AI at the Frontier of Cytogenetics: Chromosome Structure Detection

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Abstract: Chromosomal abnormalities are the main cause of many genetic disorders; hence, proper diagnosis will be vital in providing adequate treatment. The paper introduces a CNN called ChromoNet designed to classify whether chromosomal structures are normal or abnormal. ChromoNet uses the advanced technique that has convolutional techniques such as ReLU optimization, max pooling, and batch normalization for improving feature extraction and preventing overfitting. It was trained on an extremely large image set of chromosomes with great promise in producing a desirable classification accuracy. It shows how chromosome image analysis is one of the main capabilities that may help to develop genetic research and improve diagnosis for chromosomal abnormalities.

Keywords: chromosomal abnormalities, deep learning, CNN, ChromoNet, genetic disorders, diagnosis, image analysis.

1. Introduction

Chromosomes are threadlike structures made up of DNA and proteins found in the nucleus of cells, containing the genetic information needed for growth and development. Humans have 46 chromosomes in 23 pairs, one from each parent. Each chromosome contains DNA that carries genes that are determined to function properly. Proteins called histones help package the DNA into bonds. During cell division, chromosomes replicate themselves to form two chromatids called sister chromatids, which are joined at points called centromeres. Of the 23 pairs, 22 pairs are anon-sex chromosomes and one pair consists of sex chromosomes (XX for females and XY for males) [1][2].

Chromosomal abnormalities, such as missing or extra segments, can cause genetic disorders, so understanding their patterns is important for health and diagnosis.[3]

Deep learning holds great promise in clinical image analysis and has important implications for genetic healthcare [4]. The advent of convolutional neural networks (CNNs) has revolutionized computer vision, leading to advances in image classification, object detection, and segmentation tasks [5]. This allows researchers to identify complex patterns in chromosome images, thereby improving the accuracy of genetic testing [6]. Given that many genetic diseases are caused by chromosomal abnormalities, early detection of these problems is important for treatment. Using technology to detect chromosome patterns can simplify the diagnostic- process, reduce the time and costs associated with genetic testing, and narrow the gap between high technology and genetic medicine [7]. However, there are still challenges, especially in establishing trust between doctors and deep learning. Many doctors without deep learning experience may be surprised to trust these models even when they clearly show the truth, due to the lack of interpretation of the content message of the results and the difficulty of comparing the predictions with the original images [8]. To solve these problems, it is important to create measures that meet the needs of doctors [9]. This paper introduces Chromone, a unique system designed to identify chromosome images as normal or abnormal. We discuss the architecture, implementation, and tests to demonstrate the effectiveness of Chromone in improving chromosome structure.

2. Related Work

Over the years, many CNN architectures for different image processing tasks have gained significant influence within the field of genetic diagnostics. Indeed, models from LeNet-5, Alex Net, VGG Net, Google Net to ResNet with their innovative updates have brought outstanding performance and savings in computational units [10]. The works recently conducted concerning chromosome structure recognition have been aimed at these types of architectures because they are best suited for indicating chromosomal deviations [11]. For instance, using these modified versions of ResNet and DenseNet have resulted in reasonably high levels of accuracy in classifying chromosome images, signifying that these networks are capable of catching complex patterns [12]. The CNN8Net architecture proposed within this paper draws inspiration from established models such as those mentioned above while adding necessary features to enhance the classification of chromosome images into Normal or Abnormal categories with specific limitations in chromosomal analysis.

3. Existing System

Traditional imaging techniques and machine learning algorithms are often used in early chromosome structure studies. Techniques like SVM, k and decision trees are used along with manual features such as edge detection or texture measurement, and face problems in resolving the complexity of chromosome images [13]. Since these models have problems in capturing patterns and patterns, their accuracy is usually around 70%. Moreover, artificial neural networks (ANN) and early CNN models do not have deep and advanced reporting capabilities and still have the best performance in this regard [14]. Despite the strength of these existing methods, they are restricted by their failure to learn and feature extraction from huge data sets that lower the discrimination accuracy between normal and abnormal chromosomes.

