

# Artificial Intelligence and its Role in Forensic Karyotyping: A Systematic Review

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## Abstract

**Introduction:** One of the most important aspects of forensic investigations and genetic research is forensic karyotyping, which involves analyzing a person's chromosomes to find genetic anomalies and establish identification. The development of artificial intelligence (AI) technology offers a chance to improve and automate the forensic karyotyping procedure. This study examines the possible advantages and difficulties of artificial intelligence (AI) in forensic karyotyping. In forensic science, forensic karyotyping is essential for providing an accurate interpretation of genetic data for use in legal and investigative processes. It can offer useful details regarding genetic problems, such as chromosomal abnormalities or mutations, which can help with personal identification, paternity determination, or supplying proof in criminal investigations.

**Aim:** To give a general review of how artificial intelligence is used in forensic karyotyping, evaluate its possible advantages, and address any relevant issues. By being aware of Artificial Intelligence's promise and limitations in this field, we may set the stage for its efficient integration into forensic practitioner.

**Methods:** A database search we did to start the inquiry turned up 582 documents. There were 216 unique records left after duplicates were eliminated. 232 items were subsequently eliminated as a result of download problems. A final sample of 31 research was chosen from the 134 full-text papers that were evaluated (n=134), with 103 being eliminated owing to quality issues.

**Result:** The use of artificial intelligence (AI) in forensic karyotyping has several advantages, including automated chromosomal analysis, quicker abnormality discovery, and increased uniformity. For a successful application, challenges such as a lack of labelled datasets and ethical issues must be resolved.

**Conclusion:** By increasing productivity, precision, and uniformity, artificial intelligence has the potential to transform forensic karyotyping. While there are obstacles, continued study and cooperation amongst several fields might help you get through them. The ethical and appropriate use of AI in forensic karyotyping will improve forensic investigations, boost genetic research, and expand the use of genetics in the legal system.

**Keywords:** Artificial Intelligence (AI), Criminal Investigations, Forensic Karyotyping, Genetic Abnormalities, Genetic Profiles, Labelled Datasets, etc.

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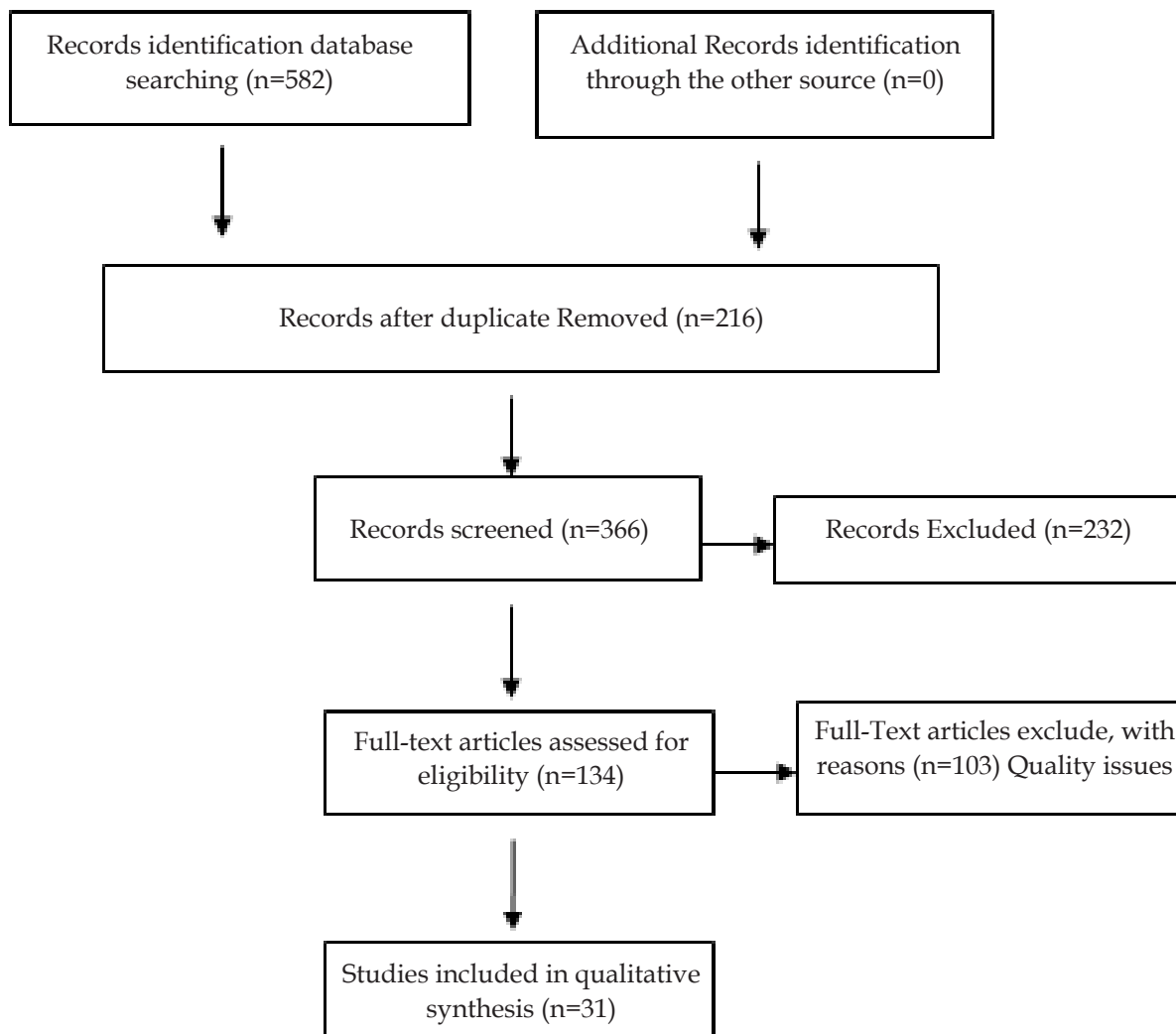
## Introduction

