Automated System For Chromosome Karyotyping Detection

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Abstract. In this study, an intelligent system specifically tailored for the meticulous task of identifying and categorizing chromosomes in the context of karyotyping, a critical process in genetics and medical diagnosis. To achieve, this project leveraged the capabilities of the YOLO (You Only Look Once) object detection framework, a sophisticated tool widely employed in computer vision. Our methodology involved training the system to recognize and categorize individual chromosomes by exposing it to a diverse set of images containing these genetic structures. Our intelligent system presents several notable advantages. Firstly, it operates with remarkable speed, significantly reducing the time required for chromosome analysis. Secondly, it demonstrates exceptional accuracy, thereby minimizing potential errors inherent in manual analysis. The implications of this system are profound, offering benefits to both clinical geneticists and researchers. Medical professionals can utilize it to gain a deeper understanding of genetic conditions, facilitating more precise diagnoses. Simultaneously, researchers can expedite their genetic studies, capitalizing on the efficiency of our automated system. The development process encompassed the creation of an extensive dataset comprising annotated chromosome images, serving as the foundational material for training our YOLO model. Through meticulous fine-tuning and optimization, we achieved outstanding results in terms of precision and recall rates, ensuring dependable chromosome detection and classification. This research delves into the technical intricacies of our system's creation, presents a comprehensive evaluation of its performance, and explores the profound implications for the field of genetics.

Keywords: YOLO, chromosome, dataset

1 Introduction

Genetic diagnostic automation has emerged as a significant area of research in recent years. The intricacies involved in diagnosing genes and chromosomes necessitate ex-

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tensive training for doctors, leading to a small margin of error. The challenges are particularly pronounced in chromosome abnormality diagnostics, where the imbalance between normal and abnormal data complicates the learning process for trainees. Gaining expertise in recognizing abnormal chromosomes becomes demanding due to the predominance of normal chromosomes. Consequently, doctors require more data and prolonged training to navigate this complexity. Chromosome abnormality encompasses anomalies, disorders, mutations, missing, extra, or irregular sections of chromosomal DNA.

Research by Gert de Graaf indicates that the incidence of Down Syndrome (DS) is 12.6 per 10,000, with an average of 5300 DS births annually in the United States between 2006-2010. An implementation of a medical project related to DS led to a 30% decrease in DS births by 2007.The process of determining an organism's karyotype involves collecting a cell, inducing cell division, arresting division in metaphase, staining chromosomes for visibility, and microscopic examination. A regular human karyotype consists of 46 chromosomes, with 22 pairs of autosomes (responsible for human characteristics) and two sex chromosomes, X and Y, determining gender. Autosomes are numbered from class 1 to class 22, with class 1 being the largest and class 22 the smallest. The remaining two chromosomes, X and Y, determine gender, with XX indicating female and XY indicating male.

1.1 Difference between Deep Learning and Machine Learning

Machine Learning (ML) requires high specificity from the user since the computer relies on manual input to interpret and search for features. Deep Learning (DL) stands out as it autonomously illustrates feature sets without manual input, ensuring high accuracy, speed, and reliability.

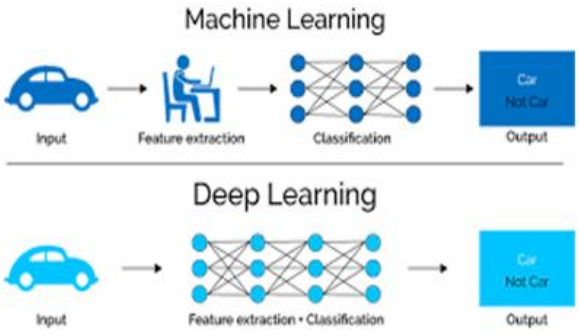

Fig.1. Machine Learning Vs Deep Learning

1.2 Neural Networks

The structure of neural networks is as follows: the algorithm gathers the data, which is then subjected to non-linear transformations. This method is essentially the same when deep learning is applied. The transformations are used to learn, and the result is obtained as a model. This process is repeated across multiple layers and numerous trials until a dependable and precise result is achieved.

