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X-STR typing for an identification casework

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Abstract. X-STRs have been proven to be useful in case of deficiency paternity testing in which the mother is available for typing, the possible X alleles of the putative father can be determined and the paternal profile can be reconstructed. In the present casework, we used X-STRs for the identification of some hair supposedly belonging to a girl who disappeared for several years, for verifying the relationship between the samples above and the mother and the sister of the girl who disappeared. It demonstrates the impact of additional X-STR markers even in identifying case works (special reverse paternity cases) that cannot be solved using autosomal markers. © 2005 Published by Elsevier B.V.

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1. Introduction

X-STRs have been proven to be useful in case of deficiency paternity testing and in effective mother–son kinship and father–daughter testing. Male individuals inherit their one X-Chr from their mother, while female individuals receive one X from the mother and the other one from the father. So, female individuals fathered by the same man share their paternal Chromosome X.

Hence in case of deficiency paternity in which the mother is available for typing, the possible X alleles of the putative father can be determined and the paternal profile can be reconstructed. In the present casework, we used X-STRs for the identification of a biological material supposed to be belonging to a girl who disappeared for several years. In fact in the house of a man suspected to be the author of another woman's murder, a headscarf similar to the one belonging to the girl and inside it some hair was found. In the absence of any biological sample belonging to the girl who disappeared, we verified the relationship between the hair above and the mother and the sister of the girl.

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Table 1
DNA profiles from the mother and the sister of the girl who disappeared

Sample	Amelogenin	GATA 175 D05	DXS8378	HRPTB	DXS7423
Mother	XX	6/8	10/11	12/12	14/15
Daughter	XX	6/9	11/11	11/12	14/16
Sample	DXS7132	DX6789	STRX1	DXS7133	DXS101
Mother	13/14	18/19	13/14	10/11	19/22
Daughter	13/16	18/20	13/14	10/10	19/25

2. Material and methods

DNA was extracted, by Instant Gene Matrix (Biorad) treatment, from blood samples drawn from the alleged mother and daughter. DNA extraction from the root of the 4 hair has been performed by the DNA IQ™ Tissue and Hair Extraction kit.

To avoid contamination, all extractions were set up in a Gelman laminar flow cabinet in a dedicated laboratory.

All samples were quantified by the Quantifiler™ Human DNA Quantification Kit using a 7300 Real-time System.

Amplification was carried out in a laboratory different from the one dedicated to the extraction, so that amplified products never entered the extraction laboratory.

For X-STR amplification, we used Mentype® Argus X-UL that is a new kit commercialised by Biotype for fast and reliable profiling of the following 5 unlinked X chromosomal STR markers DXS8378, DXS7132, HPRTB, DXS7423 and Amelogenin [1].

Additionally, we investigated in triplex DXS101, DX6789, HumSTRX1 and in duplex DX7133, GATA172D05, using MWG-Biotech primers using respectively the following amplification protocols:

1) Init. Den. 94 °C for 5 min, 25 cycles at 94 °C for 1 min, 58 °C for 45 s, 72 °C for 1 min.
2) Init. Den. 94 °C for 5 min, 30 cycles at 94 °C for 1 s, 57 °C for 1 s, 72 °C for 1 min.

For hair roots, the number of amplification cycles has been increased to 33.

DNA amplification has been performed using GeneAmp PCR Systems 9700, 2400, 2720 thermal cyclers (Applied Biosystems) [2–4].

Female and male positive controls and negative controls were used during all amplification steps.

Amplified products were analyzed by capillary electrophoresis on an ABI PRISM 3130 Genetic Analyzers (Applied Biosystems) employing GeneMapper 3.2 software.

Table 2
Reconstructed paternal X-STR profile

Sample	Amelogenin	GATA 175 D05	DXS8378	HRPTB	DXS7423
Father	XY	9	11	11	16
Sample	DXS7132	DX6789	STRX1	DXS7133	DXS101
Father	16	20	14	10	25

Table 3
Comparison among all DNA profiles

Sample	Amelogenin	GATA 175 D05	DXS8378	HRPTB	DXS7423
Mother	XX	6/8	10/11	12/12	14/15
Father	XY	9	11	11	16
Daughter	XX	6/9	11/11	11/12	14/16
Hair	XX	6/9	10/11	11/12	15/16

Table 4
Comparison between all DNA profiles

Sample	DXS7132	DX6789	STRX1	DXS7133	DXS101
Mother	13/14	18/19	13/14	10/11	19/22
Father	16	20	14	10	25
Daughter	13/16	18/20	13/14	10/10	19/25
Hair	14/16	18/20	13/14	10/11	22/25

3. Results and discussion

DNA typing of hair showed that all of them were from a female and that they showed the same X-STR profile.

In a first step by comparison between the DNA profile of the mother and that of the woman that was surely the daughter of the unavailable father, it was possible to identify the paternal X-STR profile (Tables 1 and 2).

Then, in a second step, in hair DNA profile, the presence of the paternal possible X alleles has been verified.

We found that the DNA profile of samples suspected to be coming from the alleged daughter was compatible with the reconstructed paternal DNA profile (Tables 3 and 4).

X-STR analysis surely showed that since the hair of the other woman shared the same paternal X alleles, they were coming from the girl who disappeared.

4. Conclusion

Since female individuals fathered by the same man share their paternal Chromosome X, X-STRs are useful in case of deficiency paternity testing in which the mother is available because of the ability to determine the X alleles of the putative father and to reconstruct the paternal profile.

The present case demonstrates that X-STR markers are useful even in identification caseworks (special reverse paternity test) since their analysis under certain circumstances can complement or substitute the analysis of traditional autosomal markers for solving complex kinship cases.

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