J Forensic Sci, March 2007, Vol. 52, No. 2
doi:10.1111/j.1556-4029.2006.00373.x
Available online at: www.blackwell-synergy.com

CASE REPORT

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Intentional Mixed Buccal Cell Reference Sample in a Paternity Case*

ABSTRACT: We report a case where an alleged father (AF) attempted to substitute someone else's saliva sample for his reference sample in a paternity analysis. Buccal cells were collected from the AF and the child, and DNA analysis was performed using an autosomal STR loci (Identifiler®). The profile from the AF showed extra peaks in some loci, as well as a much higher "X" allele peak relative to the "Y" allele peak at the amelogenin locus. After conducting reanalysis by another technician with another set of positive and negative controls, it was concluded that the only source of the mixed profile was by intentional introduction by the AF, at the time of sampling, of some foreign human biological material, most likely saliva from a woman. Owing to the inconclusive results, when the AF was called back to the lab and the peculiar results were explained to him, he admitted that he had introduced into his mouth saliva from another person in an attempt to be excluded as the father of the child. Although tampering with DNA reference samples is not common, some individuals may attempt to contaminate or otherwise adulterate specimens before DNA tests. Personnel responsible for sampling should be aware of this possibility and should try to establish procedures to avoid the problem.

KEYWORDS: forensic science, STRs, paternity, intentional contamination

DNA typing for human identification is a highly reliable and robust testing system, especially if international recommendations are followed (1). The results of DNA analysis are often used to exclude or "prove" the biological paternity of an alleged father (AF) (2,3). Some individuals may attempt to undermine DNA testing. There are anecdotal accounts of individuals attempting to substitute DNA donors so the biological evidence would be exculpatory (A. Eisenberg, personal communication, UNTHSC, Ft. Worth, TX). The case presented here involved an AF who attempted to switch samples by introducing foreign saliva into his mouth. We document this below.

Materials and Methods

Our standard operating procedure for reference sample collection in paternity cases is as follows: (1) donors are placed in separate rooms (in this case, AF in one room and guardian or mother and child in another); (2) after proper identification, the AF and the mother sign an informed consent form; (3) before obtaining a buccal sample, each donor is instructed to rinse the mouth with mineral water in a separated corner of the room; (4) the oral mucosa is swabbed by trained personnel; (5) the cellular material on each swab is transferred onto separate pieces of FTA® paper (Whatman, Florham Park, NJ); and (6) the samples are dried and sent to the laboratory for DNA analysis using the autosomal AmpF*l*STR Identifiler® kit (Applied Biosystems, Foster City, CA). DNA purification and analysis is performed as recommended by the manufacturers (4).

Results

Electropherograms of six STR loci and the amelogenin locus are shown in Fig. 1*a* and *b* (showing the mixed profiles for the AF) and Fig. 2*a* and *b* (showing single person profile for the AF after second buccal swab sampling).

Discussion

The first indication of a potential problem in this particular paternity case was the presence of an atypical profile of the AF, which showed evidence of a mixture of DNA (contamination). The electropherogram contained more than two peaks at some STR loci. Possible explanations for this AF profile included laboratory contamination, some biological phenomenon (such as a chimera), or tampering.

The sample handling, analytical process, and data were reviewed. The positive and negative controls were found to be appropriate. The positive and negative controls were found to produce appropriate results. The DNA extraction, amplification,

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*Partial results of this case were presented at the 58th Annual Meeting of the AAFS, Seattle, WA, February 2006.

Received 4 Mar. 2006; and in revised form 28 July 2006; accepted 30 Sept. 2006; published 6 Feb. 2007.

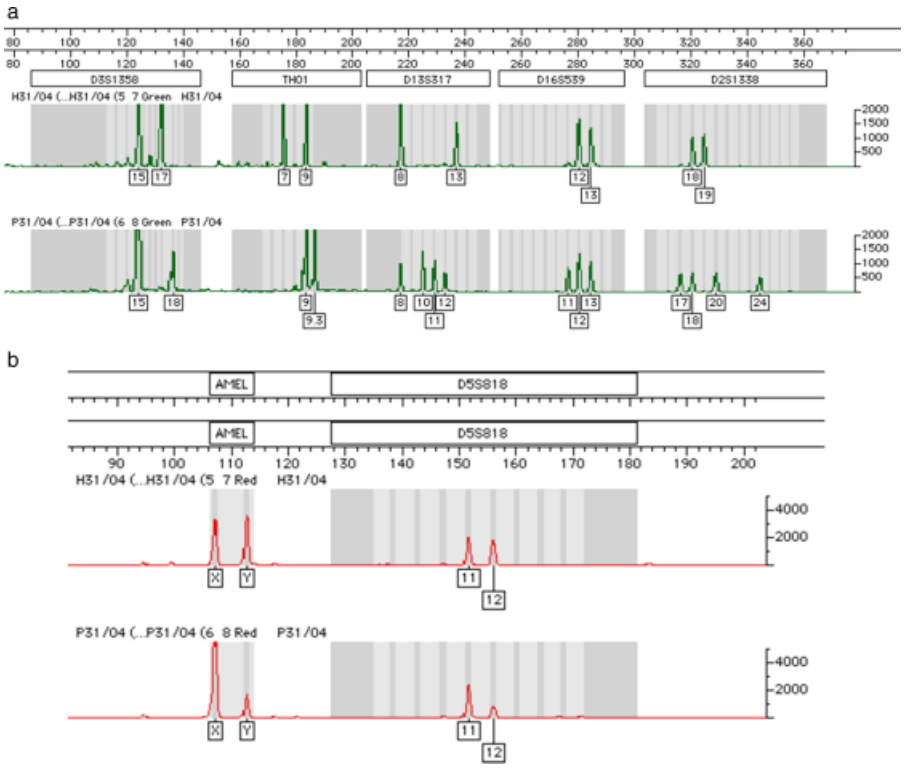

FIG. 1—*(a) A DNA mixture is evident in the profile of the alleged father (AF; bottom panel). More than three alleles are present in three of five loci; all alleles show peak over 500 relative fluorescence units (top panel, child). (b) A DNA mixture is evident in the profile of the AF (bottom panel), where the amelogenin locus shows the "X" allele peak to be much higher than the "Y" allele peak.*

and analysis were repeated by a second technician, and the same results were obtained.