1.3 YOLO Algorithm

The YOLO (You Only Look Once) algorithm, introduced in 2015 by Joseph Redmon et al., revolutionized object detection in computer vision. It framed object detection as a single-pass regression problem, predicting multiple bounding boxes and class probabilities simultaneously for improved speed and accuracy.

The YOLO family has evolved since 2016, with YOLO-v8 being the latest addition in 2023. The core concept, initiated by YOLO-v1, involved imposing a grid cell onto the image. If the center of the object fell into a grid cell, that cell would be responsible for detecting the object, enhancing efficiency in handling multiple appearances of objects.

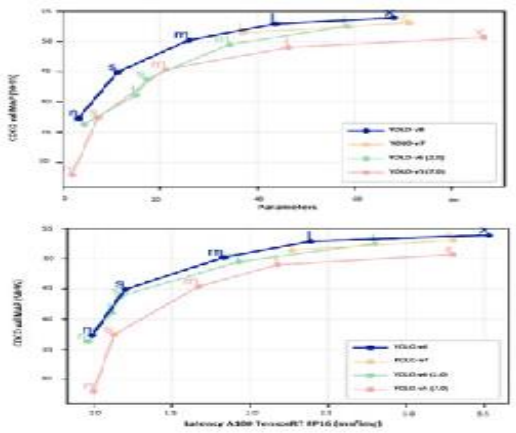

Fig.2. YOLO 8 comparison with predecessors

2 Literature Survey

The paper titled "Chromosome classification for karyotype composing applying shape representation on wavelet packet transform" by L. V. Guimaraes, A. Schuck, and A. Elbern introduces a novel method for automating the karyotyping process, focusing on classifying chromosomes into specific groups. Karyotypes, essential in genetic analysis, traditionally require manual interaction due to the random disposition of chromosomes in photos. The authors propose a technique based on the shape of chromosomes, converting it into a signature. This signature undergoes wavelet packet transform and comparison to classify chromosomes into six groups (A-G) in the karyotype. Results indicate successful chromosome classification, contributing to automation and potentially reducing the need for manual intervention.

In the pursuit of automating karyotyping, W. Zhang et al. present a CNN-based deep learning approach to classify chromosomes into 23 types. Trained on a dataset of 10,304 images, the CNN achieves an impressive 92.5% accuracy, surpassing other methods. The study introduces a "proportion of well-classified karyotype" metric, showing promise for medical professionals in genetic disorder diagnosis. This research signifies a significant step in efficient and accurate karyotyping automation.

Sharma, Swati, and L. Vig propose a Residual Convolutional Recurrent Attention Neural Network (Res-CRANN) for chromosome classification. End-to-end trainable, Res-CRANN incorporates sequence learning, attention mechanisms, and achieves superior accuracy on a dataset compared to baseline models. This demonstrates the model's efficacy in chromosome classification, emphasizing attention to band sequences.

J. Zhang et al. contribute a method for chromosome classification and straightening using an interleaved and multi-task network. Achieving high accuracy in type and polarity classification, the method expedites karyogram production, enhancing clinical chromosome diagnosis efficiency.

S. Gagula-Palalic and M. Can's unique approach involves Competitive Neural Network Teams (CNNT) and Nearest Neighbor for human chromosome classification. The committee of perceptrons achieves over 95% correct classification, and the method demonstrates effectiveness across diverse datasets.

3 Related Works

3.1 Existing System:

- Feature-Based Methods
- Template Matching
- Cascade R-CNN
- SupCAM

3.2 Limitations:

Feature-Based Methods:
Feature-based methods may face challenges in generalizing well to diverse traffic sign variations and varying lighting conditions.
Template Matching:
Template matching can be susceptible to alterations in scale, rotation, and lighting. It may exhibit suboptimal performance when traffic signs are observed from different angles or under changing lighting conditions.
Cascade R-CNN:
Although Cascade R-CNN enhances detection progressively, it can incur high computational costs due to multiple cascades. It might not deliver the same real-time performance as YOLOv8.
SupCAM:
SupCAM heavily depends on the accessibility and quality of training data. To effectively train a SupCAM model, a substantial dataset of annotated chromosome images is essential.

