

Opinion

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# Artificial intelligence in forensic genetics: applications and ethical challenges

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## Abstract

Artificial intelligence (AI) is transforming forensic genetics through groundbreaking applications in (1) population structure analysis and biogeographical ancestry inference; (2) microbial detection and body fluid identification; (3) allele recognition and mixture interpretation; (4) age inference and phenotype prediction, (5) kinship analysis; and (6) other emerging domains, such as bloodstain deposition time and transcriptomic analysis. While promising efficiency and enhanced accuracy, its integration also raises ethical, legal, and social concerns. This opinion piece critically explores both the promise and perils of AI in forensic genetics, calling for urgent action to (1) build secure and trustworthy AI systems; (2) develop agile and effective regulatory frameworks; (3) uphold ethical integrity and human-centered design; and (4) foster global collaboration to meet cross-border challenges. Together, these principles are essential to ensuring that AI's integration into forensic science advances both technological progress and the pursuit of justice.

**Keywords:** Artificial intelligence (AI), machine learning (ML), forensic genetics, ethical challenges, data security, explainable AI (XAI)

## INTRODUCTION

Forensic genetics has undergone a profound transformation, driven by advances in genomics,



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computational biology, and high-throughput sequencing technologies. Artificial intelligence (AI), particularly machine learning (ML), now plays an increasingly pivotal role in decoding complex DNA profiles, facilitating individual identification, and supporting criminal investigations<sup>[1]</sup>. Yet, this technological progress is accompanied by a range of ethical and legal challenges that remain insufficiently addressed<sup>[2]</sup>. This article offers a critical examination of both the opportunities and the risks associated with AI in forensic genetics, advocating for a measured and ethically informed approach to its implementation. We conducted a comprehensive search of PubMed and Web of Science (WoS) for publications from the past five years using the query “All Fields = [(Artificial Intelligence) OR (Machine Learning)] AND (Forensic Genetics).” Drawing on recent technological developments and representative case studies, we assess the scientific potential of AI while highlighting the sociotechnical risks it introduces in this rapidly evolving domain.

## TECHNOLOGICAL INNOVATIONS AND FORENSIC APPLICATIONS

### Population structure analysis and biogeographical ancestry inference (BGA)

AI as an auxiliary tool is transforming the resolution at which human population structure and ancestry can be explored. In studies of Chinese and East Asian populations, AI frameworks now integrate diverse molecular markers, including ancestry informative single nucleotide polymorphisms (AISNPs)<sup>[3-5]</sup>, X-chromosome insertion/deletion polymorphisms (X-InDels)<sup>[6]</sup>, Y-chromosomal short tandem repeats (Y-STRs)<sup>[7-9]</sup>, and Y-chromosomal single nucleotide polymorphisms (Y-SNPs)<sup>[10]</sup>, together with advanced ML models such as extreme gradient boosting (XGBoost)<sup>[11-13]</sup>, partial least squares discrimination analysis (PLS-DA)<sup>[14]</sup>, and soft clustering<sup>[15]</sup>. Together, these methods overcome the long-standing limitations of traditional linear models, which relied on small marker sets and offered limited discriminative power. By automatically extracting key signals from large-scale genomic data, AI-driven models<sup>[16-26]</sup> achieve significantly improved precision in distinguishing closely related groups such as northern and southern Han Chinese, Japanese, and Koreans, as well as in tracing haplogroup and surname lineages. This shift marks a methodological leap from descriptive phylogenetic analyses to scalable, data-intensive inference, redefining how population diversity and ancestral origins are characterized across East Asia.

### Microbial detection and body fluid identification

AI as an auxiliary tool is reshaping forensic microbiomics<sup>[27]</sup> and body fluid identification by enabling a multi-omics perspective. Traditional methods, which are limited by single biomarkers and low analytical resolution, are now being replaced by AI frameworks that harness ML and genetic algorithms to mine high-dimensional microbiome and molecular data. By integrating microbial signatures from 16S ribosomal RNA (rRNA) sequencing<sup>[28,29]</sup>, skin and saliva profiles<sup>[30-34]</sup>, and environmental samples such as burial soil and water<sup>[35,36]</sup>, alongside multi-omics markers including single nucleotide polymorphisms (SNPs)<sup>[37]</sup>, transfer RNA-derived small RNAs (tsRNAs)<sup>[38]</sup>, microRNAs<sup>[39]</sup>, messenger RNAs (mRNAs)<sup>[40,41]</sup>, and cytosine-guanine (CpG) methylation<sup>[42]</sup>, these approaches achieve unprecedented sensitivity and specificity. AI-driven models now deliver precise discrimination of body fluid origins<sup>[43-45]</sup>, accurate estimation of postmortem intervals (PMI)<sup>[46,47]</sup>, and geographic traceability<sup>[48,49]</sup>. With their strong robustness and interpretability, AI-driven frameworks significantly outperform conventional statistical models.

