BA/MA Thesis: Deep Learning for Inherited Retinal Diseases Detection

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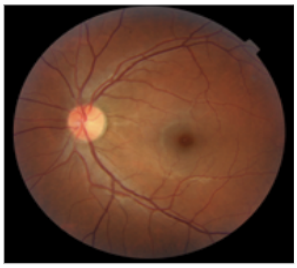
Abstract. Our project aims to improve the diagnosis of Inherited Retinal Dystrophies (IRDs), a group of rare retinal diseases impacting over 2 million people globally [1]. IRDs can lead to vision problems like night blindness, color blindness, tunnel vision, and eventual blindness, greatly affecting patients' and their families' quality of life [4]. With more than 20 different conditions falling under IRDs [1], we're dedicated to addressing the specific needs of each affected individual, taking into account the age of onset, progression rate, and causative gene(s) [3].



Fundus Image of a healthy case (wikimedia.org)

IRDs are relatively rare, with an estimated prevalence of 1 in 16,500 to 1 in 4,000, often leading to misdiagnoses and a prolonged journey for patients, especially in low and middle-income countries (LMICs) [8,9]. Among the different IRD forms, Retinitis Pigmentosa (RP) is the most common. Still, other less-known conditions include Leber Congenital Amaurosis, cone or cone-rod dystrophies, Stargardt disease, and best vitelliform macular dystrophy [2,5]. Additionally, syndromic conditions like Usher syndrome and Bardet-Biedl syndrome manifest as a combination of eye and hearing issues [6,7].

Our **goal** is to use cutting-edge deep learning techniques to **create a model that can detect abnormalities in Fundus images**. By combining images with clinical signs, we aspire to provide an accurate and timely diagnosis for IRD patients worldwide, particularly those in LMICs. This innovation has the potential to positively impact the lives of thousands of IRD patients and enhance the efficiency and accuracy of IRD diagnoses on a global scale. Join us in this exciting journey to make a meaningful difference in the lives of those affected by IRDs.

Research Questions:
Q1) Would unsupervised learning, e.g., anomaly detection models, deliver an acceptable diagnosis rate for patients with different IRDs, most importantly RP?

Q2) Does adding clinical symptoms, if available, to the fundus images improve the diagnosis rate?

Datasets. We expect to analyze more than 1,000 fundus images from healthy and IRDs. We will retrieve the healthy fundus images from the Kaggle database; namely

- (https://www.kaggle.com/datasets/andrewmvd/ocular-disease-recognition-odir5k)

while the IRDs from

- the RIPS database (https://www.icar.cnr.it/en/sites-rips-datasetrips/), (https://www.kaggle.com/datasets/linchundan/fundusimage1000), and (https://www.kaggle.com/datasets/andrewmvd/retinal-disease-classification).

We will test our algorithm on fundus images, collected from our Lebanese partner, of IRD patients who were genetically diagnosed.

Roadmap (6 months):

- Familiarize yourself with the current literature [10-12]
- Build the baseline supervised model and develop the anomaly detection model.
- Run the necessary comparisons.
- Equip the models with the Monte-Carlo Dropout for uncertainty estimation AND/OR
- Equip the models with the visualization methods, e.g., INNvitstigate
- Run extensive experiments and analysis
- Write up your thesis

Requirements:

- Solid background in Machine/Deep Learning
- Familiar with discriminative deep learning models and SOTA architectures
- Sufficient knowledge of Python programming language and libraries (Scikit-learn)
- Experience with a mainstream deep learning framework such as PyTorch.
- Machine/Deep learning hands-on experience

References:

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