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Quantitative Comparison of Deep Learning Classifiers and Human Attention in Assessing Rare Disorders

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Advisors

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Introduction

- Around 6% of the overall population is affected by genetic conditions.
- The rarity of some genetic conditions makes some diagnoses be missed if a clinician is not experienced enough with them.
- The deep learning classifiers can predict the syndromes depending on the learned facial features.
- We can use Explainable AI (XAI) methods to highlight those regions in a face that a model would have chosen to give the prediction.
- To compare the predictions of XAI, we can use heat maps obtained from an eye-tracker that is used with similar images.

Motivation

- **Rarity of genetic conditions** - makes diagnosis difficult for experienced clinicians
- **Black box between model predictions** - lack explanations
- **Check comparability** - Comparison of clinicians observations to the explanations by Artificial Intelligence (AI) model

Challenges and difficulties

- Lower performance of the classifier
- Smaller dataset and class imbalance
- Lack of methods to validating outputs

Problem statement

RQ1. What are the image saliency methods available to identify the Area Of Interest (AOI) in a given image to make predictions?

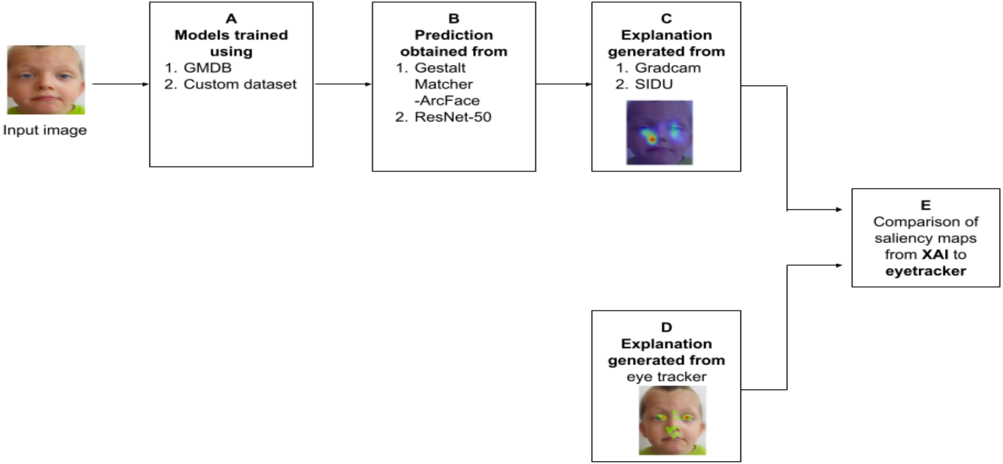
RQ2. What are the important AOI in a face to make predictions related to genetic disorders?

RQ3. Do the saliency maps of clinicians and eye-tracker saliency maps align?

RQ4. Do the clinicians and non-clinicians look at the same facial features?

Methodology

Proposed pipeline



Step A - Datasets

- GestaltMatcher DataBase (GMDB)
- Custom dataset

Figure 1: Few example of images in GMDB.

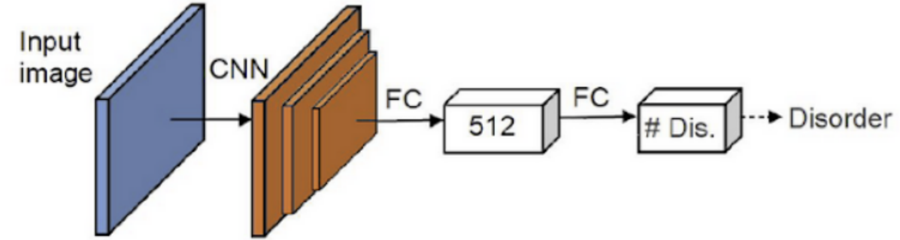
Step B - Rare disorder identification and classification methods

- GestaltMatcher-Arc[1]
- ResNet-50[2]

[1] Hustinx et al., “Improving Deep Facial Phenotyping for Ultra-rare Disorder Verification Using Model Ensembles”.
[2] Mandal, Okeukwu, and Theis, “Masked Face Recognition using ResNet-50”.

GestaltMatcher-Arc

- Extension of **DeepGestalt** approach
- Uses same architecture and face dataset (CASIA) as a base for **transfer learning**
- Each image encoded into **320**-dimensional representation vector
- Representation vectors spanned a **Clinical Face Phenotype Space (CFPS)**
- In the CFPS, patients with **rare disorders** can be matched to other similar patients
- **Clustering analysis** performed to analyze the similarity among different disorders



ResNet-50

- Is a 50 layer convolutional neural network with four main parts
  - Convolutional layers
  - Identity block
  - Convolutional block
  - Fully connected layers

