Learning Robust and Explainable Representation for Rare Disease Diagnosis From Facial Images

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Motivation

- Genomics diseases are both time and effort consuming to study especially in terms of obtaining the needed data.
- Different types of data can be used to approach the detection of genomes disorders.
- Images of patients' faces can be used as part of genomes disease and especially rare one which are challenging to spot.

Dataset

- The used dataset is facial images from GestaltMatcher (GMDB).
- It consists of a collection of facial images of patients with genetic syndromes.
- The dataset is made for research purposes for clinicians and computer scientists.
- It is an upon request dataset from https://db.gestaltmatcher.org/

Dataset Preparation / Processing

Landmark & Face Detection

Cropping

Eyes Masking
(Additional Procedure for Patients Privacy Preserving)