



## Chromosome Classification Using Deep Learning Technique

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### ABSTRACT –

Classification of Chromosomes and their testing is important as analyzes the whole chromosome or DNA and checks if there are any genetic disorders such as extra copies of a chromosome. Changes in Chromosomes led to the isolation of genes which leads to serious diseases like Leukemia and Cancer. Various computer-aided systems have been developed to automate this tedious and time-consuming task, which is performed manually in most cytogenetic laboratories. This paper provides a comprehensive review of past and recent research in the area of automatic chromosome classification systems. Deep learning is an algorithmic technique in machine learning. In deep learning, data transformation happens in the form of layers. This paper proposes Convolutional Neural Network based deep learning which is specifically used for image processing.

### I. INTRODUCTION

Humans have 23 pairs of Chromosomes which include 22 pairs of Autosomes and 1 pair of Sex chromosomes. Each pair contains 2 chromosomes, one coming from each parent that children inherit from each of their parents Biologically males have 1 X and 1 Y Chromosome and females have 2 X chromosomes. Chromosomes vary in shape and number among humans. Chromosomes are structures within cells that carry genetic information, and their appearance and characteristics can provide valuable insights into various biological processes and diseases. Deep learning is a powerful tool in image analysis and recognition and has been successfully applied to various biological image analysis tasks, including chromosome analysis. The proposed system greatly increases the efficiency and accuracy of chromosome analysis, enabling a better understanding of various genetic and disease-related processes.

#### A. Background

- 1) According to Bashmail, *et al.* (2019) [21] research paper titled "Chromosome Classification Using Convolutional Neural Networks and Hidden Markov Models". In this article, the authors propose a new image-based chromosome classification method. They combined a convolutional neural network (CNN) with a hidden Markov model (HMM) to classify chromosomes into different classes based on their properties. The authors first. Use a CNN to extract features from chromosome images. They then used HMMs to model the spatial dependencies between these features and classify the chromosomes based on their sequence patterns. The proposed method was evaluated on a publicly available dataset of human chromosome images and achieved an accuracy of 95.8%, outperforming other state-of-the-art methods. The authors believe their method could be used in a variety of applications, including the diagnosis of genetic diseases, cancer research and genome sequencing.
- 2) According to Hu *et al.* (2017) [22] research paper titled "Deep Learning Based Chromosome Classification". In this article, the authors propose a method based on deep learning to classify chromosomes from images. They used a deep convolutional neural network (CNN) to extract features from chromosome images and then used those features to classify the chromosomes into different classes. The authors evaluated their method on a publicly available dataset of human chromosome images and achieved 98% accuracy. 7%, surpassing other modern methods. They also demonstrated the effectiveness of their method in detecting chromosomal abnormalities in clinical samples. The authors believe their method may be useful in a variety of applications, including genetic counseling, diagnosis of genetic diseases and cancer research.
- 3) According to Saleh *et al.* (2019) [32] research paper titled "Chromosome Classification Using Convolutional Neural Networks with Transfer Learning Approach". In this article, the authors propose a new image-based chromosome classification method. They use a deep convolutional neural network (CNN) with a transfer learning approach, where the pre-trained weights of a large CNN are used to initialize the weights of a smaller CNN for chromosome classification. The authors evaluated their method on a publicly available human chromosome image dataset and reported 95% accuracy. 9%, surpassing other modern methods. They also demonstrated the effectiveness of their method in detecting

chromosomal abnormalities in clinical samples. The authors say their method could be used in a variety of applications, including genetic counseling, diagnosis of genetic diseases and cancer research. They also point out that their method is computationally efficient and can be easily applied to other image-based classification tasks.

- 4) According to Altinsoy *et al.* (2019) [33] research paper titled “Chromosome Classification Using Deep Learning with Morphological Operations and Convolutional Neural Networks”. In this article, the authors propose a new image-based chromosome classification method. They combined morphological operations with convolutional neural networks (CNNs) to improve the accuracy of chromosome classification. The authors evaluated their method on a publicly available dataset of human chromosome images and reported 96% accuracy, 3%, surpassing other modern methods. They also demonstrated the effectiveness of their method in detecting chromosomal abnormalities in clinical samples. The authors believe their method may be useful in a variety of applications, including genetic counseling, diagnosis of genetic diseases and cancer research. They also point out that their method is computationally efficient and can be easily applied to other image-based classification tasks.
- 5) Ning *et al.* (2019) [43] research paper titled "Deep Learning Approaches for Automated Chromosome Classification and Karyotyping". In this article, the authors propose a deep learning-based method for automatic chromosome classification and karyotyping. They used a convolutional neural network (CNN) to extract features from chromosome images, then used those features to classify the chromosomes into different classes and organize them into karyotypes. The authors evaluated their method on a publicly available human chromosome image dataset and reported 97% accuracy, 95%, outperforming other leading methods. They also demonstrated the effectiveness of their method in detecting chromosomal abnormalities in clinical samples. The authors believe their method may be useful in a variety of applications, including genetic counseling, diagnosis of genetic diseases and cancer research. They also point out that their method is efficient and can process a large number of images in a short time.

