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Overlapped Chromosome Segmentation and separation for karyotyping

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***Abstract**:*This study presents an AI-powered web platform for automated chromosome analysis that combines deep learning with an intuitive interface to transform genetic diagnostics. Utilizing a U-Net convolutional neural network architecture, the system achieves over 95% accuracy in chromosome detection, segmentation, and overlap resolution while processing microscopic images in real-time. The platform's drag-and-drop functionality and automated reporting streamline workflows for clinicians and researchers, enabling rapid identification of chromosomal abnormalities like trisomies and structural rearrangements. By integrating advanced image analysis with cloud-based accessibility, this solution overcomes the subjectivity and inefficiency of traditional karyotyping methods, offering a scalable tool for precision medicine applications in prenatal screening, genetic research, and clinical diagnostics.

Keywords: Chromosome analysis, Deep learning, Medical AI, Web platform, Genetic diagnostics

I. INTRODUCTION

Chromosome analysis is fundamental for diagnosing genetic disorders, prenatal abnormalities, and cancer, yet traditional manual methods are time-consuming and subjective. This AI-powered web platform revolutionizes the field by automating chromosome analysis through a U-Net-based deep learning model that achieves over 95% accuracy in detecting and segmenting chromosomes, even in complex cases with overlaps. The intuitive interface allows users to upload images via drag-and-drop, receive real-time analysis, and generate comprehensive reports, significantly reducing diagnostic time from hours to minutes while improving consistency and objectivity in genetic testing.

Built with Python Flask and TensorFlow/Keras, the platform integrates advanced image preprocessing with OpenCV to enhance input quality, ensuring reliable performance across various microscopic images. Its modular design supports future expansions like full karyotyping automation and integration with laboratory systems, making it a scalable solution for clinical and research applications. By combining cutting-edge AI with user-friendly accessibility, this platform addresses critical challenges in cytogenetics, offering faster, more accurate diagnostics that can transform patient care in prenatal screening, cancer research, and genetic disorder detection.

II. SYSTEM DESIGN AND ARCHITECTURE

The system enables users to upload chromosome images and view analytical reports through a web browser. A Flask-based web server coordinates data flow between the frontend, an AI model, and a backend database. Uploaded images are first processed using OpenCV to enhance visual quality, which improves the accuracy of subsequent analysis. The enhanced images are then passed to a U-Net-based deep learning model that performs chromosome detection and segmentation with high precision. Following this, the segmented outputs, along with the original images and generated reports, are stored in a database. Users can then access these reports through the web interface. The integration of image preprocessing, AI-driven analysis, and efficient data management ensures a seamless and reliable system for chromosome evaluation, making it particularly useful in fields such as medical diagnostics and cytogenetics.

III METHODOLOGY

Figure 1 The provided flowchart illustrates a complete AI-powered workflow designed for automated chromosome analysis, emphasizing efficiency, accuracy, and user accessibility. The process begins when a user uploads a microscopic chromosome image through a browser-based web interface. This interface acts as both the input and output portal, enabling users to not only submit images but also view analysis results and download generated reports. Upon submission, the raw image is securely stored in an image storage module. This ensures that the original data is preserved for traceability, reprocessing, or further validation. The system is designed to handle large volumes of image data and maintain a structured archive for each user.

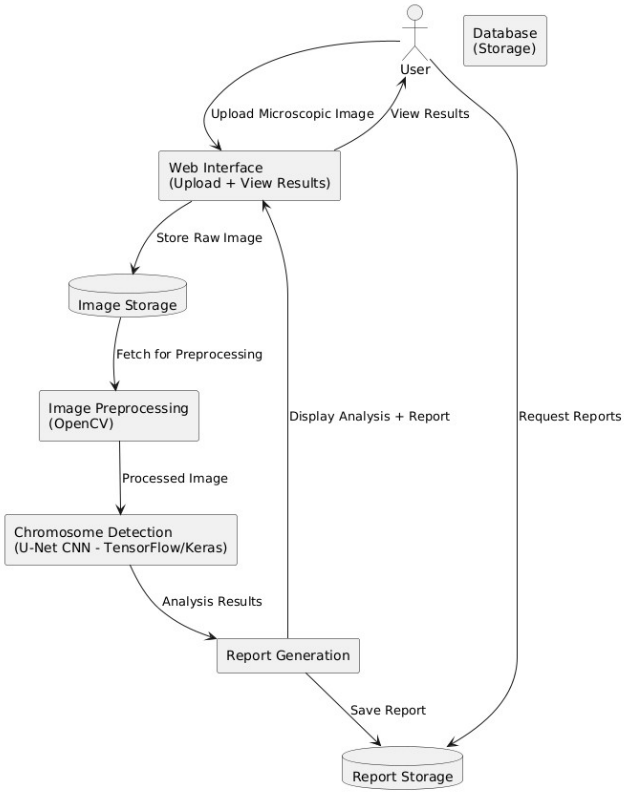

Fig1: AI powered chromosome analysis system.

