Personal Identification by DNA Typing

Yuji Yamamoto

Department of Legal Medicine, Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Okayama University, Okayama, 700-8558 Japan

Abstract: The short tandem repeat (STR) polymorphisms are highly useful tools for personal identification and paternity testing in forensic practice. Sequence analysis of mitochondrial DNA (Mt DNA) is being used to characterize forensic biological specimens, particularly when there is insufficient nuclear DNA in samples for typing. Bones, teeth and other samples that are severely decomposed may be subjected to Mt DNA analysis. We could successfully apply these techniques to investigate samples from practical forensic cases and confirming their efficacy for forensic practice.

Key words: Forensic sciences, Personal identification, Short tandem repeats (STRs), Mitochondrial DNA

Introduction

The short tandem repeats (STRs) are a rich source of highly polymorphic markers in the human genome, are relatively small in size, and can be typed with the polymerase chain reaction (PCR) in a multiplexed fashion. Thus, the STR polymorphisms are highly useful tools for personal identification and paternity testing in forensic practice. Sequencing analysis of human mitochondrial DNA (Mt DNA) is being widely used to identify forensic biological specimens, in particular there is insufficient nuclear DNA in samples for typing. Bones, teeth, hair shafts, and other samples that are severely decomposed may be subjected to Mt DNA analysis. We have applied these techniques to investigate samples from practical forensic cases and evaluated the usefulness of these methods.

Materials and Methods

DNA extraction

DNA was extracted by the phenol-chloroform protocol or using a QIAamp DNA mini kit (QIAGEN, Hilden, Germany).

PCR amplification and sequencing reaction

DNA samples were amplified using a *GenePrint*® PowerPlexTM 16 Fluorescent STR System (Promega). Multiplexed PCR of the 15 STR loci (D3S1358, TH01, D21S11, D18S51, Penta E, D5S818, D13S317, D7S820, D16S539, CSF1PO, Penta D, vWA, D8S1179, TPOX, FGA) was performed following the procedure recommended by the manufacturer with slight modifications 1). Sequences of the Mt DNA D-loop region were analyzed with the method previously described 2).

DNA fragment analysis

PCR amplified fragments were analyzed with an ABI PRISM 310 Genetic Analyzer (Applied Biosystems), a capillary electrophoresis instrument. Electrophoresis was performed using Performance Optimized Polymer 4 (POP 4) (Applied Biosystems) at 15 kV for 30 min. Then the fragments were typed based on allelic ladders with the kit using a GeneScanTM Analysis Software (Applied Biosystems). Products of the sequencing reaction were analyzed also with an ABI PRISM 310 Genetic Analyzer using Performance Optimized Polymer 6 (POP 6) (Applied Biosystems).

Correspondence to Yuji Yamamoto, Department of Legal Medicine, Okayama University Graduate School of Medicine, Dentistry and Pharmaceutical Sciences, Okayama, 700-8558 Japan. Tel: +81-86-223-7151 (Ext.7198); FAX: +81-86-235-7201. E-mail: y-yamamo@md.okayama-u.ac.jp

Results and Discussion

STR typing in paternity cases

The STR typing in 20 cases of paternity testings was performed by the present method. Results were consistent with the conclusion obtained from classical genetic markers, and in 12 non-excluded cases, the paternity index (PI) combined for 15 STR systems ranged from 575,846 to 7,054,333,894.

STR typing for a zygosity diagnosis of twins

The STR typing of twin sisters was performed for a zygosity determination. Both of the twins had the same STR types in all 15 systems and the probability of monozygosity was calculated to be 99.9996% (Fig.1).

STR typing from an abandoned infant and placenta

An infant corpse abandoned in a plastic container was discovered at a vacant lot beside a national road in September 199X. The infant was found to be mature and alive at birth by autopsy. The umbilical cord was cut sharply. A placenta in a corrugated cardboard box was discovered near the infant. STR typing from blood of the infant and a section of the cord attached to the placenta showed the identity of the infant and the placenta, and moreover the STR types of a suspected mother was obtained from putrefied blood clot of the maternal side of the placenta. After several days, a woman suspected as the mother of the infant was investigated and her blood was taken for DNA typing. As shown in Figure 2, all of the STR types of the woman were same as those obtained from the blood clot of the maternal side of the placenta and therefore we could conclude that the woman must be the mother of the infant.

STR typing from a skull immersed in a water for one year

A human skull immersed in a water of a small irrigation channel was discovered in September 199X. The skull was almost skeletonized and small masses remained in the cranial cavity seemed to be decomposed cerebrum. The lower jaw was missing. From morphological findings of the skull and the teeth, it was surmised that the deceased was a 30-50 year-old male. A 44 year-old man who had gone missing from his apartment for one year loomed up by the police investigation. The superimposition of the skull