### Allele recognition and DNA mixture interpretation

AI as an auxiliary tool is redefining the precision and automation of allele recognition<sup>[50]</sup> and mixture interpretation<sup>[51]</sup> in forensic DNA analysis. By integrating interpretable neural networks<sup>[52,53]</sup> and deep learning (DL) architectures<sup>[54]</sup>, AI systems can now resolve low-template DNA, complex electropherograms, and highly mixed biological samples<sup>[55,56]</sup> with significantly improved accuracy. These models automatically extract informative alleles, infer the number and identity of contributors<sup>[57-60]</sup> within DNA mixtures, and leverage molecular barcoding and probabilistic genotyping<sup>[61]</sup> to enhance analytical confidence and

reproducibility. Overcoming the subjective thresholds and limited resolution of traditional statistical approaches, AI-driven frameworks deliver objective and high-throughput solutions for genetic evidence interpretation. This transition marks a fundamental step toward fully automated, data-driven forensic genetics.

### Age inference and phenotype prediction

AI as an auxiliary tool is expanding the scope of forensic age inference<sup>[62]</sup> and phenotype prediction<sup>[63,64]</sup> through multimodal data fusion and nonlinear modeling. Traditional linear or single-marker frameworks often fail to capture the intricate molecular patterns underlying biological aging and external traits. AI-driven systems now integrate DNA methylation<sup>[65-72]</sup>, non-coding RNA<sup>[73-75]</sup>, and other omics-level features<sup>[76]</sup> using advanced ML and neural network architectures, enabling robust modeling even with limited samples. These approaches markedly enhance the accuracy of age prediction from blood, wound<sup>[77]</sup>, and other forensic materials, accelerating the development of intelligent epigenetic clocks<sup>[76,78]</sup>. Beyond age inference, AI models demonstrate exceptional capacity to predict eye color<sup>[79-81]</sup> and reconstruct three-dimensional facial morphology<sup>[82,83]</sup>, thereby surpassing the explanatory power of conventional genotype-phenotype association models. By addressing the complexity of forensic datasets, AI establishes a scalable and high-precision framework for molecular-level human identification.

### Kinship analysis

AI as an auxiliary tool is strengthening forensic kinship analysis by integrating precision inference with privacy-preserving computation. Conventional short tandem repeats (STRs)-based approaches often struggle to distinguish close relatives, resolve complex familial structures, or protect sensitive genetic data. AI frameworks now fuse multilayered evidence, including rapidly mutating Y-STR profiles<sup>[84-86]</sup>, facial imagery<sup>[87]</sup>, and other biometric or genomic cues, together with homomorphic encryption<sup>[88]</sup> and related privacy-preserving technologies. This integration enables high-resolution identification of complex relationships while maintaining the confidentiality of underlying genetic information. Beyond improving the discrimination of paternal lineages<sup>[84-86]</sup>, AI models have demonstrated the ability to detect incestuous relations<sup>[89]</sup>, which greatly improves the efficiency achieved through manual interpretation or classical statistical methods. By uniting accuracy with security, AI-driven kinship analysis establishes a new paradigm for reliable, ethically sound forensic genetics.

### Other emerging domains

AI as an auxiliary tool is empowering temporal inference in forensic genetics by enhancing both its precision and temporal resolution. ML models that integrate rhythmic mRNA expression profiles now enable the precise estimation of bloodstain deposition time<sup>[90,91]</sup> within a 24-h window, overcoming the static and target-dependent constraints of conventional capillary electrophoresis (CE) assays. Similarly, AI-powered transcriptomic analysis can decode complex temporal signals embedded in molecules such as microRNAs, markedly improving PMI<sup>[92]</sup>. By capturing dynamic biomarker fluctuations, these approaches address one of the central challenges in forensic genetics, providing a more reliable and biologically grounded temporal dimension to evidentiary analysis.