Unused swabs and FTA® paper from the same lots were negative by STR profiling. Although a single swab in a lot could have been contaminated during manufacturing or packaging, a more plausible hypothesis was that the AF reference sample was contaminated during transfer from the AF to the laboratory. When compared, none of the DNA profiles of individuals who handled the reference sample matched alleles in the mixed AF profile.

We hypothesized that the AF introduced into his mouth some foreign biological material (such as saliva from another person) in the short interval between rinsing his mouth with water in a small lavatory and the sampling of the buccal cells. The AF was called back to our laboratory for resampling. The AF then admitted that he had introduced into his mouth saliva from another person (his wife) that he carried in a plastic bag. He agreed to provide a new sample of buccal cells and, after DNA analysis, a typical single person profile was obtained.

The source of the contaminant DNA could not be confirmed as originating from AF's wife because no sample could be obtained from her. The AF could not be excluded as the biological father of the child, based on the DNA analysis (PI = 157415; Table 1).

After this case, we did change our sampling protocol for paternities and now a member of our staff is permanently with the donors, including the time used to rinse their mouths. DNA lab managers and personnel collecting samples should consider establishing procedures to reduce tampering in paternity cases.

References

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TABLE 1—*The genotypes from the AF and the child.*

Sample information	D3S1358	vWA	FGA	Amelogenin	D8S1179	D21S11	D18S51	D5S818
Son	15, 17	18, 18	22, 22	X, Y	15, 15	27, 29	11, 14	11, 12
AF	15, 18	17, 18	19, 22	X, Y	14, 15	28, 29	11, 17	11, 11
	D13S317	D7S820	TH01	D16S539	CSF1PO	TPOX	D2S1338	D19S433
Son	8, 13	10, 11	7, 9	12, 13	10, 12	8, 8	18, 19	13, 13
AF	8, 12	9, 11	9, 9	12, 12	12, 12	8, 8	17, 18	13, 14

The AF cannot be excluded as the biological father of the child based on the genetic evidence.
AF, alleged father.

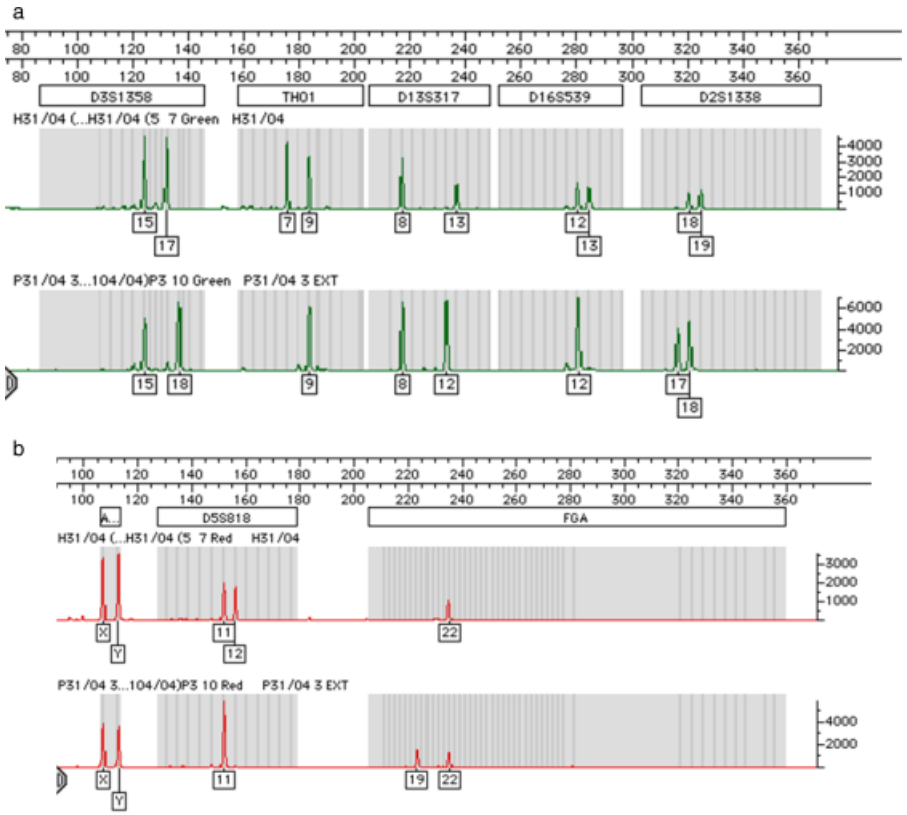

FIG. 2—*(a) DNA profiles of the alleged father (AF) from the second sampling of buccal cells (bottom panel) and the child (top panel) without contamination (from second sampling of buccal cells of the AF). (b) DNA profiles of the AF (bottom panel) and the child (top panel) without contamination (from second sampling of buccal cells of the AF). The amelogenin allele peaks show the typical male profile with balanced X and Y peaks (i.e., similar relative fluorescence units).*

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