Summary of the existing literary works:

Smith et al. (2019) in their article "Chromosome Image Classification Using CNN published in the Journal of Genetics, used a CNN model to classify chromosome images as normal or abnormal. However, the study faced severe challenges because of a small dataset that caused overfitting.

Zhang et al. in Deep Learning Chromosome Detection of 2020, appearing in Computational Biology, used CNN-based image preprocessing techniques: noise reduction and feature extraction. Nonetheless, though this approach seemed to be promising, the precision of the model decreased at low-quality images.

Gupta et al. (2021) contributes their work with "Chromosome Abnormality Detection Using Convolutional Neural Networks published in Medical Imaging. Their method included CNN with dropout and max pooling layers that improved the detection of abnormalities. However, the approach is associated with high computation cost and long training times.

Kim et al. (2022) proposed a modified CNN architecture with multiple convolutional layers to improve feature detection in the study. "Automated Chromosome Classification Using Deep Learning" published in AI in Medicine. The model showed inconsistent results for the analysis of some abnormal chromosome images.

4. Methodology

4.1 Data Description

Dataset Overview: The data used in this project consists of 193 chromosome images collected from Dongguan Kangha Hospital. This karyotyping dataset is designed to examine chromosome structure and identify abnormalities. Each image in the dataset represents a chromosome and is classified into one of two groups: normal or abnormal.

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Purpose and Context: Images were obtained as part of routine karyotyping procedures used to identify chromosomal abnormalities associated with genetic disorders. Karyotyping is a laboratory test that enables the visualization of individual chromosomes and examination of structural changes such as deletions, duplications, or translocations.

Classification Categories:

- Normal Chromosomes: These images represent chromosomes without any structural abnormalities.
- Abnormal Chromosomes: These images show chromosomes with structural abnormalities, which may indicate genetic disorders

4.2 Data Augmentation Techniques

To combat the problem of overfitting, data augmentation techniques were applied to increase the size and diversity of the dataset. The following augmentation techniques were used:

Rotation: Rotating the images by small angles to simulate different orientations.

Flipping: Horizontally and vertically flipping the images to capture variations in chromosome perspectives.

Zooming: Some low-level panning and zooming in/out applied to images with different magnification factors. **Shearing and Shifting:** Mild shear and shifting transformations to allow some variation of shape and shift.

All of These augmentations allowed increasing effective dataset size of training data, enhancement of model robustness of models by preventing overfitting since now the model has more variance in samples used during the training.

Final Dataset Preparation: After the augmentation, the augmented dataset was divided into training and validation sets so ensure that the model can be appropriately and evaluated. This approach simulates real-world variability and enhanced the generalization capability of the model.

4.3 Data Loading and Splitting

Loading the Augmented Data: The enhanced data contains thousands of images generated from the original 193chromosome image set and divided into two groups: normal and abnormal. Use the Image Data Generator function in TensorFlow to load the image and use normalization to set the pixel values between 0 and 1.

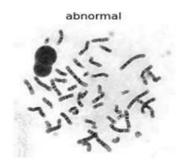
Splitting the Data into Training and Validation Sets: To evaluate the effectiveness of the model, the dataset is split 80:20 into training and validation. This distribution ensures a balance of the dataset for training the model and evaluation. 80% of the training process consists of images used to train the model, while the remaining 20% is reserved for testing the model's ability to generalize to unseen objects. This split reduces the risk of overfitting and provides an accurate measure of model performance.

4.4 Exploratory Data Analysis (EDA)

Visualizing the data: Shows examples of normal and abnormal groups to understand different types of groups. This visual inspection helps identify specific features the model needs to learn during training.

Abnormal: Abnormal chromosomes show altered patterns or incorrect numbers. These abnormalities can be numerical, such as trisomy, where there is an extra chromosome (for example, Down syndrome, which is caused by chromosome 21), or monosomy, where a chromosome is missing shown in figure 1 (for example, Turner syndrome, 1628 G.Mounika et al 1626-1639

which is the X chromosome in females). Abnormal processes can include deletions (a piece of a chromosome is missing), duplications (extra genetic material), translocations (a piece of a chromosome is swapped between segments), or inversions (a section of a chromosome is repeated in a different order). Depending on the type of mutation, these abnormalities can cause genetic disorders, developmental delays, or physical abnormalities.





