International Journal of Medical Toxicology & Forensic Medicine
2026, Volume 16, E51792

Research Paper

Automatic Chromosomal Abnormality Detection Using Varifocal-Net with CNN

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Citation Kannabiran K, Thangapalani L, Santhanalakshmi M. Automatic Chromosomal Abnormality Detection Using Varifocal-Net with CNN. International Journal of Medical Toxicology and Forensic Medicine. 2026; 16:E51792.

https://doi.org/10.22037/ijmtfm.v16.51792

Article info:
Received: 20 Feb, 2026
First Revision: 25 Feb, 2026
Accepted: 04 Mar, 2026
Published: 25 Apr, 2026

Keywords:
Chromosomal abnormality detection, Karyotyping, Convolutional neural network, Varifocal-Net, Deep learning, Cytogenetics

ABSTRACT

Background: Chromosomal abnormalities are a major cause of hereditary disorders, congenital anomalies, and developmental impairments. Conventional karyotype analysis relies on manual inspection by cytogenetic experts, making it time-consuming and prone to subjective interpretation.

Methods: To overcome these shortcomings, this article illustrates an automated chromosomal abnormality detection framework based on a Varifocal-Net–integrated Convolutional Neural Network (CNN) architecture. The proposed model adopts a dual-scale learning strategy, where a global-scale network captures overall chromosomal morphology, and a local-scale network extracts fine-grained structural features such as banding patterns and centromere regions. Deep feature learning is achieved using dual VGG-16 backbones enhanced with residual connections and multi-task learning. The system has undergone training and validation on a curated and expert-verified karyotype image dataset.

Results: Significant classification of performance is demonstrated by findings on experiments achieving 99.04% accuracy, 98.63% sensitivity, and 100% specificity, outperforming baseline CNN and residual architectures.

Conclusion: The inferences depicts that the proposed framework offers a reliable and efficient solution for automated analysis of karyotype evidencing promising potential for clinical decision-support applications.

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Introduction

Chromosomal abnormalities arise from numerical or structural alterations in chromosomes .The above-mentioned factors are associated with congenital diseases, congenital anomalies, infertility, and various cancers [1]. Numerical abnormalities involve deviations from the normal diploid chromosome number, deletions, duplications, inversions, and translocations are examples of structural anomalies, that can have major impact on gene expression and phenotypic outcomes.

Conventional karyotype analysis is primarily performed through manual inspection of chromosome spreads by trained cytogenetic experts. Although widely adopted in clinical practice, this process is labor-intensive, time-consuming, and prone to interobserver variation. Factors such as chromosome overlap; curvature, non-uniform staining, and image noise make precise interpretation, thereby motivating the necessity for automated and reliable computational approaches [2].

Early attempts at automated chromosome analysis relied on handcrafted features such as chromosome length, area, centromere position, and intensity distribution. While these methods provided partial automation, their performance was limited due to sensitivity to staining variations and limited generalization amidst datasets [3]. Medical image analysis has been impacted by the rapid advancement of deep learning, particularly through convolutional neural networks (CNNs), which enable automatic hierarchical feature extraction directly from raw image data [4].

Multi-scale CNN architectures and attention-guided learning frameworks can improve chromosome classification and abnormality detection by capturing both global structural context and local discriminative patterns [5], [6]. Residual and densely connected networks have further enhanced feature reuse and training stability in deep architectures [7]. Additionally, weakly supervised learning methodologies explored to reduce annotation dependency in cytogenetic image analysis [8]. Despite these advances, accurate detection of subtle chromosomal abnormalities remains challenging due to fine-grained visual similarities among chromosome classes.

Varifocal-Net, originally developed for fine-grained visual recognition, effectively integrates global contextual information with localized discriminative features, making it well-suited for chromosome image analysis [9]. Motivated by its proven effectiveness, this work extends Varifocal-Net to automated chromosomal abnormality detection by integrating it with deep CNN feature extractors.

