



## Are DNA tests infallible?

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### Abstract

Modern technology has strongly influenced most fields of knowledge and forensic sciences do not escape this reality. With the advent of the application of molecular biology to human identification by means of DNA typing, conceptual conflicts were introduced. After over 15 years of worldwide experience, the robustness and reliability of DNA analysis was demonstrated. However in Latin America, judges, prosecutors and defenders, due in part to different educational background as compared to scientists, may ignore potential restrictions concerning DNA profiling results. Since its beginnings, DNA testing was surrounded by an aura of infallibility. Nowadays, a big number of highly polymorphic genetic markers, included in commercial kits, as well as automated devices for DNA extraction and purification, PCR amplification, electrophoresis and data analysis are available. Nevertheless, errors may occur. This work describes the main causes for errors in DNA testing in forensic laboratories that simultaneously processes hundreds or thousands of samples a month and makes suggestions to prevent each particular problem. It is important to underscore that DNA testing should be considered one more piece of evidence within the context of a criminal or forensic investigation, and that the judicial sentences should be based on the evidence as a whole and not just on the genetic studies.

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### 1. Introduction

The results of scientific testing of physical evidence in criminal cases have been presented in courts for over 100 years. Scientific testimony has frequently played a significant role in the conviction of individuals responsible for the commission of both

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violent and non-violent offenses. Equally significant, scientific evidence has repeatedly resulted in the exoneration of potential suspects in criminal cases. DNA identification testing methods present the forensic examiner with a modern and powerful tool for the resolution of such crime.

More than a decade has passed since DNA typing methods were first used in Latin America in criminal investigations and trials. Law enforcement agencies and universities have committed substantial resources to the technology; prosecutors, defense counsel and judges have struggled with the terminology and ideas of molecular biology, genetics and statistics. Our lab offered suggestions for improving forensic DNA testing and its use in law enforcement.

Nowadays, DNA can be obtained in substantial amounts and in optimal condition when blood or tissue is obtained from a person, or in limited amounts, degraded, or contaminated, as in some samples from crime scenes. But even with the best laboratory technique, there is intrinsic, unavoidable variability in the measurements that introduces uncertainty that can be compounded by poor laboratory technique, faulty equipment or human error. The occurrence of errors can be minimized by scrupulous care in evidence collection, sample handling, lab procedures and case review.

The present work contains recommendations for the novel forensic labs to prevent each particular problem.

## **2. Collection, transportation and storage**

The DNA laboratory must have clear procedures which address handling and preserving the integrity of evidence. Key components of such an evidence control procedure include proper labeling and sealing of evidence, a documented chain of custody record, and a secured area designated for evidence storage. Each item of evidence (or its container) must be marked with a unique identifier.

A written chain of custody record must include the signature of each individual receiving or transferring evidence, with the corresponding identifier which specifies each evidentiary item. This record must provide a comprehensive, documented history for each evidence transfer over which the laboratory has control. Electronic tracking of evidence is an acceptable alternative to a written record as long as the computerized data are sufficiently secure, detailed and accessible for review and can be converted to a hard copy when necessary.

The laboratory must ensure that evidence stored under its custody is properly sealed and protected from loss, contamination and/or deleterious change. An evidence container is properly sealed if its contents cannot readily escape and if entering the container results in a detectable alteration to the container or seal. It is highly desirable for the seal to be labeled in a manner which identifies the individual responsible for sealing the evidence.

Secure areas for evidence storage must exist within the lab. This may include the use of temporary or short-term storage, demonstrating proper security through defined, controlled access to the evidentiary storage area. Short-term storage areas may vary from a locked file cabinet to an entire examination room.

### **3. Handling within the lab**

#### *3.1. Contamination prevention*

PCR is widely used in almost all the forensic labs, but its great sensitivity poses the concern that a sample contaminated with extraneous human DNA might yield spurious amplification products. Concern has been expressed, for example, that in the handling of evidence samples, DNA might be transferred from the handler to the sample. This might become a significant problem when the sample contains very little DNA, like skeletal remains. A greater concern is the contamination of a DNA sample with a PCR product from a previous amplification; this is termed carry-over contamination. Good lab procedure minimizes the risk of this problem working in different area pre and post amplification, with separation in time or physical space for each activity, and using proper controls.