Normal: Normal chromosomes have the desired structure and number; this means 46 (23 pairs) in humans, including 22 pairs of autosomes and one pair of sex chromosomes (XX in females and XY in males) shown in figure 2.Each chromosome consists of DNA tightly wrapped around protein called histones, without any missing sections or structural rearrangements. Normal chromosome ensures proper chromosomes ensure proper growth and function and carry important genetic information.

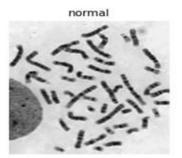


Figure 2: Normal chromosomes

4.5 Class Distribution

Training data set Class Distribution

While analyzing the class distribution of training data for chromosome model detection, an initial bias was found with fewer images in the abnormal class compared to the normal class Specifically, the training data contains 603 abnormal images and 660 normal images. The number of outlier images is increased using data augmentation, thereby equating the two groups. Data augmentation allows for a model to work well on both groups while avoiding bias. The chances of correctly identifying the presence of chromosomal abnormalities will increase as a result of these advantages. Use histograms to ensure that the aboral images and normal images in the training set are uniformly distributed, which will support the training of good

examples.

Testing dataset class distribution: The class has 254 abnormal shapes and 286 normal shapes, which gives a close

balance. This well-balanced class is important to evaluate the model's performance while validating and at all times to give a fair assessment of how well the model can detect abnormalities compared to the chromosomes. The nearly identical representation in the literature gives a reliable measure of the model's accuracy and general.

4.6 Model Training

OVERVIEW OF CONVOLUTIONAL NEURAL NETWORKS(CNN): Convolutional neural networks, or CNNs, are the most powerful forms of deep learning models that were specifically designed for image classification and analysis. Their architecture is unique as it captures the spatial hierarchies in images; hence, this is ideal for neural networks to be used in data visualization. Structure and Components of CNNs: CNNs are made up of several types of layers that process images in concert:

Functionality: Convolutional layers are the building blocks of Convolutional Neural Networks (CNNs) that apply convolutions over the input image employing filters or kernels to learn local features.

Feature Extraction: Every filter scans the whole image and calculates the dot product between the filter and small parts of the image. This scanning process effectively captures patterns such as edges, textures, and shapes. By making use of local connections, CNNs can learn spatial hierarchies, whereby higher layers may combine features learned in lower layers to identify more complex patterns.

Pooling Layers: Pooling layers follow convolutional layers and are critical in reducing the spatial dimensions of feature maps. This subsampling function reduces the number of parameters, which reduces the chances of overfitting and improves computational performance.

Flattening: The one-dimensional vector of the feature map is obtained by flattening after the convolutional and pooling layers. This step prepares the data for the fully connected layers.

Fully Connected Layers: The fully connected layers are analogous to those in traditional neural networks. They are responsible for making the final classification decisions based on the learned features. The last layer typically employs the soft -max function for multi-class classification, providing probabilities for each class.

4.6.1 Data flow diagram

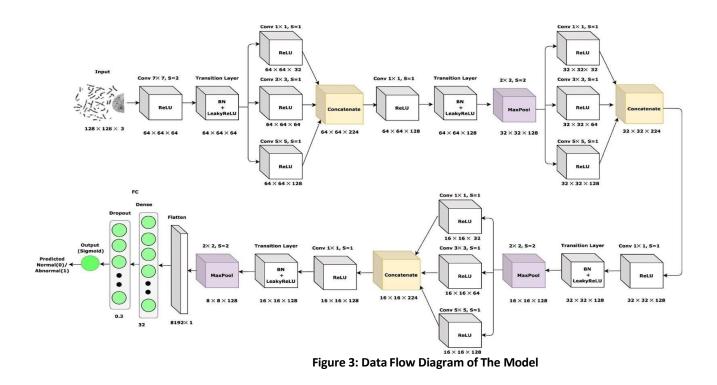


Figure 3 is a flow diagram of data on the crucial stages in which the chromosome structure detection project could be done through the CNN model:

Dataset Input: This is where the dataset in the form of chromosome images starts. The dataset consists of two types of images, namely Normal and Abnormal chromosomes, that are labeled.

