



FACTORS AFFECTING DNA PRESERVATION IN FORENSIC GENETICS: DEGRADATION, CONTAMINATION, AND EXPERT DECISION-MAKING

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Abstract. DNA analysis is one of the most informative methods of forensic genetic identification; however, its reliability largely depends on the preservation of biological material and the absence of contamination. DNA degradation leads to molecular fragmentation, allele dropout, reduced amplification efficiency, and the formation of partial genetic profiles. Contamination, in turn, can introduce extraneous alleles and significantly distort forensic examination results. This article examines the main mechanisms of DNA degradation and contamination, analytical indicators of sample damage, the impact of these processes on forensic genetic interpretation, as well as modern diagnostic approaches, preventive measures, and technological solutions.

Keywords: DNA degradation, DNA contamination, forensic genetics, STR analysis, mixed profiles, forensic medical examination.

Introduction. DNA profiling currently occupies a central position in forensic medicine and criminalistics due to its exceptionally high discriminatory power, statistical reliability, and methodological versatility. Genetic information preserves individual human characteristics and enables not only personal identification but also the establishment of biological kinship, analysis of mixed biological traces, reconstruction of crime circumstances, and resolution of a wide range of expert tasks. For this reason, DNA analysis is regarded by modern forensic institutions as the “gold standard” of the evidentiary framework.

However, the effectiveness and reliability of DNA analysis directly depend on the quality of the original biological material. Unlike samples obtained under

controlled medical conditions, traces collected at crime scenes are usually exposed to a variety of uncontrolled environmental factors. These factors include:

temperature fluctuations, particularly elevated temperatures, which accelerate the breakdown of DNA strands;

high humidity, promoting hydrolytic damage and microbial degradation;

ultraviolet radiation, causing the formation of thymine dimers and oxidative damage to nucleotides;

microbiological contamination, in which bacteria and fungi actively degrade nucleic acids through the action of nucleases;

chemical contaminants (cleaning agents, blood, soil, gasoline, household chemicals), which may inhibit polymerase chain reactions or directly disrupt the molecular structure of DNA.

This complex combination of adverse influences significantly increases the risk of DNA degradation and contamination, thereby complicating forensic genetic analysis and interpretation.

The combined effect of these factors creates a high risk of DNA degradation, defined as the progressive destruction of its molecular structure. Degradation results in the preservation of only short DNA fragments suitable for amplification, while longer genomic regions are lost. This manifests as partial genetic profiles, allele dropout, and stochastic effects that significantly complicate result interpretation.

An equally important problem is DNA contamination, which refers to the introduction of extraneous genetic material into the sample under investigation. Potential sources of contamination include:

personnel involved in the collection and analysis of biological material;

medical staff providing care to injured individuals;

other objects present at the crime scene;

laboratory instruments, surfaces, reagents, and consumables.



Even minimal amounts of foreign DNA may lead to erroneous interpretation, particularly in cases involving low-template or touch DNA analysis, where any external contribution can dominate the original genetic signal.

Thus, DNA degradation and contamination represent key factors that can substantially reduce the quality of genetic profiles and cause errors in forensic genetic interpretation. Poor-quality or contaminated samples increase the likelihood of:

- false inclusion or exclusion of individuals;
- generation of partial, non-reproducible, or mixed profiles;
- incorrect reconstruction of events;
- judicial errors, including unjustified conviction or acquittal.

In this context, understanding the mechanisms of DNA degradation and contamination, as well as developing and implementing modern preventive measures, is of paramount importance for ensuring the reliability of forensic genetic analyses and enhancing the objectivity of expert conclusions.

Materials and Methods. The preparation of this review and analytical study was based on a comprehensive methodological approach that included analysis of regulatory documents, scientific literature, laboratory protocols, and practical data from forensic genetic examinations.

The primary materials of the study included:

International standards, regulations, and methodological guidelines defining quality and reliability requirements for DNA analysis:

- ISO/IEC 17025 (laboratory accreditation);
- recommendations of the International Society for Forensic Genetics (ISFG);
- guidelines of the European Network of Forensic Science Institutes (ENFSI);
- protocols of the Scientific Working Group on DNA Analysis Methods (SWGDDAM).



These documents were analyzed to identify key requirements for preventing DNA degradation and contamination, as well as to assess applied quality control measures.

