<Conference abbreviation>
<Conference Series name> <Volume number and Year> <DOI Number>

Advancing Cytogenetic Diagnostics with PO-CNN: Robust Population-Based Optimization for Chromosome Classification

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Abstract.

In this study, we propose a novel Population- based Optimized Convolutional Neural Network (PO-CNN) for automated chromosome classification. The framework integrates a population-based optimization strategy to efficiently tune CNN hyperparameters, addressing the limitations of conventional optimization methods such as Genetic Algorithm (GA), Whale Optimization Algorithm (WOA), and Particle Swarm Optimization (PSO). Experimental evaluation across multiple metrics demonstrates that PO-CNN achieves superior performance, with an accuracy of 96.33%, sensitivity of 95.97%, specificity of 96.64%, PPV of 96.48%, NPV of 96.19%, and F-score of 96.23%. Comparative analyses, including heatmaps, time-series plots, and hierarchical clustering, confirm the robustness and balanced reli- ability of PO-CNN against baseline models.

Keywords. Chromosome classification; Karyotype analysis; Convolutional Neural Networks (CNN); Metaheuristic optimization; Medical image analysis

1. INTRODUCTION

The leading carriers of genetic information are chromosomes, which are the basis of cytogenetic investigation. One of the foundations of medical genetics is karyotype analysis, which implies the systematic study of the number and structure of chromosomes. It is essential for the diagnosis of congenital disorders, hepatological malignancies, repro- ductile abnormalities, and prenatal genetic screening. The fact that patient numbers are increasing in cytogenetics labs serves to exacerbate the need for scalable solutions. Thus, the concept of automating chromosome classification using intelligent computational tools has emerged as a key field of study to minimize human influence in the process and enhance accuracy and scalability. Deep learning has become a disruptive technology in the healthcare sector,

especially in medical image analysis, in the last few years. CNNs have been incredibly successful in different areas of diagnosis, including cancer detection, radiology, pathology, and genetic imaging [1].

Their hierarchical feature representations, due to their capability to learn them, make them particularly applicable in chromosome classification, where minute differences in size, shape, and banding need to be detected with accuracy. Furthermore, advancements in metaheuristic optimization have enabled the easy optimization of CNN architectures and hyper- parameters, thereby guaranteeing greater accuracy, sensitivity, and specificity in the results. Deep learning in cytogenetics promises to enable automated systems to analyse karyotypes that are not only fast and accurate but also scalable to clinical scales [2]. Although multiple CNN-based frameworks have been proposed for chromosome classification, several gaps re- main. First, many existing models rely heavily on handcrafted preprocessing and large annotated datasets [3], which are often scarce in clinical practice. Second, optimization strategies such as Genetic Algorithms (GA), Whale Optimization Algorithm (WOA), and Particle Swarm Optimization (PSO) have been integrated with CNNs to enhance classification performance [4]. However, they still struggle to achieve an optimal trade-off between accuracy, sensitivity, and specificity. As reflected in our comparative experiments, GA-CNN and WOA-CNN achieved relatively modest accuracies of 91.26% and 91.89%, respectively. PSO-CNN achieved an improved performance of 92.74%, while GWO-CNN performed even better at 93.77%. However, all of these models fell short in terms of sensitivity and F-score, rendering them less reliable for clinical decision-making. These limitations underline the necessity for more advanced optimization frameworks that can consistently improve CNN-based chromosome classification while maintaining generalizability across datasets and imaging conditions [5].

2. LITERATURE REVIEW

Chromosome recognition plays a pivotal role in diagnosing haematological malignancies and genetic diseases, yet the manual karyotyping process remains repetitive and time-consuming. To address this, several studies have proposed artificial intelligence (AI) and deep learning-based frameworks aimed at improving efficiency, accuracy, and clinical applicability. One significant advancement is KaryoNet, an end-to-end differentiable combinatorial optimization method that intro- duces the Masked Feature Interaction Module (MFIM) and Deep Assignment Module (DAM). This system effectively captures long-range chromosome interactions and predicts both type and polarity. It achieved state-of-the-art accuracy rates of 98.41% on R-band and 99.58% on G-band chromosomes, demonstrating strong applicability for diagnosing numerical abnormalities [6].

Another approach utilizes a deep convolutional neural net- work (DCNN) optimized through Hybrid Moth-Flame Optimization with Hill-Climbing (HMFOHC). To address dataset limitations, generative adversarial networks (GANs) are employed to generate synthetic training samples. This method demonstrated high performance in distinguishing five common chromosomal abnormalities (e.g., Trisomy 13, Trisomy 21, and Monosomy X), achieving an accuracy of 98.65% with reduced inference time [7]. The Model Architecture Search System (MASS) automates the selection of optimal CNN-based architectures for chromo- some classification tasks by leveraging Bayesian optimization through the Tree-structured Parzen Estimator (TPE). Experiments on two datasets confirmed its superior adaptability and performance without the need for manual hyperparameter tuning [8].