3.3 Proposed System:

Data Collection:
Collect an extensive dataset of images containing chromosome spreads. These images should have annotations with bounding boxes around individual chromosomes, indicating their respective locations.
Data Preprocessing:
Resize and normalize the images to ensure uniformity in size and format.Convert the annotation data into YOLO-compatible format, typically involving class labels (chromosome type) and bounding box coordinates relative to the image size.
Model Selection:
Opt for a pre-trained YOLO model suitable for object detection tasks. YOLOv8 is a recommended choice.
Model Fine-Tuning:
Fine-tune the selected YOLO model using your chromosome dataset. This process involves retraining the model on your dataset to adapt it specifically to chromosome detection.
Training:
Partition your dataset into training, validation, and test sets.
Train the YOLO model on the training set, continually monitoring its performance on the validation set. Adjust hyperparameters and model architecture as necessary.
Evaluation:
Assess the trained model on the test set, employing relevant metrics such as precision, recall, and F1-score to gauge its accuracy.
Visualization:
Develop a visualization system to exhibit the detected chromosomes on the original images, facilitating user interpretation of the results.

Deployment:
Integrate your model into an automated system. This may entail creating a user-friendly interface for users to upload images and receive karyotyping results seamlessly.

4 Methodology

4.1 Data Collection:

The dataset for this study comprises 453 images in the training set, 22 images in the test set, and 3 images in the validation set. The dataset is labeled with numerical labels and corresponding class names such as ['1', '10', '11', ..., 'Y'], which are essential for proper annotation, configuring the model's output layer, and interpreting predictions during evaluation. The dataset structure appears well-organized, making it suitable for a multi-class image classification task, specifically focused on identifying various types of chromosomes.

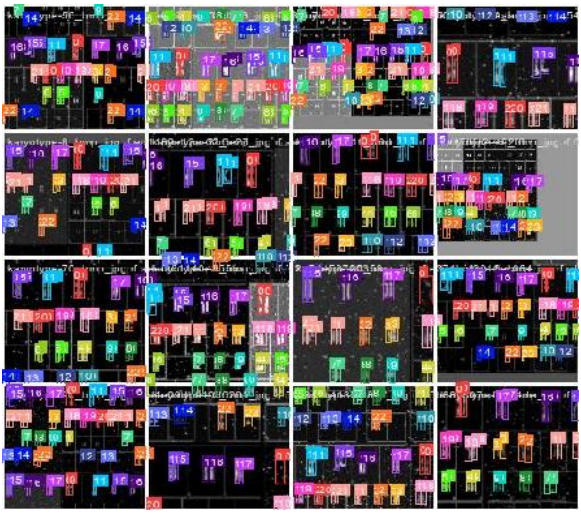

Fig.3. Train Batch

4.2 Annotation and Labelling:

The annotation process involves several tools, including Drag and Select, Bounding Box Annotation Tool, Polygon Annotation Tool, Smart Polygon, Label Assist, and Zoom Tool. These tools facilitate the precise selection, editing, and annotation of images. The Bounding Box Annotation Tool, represented by a rectangular box icon, allows for the creation of new bounding-box annotations by clicking and dragging across an image. This tool, along with others, ensures the accurate representation of objects within the dataset.

