

DNA mixtures in forensic casework resolved with autosomic STRs

Fabricio González-Andrade^{a,*}, Miguel Bolea^b,
Begoña Martínez-Jarreta^b, Dora Sánchez^a

^a DNA Laboratory, GENética, Hospital Metropolitano, Quito, Ecuador

^b Legal Medicine Department, University of Zaragoza, Spain

Abstract. To assess the technical and judicial consequences resulting from the practical application of *DNA testing* in forensic research in the numerous sex crimes in Ecuador. The aim of this work is to review our casework results obtained with the mixed genetic STR profiles encountered in our laboratory and evaluate the problems in the interpretation of the results. © 2006 Published by Elsevier B.V.

Keywords: DNA mixture; STR profile; Sex offence; Forensic casework; PCR

1. Introduction

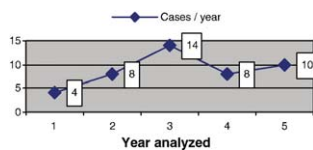
When a sample contains DNA from more than one contributor, the interpretation of its genetic profile becomes complicated. The incidence, complexity, and importance of mixed profiles is increasing due to the sensitivity of polymerase chain reaction (PCR)-based typing methods.

2. Material and methods

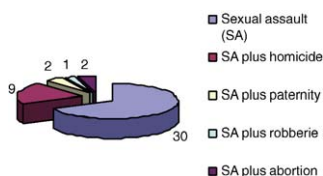
Samples: 123 samples precedent of 44 four cases of sexual offences were studied by DNA technique and analysed by technical team from Molecular Genetics Laboratory, between 2001 and 2005 (August). The origin of the request, the samples and the unquestioned samples was determined for contrast with the exhibits; the results obtained and each case were followed up judicially. To record the identity of those involved in

* Corresponding author. Tel./fax: +593 2269354.

E-mail address: fabriciogonzaleza@yahoo.es (F. González-Andrade).



Graphic 1.

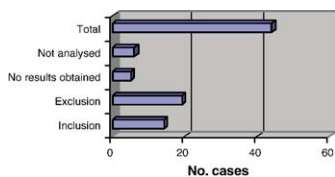


Graphic 2. Types of offences.

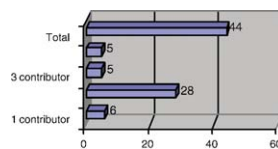
each case, photocopies of identity documents were made, the respective fingerprints were taken and *Informed Consent* was requested in writing, as previously published [1,2]. DNA extraction: Phenol–Chloroform–Isoamiliic Alcohol and proteinase K were used for the remains. For the unquestioned samples, we used the Wizard Genomic DNA Purification Kit System[®] method (Promega Corporation, Madison WI, USA). The DNA was quantified by means of UV absorbance in Gene Quant Calculator[®]. For the tissue mixtures, we made a differential extraction using a separation solution for the female fraction Tris/Edta/NaCl 400 µL, sarkosyl 25 µL, proteinase K (20 mg/mL), and for the male fraction Tris/Edta/NaCl 150 µL, sarkosyl at 20%, proteinase K (20 mg/mL) and DTT 0.39 M, in accordance with our protocols [1,2]. PCR: Amplification was carried out in a Genius[®] thermocycler; the manufacturer’s recommendations were followed. Typing: We used an ABI 310 sequencer. The fragment size and the allelic designation of the different loci were established by comparison with the allelic ladders of the PowerPlex 16 kit (Promega) and they were subsequently interpreted with the Gene Scan Analysis Software[®] programme [3,4]. We followed the recommendations of the *DNA Commission of the International Society of Forensic Genetics* for STR analysis [5]. Control of procedures: For quality control purposes, blank controls were processed in the extraction and amplification of each reaction. A positive human DNA control of cell line 9947 A, which had previously been typed for all the systems, was used. For external quality control, the laboratory takes part in an annual proficiency test with the *International Society for Forensic Genetics* [6]. The examination was carried out in duplicate, with a DNA control of known genetic information. Fifteen autosomic microsatellites and the sex-typing marker Amelogenin were studied. Analysis of data: The Grape 1.1 software, DNA fingerprinting, Statistical Evaluation was used for analysis of the mixtures [7].

3. Results and discussion

Traditionally, the finding of semen, spermatozoids or acid phosphatase in cervico-vaginal samples has been considered as sufficient evidence to prove recent sexual contact.



Graphic 3. Results obtained.



Graphic 4. Mixed profile and number of contributors.

Table 1
Occurrence of mixed profiles depending on the nature of the sample

| Origin of the samples | Blood | Semen | Saliva | Vaginal swabs on slides | Victim's underwear (clothes/bedding) | Hair and nails |
|-----------------------|-------|-------|--------|-------------------------|--------------------------------------|----------------|
| Aggressor | 40 | 2 | 2 | | | |
| Victim | 21 | | | 28 | 18 | 2 |
| Scene of crime | 8 | 2 | | | | |
| Total | 69 | 4 | 2 | 28 | 18 | 2 |
| <i>N</i> =123 | 56% | 3.3% | 1.6% | 22.8% | 14.7% | 1.6% |

Table 2
Genotypes in mixed samples and contributors' profile (observations=57)

| | | | |
|-----------------|---|----|-------|
| Four peaks | Heterozygote+heterozygote (non-overlapping alleles) | 5 | 8.7% |
| Three peaks | Heterozygote+heterozygote (non-overlapping alleles) | 15 | 26.4% |
| | Heterozygote+homozygote (non-overlapping alleles) | 7 | 12.3% |
| Two peaks | Heterozygote+heterozygote (non-overlapping alleles) | 11 | 19.2% |
| | Heterozygote+homozygote (two overlapping alleles) | 5 | 8.7% |
| | Homozygote+homozygote (non-overlapping alleles) | 4 | 7.1% |
| One peak | Homozygote+homozygote (overlapping alleles) | 3 | 5.3% |
| Allele dropouts | | 7 | 12.3% |

Currently, the presence of a mixture of cells from the victim and the suspect, in the evidence found, makes it possible to identify each individual by means of their DNA. Analyses have mainly been useful for the exclusion of suspects and to clarify the role of each person involved in a sexual offence. In all inclusion cases, the Likelihood Ratio was over 1 million. In many cases, there is no biological evidence left by the perpetrator at the scene of the crime. This is clearly a disadvantage when it comes to solving the case. In our activity, the cell mixture obtained from articles of clothing or vaginal swabs has mainly female component (Graphics 1–4; Tables 1 and 2).

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