#### *3.2. Proficiency testing and quality controls*

These tests can be open or blind. In open proficiency tests, the analyst knows that a test is being conducted; in the blind one, the analyst does not know that the test is being conducted.

A blind test is therefore more likely to detect such errors as might occur in routine operations. However, the logistic of constructing fully blind tests are formidable. The “evidence” samples have to be submitted through an investigative agency so as to mimic a real case, and unless that is done very convincingly, a lab might well suspect that it is being tested.

Whichever kind of test is used, the results are reported and, if errors are made, needed corrective action is taken. Several tests per year must be implemented.

Our lab coordinated the first and the second Latin American quality controls, with the participation of more than 30 labs from almost all the countries of the region.

#### *3.3. Training courses*

A laboratory’s training program must emphasize and teach the skills and knowledge required to achieve the minimum standards of competence and good laboratory practice within a specific area of work.

It is management’s responsibility to establish the adequacy of the training of any staff member who has not completed the laboratory’s formal training program. Examples may include the acquisition of fully trained personnel from a separate organization or the assignment of experienced forensic DNA case working examiner/analysts to validate a new DNA testing procedure. All individuals, regardless of previous training and experience, must successfully complete a qualifying test for the specific DNA technology to be used at the current lab, prior to assuming casework responsibilities.

Since 1996, our lab organizes theoretical and practical post-graduate courses every year; in 2000 and 2001, two international courses supported by the University of the United Nations and United Nations Biotechnology for Latin America and the Caribbean

Program. Both of them were organized in Venezuela. More recently, a basic training course through Internet has been offered, that is available in Spanish (<http://www.ubanet.com.ar>).

### 3.4. Internal validation of procedure

Validation is the process used by the scientific community to acquire the necessary information for accessing a procedure's reliability to obtain a specific, desired result. The validation process also serves to identify critical aspects of a procedure which must be controlled and monitored, while defining the limitations of the procedure.

But prior to implementing an existing DNA procedure, developmentally validated by another lab, the forensic lab must first demonstrate the reliability of the procedure internally. The internal validation studies conducted by the forensic lab should be sufficient to document the reliability of the technology as practiced by that lab.

### 3.5. Independent retest

No amount of attention to detail, auditing and proficiency testing can completely eliminate the risk of error, so wherever feasible, evidence material should be separated into two portions, with one portion reserved for possible duplicate testing. The best protection an innocent suspect has against an error that could lead to a false conviction is the opportunity for an independent retest.

## 4. Results interpretation

### 4.1. Spurious signals

In some cases, we observe faint signals in addition to two well-defined bands or peaks. It does not cause difficulties in the interpretation of results of a paternity testing (except in homozygous), but potentially mixed samples (such as vaginal swabs) can be a problem.

### 4.2. Allele drop-out

In some forensic samples, with very tiny or highly degraded amounts of DNA, like single hairs, saliva traces or skeletal remains, some alleles can be missing, showing a heterozygote as a homozygote. We suggest trying with different dilutions of the DNA sample, and never exclude a paternity based only in apparently homozygous loci.

### 4.3. Mutations

Germline mutation will produce apparent exclusions in paternity testing. For example, for the most variable human minisatellite, MS1, the mutation rate is an extraordinary 0.05 per gamete. Offspring are as likely to inherit a mutant allele from their mother as from the father, and mutant alleles can be either larger or smaller than their progenitor allele.

Moderate mutation rates do not significantly interfere with the use of these mini- or microsatellites in paternity analysis, provided that mutation rates are known and can be incorporated into statistical likelihood ratio analyses of paternity against non-paternity. However, the new forensic labs could be confused about that, and consider the mutation as an exclusion.

## **5. Conclusions**

Even the strongest evidence will be worthless or worse, might possibly lead to a false conviction, if the evidence sample did not originate in connection with the crime. Given the great individualising potential of DNA evidence and the relative ease with which it can be mishandled or manipulated by the careless or unscrupulous, the integrity of the CHAIN OF CUSTODY is the paramount importance.

The occurrence of errors can be minimized by scrupulous care in evidence collecting, sample handling, good lab procedures, independent retest and case review, but NO AMOUNT OF CARE CAN ELIMINATE THE POSSIBILITY OF ERROR, so we suggest that DNA TESTING should be considered one more piece of evidence within the context of a criminal or forensic investigation, and that the judicial sentences should be based on the evidence as a whole and not just on the genetic studies.