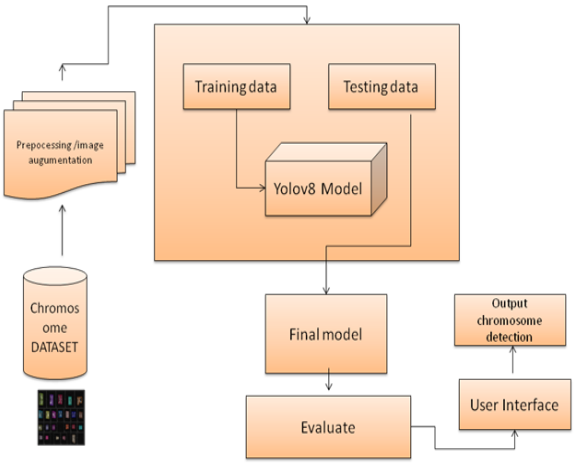

Fig.4. Architecture

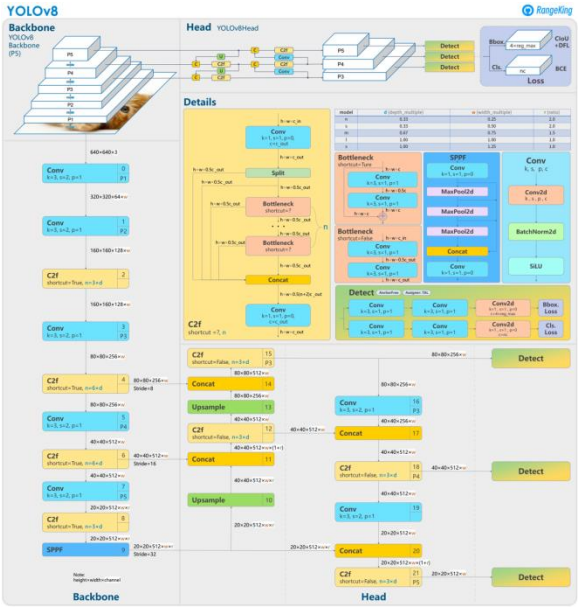

Fig.5. YOLOv8 Architecture

YOLOv8 introduces a new backbone network, anchor-free detection head, and loss function. Notable changes include replacing the C3 module with the C2f module, altering convolution layers, and implementing a decoupled head. Additionally, YOLOv8 features anchor-free bounding boxes, eliminating the need for manually identified anchor boxes. The Mosaic Augmentation process, utilized during training epochs, is stopped before the end to optimize training patterns. The efficiency and accuracy of

YOLOv8 surpass previous versions, as indicated by mean Average Precision, size, and latency during training.

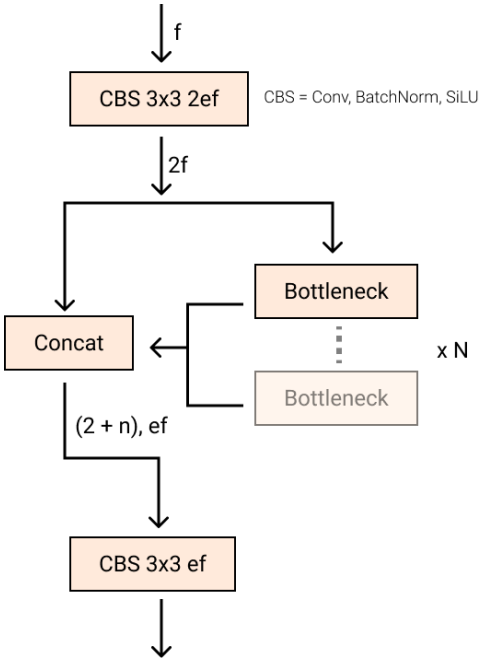

Fig.6. The C2f Module

4.3 Figure - Comparison - Efficiency and Accuracy

The efficiency and accuracy of YOLOv8 are demonstrated through a comparison with YOLOv7, YOLOv6-2.0, and YOLOv5-7.0, showcasing superior mean Average Precision, size, and latency during training. Statistical comparison tables highlight the performance improvements across different-sized YOLOv8 models. The results underscore YOLOv8 as a significant advancement, consistently outperforming YOLOv5 and other frameworks.

In summary, the methodology involves a well-structured dataset, precise annotation tools, and the utilization of YOLOv8 architecture with improvements in backbone structure and bounding box methodology, leading to enhanced efficiency and accuracy in image classification tasks.