## CURRENT CHALLENGES AND FUTURE DIRECTIONS

### Scientific and technical aspects

Forensic AI systems have technical limitations such as algorithmic bias, model opacity and vulnerability to adversarial manipulation<sup>[93]</sup>; models trained on Eurocentric datasets often misclassify underrepresented populations, and the lack of transparency in “black box” algorithms undermines judicial credibility, requiring explainable AI (XAI) and robust cybersecurity for traceability and reliability<sup>[94]</sup>. Future progress should emphasize explainability and accountability: adopting XAI models such as decision trees and

Bayesian networks<sup>[95]</sup>, using hybrid systems with human oversight and automation, applying validation protocols assessing accuracy and interpretability, and expanding datasets via inclusive genomic initiatives can boost transparency, evidentiary reliability and fairness<sup>[96]</sup>.

### **Governance and regulatory aspects**

The rapid adoption of AI has outpaced the creation of dedicated forensic legislation. Most jurisdictions lack clear rules on accountability, admissibility and data governance. While frameworks such as the European Union (EU) General Data Protection Regulation (GDPR) stress explicit consent, such safeguards are often overridden in the name of public safety, causing regulatory ambiguity<sup>[97]</sup>. Robust governance frameworks need to include transparency requirements and back privacy-preserving technologies such as federated learning and homomorphic encryption<sup>[98]</sup>. Dynamic consent mechanisms should enable individuals to maintain continuous control over their genetic data, matching data governance with changing ethical and legal expectations. Effective oversight requires flexible, risk-adaptive regulatory frameworks supported by mandatory auditing, certification and clearly defined legal responsibilities, and establishing accountability mechanisms is key to ensuring compliance and public trust<sup>[99]</sup>.

### **Ethical and societal aspects**

Extensive use of public and commercial genetic databases sparks concerns about informed consent, data reuse and privacy. Incidents such as unauthorized access to 23andMe data highlight the urgency of transparent data stewardship and ethical oversight<sup>[100]</sup>. Ethical deployment of such databases should follow a “privacy by default” principle, reducing exposure risks at every analytic stage. Prioritizing data equity and ensuring responsible use of population-scale genetic resources are essential to maintain public trust and fairness<sup>[101]</sup>. Meanwhile, AI deployment in this field must uphold fairness, inclusivity and human dignity, with strategies including systematic bias detection, fairness assurance protocols, proactive public engagement, relevant education and human-centered system design<sup>[93]</sup>.

### **Global collaboration aspects**

Disparities in legal, ethical and technical standards across regions hinder equitable, consistent AI application. International bodies such as the International Society of Forensic Genetics (ISFG) need to promote unified guidelines, fairness audits and cross-border cooperation, including creating harmonized standards, sharing safety research findings in non-competitive AI domains and establishing global monitoring networks to ensure responsible global deployment<sup>[102]</sup>. Bridging gaps among scientists, forensic practitioners, legal experts and ethicists requires sustained cross-sector engagement; continuous multistakeholder dialogue, joint training and collaborative policy efforts can drive socially responsible, globally consistent implementation of forensic AI<sup>[103]</sup>.

## **CONCLUSION**

AI is profoundly reshaping forensic genetics across a spectrum of critical applications. These include population structure analysis, BGAI, microbial detection, body fluid identification, allele recognition, mixture interpretation, age inference, phenotype prediction, kinship analysis, and other emerging frontiers such as bloodstain deposition time and transcriptomic analysis. By integrating multi-omic, imaging, and dynamic biological signals, AI transforms static genetic profiling into a multidimensional and predictive science. Yet the power of these technologies also demands foresight: data security, algorithmic bias, and ethical accountability must evolve in parallel with technical innovation. The future of forensic AI depends not only on model accuracy but also on transparency, robustness, and governance by design, which embeds ethical and legal safeguards from the outset. Through interdisciplinary collaboration, open international standards, and public engagement, AI can mature into a responsible scientific partner that enhances human identification and understanding, ultimately strengthening both the rigor and the humanity of modern forensic genetics.

## DECLARATIONS

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The author contributed solely to the article.

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The author declared that there are no conflicts of interest.

### Ethical approval and consent to participate

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### Consent for publication

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