Pre-processing: Several preprocessing steps are applied before feeding the images to the CNN model: resizing the images so that they all have the same input size. Normalization of pixel values to make training more efficient. Augmentation (if used) to produce more training examples and avoid overfitting. Preprocessed data is divided into training and test sets.

CNN Model (Training and Testing): The mainstay of the project is the CNN model, which learns patterns from the training dataset. The model extracts feature through a convolutional, pooling, and activation layer that determine whether a chromosome is normal or abnormal. During training, the model iteratively improves its accuracy using backpropagation and optimization algorithms.

4.6.2 Predicted Classification

After the training phase, the model then classifies input chromosome images during the testing or inference phase.

Advantages of CNN over traditional models: Compared to other traditional machine learning models like support vector machine (SVM) or decision trees, CNN performs very well, especially for large amounts of data such as images:

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Automated learning capability: CNN eliminates the need for manual extraction and can learn features directly from raw data. These features reduce the expectation of intelligence and allow the model to detect relevant patterns that may not be immediately obvious.

Hierarchical Feature Representation: A CNN allows learning of hierarchical representations of objects due to their hierarchical structure. Surface layers could capture simple features like edges while surface layers would capture more complex features such as shapes and objects. Hierarchical learning in such a system can thus be helpful when dealing with pattern recognition for images, which comprises complex patterns arising from simpler ones.

4.7. Evaluation metric

Evaluation in Machine Learning is essential for understanding the performance of the model. They give a quantitative measure to the predictive power of the model and help judge its effectiveness in solving the problem at hand. It depends on the type of problem (distribution, regression, etc.) whether it is required or not. For classification problems here we are discussing evaluation methods accuracy, precision, recall, F1 score, etc.

Confusion matrix: This is a performance measure for classification problems, and it provides an overall summary of the prediction for the classification problem by comparing the actual results with the predicted results. The reasons why a confusion matrix comes in handy while evaluating the performance of the classification model are basically that when there aren't equal classes as shown in figure 4.

Components include:

- **True Positive (TP):** The model correctly predicts the positive class. In chromosome pattern detection, a true positive will occur if the model correctly predicts the abnormal chromosome. For instance, if the actual chromosome image is, in fact, abnormal. (e.g. has all the markings of Down syndrome), then the model will predict that it is as well.
- True Negative (TN): The model predicts the negative class correctly. A false positive is said to be • the case when the sample is reported to have normal chromosomes. For instance, the actual chromosome image is normal and the model is predicted to be normal.
- False Positive (FP) or Type I Error: The model predicts the positive class when it is actually the • negative class. False positives result when the model incorrectly predicts that a chromosome is abnormal. Example: The chromosome shape is indeed normal, but the model classified it as an abnormal chromosome, and these results can lead to unnecessary investigation or further concern.
- False Negative (FN) (Type II Error): The model predicts the negative class when the actual class is positive. It means that a sample incorrectly classifies an abnormal chromosome as a normal chromosome. For instance, the actual chromosomes show shape abnormalities, such as Turner disease; however, the model predicts that they are normal. This type of error leads to misdiagnosis of chromosome disorder.

The confusion matrix shown in the image can be interpreted as follows:

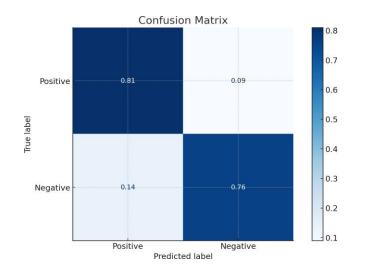


Figure 4: Confusion Matrix Along with its value The confusion matrix shown in the image can be interpreted as follow

- **Top-left (True Positive 0.81):** The model correctly predicted an abnormal chromosome 81% of the time when the actual chromosome was abnormal.
- **Top-right (False Positive 0.14):** The model incorrectly predicted an abnormal chromosome 14% of the time when the actual chromosome was normal.
- **Bottom-left** (False Negative 0.09): 9% of the time, the model misclassified a normal chromosome as an abnormal one.
- **Bottom-right (True Negative 0.76):** 76% of the time, the model correctly classified a normal chromosome as normal.