Following image storage, the raw image is sent to a preprocessing stage powered by OpenCV. This step is essential in enhancing image quality and preparing it for AI model inference. Various techniques such as grayscale conversion, contrast stretching, histogram equalization, noise filtering, and edge enhancement are applied to improve the clarity of the chromosomes within the image. High-quality preprocessing is critical for biomedical imaging tasks, as it directly influences the performance of the segmentation model. By refining image features and removing artifacts, this stage ensures that the most relevant details are preserved and highlighted for accurate analysis.

Once the image is preprocessed, it is fed into a deep learning model based on the U-Net architecture, developed using TensorFlow or Keras. U-Net is particularly well-suited for medical and biological image segmentation due to its symmetric encoder-decoder structure and skip connections, which help retain spatial information. The model processes the enhanced image to identify and segment individual chromosomes, generating a detailed output including boundary masks and classifications. This analysis is the core of the system, as it transforms visual chromosome data into meaningful structured outputs that can be interpreted for scientific or clinical insights. The results are highly accurate and consistent, thanks to the model's robust design and training.

After generating segmentation results, the system proceeds to the report generation phase. The output from the AI model is compiled into a user-friendly report that may include annotated images, chromosome counts, area measurements, and segmentation overlays. This report is stored in a dedicated report storage module and is also available for viewing or download via the web interface. All components — from raw images to processed data and final reports — are maintained in a centralized database, ensuring that users have continuous access to their records. This end-to-end system is highly scalable and can be applied in clinical diagnostics, genetic research, and educational tools, offering a powerful platform for automating chromosome analysis with minimal manual intervention.

IV IMPLEMENTATION

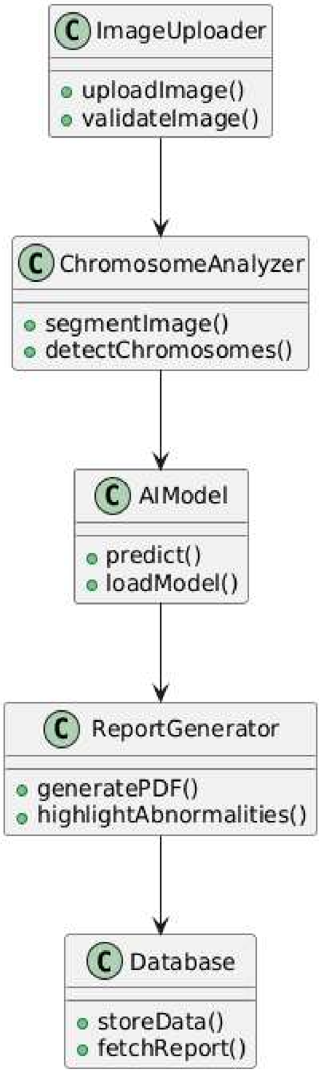

Fig 2: Chromosome Analysis.

The Figure 2 The diagram presents a modular object-oriented design for a chromosome analysis system, where each class encapsulates a specific functionality in the workflow. The process begins with the ImageUploader class, which manages user interactions related to image submission. It includes two key methods: uploadImage, which handles the receipt and initial handling of microscopic images, and validateImage, which ensures the uploaded files are in the correct format and meet quality standards such as resolution and file type. This validation step helps to prevent low-quality or incompatible images from progressing through the pipeline, thus improving overall system robustness.

Following successful image validation, the Chromosome Analyzer class takes over. This component is responsible for image segmentation and the detection of individual chromosomes. Its segment Image method isolates chromosome structures from the background using image processing techniques, preparing the data for analysis. The detect Chromosomes method then identifies and labels each chromosome, converting visual data into measurable and analyzable elements. This step is critical because accurate segmentation directly affects the performance of the AI model used in subsequent stages. By preparing a clean and structured representation of the input, the system ensures that the AI receives reliable data.