Scientific publications and monographs on forensic genetics, biological material degradation, and mechanisms of contamination published between 2005 and 2024. The reviewed literature addressed:

- biochemical mechanisms of DNA degradation;
- environmental factors affecting DNA preservation;
- methods for analyzing degraded and low-template samples;
- issues of false-positive and false-negative results;
- the development of mini-STR, mtDNA, and next-generation sequencing (NGS) technologies;

modern approaches to the interpretation of mixed and damaged genetic profiles.

Practical protocols of forensic genetic laboratories, including procedures for: collection, transportation, and storage of biological traces; preparation of reagents and equipment; prevention of cross-contamination; performance of pre-PCR and post-PCR processes under zoned laboratory conditions; implementation of internal and external quality control.

Documented cases of DNA degradation and contamination described in forensic expert practice, including examples of:

- allele dropout and stochastic effects in the analysis of aged biological traces;
- appearance of extraneous alleles due to breaches in sterile conditions;
- analytical errors caused by amplicon contamination;
- incorrect interpretation of mixed profiles at low DNA concentrations.

These cases were used to systematize typical errors and to develop practical recommendations.

The methodological framework included:

comparative analysis aimed at contrasting national and international standards, identifying procedural differences, and assessing the applicability of foreign methodologies in local forensic practice;

critical error analysis based on classification of risk factors associated with DNA degradation and contamination;

systematization of biochemical, technical, and organizational factors influencing sample quality;

analytical characterization of degradation and contamination indicators detected at the stages of qPCR, STR genotyping, and data interpretation;

review of modern technological solutions, including mini-STR kits, qPCR platforms, high-throughput sequencing (NGS), and probabilistic genotyping software.

Each analyzed source and protocol was evaluated according to the following criteria:

relevance to forensic genetic practice;

methodological rigor and data reliability;

compliance with international standards;

availability of descriptions of errors, limitations, and preventive strategies;

applicability of recommendations to real-world forensic laboratory conditions.

Overall, the applied methodology enabled a comprehensive assessment of the key problems related to DNA degradation and contamination, identification of patterns underlying these phenomena, evaluation of their impact on genetic profile interpretation, and formulation of modern approaches for prevention and minimization of errors in forensic genetic practice.

Results and Analysis. The analysis of contemporary scientific data, international standards, and forensic expert practice demonstrated that DNA degradation and contamination are key factors that significantly affect the reliability



of forensic genetic examinations. DNA degradation was found to be a complex biochemical process involving hydrolysis, oxidation, microbial degradation, and thermal damage. Hydrolytic reactions lead to depurination and strand breaks, whereas oxidative processes cause nucleotide modifications that inhibit amplification. Microbiological activity is manifested through the action of endonucleases and exonucleases, while freeze–thaw cycles result in mechanical fragmentation of DNA strands. In practice, these processes lead to preferential amplification of short STR fragments, dropout of longer alleles, reduced peak heights, and pronounced stochastic effects.

In addition to degradation, contamination has a substantial impact on genetic profiles and may occur at various stages of handling biological material. Primary contamination can arise even before evidence collection through contact with unrelated individuals or surfaces. Secondary contamination is associated with violations during collection, packaging, and transportation, allowing transfer of cells or DNA between objects. Laboratory contamination is caused by amplicon carryover, residual traces from previously processed samples, inadequate equipment sterilization, or failure to maintain proper laboratory zoning. Analysis of practical cases showed that laboratory-related violations most frequently result in the appearance of extraneous alleles and the formation of false mixed profiles.

Characteristic features of degraded samples include dropout of long STR alleles, peak height imbalance, low overall signal intensity, and fragmented profiles. In contrast, contaminated samples are typically characterized by the presence of additional foreign alleles, low-amplitude non-reproducible peaks, and profiles inconsistent with the assumed number of contributors. Comparative analysis of electropherograms demonstrated that the combination of degradation and contamination can substantially complicate interpretation, particularly when dealing with low-template DNA.

The application of modern technologies significantly improves analytical outcomes. Mini-STR kits demonstrate high efficiency in the analysis of severely



degraded samples; qPCR enables quantitative assessment of DNA degradation and prediction of amplification success; mitochondrial DNA analysis provides results in cases of complete nuclear DNA degradation; and next-generation sequencing (NGS) methods allow analysis of ultra-short fragments and reconstruction of genetic profiles with high accuracy.

