8% agarose gel electrophoresis. The autosomal STR DNA was amplified using Power plex 21 PCR amplification kit as per standard protocol (Power Plex® 21 System, Promega, USA). The Y-STR was amplified using Power plex Y23 PCR amplification kit as per standard protocols (Power Plex® Y23 System, Promega, USA). The PCR amplification was carried out on ABI Verity Thermal Cycler (Thermo Fisher Scientific). The amplified PCR products were analyzed with automated Genetic Analyzer ABI 3130. The data was analyzed using Gene Mapper software to generate DNA profiles. The DNA profiles were analyzed through Gene Marker HID v 3.0.0 for relationship testing for 13 CODIS markers viz. CSF1PO, TPOX, TH01, vWA, D16S539, D7S820, D13S317, D5S818, FGA, D8S1179, D18S51, D21S11, D3S1358 using General Asian Population database for allele frequency.

Results and Discussions

An autosomal STR DNA profile pertaining to a female was observed based on amelogenin marker from the randomly sampled tissue of fetus (Table 1) whereas the source tissue was of a male fetus as identified on the basis of developed genitals. There are reports in the Indian population that showed rare Y-deletion in the amelogenin markers [4] and the possibility of rare Y-deletion in the population was required to be ascertained to rule out any ambiguity in sex determination. No Y-STR DNA profile could be obtained on analysis confirming the female gender of the tested fetal tissue. In order to rule out any fallacy in the test, the remaining tissue was analyzed and yielded a male DNA profile (Table 2) which was in agreement with the observation of male fetus mentioned in the postmortem report but was in contrast to the first DNA findings that of female origin. In this case study, we explored further to solve the mystery of two different DNA profiles from the same tissue. DNA profiles were analyzed through Gene Marker software to establish the linkage. In this experiment, the Likelihood Ratio (LR) was calculated for different relations like Parent-Child, Full-Siblings, Half-Siblings & Cousins (Table 3).

Table 1: Allelic table showing DNA profile of Female origin.

Markers	Allele 1	Allele 2
AMEL	X	X
D3S1358	15	16
D1S1656	9	15
D6S1043	11	14
D13S317	8	10
Penta E	9	15
D16S539	9	11
D18S51	14	15
D2S1338	18	22
CSF1PO	12	12
Penta D	10	12
TH01	6	9.3
vWA	16	19
D21S11	31.2	32.2
D7S820	11	12
D5S818	12	13
TPOX	8	12
D8S1179	10	11
D12S391	18	22
D19S433	14	16
FGA	21	21

Table 2: Allelic table showing DNA profile of Male Origin.

Markers	Allele 1	Allele 2
AMEL	X	Y
D3S1358	15	16
D1S1656	12	15
D6S1043	12	14
D13S317	10	11
Penta E	9	15
D16S539	9	9
D18S51	14	15

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D2S1338	18	19
CSF1PO	12	12
Penta D	11	12
TH01	6	6
vWA	14	16
D21S11	30	31.2
D7S820	11	11
D5S818	12	13
TPOX	8	8
D8S1179	10	11
D12S391	18	22
D19S433	13	14
FGA	21	23

Table 3: Likelihood ratio (LR) for 13 CODIS markers in respect of different relationships of profile 1 and profile 2.

Marker	Profile1		Profile2		Parent/Child (LR)	Full Sibling (LR)	Half Sibling (LR)	Cousins (LR)
CSF1PO	12	12	12	12	2.60E+00	3.23E+00	1.80E+00	1.40E+00
TPOX	8	12	8	8	1.01E+00	7.55E-01	1.01E+00	1.00E+00
TH01	6	9.3	6	6	2.88E+00	1.69E+00	1.94E+00	1.47E+00
vWA	16	19	14	16	1.46E+00	9.81E-01	1.23E+00	1.12E+00
D16S539	9	11	9	9	2.28E+00	1.39E+00	1.64E+00	1.32E+00
D7S820	11	12	11	11	1.59E+00	1.05E+00	1.30E+00	1.15E+00
D13S317	8	10	10	11	2.33E+00	1.42E+00	1.67E+00	1.33E+00
D5S818	12	13	12	13	2.99E+00	5.70E+00	1.99E+00	1.50E+00
FGA	21	21	21	23	3.16E+00	1.83E+00	2.08E+00	1.54E+00
D8S1179	10	11	10	11	4.40E+00	1.17E+01	2.70E+00	1.85E+00
D18S51	14	15	14	15	2.57E+00	4.82E+00	1.78E+00	1.39E+00
D21S11	31.2	32.2	30	31.2	4.66E+00	2.58E+00	2.83E+00	1.92E+00
D3S1358	15	16	15	16	1.57E+00	2.26E+00	1.28E+00	1.14E+00
Product Score:					7.29E+04	2.88E+04	1.11E+03	6.04E+01

The LR data clearly revealed the Parent-Child relationship between the generated DNA profiles. The emergence of genetic genealogy in recent times has proved a boon for resolving human identity problems. The investigative genetic genealogy is a highly effective tool for using DNA to determine the identity of unknown individuals [5]. The development of two types of DNA profiles from fetal tissue could be attributed to non-adherence of proper sampling protocols in handling the fetal tissue. The presence of female DNA profile could be attributed to maternal DNA contamination as both the DNA profiles were found genetically related. A mechanism is required in place with strong emphasis to ensure the validity of results by way of replicate testing or retesting (ISO: IEC 17025:2017) as a measure of quality assurance in the laboratory so that erroneous conclusions are not drawn to avoid incorrect reporting of results besides adopting sampling protocols and can prove beneficial especially in a case where fetus is of female origin. on account of lack of understanding of DNA sampling protocol to be employed at the time of sampling had led to a bizarre situation but due caution exercised in the laboratory did save from erroneous conclusions and reported the correct DNA results.

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