

AI Approach for Enhanced Thalassemia Diagnosis Using Blood Smear Images

Daniela MAZZUCA ^{a,b}, Fulvio BERGANTIN ^c, Davide MACRÌ ^c,

Francesco ZINNO ^a, and Agostino FORESTIERO ^{c,1}

^a *Immunohaematology Section, Annunziata Hospital, Via F. Migliori, CS, Italy* ^b

Department of Clinical and Experimental Medicine, University Magna Græcia of Catanzaro, Italy

^c *Institute for High-Performance Computing and Networking, National Research Council, Rende (CS), Italy*

Abstract. This paper aims to propose an approach leveraging Artificial Intelligence (AI) to diagnose thalassemia through medical imaging. The idea is to employ a U-net neural network architecture for precise erythrocyte morphology detection and classification in thalassemia diagnosis. This accomplishment was realized by developing and assessing a supervised semantic segmentation model of blood smear images, coupled with the deployment of various data engineering techniques. This methodology enables new applications in tailored medical interventions and contributes to the evolution of AI within precision healthcare, establishing new benchmarks in personalized treatment planning and disease management.

Keywords. Artificial Intelligence, Thalassemia, U-Net architecture, Personalized Medicine

1. Introduction

Thalassemia is a genetic disorder characterized by lower than normal hemoglobin levels due to the absence or malfunction of genes vital for hemoglobin production. Hemoglobin, a protein in red blood cells, is crucial for oxygen transport. Thalassemia is categorized into alpha and beta types based on the specific genes affected. Its symptoms vary from none to severe anemia, and in the most severe cases, it can lead to fetal mortality. Symptoms may manifest early in life, potentially impacting lifespan. Although incurable, treatments are available to alleviate symptoms. The application of artificial intelligence in diagnosing and managing thalassemia has seen significant advancements, as evidenced by various studies. [1] explores the effectiveness of AI in diagnosing and classifying thalassemia. It discusses the challenges of distinguishing thalassemia from other causes of microcytic anemia and how AI can aid in accurate diagnosis and treatment planning through analysis of complete blood count (CBC) parameters. [3] introduces a novel AI

¹Corresponding Author: Agostino Forestiero, agostino.forestiero@icar.cnr.it

framework for diagnosing thalassemia using deep learning and medical imaging. It focuses on developing a supervised semantic image segmentation model, leveraging data engineering methods for improved diagnosis accuracy, and demonstrating significant advancements in medical imaging for thalassemia diagnosis. The utilization of AI to analyze erythrocyte morphology and machine learning to identify thalassemia gene carriers in non-anemic populations were proposed in [4]. By quantitatively analyzing abnormal erythrocytes, the study developed a prediction model, demonstrating AI's potential to enhance thalassemia carrier identification.

2. Proposed Approach

We propose a novel application of AI methodologies for the detection and classification of erythrocyte morphology, facilitating the diagnostic process for thalassemia. This approach relies on the application of the U-net architecture [2] for semantic image segmentation of blood smear images. The U-net architecture, renowned for its effectiveness in semantic image segmentation, especially in medical imaging, includes a contraction path (encoder) that captures context and an expansion path (decoder) for accurate localization. The architecture's innovative use of skip connections bridges the encoder and decoder, ensuring detailed feature preservation and accurate segmentation. Initially designed for biomedical image segmentation with limited data, U-net's structure and functionality have significantly impacted medical imaging and beyond, inspiring a range of modifications and applications in various segmentation tasks. This precision is crucial for identifying subtle morphological variations indicative of thalassemia. By automating the segmentation and classification process, the proposed approach allows for reducing the time required to diagnose thalassemia, with notable benefits for patient care. The implementation of U-net for erythrocyte morphology classification also provides an avenue for exploring explainable AI (XAI) principles in medical diagnosis. By analyzing the network's attention and feature maps, researchers can gain insights into the model's decision-making process, contributing to the development of more transparent and interpretable AI tools in personalized healthcare.

References

- [1] Khaled Ferih, Basel Elsayed, A. Elshoeibi, A. Elsabagh, Mohamed Elhadary, Ashraf Soliman, Mohammed Abdalgayoom, and Mohamed Yassin. Applications of artificial intelligence in thalassemia: A comprehensive review. *Diagnostics*, 13, 2023.
- [2] O. Ronneberger, P. Fischer, and T. Brox. U-net: convolutional networks for biomedical image segmentation. *Lecture Notes in Computer Science*, pages 234–241, 2015.
- [3] A. Zaylaa, Mohammad Makki, and Rola Kassem. Thalassemia diagnosis through medical imaging: A new artificial intelligence-based framework. In *2022 International Conference on Smart Systems and Power Management (IC2SPM)*, pages 41–46, 2022.
- [4] Fan Zhang, Jieyu Zhan, Yang Wang, Jing Cheng, Meinan Wang, Peisong Chen, J. Ouyang, and Junxun Li. Enhancing thalassemia gene carrier identification in non-anemic populations using artificial intelligence erythrocyte morphology analysis and machine learning. *European Journal of Haematology*, 2023.