The primary findings of this work are summarized as follows:

- A dual-scale Varifocal-Net-based framework for comprehensive chromosome feature extraction.
- Integration of VGG-16 deep feature learning with residual connections for improved robustness.
- Application of multi-task learning to enhance classification accuracy and generalization.

Comprehensive experimental confirmation showcasing enhanced effectiveness over baseline deep learning models.

Materials and Methods

This segment outlines the dataset, preprocessing strategy, network architecture, learning mechanisms, and evaluation criteria used in the proposed chromosomal abnormality detection framework. The overall methodology is designed to ensure robustness, generalization, and clinical relevance by combining dual-scale feature learning with deep convolutional representations. The proposed system follows a structured pipeline consisting of dataset preparation, feature extraction using Varifocal-Net and CNN backbones, multi-task learning, and comprehensive performance evaluation (Figure 1) illustrates dual-scale feature extraction using global and local subnetworks, deep feature learning with VGG-16 backbones, residual connections, and multi-task classification.

Dataset Preparation

The dataset employed in this research consists of 3,200 karyotype chromosome images collected from clinical cytogenetic laboratories and validated by experienced domain experts to ensure annotation reliability and diagnostic correctness. The dataset includes a balanced representation of normal and abnormal chromosomes, enabling unbiased learning and evaluation.

- Normal chromosomes: 1,600
- Abnormal chromosomes: 1,600

The abnormal class includes both numerical abnormalities, such as trisomy, and structural chromosomal anomalies, including deletions, duplications, and translocations. This diversity allows the proposed methodology to train a wide range of chromosomal variations encountered in real clinical scenarios.