5 Results and Discussions

5.1 Output:

0	4.7889	5.3295	1.3579	0	0	0	0	3.5788	5.211	1.0403	1.00E-04
	1.00E-04	1.00E-04									
1	3.5485	4.9611	0.98645	0	0	0	0	3.1473	4.9745	0.90292	0.000203
	0.000203	0.000203									
2	3.1183	4.5979	0.90604	0.01507	0.04696	0.01078	0.00245	3.1511	4.3364	0.8675	0.000306
	0.000306	0.000306									
3	2.9512	4.3823	0.88097	0.02288	0.14388	0.02254	0.00655	2.907	3.846	0.84819	0.000355
	0.000355	0.000355									
4	2.89	4.1274	0.8599	0.02969	0.17675	0.02941	0.00852	2.9606	3.6399	0.8601	0.000355
	0.000355	0.000355									
5	2.8025	3.8958	0.84959	0.04764	0.28729	0.06391	0.01932	2.6329	3.3575	0.84231	0.000354
	0.000354	0.000354									
6	2.7321	3.6599	0.83681	0.05404	0.32826	0.08557	0.02903	2.5167	3.1837	0.83179	0.000353
	0.000353	0.000353									
7	2.6562	3.418	0.82737	0.06387	0.36708	0.11789	0.04134	2.4767	3.023	0.83031	0.000353
	0.000353	0.000353									
8	2.6037	3.1948	0.8188	0.10437	0.29173	0.12965	0.04573	2.5276	2.6129	0.83769	0.000352
	0.000352	0.000352									
9	2.5341	2.9727	0.81272	0.06755	0.44998	0.14355	0.05701	2.1768	2.5902	0.81791	0.000351
	0.000351	0.000351									
10	2.4853	2.8053	0.80088	0.50016	0.21361	0.16505	0.06893	2.1674	2.3286	0.8153	0.000351
	0.000351	0.000351									
11	2.4191	2.6365	0.79878	0.3202	0.29388	0.17452	0.074	2.1551	2.3753	0.81279	0.00035
	0.00035	0.00035									
12	2.4391	2.48	0.8021	0.19954	0.37448	0.20829	0.08733	2.13	2.3689	0.80984	0.000349
	0.000349	0.000349									

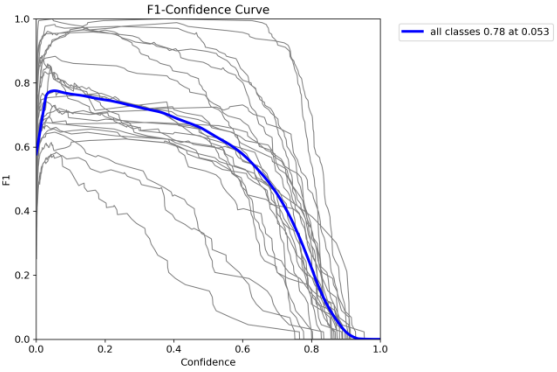

Fig.7. Confidence curve

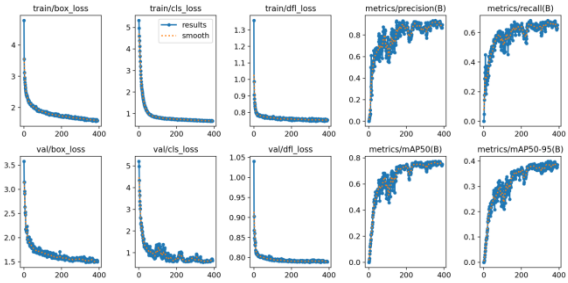

Fig.8. score, precision and recall graph

6 Conclusion

In conclusion, our research has successfully delivered an intelligent system designed to streamline the intricate process of identifying and categorizing chromosomes within the domain of karyotyping. This process holds critical significance in genetics and medical diagnosis. Harnessing the power of the YOLO (You Only Look Once) object detection framework, a formidable tool in computer vision, our system has been trained to autonomously recognize and classify individual chromosomes. This training involved exposure to a diverse set of annotated images, resulting in a system that offers notable advantages, including heightened speed and accuracy.

The YOLO-based system significantly reduces the time required for chromosome analysis, offering a more efficient alternative to manual methods. The heightened accuracy mitigates errors that are inherent in manual analysis. The implications of our work extend across clinical genetics and research, providing medical professionals with a reliable tool to better understand genetic conditions and facilitate more precise diagnoses. Additionally, researchers can leverage the system's efficiency to accelerate genetic studies, contributing to advancements in the field.

The development process included the creation of a comprehensive dataset consisting of annotated chromosome images, forming the foundation for training our YOLO model. Through rigorous fine-tuning and optimization, this research achieved outstanding precision and recall rates, ensuring robust chromosome detection and classification. In essence, our intelligent system represents a valuable contribution to the field, with the potential to transform and expedite chromosome analysis in both clinical and research settings.

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