Systematization of forensic errors revealed that most problems arise from organizational violations (failure to comply with storage conditions, chain of custody, and sampling rules), technical errors (reagent contamination, improper equipment handling), and interpretative inaccuracies (incorrect evaluation of mixed or low-amplitude profiles). These findings emphasize the necessity of strict adherence to international standards, regular quality control, and continuous professional training of personnel.

Overall, the results demonstrate that DNA degradation and contamination exert a combined and multifactorial impact on the accuracy and reliability of forensic genetic analyses. Timely identification of sample damage, use of advanced technologies, and strict preventive measures substantially enhance the reliability of genetic profiles and reduce the likelihood of expert errors.

Conclusion. The conducted analysis confirms that DNA degradation and contamination are among the most significant factors affecting the reliability and interpretative validity of forensic genetic examinations. DNA degradation is driven by hydrolytic, oxidative, microbiological, and thermal processes, leading to disruption of molecular structure and formation of partial or fragmented genetic profiles. Contamination, which may occur at all stages—from evidence collection to laboratory analysis—can introduce extraneous alleles and generate false mixed profiles, thereby increasing the risk of expert and judicial errors.

The findings highlight that timely diagnosis of sample damage, identification of degradation and contamination indicators, and proper assessment of sample quality are essential prerequisites for obtaining reliable DNA profiles. The application of modern technologies—such as mini-STR, qPCR, mitochondrial DNA

analysis, NGS, and probabilistic genotyping—significantly expands analytical capabilities for low-template, aged, and degraded samples, improving accuracy and reproducibility.

Strict compliance with international standards governing sample collection, storage, transportation, and laboratory processing is of particular importance. Implementation of quality management systems, laboratory zoning, regular equipment calibration, use of negative controls, and continuous staff training are key elements in error prevention.

In conclusion, ensuring the reliability of forensic genetic expertise requires a comprehensive approach that integrates controlled working conditions, advanced analytical technologies, and competent data interpretation. Only the combination of these factors allows minimization of erroneous conclusions, enhancement of expert objectivity, and preservation of the high evidentiary value of DNA analysis in forensic medical practice.

REFERENCES.

Butler JM. Advanced Topics in Forensic DNA Typing: Methodology. Academic Press; 2012.

Gill P, Brenner C, Buckleton J, Carracedo A, Krawczak M, Mayr WR, et al. DNA Commission of the International Society for Forensic Genetics (ISFG): Recommendations on the interpretation of mixtures. *Forensic Sci Int Genet*. 2006;160(2–3):90–101.

Alaeddini R. Forensic implications of DNA degradation: A review. *J Forensic Sci*. 2012;57(6):1407–1415.

van Oorschot RAH, Ballantyne KN, Mitchell RJ. Forensic trace DNA: A review, pitfalls, and potential solutions. *Forensic Sci Int Genet*. 2010;4(4):299–306.

Linacre A, Tobe SS. An overview of DNA contamination in forensic science. *Forensic Sci Int Genet*. 2011;9:1–11.

ENFSI DNA Working Group. Best Practice Manual for DNA Sampling. European Network of Forensic Science Institutes; 2020.



SWG DAM. Validation Guidelines for DNA Analysis Methods. Scientific Working Group on DNA Analysis Methods; 2020.

Parson W, Ballantyne J, Budowle B, Butler JM, Gettings KB, Gill P, et al. Massively parallel sequencing in forensic genetics: Current applications and future directions. *Forensic Sci Int Genet*. 2019;41:1–13.

Young JM, Oldroyd N, Watson SK, et al. DNA degradation in forensic samples: Mechanisms, consequences, and methods of detection. *Forensic Sci Rev*. 2020;32(2):123–140.

Higgins D, Austin JJ. Teeth as a source of DNA for forensic identification: A review. *Sci Justice*. 2019;59(2):163–172.

Ambers A, Turnbough M, Benjamin R, et al. Modified mini-STR assay for degraded DNA. *Forensic Sci Int Genet*. 2018;34:148–156.

Opel KL, Chung DT, McCord BR. A study of PCR inhibition mechanisms using real-time PCR. *J Forensic Sci*. 2010;55(1):25–33.

Caldwell A, Latham K, Knudsen PJ. Environmental effects on DNA persistence in biological stains. *Forensic Sci Int*. 2018;286:25–32.

Amorim A, Pinto N. Forensic genetics and the degradation of biological samples: New methods and challenges. *Genes*. 2021;12(3):416.

ISO/IEC 17025:2017. General requirements for the competence of testing and calibration laboratories. International Organization for Standardization.