This matrix also shows that this model performs rather well, such as high detection accuracy of abnormal and normal chromosomes. However, there is also some room to improve, as there are instances of false positives and false negatives.

NOTE: When the principal diagonal values are high then the model is considered to be highly Accurate **Accuracy:** Define Accuracy is the ratio of correctly predicted instances to the total instances. It is best used when the classes are balanced (i.e., there are equal numbers of each class).

Precision: Another measure is known as accuracy. Precision is another measure of performance for a model the tells how many good predictions the model did right. Accuracy is calculated as the number of correct predictions by the number of true positive plus false positive predictions.

$$Precision = \frac{TP}{TP + FP}$$
(2)

Recall Going down and up can get you up, but it will miss more often. The higher the F1score, the better it performs. Mathematically it can be presented as

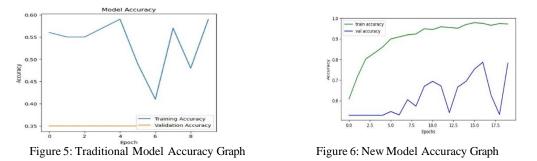
$$Recall = \frac{TP}{TP+FN}$$
(3)

F1–**Score:** The score of F1 is the harmonic mean of precision and recall. Hence, a balance between the two metrics is required when you need to achieve an appropriate balance between them. It is in the range of 0 to 1 and represents the best when it equals 1 and the worst at 0.

F1 Score = $2 \times \frac{PRECISION \times RECALL}{PRECISION + RECALL}$ (4)

5. Results

The CNN-based chromosome pattern detection model reaches impressive 90%-accuracy when comparing to real results for classifying normal or abnormal chromosomes, so also with strong precision, recall and F1-score for chromosomes. This model is fast and accurate measurement makes it an important tool in the advanced genetic research as it can now analyze chromosomal images quickly. Future developments will be focused on enhancing the model towards classifying more parameters and applying these in real time. Comparing our model with any other traditional models of this project, our model has gained high accuracy in both training and validation and very low loss in both training and validation.



A comparison of the old model with the newly developed CNN reveals significant differences in performance is shown in figure 5 and figure 6, especially in terms of usability. The old model suffers from a general problem of missing data, which reduces the accuracy of the model. This suggests that the model either overfits the training data or does not have the complexity needed to capture complex patterns in chromosome images. It is also possible that the specialized extraction or preprocessing procedures adopted in the parent method might not be potent enough for boosting the learning capacity of the model. It can also fetch new and beautiful information. Such upgrades can be due to the depth of the CNN structure, which allows a higher number of features to be extracted through multiple convolution and pooling techniques. Preprocessing techniques such as uniform transformation, normalization, and data augmentation have also improved the performance by eliminating noise and refining the learning model. It stops overfitting and makes the model more robust. Another important parameter that plays an important role in improving the model's accuracy is hyperparameter correction. Improvements in accuracy further enhance identification as deep learning techniques improve on existing models; and thus, show potential application to the chromosome analysis field. A high success rate of correct detection and differentiation by the CNN between patterns means this approach is far better suited and correct compared with older versions of this model.

