

## Instant Journal of Forensic Science

Case Report

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### Short tandem repeat typing technologies used in paternity testing: a case study

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**Received Date:** Apr 17, 2020 / **Accepted Date:** Apr 21, 2020 / **Published Date:** Apr 23, 2020

#### Abstract

**Background:** In forensic field, for establishing the paternity of disputed offspring, STR typing (autosomal, and Y typing) are commonly used to dissolve the cases. Short tandem repeat (STR) technology is used to evaluate specific regions (loci) within nuclear DNA. Variability in STR regions can be used to distinguish one DNA profile from another. The Federal Bureau of Investigation (FBI) uses a standard set of 13 specific STR regions for CODIS.

**Aim of the study:** The present work was done to find out the biological father of child in a case where mother was killed by her husband for the doubt of her pregnancy.

**Methods:** After taking blood samples from the murdered mother, the baby and the killer husband, Autosomal STRs Using AmpFISTR Identifier Plus PCR Amplification Kit and Y-STR analysis using AmpFISTR Yfiler PCR Amplification Kit were done to solve the disputed paternity case.

**Results:** After the analysis of DNA STRs profiles of autosomal and Y-chromosome markers, the dead fetus was proved to be the son of the killer father.

**Conclusions:** Autosomal STR multiplex analysis combined with Y-STR analysis help to reach a conclusion in disputed paternity cases with greater confidence.

**Keywords:** Disputed; Paternity; Multiplex; Short tandem repeats; Y-STR; Analysis

**Cite this article as:** Amany Abdel Hakim Mousa, Dena Mohamed Naguib Abdel Moawed, Adel Mohamed Abdallah Elberry. 2020. Short tandem repeat typing technologies used in paternity testing: a case study. Int J Forensic Sci. 2: 04-09.

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#### Introduction

Modern DNA typing technology has used new methods to perform human identity testing.

This is of great importance in a number of situations including the determination of assailants of violent crimes such as murder and sexual assault, and identification of remains of missing persons or victims of mass disasters

[1]. Another field of application of DNA typing is disputed paternity cases and paternity testing using autosomal, mitochondrial or Y-chromosomal DNA markers [2-3].

DNA regions with repeat units that are 2-6 bp in length are called microsatellites or short tandem repeats (STRs). The number of repeats in STR markers is highly variable among individuals, making these markers effective for use in forensic applications [4-5]. Polymorphic STR loci can be analyzed simultaneously by multiplex PCR. The Federal Bureau of Investigation (FBI) uses a standard set of 13 specific STR regions for CODIS [6]. Also, the analysis of short tandem repeat (STR) markers located on the Y chromosome is an important method in forensic casework analysis. Y-STR analysis is able to detect the presence of minute amounts of male DNA of one or multiple donors and resulting genetic profiles can be compared to known reference samples [7]. This study reports one example where autosomal STRs combined with Y-STRs helped to reach a conclusion in disputed paternity case with greater confidence.

## Case

The story began in a local district in Cairo by reporting the death of a pregnant woman at the end of pregnancy as the result of severe beating from her husband, as he was suspicious of her behavior. The woman was taken to the morgue after inspecting the crime scene to collect evidences and equipment used causing her injuries.

## External examination

In morgue, X-rays revealed no fractures all over the body. The corpse was fully dressed and after taking off of her clothes, there were injuries in the form of double striped bruises around the trunk, thighs, chest and back (Figure 1). There was a lacerated wound on the head. She had enlarged abdomen because of her pregnancy (Figure 2).



**Figure 1:** Double striped bruises all over the victim's body.



**Figure 2:** Lacerated wound in the victim's head.

## Autopsy examination

Autopsy revealed fissure fracture of the skull with subarachnoid hemorrhage in the left and right frontal part combined with cerebral edema. Dead full term male fetus was delivered from her uterus (Figure 3). Blood samples were taken from the dead mother, as well as from the dead fetus. Control blood samples were from the husband for doing paternity test.



**Figure 3:** Dead full term fetus.

## Paternity testing

- **DNA extraction**

Extraction of DNA from all collected blood samples was done by QIAamp DNA Mini kit (Applied Biosystem, USA).

- **DNA quantification**

Degree of extract contamination by polymerase chain reaction (PCR) inhibitors was evaluated using Real time PCR based kit, Quantifiler™ Human DNA Quantification Kit (Applied Biosystems, USA) according to the manufacturer's protocol.

- **STR genotyping**

Fifteen STR markers in addition to a gender identification marker were examined for human identification Using Amp FISTR Identifier Plus PCR Amplification Kit (Applied Biosystems, USA) and AmpFISTR Yfiler PCR Amplification Kit (Applied Biosystems, USA) for Y-STR analysis. Amplicons were separated using a genetic analyzer (ABI 3130x; Applied Biosystems, USA) and analyzed with the appropriate software (Gene Mapper *ID*, version 3.2; Applied Biosystems) using standard procedures.

## Results

The cause of death of mother was the subarachnoid hemorrhage and the fetus died due to stoppage of the nutrition from his mother after her death. The DNA extracted was in a good quality and in a good acceptable quantity for making complete DNA profile .The sample (A) was from the dead mother of, sample (C) was from the dead fetus and sample (B) was from the killer husband. After the analysis of all DNA STRs profiles of autosomal and Y-chromosome markers, the dead fetus was proved to be the son of the killer father who was the husband of the dead mother (Table 1,2).