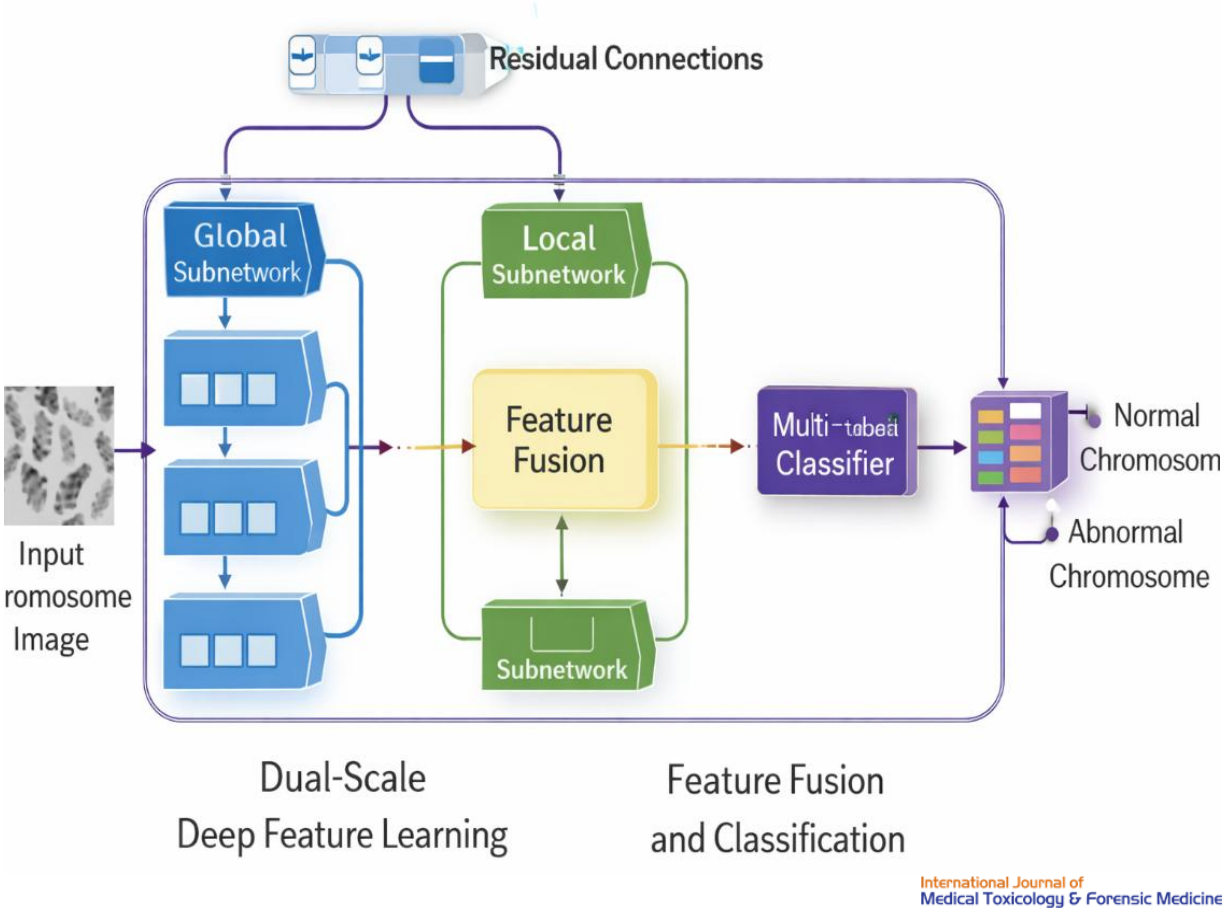

Figure 1. Proposed Varifocal-Net–integrated CNN framework.

The data collection is segregated into 70% training, 15% validation, and 15% testing sets to ensure proper training of the model, tuning of hyper parameter and unbiased performance evaluation.

Images are rescaled to a uniform resolution and intensity-normalized to reduce inter-sample variability caused by staining and imaging conditions before training. Data augmentation methods, including rotation, horizontal and vertical flipping, and contrast enhancement, are utilized during training to enhance model generalization and minimize over fitting. These augmentations simulate real-world variations in chromosome orientation and appearance.

Varifocal-Net Architecture

The proposed framework is based on a dual-scale Varifocal-Net architecture, which is specifically designed to encompass global and local chromosomal characteristics. This architecture consists of two complementary subnetworks:

Global-Scale Network (G-Net):G-Net processes the entire chromosome image to extract holistic features related to overall morphology, shape, size, and orientation. These characteristics offer crucial contextual details for distinguishing chromosome categories.

Local-Scale Network (L-Net):L-Net focuses on localized and discriminative regions such as centromeres, banding patterns, and structural irregularities. By refining the regions identified by G-Net, L-Net enables accurate detection of subtle abnormalities.

Feature fusion is performed using channel-wise concatenation followed by attention-weighted aggregation, enabling the model to highlight diagnostically significant areas while diminishing irrelevant background information. This fusion strategy results in a resilient and seelctive feature representation for classification.

Deep Feature Extraction Using VGG-16

Deep feature extraction is performed using VGG-16, an effective architecture with proven capability in hierarchical feature learning is selected. In the proposed model:

- Pre-trained VGG-16 networks are fine-tuned using chromosome images to adapt learned features to the cytogenetic domain.

