

## THE LIMITS OF DNA TESTING USED AS EVIDENCE IN COURT. A REPORT BASED ON THE PRACTICE OF THE INSTITUTE OF LEGAL MEDICINE CLUJ-NAPOCA

*Dan PERJU-DUMBRAVĂ\**

*Ioana MIC\*\**

*Ovidiu Sorin CHIROBAN\*\*\**

### Abstract

*The evaluation of the evidentiary value of scientific evidence is the assessment of the strength of the link between a finding and a person. It is usually a statistical assessment but its presentation is full of pitfalls. The evaluation of scientific evidence must be based on an established methodology to both evaluate, expose and interpret the evidence. The formidable expansion in the use of DNA has not only increased the extent of interaction between forensic scientists and lawyers but more importantly, it has increased the relevance of socio-legal and ethical perspectives in strategies for applying forensic DNA techniques. Since its beginnings, DNA testing was surrounded by an aura of infallibility. Nevertheless, errors may occur. 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. However judges, prosecutors and defenders, due in part to different educational background as compared to scientists, may ignore potential restrictions concerning DNA profiling results.*

Keywords: *forensic science; DNA test; DNA database; Chimerism; Criminal law*

### **Introduction**

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 25 years of worldwide experience, the robustness and reliability of DNA analysis was demonstrated. However 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,

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\* M.D., Ph. D., Professor at "Dimitrie Cantemir" Christian University, Faculty of Law Cluj-Napoca.

\*\* M.D., Resident Doctor at „Iuliu Hatieganu” University of Medicine and Farmacy Cluj-Napoca.

\*\*\* M.D., Assistant Professor at „Iuliu Hatieganu” University of Medicine and Farmacy Cluj-Napoca.

PCR amplification, electrophoresis and data analysis are available. Nevertheless, errors may occur. 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.

DNA profiling has become the established forensic procedure when dealing with biological stains in crime investigation or when paternity is in question. In recent years in our country the technical methodology underwent several modifications and dramatic improvements. The reduction of the analysis time due to automation of some of the analytical steps and the availability of multiplex kits are additional important factors for the success of the technique.<sup>1</sup> Since then a new market of quick and low-cost offers by internet advertisement has opened. However, no official body controls the validity of typing procedures as well as possible violation of personal rights of individuals included in the genetic analysis.<sup>2</sup> The aim of this brief communication is to outline the scope of expert's responsibility for a good medico-legal practice of DNA testing.

Certain points have been raised in the legal literature about the discipline of forensic DNA profiling which forensic scientists should give their attention to. In essence, they relate to the way in which forensic science needs to operate in doing its research, validating its findings, and promoting high professional standards among its practitioners both in the laboratory and in the courtroom. All of this must occur in the context of an increasing trend to treat forensic science as an arm of the police forces or justice, or if not that, then as part of a privatised commercial organisation. Acknowledging this may indeed assist forensic scientists to rise to a better sense of their own capabilities.

Forensic scientists should accept fully their need to operate as part of an investigatory team and draw the appropriate professional conclusions; thereby confronting some of the proposed ethical complexities in the process. A forensic scientist may not immediately see the relevance of considering the perceptions of those beyond the sanctum of their discipline (in this case the legal community), especially when the content is less than congratulatory. Indeed it is important to clearly demarcate professional boundaries, one professional field cannot totally subjugate its values to those of another. Scientists with a forensic practice should not allow legal law enforcement professionals to dominate their manner of operation and their conduct. Otherwise forensic scientists give up any claim to identify themselves as a professional group. This is a strong point that needs to be remembered always.

In addition, the formidable expansion in the use of DNA has not only increased the extent of interaction between forensic scientists and lawyers but more importantly, it has increased the relevance of socio-legal and ethical perspectives in strategies for applying forensic DNA techniques. This field that may have once been

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<sup>1</sup> Walter Bar, *DNA Profiling: Evaluation of the Evidentiary Value*, in "Legal Medicine", 5, 2003, S41-S44.

<sup>2</sup> Christian K. Rittnera, Peter M. Schneidera, Gabriele Rittnerb, *Expert Witness in Paternity Testing in Germany*, in "Legal Medicine", 5, 2003, p. 65-67.

regarded as a more scientific exentricity has evolved into a far-reaching public tool. Improving the utility of forensic DNA profiling is as likely to occur through operational or public policy decisions as it is through technological advances. This broadening scope of forensic DNA profiling must be seen as making the case for scientific probity and multidisciplinary awareness all the stronger.