In forensic investigations, forensic karyotyping is an essential part of the genetic study to identify an individual and identify genetic anomalies.<sup>[1]</sup> It includes the investigation and interpretation of a person's chromosomes, usually by manual inspection by forensic professionals.<sup>[2]</sup> But this procedure could involve a lot of work, take a long time, and involve mistakes made by people.<sup>[3]</sup> The use of AI approaches to automate and improve the process of forensic karyotyping is becoming more and more popular as a result of the fast breakthroughs in AI.<sup>[4]</sup> Artificial intelligence (AI) is the field of study that deals with the creation of computer systems that are capable of doing activities that normally require human intellect, including perception, reasoning, learning, and problem-solving.<sup>[5][6]</sup> The use of AI methods in forensic karyotyping has considerable promise for enhancing effectiveness, accuracy, and uniformity.<sup>[7]</sup> AI techniques, like machine learning and deep learning, have demonstrated significant potential in a variety of domains.<sup>[8]</sup> The use of AI in forensic karyotyping is multidimensional and has several important advantages.<sup>[9]</sup> First, AI algorithms can automate the time-consuming operation of chromosomal analysis, greatly cutting down on the amount of time and effort needed by forensic professionals.<sup>[10]</sup> AI can acquire the capacity to precisely detect and categorize chromosomal abnormalities by utilizing machine learning algorithms to learn from labelled information.<sup>[11]</sup> This can help to identify genetic abnormalities, provide genetic profiles for criminal investigations, and offer insightful data for genetic research. Additionally, uniformity in forensic karyotyping is a problem that AI can solve.<sup>[12][13]</sup> Traditional karyotyping techniques might be vulnerable to subjectivity and inter-observer variability, producing erratic findings.<sup>[14]</sup> Deep learning methods allow AI computers to learn from enormous datasets and provide uniform standards for classifying chromosomal anomalies.<sup>[15]</sup> The overall quality of forensic karyotyping analyses can be improved by this standardization, which can increase the results' dependability and repeatability.<sup>[16]</sup> Despite the potential advantages, incorporating AI into forensic karyotyping also poses certain difficulties. The lack of labelled datasets with annotated karyotypes for AI model training is a