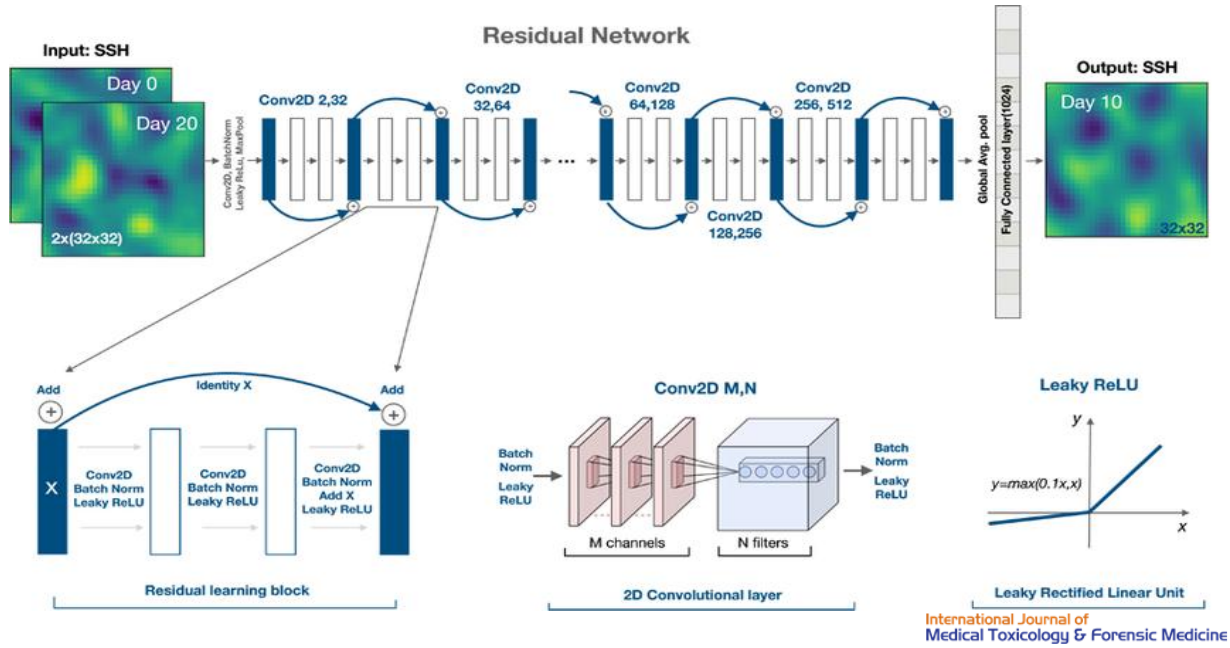

Figure 2. Proposed Varifocal-Net–based residual CNN architecture for chromosomal abnormality detection.

- Dense layers are eliminated to enable flexible feature extraction and reduce model complexity.

- Dual VGG-16 backbones are employed to separately learn features relevant to autosomal and sex chromosome abnormalities, enabling specialized representation learning.

This design enhances the ability to distinguish the network while maintaining stable and efficient training behavior. Figure 2 illustrates Architecture of the residual CNN used in the proposed framework. The network consists of stacked 2D convolutional layers with batch normalization and Leaky ReLU activation, integrated through residual learning blocks with identity skip connections. Global average pooling and dense layers are employed for feature aggregation and output prediction, enabling stable training and effective deep feature representation.

Residual and Multi-Task Learning

To enhance the stability of training and reuse of features, Resnet architecture are incorporated into the network. The connections enable efficient backpropagation of gradients, eliminate vanishing gradient problems, and enable the learning of deep feature representations without compromising performance.

Furthermore, multi-task learning is utilized to predict jointly:

1. Chromosome abnormality status (normal/ abnormal)
2. Chromosome type and sign

The model leverages shared representations to enhance generalization and robustness. A weighted multi-task loss function is applied to control the contribution of tasks during training, preventing any task from dominating the training process.

Performance Measures

The effectiveness of the suggested chromosomal abnormality detection system is evaluated using multiple quantitative metrics to guarantee a through and clinically meaningful assessment. The total number of correctly identified chromosome images divided by the total sample count determines the accuracy which measures the complete correctness of the classification system. The test evaluates how well the model detects abnormal chromosomes which serves as an essential requirement to reduce clinical diagnosis false-negative results. The system uses specificity to assess its ability to correctly identify normal chromosomes which helps eliminate false-positive results that might cause unnecessary medical alarm.

The precision metric shows how trustworthy abnormality predictions are by showing which percentage of abnormal chromosomes was accurately detected from all samples that were classified as abnormal. To provide a balanced evaluation of precision and recall the F1-score calculates their harmonic mean which produces one performance

metric that helps assess situations with class imbalance. The model evaluation includes Positive Predictive Value (PPV) and Negative Predictive Value (NPV) which measure how likely the model will make correct predictions about abnormal and normal cases.