3. DATASET DESCRIPTION AND PREPROCESSING

The data were produced by using a camera mounted on top of a microscope to capture images of the chromosomes. The lens and angle were adjusted under the careful supervision of professional cytogeneticists to achieve the best staining patterns. The images obtained were subsequently directed to a computer system, where additional analysis and preprocessing were performed. This would ensure that the banding information, as well as the general morphology of the chromosomes, was maintained in the digital data. This paper employed the in-situ harvest technique and the trypsin /Wright stain technique to prepare metaphase-stage chromosome spreads for G-banding.

4. OPTIMIZED DEEP LEARNING RESULTS

This performance gap of nearly five percentage points over GA-CNN reflects the robustness of the proposed optimization strategy in consistently identifying both normal and abnormal chromosomes. Sensitivity, which measures the ability of a model to correctly detect abnormal chromosomes, was also highest for PO- CNN (95.97%). This means that the model is highly effective in minimizing false negatives, an essential requirement for medical diagnosis where missing an abnormality could delay treatment or misinform clinical decisions. By comparison, the sensitivity of GA-CNN and WOA-CNN plateaued at 91.84%, showing that these models are more prone to over- look abnormalities. Similarly, PSO-CNN, although slightly better, achieved only 91.84%, while GWO-CNN performed reasonably well at 93.02% but still fell short of PO-CNN. This finding is particularly important, as sensitivity is often prioritized in screening systems where early detection of genetic disorders such as Trisomy 21 or Turner syndrome is critical.

TABLE I :Performance comparison CNN models for chromosome classification.

Models	Accuracy	Sensitivity (TRP)	Specificity (TNP)	PPV	NPV	F-score
PO-CNN	0.9633	0.9597	0.9664	0.9648	0.9619	0.9623
GWO-CNN	0.9377	0.9302	0.9443	0.9398	0.9358	0.9350
PSO-CNN	0.9274	0.9184	0.9373	0.9398	0.9145	0.9290
WOA-CNN	0.9189	0.9184	0.9194	0.9231	0.9145	0.9207
GA-CNN	0.9126	0.9184	0.9050	0.9231	0.8989	0.9207

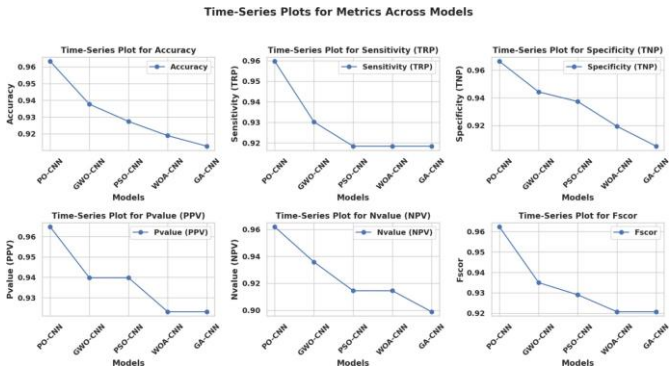

Fig. 1. Time-series plots of performance metrics across optimization-based CNN models.

CNN and PSO-CNN achieved only 93.50% and 92.90%, respectively. Both WOA-CNN and GA-CNN stagnated around 92.07%. A high F-score confirms that PO-CNN avoids the common trade-off between sensitivity and precision, instead delivering equally strong performance across both.

To further illustrate the comparative performance trends of the evaluated models, Figure 1 presents time-series style plots for each of the six-performance metrics (Accuracy,

Sensitivity, Specificity, PPV, NPV, and F-score). Unlike the tabular presentation, these plots clearly show the gradual decline in performance from PO-CNN to GA-CNN across all indicators. The sharp separation between PO-CNN and the baseline models highlights its superiority, while the relatively clustered performance of PSO-CNN, WOA-CNN, and GA-CNN indi- cates their limited ability to achieve consistent improvements. The dendrogram at the top of the figure highlights how metrics group together, with Accu- racy, PPV, and F-score forming a tightly correlated cluster, while Sensitivity and Specificity exhibit relatively distinct behaviour.

5. CONCLUSION

This study presented a population-based optimized convolutional neural network (PO-CNN) for automated chromo- some karyotype classification. Across six clinically meaning- ful metrics—Accuracy, Sensitivity (TRP), Specificity (TNP), PPV, NPV, and F-score—PO-CNN consistently outperformed four strong metaheuristic baselines (GWO-CNN, PSO-CNN, WOA-CNN, GA-CNN). In particular, PO-CNN achieved an Accuracy of 0.9633, Sensitivity of 0.9597, Specificity of 0.9664, PPV of 0.9648, NPV of 0.9619, and F-score of 0.9623, establishing a balanced improvement that reduces both false negatives and false positives.

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