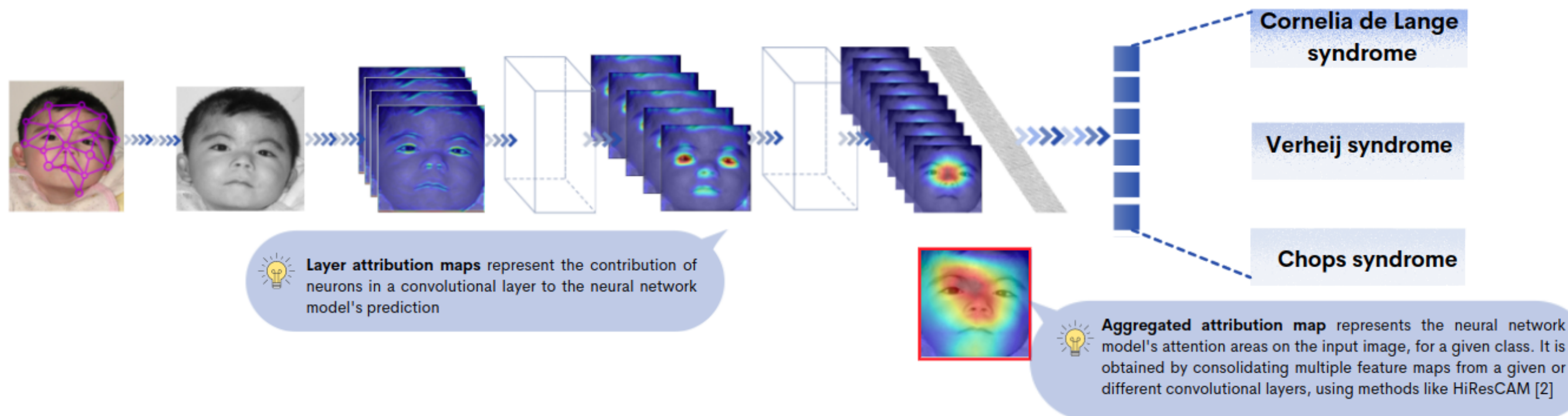


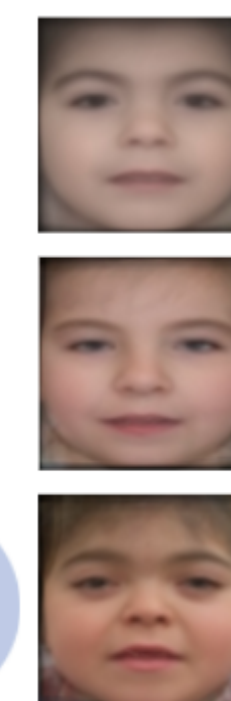
Problem Statement

GestaltMatcher [1] is a deep convolutional neural network (CNN) based next-generation phenotyping (NGP) tool, which claims to surpass the performance of clinical practitioners in the identification of certain rare genetic syndromes, from frontal facial images of patients. Despite its success in terms of predictive performance, the black box nature of its underlying neural network model makes the tool's decisions uninterpretable, limiting it from getting deployed in a clinical setting.

GestaltMatcher Classifier



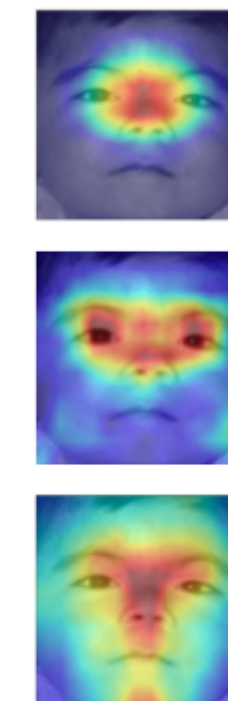
Composite Faces



Qualitative evaluation of the GestaltMatcher's top-k predictions can be performed by visually comparing the syndromes' respective composite faces with the patient's face

Quantitative evaluation of predictions can be performed using similarity maps, which highlight the regions of phenotypical feature similarity between each of the top-k syndrome's composite face and the patient's face

Similarity Maps



Note: Images shown under Similarity Maps are for representative purposes only.

Dataset and Methods

- GestaltMatcher database (GMDB) dataset** [1] was used to train the classifier model, obtain attribution maps and generate composite faces.
- HiResCAM** (Class Activation Mapping) [2] method was used to generate layer visualizations and attribution maps.
- Composite faces** were generated by aligning, warping, and averaging facial images from the train split of the GMDB dataset.

Summary

- Our work proposes three different features that can be used CNN-based NGP tools like GestaltMatcher [1], which enable clinicians to review the basis of their predictions independently, and also support them in diagnosing rare genetic syndromes.
- Analyzing artifacts like composite faces and attribution maps offers a way for the scientific community to discover facial regions containing novel phenotypic features.

References

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