The Receiver Operating Characteristic (ROC) curve together with its Area Under the Curve (AUC) measurements serve as tools to evaluate Robust Feature learning across various classification threshold levels. Higher AUC values show that normal and abnormal classes have significant class separation. The combination of performance measures shows how well the proposed chromosomal abnormality detection system works for both medical diagnosis and clinical use (Figure 2).

Results

The experimental evaluation of the submitted chromosomal abnormality detection architecture was performed using a five-fold cross-validation strategy to ensure robustness and reliable generalization.

In each fold, training was conducted on 70% of the test dataset which left 15% for validation and 15% for testing purposes. The validation set was used to find optimal model parameters while performance metrics were calculated by normalizing results across five different folds.

The proposed Varifocal-Net-based CNN system uses supervised learning together with mini-batch optimization for its training process. The training set used data augmentation to improve generalization while the validating and testing data sets remained intact. The trained model was assessed using standard performance metrics that are applicable to clinical diagnostic assessments.

The results shown in Table 1 indicate outstanding classification results with high sensitivity and perfect specificity, demonstrating the model's capability to reliably distinguish normal and abnormal chromosomes while avoiding false-positive detections. The uniform elevated values across all metrics confirm the stability and efficacy of the proposed approach.

Table 1. Performance Measures of Varifocal-Net–based CNN.

Metric	Value (%)
Accuracy	99.04
Sensitivity	98.63
Specificity	100
Precision	100
Recall	98.63
F1-score	99.31
PPV	96.92

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Discussion

To further enumerate the efficacy of the proposed framework, experimental results were compared against commonly used chromosomal analysis methods, including conventional CNN, ResNet, and DenseNet models.

The results depict the outstanding performance of proposed Varifocal-Net–based model with reference to conventional and advanced CNN architectures across all evaluated metrics, particularly considering sensitivity and specificity, which are vital for clinical applications.

Figure 3 illustrates the comparative accuracy, sensitivity, and specificity of different chromosomal abnormality detection methods. The proposed Varifocal-Net CNN achieves the maximum performance in terms of accuracy, sensitivity and specificity as depicted in the bar graph shown in Figure 3. In particular, the specificity of the method reaches 100%, indicating zero false-positive detections. The graphical representation demonstrates how learning both global and local features together leads to better diagnostic performance.

The dual-scale feature learning capability of the Varifocal-Net architecture enables the proposed chromosomal abnormality detection framework to achieve its superior experimental results. The proposed method effectively combines global morphological information with localized structural features such as banding patterns and centromere regions in contrast to conventional single-scale feature extraction of CNN model. The system achieves reliable results through its ability to detect minor chromosomal abnormalities which traditional methods find difficult to identify.

Table 2 presents numerical data which demonstrates that the proposed method outperforms conventional CNN and ResNet and DenseNet architectural designs. The model shows better sensitivity which helps to detect abnormal chromosomes and the model achieves perfect specificity which enables it to identify normal samples without any false alerts. The combination of residual learning with multi-task learning creates a training framework that delivers both effective training results and stable performance across different classification tasks through shared representation learning.

The model maintains strong robustness against chromosome orientation changes and staining intensity variations and image noise. The proposed

system functions as a computer-aided decision-support tool for cytogenetic analysis because it provides rapid inference and accurate diagnostic results which help clinicians maintain consistent assessment procedures.

framework maintains strong protection against changes in image content while providing a dependable computer-aided decision-support system that assists cytogenetic research. Future research will explore two areas which include expanding the dataset