### ***The evidentiary value of scientific evidence***

The evaluation of the evidentiary value of scientific evidence is the assessment of the strength of the link between a finding and a person. It is usually a statistical assessment but its presentation is full of pitfalls. The evaluation of scientific evidence must be based on a established methodology to both evaluate, expose and interpret the evidence. The information of the scientist is usually a numerical one, e.g. the probability of observing two matching profiles. Assessments of simple stains as well as of stain mixtures with contributors of different ethnic origin and/or with related contributors or reference persons and even artifacts can be quantified and expressed by likelihood ratio(s). Thereby, scientific evidence can be integrated by juries into the continuous process of evaluating prior odds and changing them into posterior odds by new information in the case.<sup>3</sup>

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.

These errors can include sample switches, inappropriate testing and reporting, malfunctioning equipment or reagents, and testimony inconsistent with written reports. A full disclosure should be made if a sample switch does occur or a mistrial could result.

In our paternity analysis, blood samples which usually yield a high amount of high molecular weight DNA are used. In forensic casework, it is well known that very often the biological trace is not in perfect conditions, and therefore frequently yielding only highly degraded DNA in additionally very low amounts.

Furthermore, the amplification process is often disturbed due to the presence of inhibitors that are co-extracted with the DNA<sup>4</sup>. These circumstances can lead to trouble in obtaining complete genetic profiles. When a full genetic profile cannot be detected, e.g. due to PCR inhibition or DNA degradation, the corresponding matching probability might still be high enough to serve as significant evidence in forensic casework. Especially longer fragments often fail to be amplified since no template of such fragment length is available in the original DNA extract. An STR multiplex

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<sup>3</sup> G. Penacino, A. Sala, D. Corach, *Are DNA Tests Infallible*, in "International Congress Series", 1239, 2003, p. 873-877.

<sup>4</sup> N. von Wurmb, D. Meissner, R. Wegener, *Influence of Cyanoacrylate on the Efficiency of Forensic PCRs*, in "Forensic Sci Int", 124, 2001, p. 11-16.

pattern with only the short fragments being detected and increasing failure of the longer targets is known as the typical STR pattern of ancient DNA and is also common in forensic trace analysis.<sup>5</sup>

An other problem occurs when a close relative of the suspect is a potential alternative donor of a recovered stain it is not possible to assess and report the evidentiary strength independently of how probable each of the potential donors are to be the source prior to considering the forensic findings (the “blaming-on-the-brother” syndrome) <sup>6</sup>. When the DNA profile typed lacks information in a number of loci, the evidentiary strength of the profile decreases in general and other relatives than a full sibling increase their potentials to be the source of the stain. In forensic laboratories today, a reservation is made in the statement that the evidentiary strength holds provided no close relative was the source. If such a statement is met in court, by questions like “What if it was the suspect's brother who left the stain”, the expert witness may reassess the value of evidence and answer with a value that is much lower than the initial one. This will of course confuse the court, and a better alternative is to have the brother swabbed (provided he is available for swabbing), type his DNA, and report a new result. Such a result would either identify the brother as an equally likely source of the stain or exclude him as a source.

Perhaps the most challenging aspect of performing case review for defense attorneys is determining what to do when errors are identified. Each attorney has their own individual strategy for best assisting their client with this information. Some choose to use it for gaining sentence reduction for their client in a plea bargain rather than disclose the error at trial. Others prefer to go forward with a trial and disclose the errors openly in a reasonable effort to discredit the investigator or scientist who made the error.<sup>7</sup>

### ***Chimerism and its importance on interpreting results***

The term chimera is used when an organism contains cells originating from two or more zygotes.<sup>8</sup> Chimeric individuals are characterized by having cells with different genetic patterns originating from two or more zygotes. Chimerism can be divided into distinct classes. A partial or a whole-body chimerism can be distinguished on the basis of causal mechanism. The partial chimerism is detectable in only one organ system, predominantly in the hematopoietic system and can be artificial, develop by transfusion or transplantation of allogenic hematopoietic stem cells or a solid organ, or congenital. Partial hematopoietic chimerism can be found in

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5 S. Hummel, B. Bramati, T. Schultes, M. Kahle, S. Haffner, B. Herrmann Megaplaus, *DNA typing can provide a strong indication of the authenticity of ancient DNA amplifications by clearly recognizing any possible type of modern DNA*, in “Anthropol Anz”, 58, 2000, p. 15–21.

6 A. Nordgaard, R. Hedell, R. Ansell, *Assessment of forensic findings when alternative explanations have different likelihoods—“Blame-the-brother”-syndrome*, in “Science and Justice”, 52, 2012, p. 226–236.

7 H. M. Coyle, *The importance of scientific evaluation of biological evidence - Data from eight years of case review*, in “Science and Justice”, 52, 2012, p. 268–270.

