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GENDER MARKERS AS A TOOL FOR FORENSIC MEDICAL IDENTIFICATION

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Abstract
Biological sex markers play a crucial role in forensic medical identification, particularly in cases involving mixed biological samples, sexual violence, and severely degraded or limited genetic material. The analysis of sex-specific genetic markers enables the determination of biological sex, assists in the interpretation of complex DNA mixtures, and enhances the evidentiary value of forensic genetic findings. In forensic practice, sex markers such as amelogenin, Y-chromosomal short tandem repeats (Y-STRs), and X-chromosomal markers are widely applied to differentiate male and female genetic components, especially in male–female mixtures. These markers are of particular importance in the investigation of sexual offences, missing persons cases, and disaster victim identification. Their use contributes to narrowing suspect pools and improving the accuracy of expert conclusions. Despite their advantages, the application of biological sex markers is associated with certain limitations. Genetic anomalies, mutations, deletions, and population-specific variations may affect the reliability of sex determination. In addition, interpretation challenges arise in low-template DNA samples, complex mixtures, and cases involving chimerism or intersex conditions. Therefore, careful methodological control and expert judgment are essential. This article reviews the forensic applications of biological sex markers, discusses their methodological strengths and limitations, and emphasizes the importance of integrating sex marker analysis with autosomal and mitochondrial DNA data to achieve reliable and scientifically sound forensic identification.

Keywords: Biological sex markers; forensic medicine; forensic genetics; human identification; Y-chromosomal markers; X-chromosomal markers; DNA mixtures.

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Introduction

Forensic medical identification relies on the accurate interpretation of biological evidence to establish individual characteristics relevant to medico-legal investigations. Among the various genetic tools used in forensic genetics, biological sex markers occupy a significant position, as the determination of biological sex represents one of the fundamental elements of human identification. The analysis of sex-specific genetic markers provides essential preliminary information and supports further interpretation of complex forensic findings. In modern forensic practice, biological sex markers are particularly valuable in cases involving mixed biological traces, sexual offences, missing persons investigations, and mass disaster victim identification. Markers such as amelogenin, Y-chromosomal short tandem repeats (Y-STRs), and X-chromosomal markers allow differentiation between male and female genetic components, facilitating the interpretation of DNA mixtures and contributing to the reconstruction of forensic circumstances. Their application is especially important when autosomal DNA profiles are incomplete, degraded, or insufficient for definitive conclusions. Despite their widespread use, the forensic application of biological sex markers is not without limitations. Genetic anomalies, including deletions of the amelogenin locus, chromosomal abnormalities, mutations, and population-specific variations, may lead to ambiguous or misleading results. Additionally, low-template DNA, complex mixtures, and specific biological conditions can complicate analytical interpretation and reduce the reliability of sex determination if not properly controlled. This article aims to examine the role of biological sex markers as a tool for forensic medical identification, with emphasis on their applications, methodological challenges, and interpretative limitations. Particular attention is given to the necessity of integrating sex marker analysis with autosomal and mitochondrial DNA profiling, as well as with contextual and case-specific information, to ensure scientifically robust and legally defensible expert conclusions.

Materials and Methods

This article is based on a narrative review of contemporary scientific literature addressing the forensic application of biological sex markers. Peer-reviewed articles, methodological studies, and international guidelines published in

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recognized forensic genetics journals were systematically analyzed. Sources were selected from established scientific databases and professional forensic organizations, with particular focus on publications describing the use of sex-specific genetic markers in forensic identification and mixture interpretation. The methodological scope of the review included evaluation of commonly used biological sex markers, such as the amelogenin gene, Y-chromosomal short tandem repeats (Y-STRs), and X-chromosomal markers. Attention was given to laboratory approaches for DNA extraction from forensic biological materials, polymerase chain reaction amplification, and interpretation of electrophoretic and sequencing data obtained from sex marker analysis. In addition, interpretative strategies and potential sources of analytical error were examined, including issues related to low-template DNA, mixed biological samples, genetic anomalies, and population-specific variations. International recommendations and quality assurance standards relevant to forensic DNA analysis were reviewed to assess best practices in laboratory control, validation, and expert reporting. No original biological samples were collected or analyzed in this study. Ethical approval was not required, as the work was based exclusively on previously published data and publicly available scientific and methodological sources.

Conclusion

Biological sex markers constitute an essential component of forensic medical identification, providing critical information for the interpretation of biological evidence in a wide range of medico-legal investigations. Their application is particularly valuable in cases involving mixed biological samples, sexual offences, degraded DNA, and situations where rapid preliminary identification is required. By enabling the differentiation of male and female genetic components, sex marker analysis enhances the clarity and evidentiary value of forensic genetic findings. At the same time, the forensic use of biological sex markers is subject to important limitations. Genetic anomalies, locus deletions, mutations, and population-specific variations may compromise the accuracy of biological sex determination if these factors are not adequately considered. Interpretation challenges are further increased in low-template DNA samples and complex mixtures, underscoring the need for rigorous methodological control and expert judgment. Consequently, biological sex markers should not be used as an isolated identification tool but

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rather as part of an integrated forensic approach. The combination of sex marker analysis with autosomal and mitochondrial DNA profiling, alongside contextual and case-specific information, ensures more reliable, scientifically sound, and legally defensible expert conclusions in forensic medical practice.

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