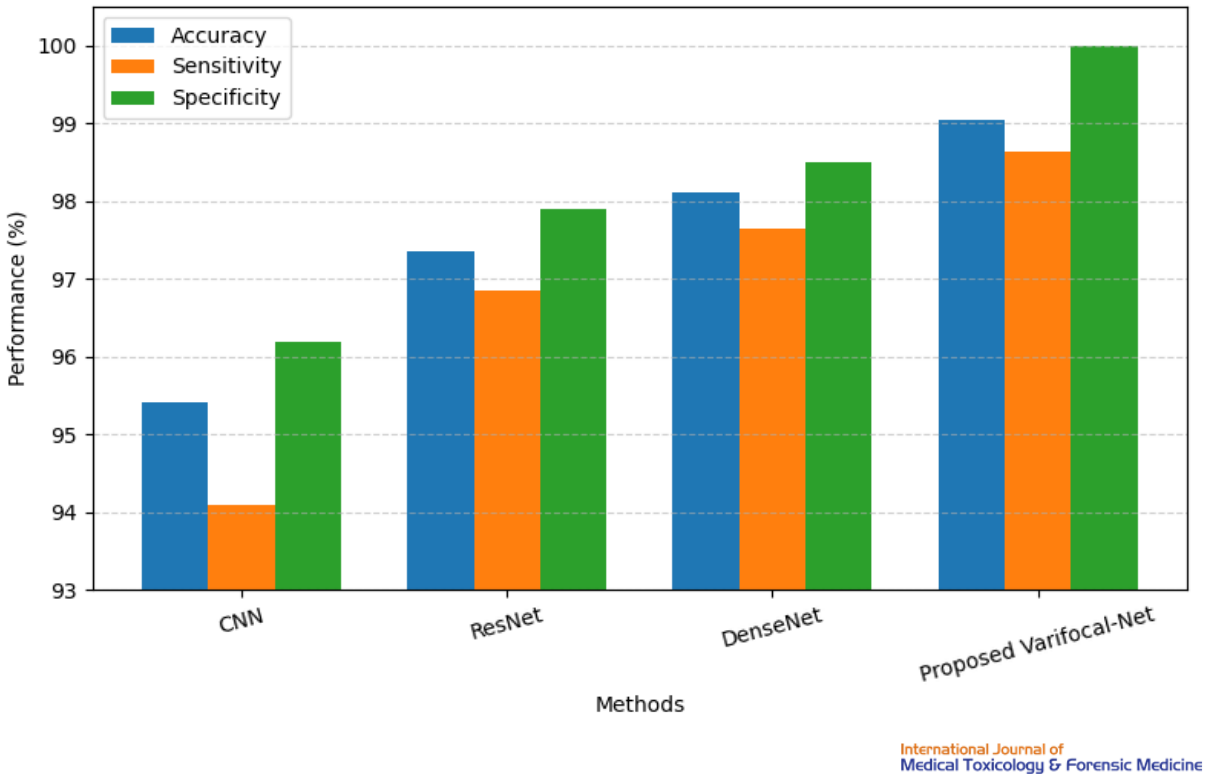

Figure 3. Comparative performance analysis of chromosomal abnormality detection methods.

Table 2. Comparison with Existing Methods.

Method	Accuracy (%)	Sensitivity (%)	Specificity (%)	F1-score (%)
Conventional CNN	95.42	94.10	96.20	94.85
ResNet	97.36	96.85	97.90	97.10
DenseNet	98.12	97.65	98.50	97.88
Proposed Varifocal-Net CNN	**99.04**	**98.63**	**100**	**99.31**

Conclusion

The automated chromosomal abnormality detection system uses a Varifocal-Net–integrated CNN framework to detect chromosomal abnormalities. The proposed method uses global chromosomal morphology together with localized structural feature learning to address the limitations found in traditional karyotype analysis methods and single-scale deep learning systems. The five-fold cross-validation testing showed excellent diagnostic accuracy because the system achieved 99.04% accuracy and 98.63% sensitivity and 100% specificity without producing any false-positive results. The

and developing learning methods based on explainable artificial intelligence and attention mechanisms. The suggested method achieves better performance because its dual-scale Varifocal-Net architecture enables learning of global and local features and it uses residual connections together with multi-task learning to strengthen training processes and improve model performance. The model maintained its ability to function despite different chromosome orientations which included variations in staining intensity and image noise thus demonstrating its suitability for use in practical cytogenetic processes.

Acknowledgment

The authors gratefully acknowledge the technical support provided by the NPR College of Engineering and Technology.

Funding

No funding received.

Conflicts of Interest

The authors report there are no competing interests to declare.

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