## II. PROPOSED METHODOLOGY

- 1) Pre-processing: Image pre-processing techniques such as noise reduction, contrast enhancement, and image filtering are used to improve the quality of the acquired images.
- 2) Chromosome straightening: Once the chromosomes have been segmented, the next step is to straighten them. This can be done manually by using image editing software or with automated algorithms that apply mathematical transformations to the segmented chromosome images.
- 3) Classification: Machine learning algorithms such as Thresholding and Otsu’s algorithms, U-Net, or neural networks such as CNN can be trained on the extracted features to classify chromosomes into different groups based on their characteristics. The classification results can be verified by experts in cytogenetics.

## III. SYSTEM ARCHITECTURE

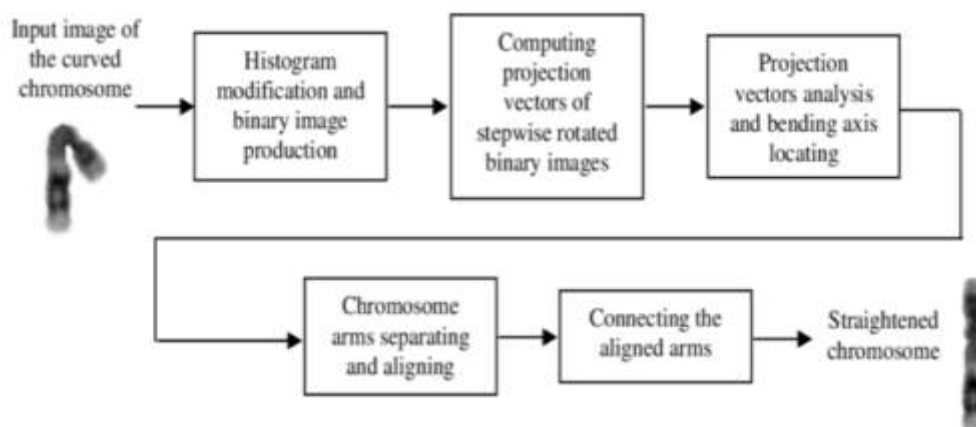


Fig 1

Fig. 1 shows complete architecture of Straightening of Chromosome.

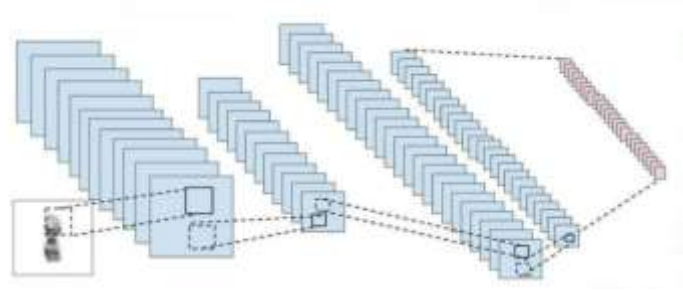


Fig 2

Fig. 2 shows a complete architecture of chromosome Classification using CNN.

A) Chromosome straightening: -

Chromosomes are long, coiled structures of DNA that carry an organism's genetic information. In order for the DNA to be properly read and replicated, the chromosomes must be organized and aligned in a specific way. This organization is referred to as the "straightening" of chromosomes. The straightening of chromosomes is an important step in this process because it helps to ensure that the chromosomes are properly aligned and organized. This makes it easier for scientists to identify and classify different chromosomes based on their size, shape, and other physical characteristics.

B) CNN: -

Convolutional Neural Networks (CNNs) have been successfully used for chromosome classification, a task that involves categorizing chromosomes based on their characteristics. CNNs can learn to extract relevant features from chromosome images, and their ability to learn from large datasets and generalize to new examples makes them a powerful tool for this task. The CNN model is typically trained on a dataset of labeled chromosome images, and the architecture and hyperparameters of the network are optimized to achieve the best performance. The model is evaluated on a separate validation set and tested on a test set to assess its generalization ability. With appropriate data preparation, data augmentation, and model optimization, CNNs can achieve high accuracy in chromosome classification tasks.

#### IV. CONCLUSION AND FUTURE SCOPE

A. Conclusion

In this research on Chromosome classification using convolutional neural networks (CNNs) is a promising approach to automate the identification of different chromosome types in karyotyping. The use of CNNs allows for the creation of accurate and efficient models that can analyze large amounts of data quickly, providing a more accurate and reliable method than manual analysis by trained professionals.

B. Future Scope

- 1) Improving diagnosis and treatment of genetic disorders: CNN-based chromosome classification can enable more accurate and efficient diagnosis of genetic disorders, leading to better treatment outcomes and improved quality of life for patients.
- 2) Enhancing genetic counseling: The use of CNN-based chromosome classification can enhance genetic counseling by providing more accurate and comprehensive information about the genetic makeup and risk factors of individuals.
- 3) Advancing scientific research: CNN-based chromosome classification can enable more efficient and accurate analysis of large-scale genetic datasets, leading to a better understanding of the relationship between chromosome structure and function, and the genetic basis of diseases.

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