**Table 1:** The Identifier plus STR profiles of victim, husband, and the fetus.

| STR Loci   | Victim (A) | Husband (B) | Fetus (C) |
|------------|------------|-------------|-----------|
| D3S1358    | 15,16      | 15,17       | 15,16     |
| CSF1PO     | 10,11      | 10,13       | 10,13     |
| D7S820     | 8,10       | 11,13       | 10,11     |
| D21S11     | 27,24      | 28,30       | 24,28     |
| D8S1179    | 13,14      | 14,15       | 13,15     |
| D19S433    | 12,14      | 15,15.2     | 12,15.2   |
| D2S1338    | 17,17      | 18,18       | 17,18     |
| D16S539    | 9,11       | 11,12       | 11,12     |
| D13S317    | 10,13      | 11,12       | 10,12     |
| TH01       | 6,10       | 7,8         | 6,8       |
| FGA        | 22,28      | 21,26       | 22,26     |
| D5S818     | 10,14      | 12,13       | 12,14     |
| D18S51     | 13,13      | 15,17       | 13,15     |
| TPOX       | 8,12       | 8,11        | 8,8       |
| VWA        | 14,19      | 16,18       | 14,16     |
| Amelogenin | XX         | XY          | XY        |

**Table 2:** Y-STR profiles of both the fetus and the husband.

|            | <b>DYS456</b>   | <b>DYS3891</b> | <b>DYS390</b>    | <b>DYS3911</b> |
|------------|-----------------|----------------|------------------|----------------|
| Fetus(C)   | 14              | 13             | 23               | 29             |
| Husband(B) | 14              | 13             | 24               | 30             |
|            | <b>DYS458</b>   | <b>DYS19</b>   | <b>DYS385a b</b> | <b>DYS393</b>  |
| fetus(C)   | 19              | 15             | 13,15            | 15             |
| husband(B) | 19              | 15             | 13,15            | 15             |
|            | <b>DYS391</b>   | <b>DYS439</b>  | <b>DYS635</b>    | <b>DYS392</b>  |
| fetus(C)   | 10              | 12             | 21               | 11             |
| husband(B) | 10              | 12             | 21               | 11             |
|            | <b>YGATA H4</b> | <b>DYS437</b>  | <b>DYS438</b>    | <b>DYS448</b>  |
| fetus(C)   | 11              | 16             | 10               | 20             |
| husband(B) | 11              | 16             | 10               | 20             |

## Discussion

Most of cases regarding disputed paternity arise in many legal aspects like affiliation orders, divorce proceedings and questioned legitimacy. It may also be used to find out paternity in cases of inheritance, guardianship, maintenance, legitimacy, adultery or fornication [8]. In the present study the STR typing aimed to solve the doubt of the husband in being the father of his son. Blood sample 'C' of baby had one of the alleles in the genotype profile alike to one of the alleles in the genotype profile of the mother 'A'. All the non-maternal alleles of the amplified identifier STR loci of the liquid blood sample of baby 'C' were alike to one of the alleles in the genotype profile of the husband 'B'. In the present study, 15 tetranucleotide markers or STR loci in addition to a gender identification marker were examined for human identification purposes in the paternity testing. This multiplex STR analysis was used in other similar studies using the same STR markers to solve cases of disputed paternity [6, 9-10].

In paternity testing, 99% or greater probability of a biological relationship is considered proof of paternity. When the biological father of a child is in doubt, a PCR-based STR assay is the most accurate choice available for paternity testing [10]. The Y STR profiles were identical in the child and father. These observations revealed that person 'B' is the biological father

of baby 'C'. The male-specific part of the human Y chromosome is widely used in forensic DNA typing, particularly in cases where standard autosomal DNA profiling is not informative. Because the fact of that Y-STR haplotypes are shared between paternally related men belonging to the same paternal lineage; Y-STR haplotyping is suitable for solving paternity disputes of male offspring [11]. Alshamali et al. (2004) used also Y-STR markers for solving four cases of paternity testing [12]. Burgarella and Navascues (2010) as well as Vanek (2009) revealed that the Y-STR testing can be used for identification of legitimate male family members among the doubtful pedigree lineages which is failed to be identified by autologous STR markers [13-14]. Ge et al. (2012, 2014) revealed that Y-STR analysis can be used to separate male DNA from other male or female DNA whose autologous STR markers match at certain loci. And they mentioned that, the Y-STR data has been approved by the FBI, for use by forensic laboratories generating DNA profiles for inclusion to the databases such as the US National DNA Index System (NDIS) and the Combined DNA Index System (CODIS) [15-16].

## Conclusion

DNA fingerprinting is a technique, in which virtually unique sequence of bases in the DNA



strands are used to compare one biological sample with another to investigate genetic relationship. Multiplex STR typing is a reliable tool to solve problems of family relationships and forensic caseworks. We recommend the use of Y STRs multiplex analysis combined with autosomal STRs in cases of complicated disputed paternity. Y STRs multiplex analysis combined with autosomal STRs help to reach a conclusion in complicated cases with greater confidence.

## Key Points

1. Disputed paternity is an important problem arises in many legal aspects and need to be solved by reliable methods.
2. Technologies used for performing forensic DNA analysis differ in their ability to differentiate two individuals and in the sample processing speed.
3. Short tandem repeats (STRs) are widespread polymorphic markers throughout the human genome and can be detected by PCR.
4. STR typing is a reliable genetic tool which has an important to solve problems of disputed paternity.
5. The male-specific part of the human Y chromosome is widely used in forensic DNA typing, particularly in cases where standard autosomal DNA profiling is not informative.

## Compliance with ethical standards

Ethical approval: This article doesn't contain any studies involving human participants or animals performed by the authors. Informed consent: Informed consent was obtained from the victim's father.

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