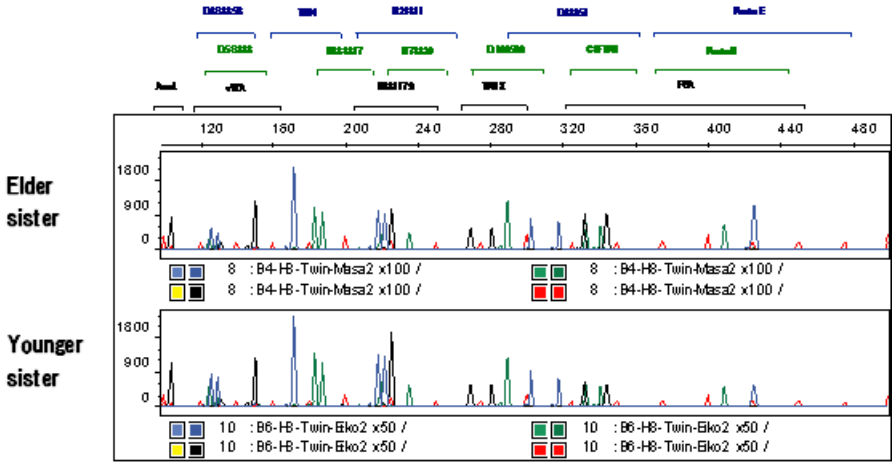

Fig. 1 STR typing for a zygosity diagnosis of twin sisters.

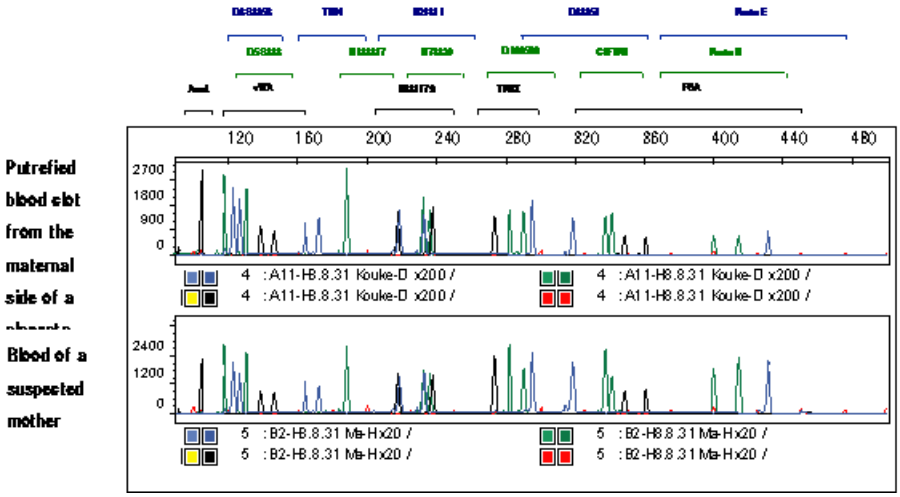

Fig. 2 STR typing of a putrefied blood clot from the maternal side of a placenta and blood of a suspected mother.

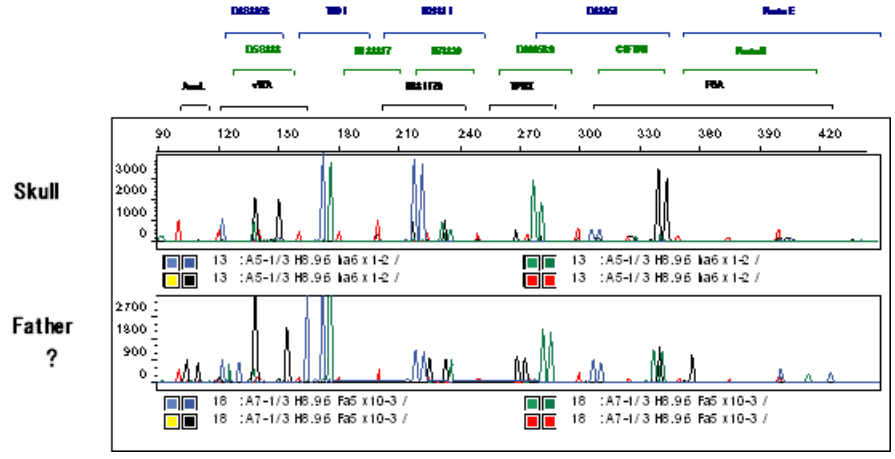

Fig. 3 STR typing from a tooth of the skull and blood of the father of a missing man.

and a photograph of the missing man supported the identity but there was need for more conclusive evidence. Therefore, the DNA analysis was carried out from a molar of the upper jaw of the skull and blood of the father and elder brother of the missing man. STR types and the Mt DNA sequences detected in these specimens strongly suggested the identity between the skull and the missing man (Fig.3).

The STR typing by the multiplexed PCR reaction and Mt DNA analysis were extremely effective for the forensic practice.

References

1. Okamoto O, Yamamoto Y, Inagaki S, Yoshitome K, Ishikawa T, Imabayashi K, Miyaishi S and Ishizu H. Analysis of short tandem repeat (STR) polymorphisms by the PowerPlex 16 system and capillary electrophoresis: application to forensic practice. Acta Med Okayama 57(2): 59-71, 2003

2. Horai S, Hayasaka K, Murayama K et al. DNA amplification from ancient human skeletal remains and their sequence analysis. Proc Japan Acad 65B: 229-233, 1989