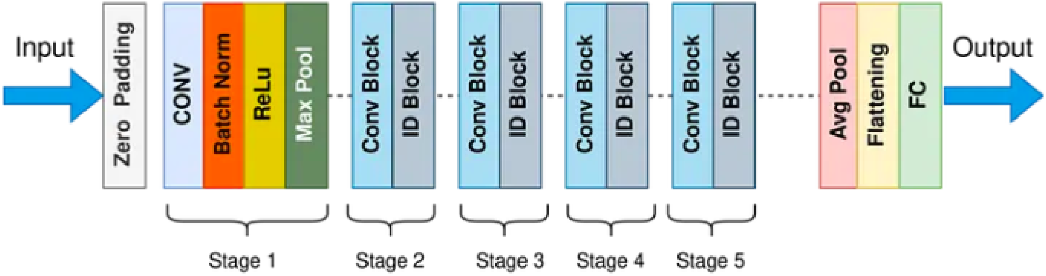


Figure 3: ResNet50 architecture

Experimental setup

Network architectures

Hyperparameters	Values	
	GestaltMatcher-Arc	ResNet-50
Input image size	100×100	100×100
Learning rate	0.001	0.01
Loss Function	Cross-entropy	Categorical cross-entropy
Epochs	50	50
Batch size	32	32

Table 1: Details of training of GestaltMatcher-Arc and ResNet-50.

Step C - Explanation methods

- Gradient-weighted Class Activation Mapping (Grad-CAM)[3]
- Similarity Difference and Uniqueness (SIDU)[4]

[3] Selvaraju et al., **Grad-CAM: Why did you say that?**

[4] Muddamsetty et al., “Visual explanation of black-box model: Similarity Difference and Uniqueness (SIDU) method”.

Grad-CAM

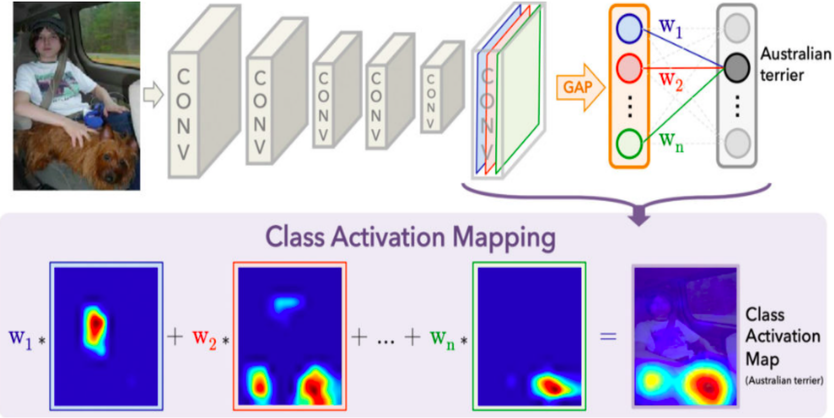


Figure 4: CAM architecture. Image source[5]

[5] Zhou et al., **Learning Deep Features for Discriminative Localization.**

SIDU

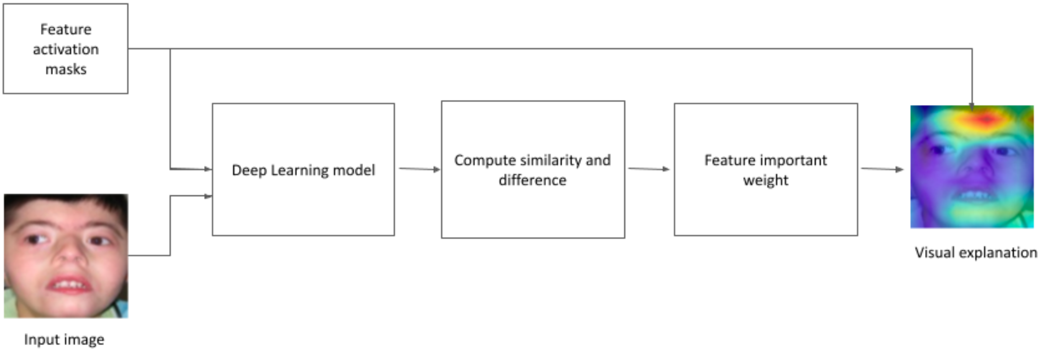


Figure 5: Overall architecture of SIDU in our case.

Implementation details

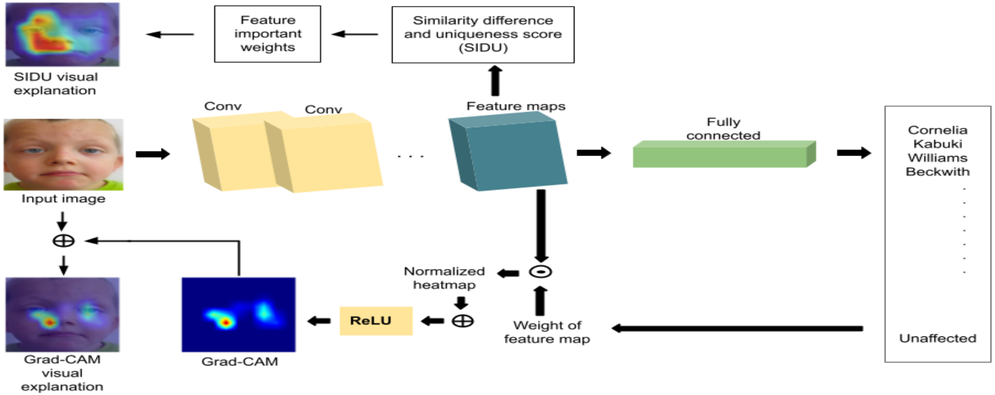


Figure 6: Grad-CAM and SIDU for GestaltMatcher-Arc and ResNet-50 in our research.