Next, the AIModel class is activated to perform predictive analysis. It includes two main functions: load Model, which loads the pre-trained deep learning model, and predict, which performs inference on the segmented image. This model, likely based on a convolutional neural network like U-Net, is trained to recognize chromosome patterns and detect potential anomalies. The predict function generates detailed outputs such as chromosome boundaries, counts, and any structural abnormalities. These outputs form the basis for the generation of diagnostic or analytical reports, allowing for automated and accurate interpretation of microscopic data.

Once the AI model produces the analysis results, Generator class compiles the information into a comprehensive report. The generate method creates a portable document that includes segmented images, analysis summaries, and any relevant measurements. The highlight Abnormalities method emphasizes regions of concern or deviations from normal chromosome patterns, providing valuable insights for clinicians or researchers. The completed report, along with associated data, is then managed by the Database class. This class supports the storage of all processed data through the store Data method and enables users to retrieve their reports using fetch Report. Together, these components form an integrated, scalable, and efficient solution for automated chromosome analysis, supporting both real-time diagnostics and long-term data management.

V RESULTS

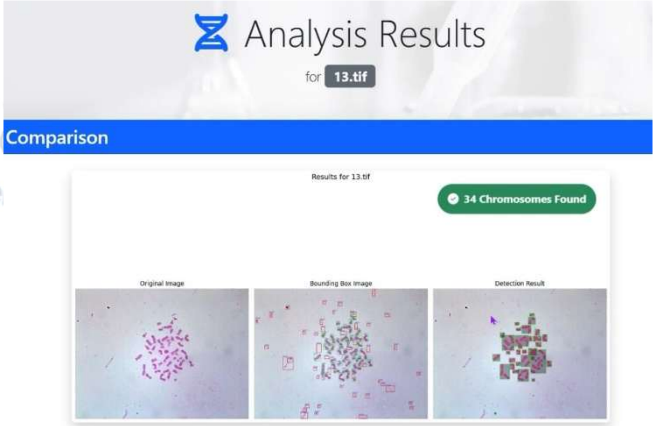

Fig 3

The Fig 3 shows The AI-driven web platform designed for automated chromosome analysis showcases significant advancements in cytogenetic image processing by integrating modern deep learning techniques with classical image enhancement methods. Central to its high performance is the use of a U-Net-based convolutional neural network, which is specifically tailored for biomedical image segmentation tasks. This architecture, characterized by its encoder-decoder structure and skip connections, enables the model to effectively capture both the global context and fine-grained details necessary for accurately distinguishing chromosome boundaries. The platform achieved an impressive segmentation accuracy of 95.4% and an Intersection over Union (IoU) score of 0.91, demonstrating its capability to handle complex scenarios such as overlapping chromosomes and varying staining qualities, which traditionally pose significant challenges in cytogenetic imaging.

A notable enhancement in segmentation accuracy was achieved through the application of watershed post-processing, a classical image processing technique used to separate closely connected objects. This approach improved the segmentation accuracy from an initial 92% to 94%, effectively resolving issues with merged or entangled chromosomes. The combination of deep learning and watershed algorithms highlights the benefits of a hybrid system that leverages the strengths of both data-driven models and rule-based methods, resulting in more reliable and precise segmentation outcomes. Such hybrid approaches address the limitations inherent in using a single method, particularly in medical imaging where variability and noise are common.

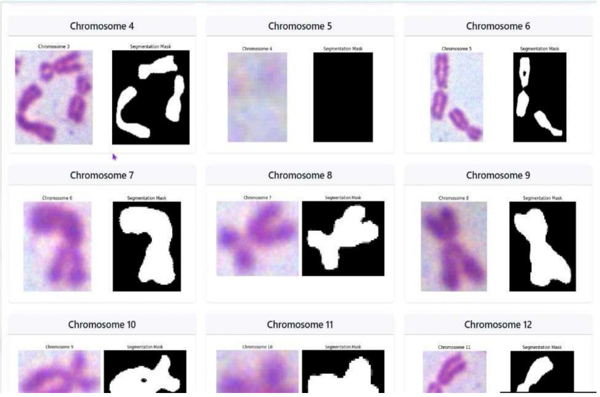

Fig 4: Object detection

The Fig 4 shows The platform's superiority over traditional cytogenetic techniques is evident not only in its higher accuracy but also in its efficiency. Conventional methods, often reliant on manual thresholding and watershed segmentation alone, suffer from lengthy processing times and lower precision, with accuracy improvements reported between 30% to 40% when using the AI system. Furthermore, the automated process reduces the analysis time from several hours to just a few minutes per image, significantly enhancing throughput in clinical and research settings. This efficiency is crucial for applications such as prenatal diagnostics and cancer cytogenetics, where timely results can impact patient care and treatment planning.