8 X. Hong et al., *A dispermic chimera was identified in a healthy man with mixed field agglutination reaction in ABO blood grouping and mosaic 46, XY/46, XX karyotype*, in “Transf Apheres Sci”, 2012, <http://dx.doi.org/10.1016/j.transci.2012.10.002>.

dizygotic twins where hematopoietic cells have been exchanged in a context of twin-twin transfusion over vascular anastomoses between the two dichorionic placentas. A mixed or complete hemopoietic chimerism can result in the patient's blood after the transplantation of allogenic marrow or hematopoietic progenitor cells. Whole body chimerism can be observed in tetragametic chimeras as a consequence of the fertilization of two oocytes by two spermatozoas and the fusion of these products into one body.<sup>9</sup> Within permanent chimeric individuals, "bloodchimeras" (twin chimeras), which result from blood vessel junctions of dizygotic twins, can be distinguished from "whole body chimeras", where the coexistence of the different cell lines is not restricted to haemopoietic cells but spread over various tissues.

Whole body chimerism (tetragametic or dispermic chimerism) is characterized by double parental or double paternal contribution of markers in all tissues. In very rare cases, double maternal contribution has also been detected.<sup>10</sup>

Chimerism may be a pitfall in forensic investigations like paternity testing and crime cases. Different cells from different tissues will have different DNA, the consequence is that an expertise of a biological sample, say blood, can give false negative results when compared with DNA from other cell populations (eg. from saliva or sperm). For example if we have a sperm sample from a crime scene that we compare with a saliva sample taken from a suspect these could lead to false negative result if the suspect is a case of chimerism. In this case the only way to exclude a suspect is to test the same biological product as that collected at the scene crime.

### ***Databases in E.U.***

Since the creation of the United Kingdom National DNA Database in 1995, many European countries have legislated laws for initiating and regulating their own databases.<sup>11</sup> In 2008, the Council of the European Union converted the Treaty of Prüm into EU legislation, according to which, every EU country is required to establish a forensic DNA database which should be available for automated searches by other EU member states. Dealing with databases and their regulation is a matter of what local communities are willing to accept, since such decisions can affect the whole community.<sup>12</sup> There are currently three available approaches concerning forensic DNA databases, each with its advantages and drawbacks.<sup>13</sup> The first one,

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<sup>9</sup> V. L. Souter et al., *A case of true hermaphroditism reveals an unusual mechanism of twinning*, in "Hum. Genet.", 121, 2007, p. 178-185.

<sup>10</sup> B. Glock et al., *Investigation of chimerism in a healthy, adult female by means of minisatellite and microsatellite typing*, in "International Congress Series", 1239, 2003, p. 561-563.

<sup>11</sup> P. M. Schneider, P. D. Martin, *Criminal DNA databases: the European situation*, in "Forensic Sci. Int.", 119, 2001, p. 232-238.

<sup>12</sup> ENFSI DNA Working Group: ENFSI document on DNA database management <http://www.enfsi.eu/page.php?uid=98>.

<sup>13</sup> M. Guillen, M.V. Lareu, C. Pestoni, A. Salas, A. Carracedo, *Ethical-legal problems of DNA databases in criminal investigation*, in "J. Med. Ethics", 26, 4, 2000, p. 266-271.

which is the most permissive, which is based on the genotyping and the inclusion of the general population on the database. The second one, which is more conservative, which permits DNA fingerprinting and the inclusion of profiles on the database only for a specific list of crimes and only for individuals linked to a high degree with a crime. Finally, the third one, which is against the preparation of DNA databases for criminal investigation. According to this approach, the forced subjection of the individual to testing is ordered only when there is clear proof that the individual is closely linked to the crime committed. Nevertheless, the results of the DNA analysis are not stored in a database, and, according to this approach, this guarantees that they will not be used for purposes other than those for which they were initially carried out.

DNA databases give rise to several ethical-legal problems and scientists are skeptical about their benefits versus their social and ethical costs.<sup>14</sup> The launching and regulation of forensic DNA databases is in accordance with the European Court of Human Rights only if they guarantee that the principle of proportionality is respected.<sup>15</sup>

### **Conclusions**

- The expansion in the use of DNA has not only increased the extent of interaction between forensic scientists and lawyers but more importantly, it has increased the relevance of socio-legal and ethical perspectives in strategies for applying forensic DNA techniques
- The errors that can occur in DNA analysis include sample switches, inappropriate testing and reporting, malfunctioning equipment or reagents, and testimony inconsistent with written reports.
- 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.
- Chimerism can be a pitfall in forensic investigations like paternity testing and crime cases and should be taken into discussion.
- In Romania no official body controls the validity of DNA typing procedures as well as possible violation of personal rights of individuals included in the genetic analysis.

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<sup>14</sup> M. Levitt, *Forensic databases: benefits and ethical and social costs*, in "Br. Med. Bull.", 83, 2007, p. 235–248.

<sup>15</sup> M. Guillen Vazquez, C. Pestoni, A. Carracedo, *DNA databases for criminal investigation purposes: technical aspects and ethical-legal problems*, in "Law Hum. Genome Rev.", 8, 1998, p. 137–158.