serious obstacle.<sup>[17]</sup> Building reliable and precise AI systems requires acquiring and curating such datasets that reflect diverse chromosomal abnormalities.<sup>[18]</sup> To overcome this issue and provide comprehensive datasets that can propel the development of AI in forensic karyotyping, a collaboration between geneticists, forensic specialists, and computer scientists is crucial.<sup>[19]</sup> The application of AI in forensic karyotyping also heavily relies on ethical issues. To guarantee the appropriate and ethical usage of AI algorithms, considerations for patient privacy, informed permission, and data security must be taken.<sup>[20]</sup> In the course of the forensic karyotyping procedure, security measures should be in place to safeguard private genetic data and guarantee that people's rights are upheld.<sup>[21]</sup> Artificial intelligence has the power to transform forensic karyotyping by increasing productivity, precision, and consistency.<sup>[22]</sup> Chromosome analysis automation and the creation of AI algorithms capable of precisely identifying and categorizing chromosomal anomalies can greatly improve forensic investigations and genetic research.<sup>[23]</sup> To integrate AI in forensic karyotyping responsibly, however, issues with the accessibility of labelled datasets and ethical issues must be resolved.<sup>[24]</sup> To overcome these obstacles and realize the full potential of AI in this area, further study and collaboration between computer scientists, geneticists, and forensic specialists are essential.<sup>[25]</sup> The effective use of AI-driven forensic karyotyping technologies can have wide-ranging effects, enhancing forensic investigations, expanding our understanding of genetics, and promoting the use of genetics in the legal system.<sup>[26]</sup>

## Methodology

We began our investigation by searching the database for a specific term associated with our subject, which revealed 582 documents. We eliminated duplicate entries to confirm the veracity of our findings, leaving 216 unique records. Following a thorough review, 232 of these recordings were eliminated because of download problems. After the first screening, we continued to evaluate the full-text articles' suitability, reducing our selection to 134 articles for additional study. 103 items were eliminated after a comprehensive review due to quality issues. A final selection of 31 papers that satisfied our criteria and were included in the qualitative synthesis was obtained through this procedure.



**Prisma Flow chart:**

## Result

There are several advantages to using artificial intelligence (AI) in forensic karyotyping. Automating the time-consuming process of chromosome analysis, it helps hasten the discovery and categorization of chromosomal abnormalities. AI has the ability to increase consistency by creating uniform interpretational standards and minimizing subjectivity among observers. For successful implementation, there are a few obstacles that must be overcome. The scarcity of labelled datasets, which are essential for training AI systems, is one issue. Furthermore, ethical issues related to AI in forensic karyotyping need to be thoroughly investigated and settled. It is essential to overcome these obstacles to properly utilize AI in forensic karyotyping and obtain improved consistency and efficiency.

## Discussion

An overview of artificial intelligence and its uses in forensic science, particularly in the area of karyotyping, will be covered in this talk. It would investigate the possible advantages of automating and improving forensic karyotyping procedures using AI.<sup>[27]</sup> This debate would dive into the use of AI in forensic karyotyping to analyse chromosomal abnormalities. It would investigate how AI algorithms may help in the detection and classification of different abnormalities, including duplications, translocations, inversions, and deletions. The benefits and drawbacks of AI-assisted analysis would be looked at.<sup>[28]</sup> The use of machine learning approaches for pattern detection in karyotyping will be the main topic of this lecture. It would investigate the training of AI algorithms on

massive datasets to find trends and more precisely characterize chromosomes. We would also talk about the difficulties in creating reliable machine-learning models for karyotyping.<sup>[29]</sup> Karyotyping Workflow Automation Using AI. This conversation would focus on using AI technology to automate the karyotyping routine. It would investigate how automating time-consuming and repetitive operations like picture capture, chromosomal segmentation, and pairing may be aided by AI. The potential benefits of automation powered by AI for increasing productivity and lowering human mistakes will be highlighted.<sup>[30]</sup> This conversation would cover the moral questions raised by the use of AI in forensic karyotyping. It would include issues including privacy worries, the requirement for human monitoring of decision-making, and potential biases in AI algorithms, among others. We would talk about ways to guarantee openness, justice, and accountability in AI-driven karyotyping procedures.<sup>[31]</sup>

### Conclusion

Through improved productivity, accuracy, and consistency, artificial intelligence (AI) has the potential to significantly improve forensic karyotyping. Even if there are difficulties, continued multidisciplinary research and cooperation can help remove these barriers. It is possible to enhance forensic investigations, promote genetic research, and expand the use of genetics in the legal system through the ethical and responsible use of AI in forensic karyotyping, opening the door for more effective and efficient methods in this important area.

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