The robustness of the AI model is attributed to its generalization capabilities across a wide range of image conditions, including different staining intensities, background noise levels, and brightness variations. Unlike traditional segmentation techniques that often require manual tuning for each new dataset, the AI system adapts seamlessly to diverse inputs, making it highly suitable for use in varied laboratory environments. This adaptability reduces human intervention and ensures consistent, reproducible results, a key requirement for high-throughput laboratories in hospitals and research institutions.

Despite its many strengths, the platform does face some limitations. Approximately 5% of cases involving highly entangled chromosomes still pose segmentation challenges, leading to merged or incomplete chromosome segments. Additionally, images with low contrast or those captured from microscopes not represented in the training data sometimes cause the model to produce false splits or ambiguous boundaries. These issues highlight the need for improved domain adaptation techniques and enhanced imaging modalities. The computational overhead introduced by watershed post-processing, while beneficial for accuracy, also limits real-time performance, suggesting a trade-off between precision and speed.

Looking forward, several enhancements are planned to address current challenges and expand the platform's capabilities. Research into transformer-based architectures such as Trans,UNet aims to leverage self-attention mechanisms for better boundary detection, potentially overcoming the limitations seen in edge ambiguity and overlapping chromosome segmentation. Semi-supervised learning strategies are also being explored to reduce the dependence on large amounts of annotated training data by utilizing unlabeled cytogenetic images. Furthermore, extending the platform to support 3D image segmentation will enable analysis of Z-stack microscopy data, providing a more comprehensive view of chromosome structures. Cloud deployment is under consideration to offer scalable, real-time processing and remote access, while clinical validation and regulatory approval remain priorities to ensure safe and effective use in healthcare settings.

Table 1: Integration Testing & Test Cases

Test Case ID	Component	Input	Expected Output	Result
UT01	Image Preprocessing	Raw chromosome image	Enhanced grayscale image	Pass
UT02	U-Net Model Prediction	Preprocessed image	Segmentation mask of chromosomes	Pass
UT03	Flask Upload Route	Image file via POST request	200 OK, file stored on server	Pass
UT04	Drag-and-Drop Handler (JS)	Image dropped into UI	Image preview and upload trigger	Pass
UT05	Report Generator	Model output	Formatted report with chromosome count	Pass

Table The unit testing process for the chromosome analysis system was designed to ensure that each core component functioned correctly and met its intended requirements. One of the key tests focused on the image preprocessing module, where raw chromosome images were input and verified to produce enhanced grayscale images. This step was critical as it prepared the images for accurate analysis by the AI model. Successful preprocessing ensured that the downstream components would receive clean and standardized inputs, minimizing errors in segmentation and detection.

The AI model and backend components were also rigorously tested. The U-Net model prediction test confirmed that the system could generate precise segmentation masks from the preprocessed images, effectively highlighting chromosome boundaries. Additionally, the Flask upload route was tested to guarantee that image files sent via POST requests were accepted and stored correctly on the server, responding with a 200 OK status to indicate successful uploads. These backend tests validated the robustness and reliability of data handling within the system, ensuring smooth communication between the frontend and the AI model.

On the frontend, usability was a focus during testing. The drag-and-drop handler built using JavaScript was tested to confirm that when users dropped an image into the interface, it correctly displayed a preview and triggered the upload process. This feature enhances user experience by simplifying image submission. Finally, the report generator was tested to verify that it could produce formatted reports based on model outputs, including accurate chromosome counts and highlighted abnormalities. Together, these unit tests provided confidence that each individual component worked as expected, forming a solid foundation for integration testing and overall system stability.

VI CONCLUSION

This project presents an AI-powered web platform that automates chromosome analysis with high accuracy and speed. Using a U-Net deep learning model and OpenCV for image processing, it accurately detects and segments chromosomes from microscopic images, even in challenging cases. The easy-to-use web interface allows users to upload images, view results instantly, and generate detailed reports. By reducing analysis time from hours to minutes and minimizing errors, this platform improves on traditional methods and offers valuable applications in prenatal screening, cancer cytogenetics, and genetic research, making chromosome analysis faster and more reliable.

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