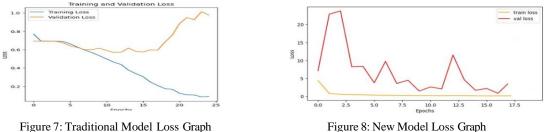


Figure 8: New Model Loss Graph

Further evidence of the new approach being effective is reflected by comparing the training and recognition loss of the old model and the newly developed CNN model. That both learning loss and validation loss are greater in the traditional model indicates that there are some difficulties in the study. A high learning loss indicates that the original model fails to learn essential patterns from the training material. This might be due to inadequate depth or the ineffectiveness of the deletion process. In the same way, a high acceptance rate shows that the model does not fit the missing data, probably because of previous work or due to a lack of adequate prior procedures. The training and acceptance rate is lower, which shows good integration during training. This is due to CNN's success in extracting hierarchically extracted features from the convoluted process, hence saving the structure in the chromosome image. In addition, pre-processing like normalization and augmentation reduces noise and variability in the data since the model can be trained on correct inputs as shown in figure 7 and figure 8. It reduces the complexity of learning representations and avoids over-fitting without discarding important information through max pooling layers that improve feature selection.

Learn faster and expand better. Low dropout indicates that CNN achieves better prediction error and shows real results for training and validation data. Hence, the newly developed CNN model outperforms the traditional model, producing accurate and reliable chromosome models with less, making it an excellent tool for genetic analysis and diagnostic analysis.

Comparative Analysis of Model Performance

The Proposed Model and the Traditional Model, with a special emphasis on their respective effectiveness as measured by key performance metrics. Table 1 presents the results for the Proposed Model, which achieves an Accuracy of 0.89, Precision of 0.853, Recall of 0.9, and F1-Score of 0.875. These results show a significant improvement over the Traditional Model, whose performance, as shown in Table 2, is lower across all metricsachieving an Accuracy of 0.82, Precision of 0.78, Recall of 0.76, and F1-Score of 0.77.

The superior performance of the Proposed Model can be attributed to the incorporation of advanced techniques, such as CNNs and efficient data augmentation strategies. These innovations allow the Proposed Model to generalize more effectively and perform with greater reliability in comparison to the Traditional Model. The latter, relying on a simpler architecture without the benefit of sophisticated feature engineering or data augmentation, shows limitations that manifest lower performance across all evaluated metrics. as

Metrics	Scores
Accuracy	0.89
Precision	0.853
Recall	0.9
F1-Score	0.875

Metrics	Scores
Accuracy	0.82
Precision	0.78
Recall	0.76
F1-Score	0.77

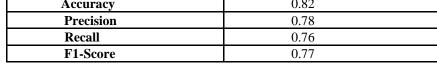


Table 2: Metric Scores of the Traditional Model

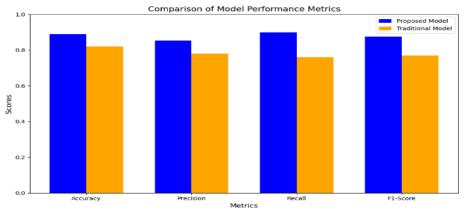


Figure 9: Model performance comparison

This comparison underlines the importance of the adoption of sophisticated methodologies such as CNNs and data augmentation during model development. It underlines their capacity to enhance both accuracy and generalization ability of predictive models. The bar chart shows that there is a performance advantage of the Proposed Model that uses a Convolutional Neural Network (CNN) with efficient data augmentation techniques against the Traditional Model. All key metrics, including accuracy, precision, recall, and F1-Score, show higher scores in the Proposed Model. The use of data augmentation techniques like rotation, flipping, and scaling increases the diversity of the training dataset, allowing the CNN to generalize better and perform more reliably. In contrast, the Traditional Model, probably based on a less complex machine learning approach with minimal feature engineering and no advanced augmentation, shows lower effectiveness, especially in precision and recall. This performance gap illustrates the effectiveness of combining modern deep learning architectures with robust data augmentation strategies to deliver superior results.

6. Conclusion

It designed a chromosome structure detection system meant to classify the chromosomes as either normal or abnormal. Using CNN, this architecture led to its great success, in that high accuracy is displayed and therefore used in identifying anomalies within the structure of chromosomes. Reliably produced predictions mean this model might find further applicability in assisting diagnostics within medical applications as well as in analyzing genes.

The results show that deep learning techniques can be of great value in cytogenetics, providing a tool to assist experts in the early detection of chromosomal abnormalities. Future work could include an extension of the dataset, multi-class classification to identify specific syndromes, and deployment of the model in real-world clinical environments for further validation and enhancement.

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