Step D - Eye-tracking experiment

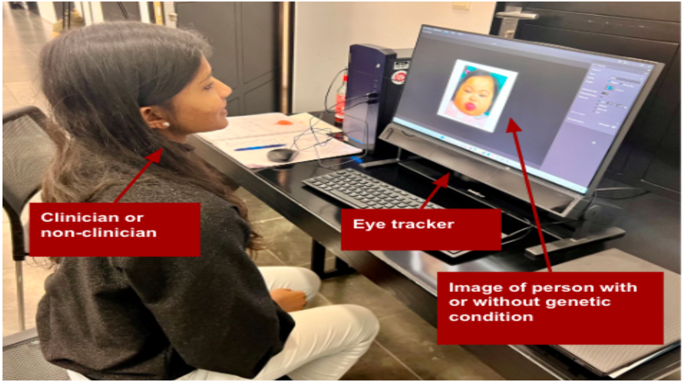


Figure 7: Eye-tracking experiment performed at IGSB Bonn on clinicians and non-clinicians in association with NIH.

Evaluation and results

- Evaluation
  - Experiment 1: Explanation generated by visual explanation methods
  - Experiment 2: Human-grounded evaluation
- Comparison metrics - Evaluation of eye-tracking to XAI methods

Experiment 1: case 1

Explanation generated by visual explanation methods

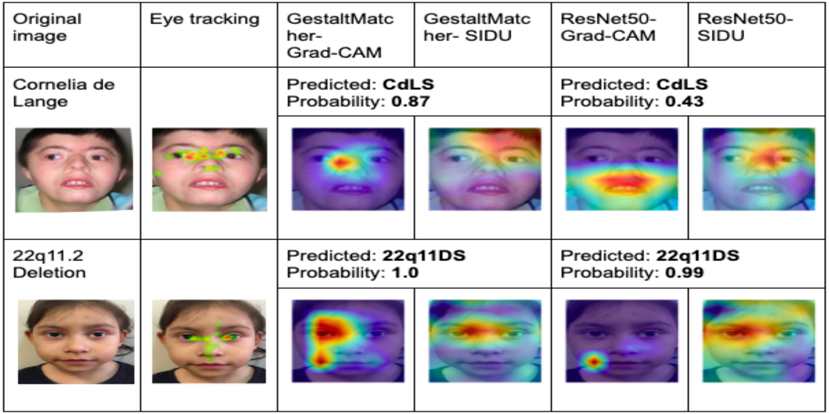


Figure 8: Heat maps of correctly predicted

Experiment 1: case 2

Explanation generated by visual explanation methods

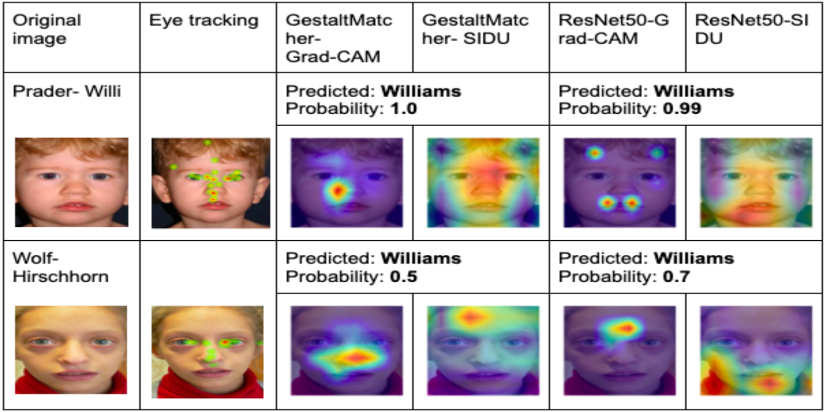


Figure 9: Heat maps of wrongly predicted

Experiment 2

Evaluations from clinicians based on the AOI in face

Condition	Facial features				
	Forehead	Eyes/Periorbital	Nose	Mouth	Ears
Cornelia de Lange		+	+	+	+
22q11.2 deletion			+		
Down		+	+	+	+
Noonan		+			+
Prader-Willi		+			
Wolf-Hirschhorn	+	+	+		
Williams-Beuren		+	+	+	

Table 2: Syndrome-specific facial features identified by a clinician. + sign shows the feature important in a face.

Evaluation: comparison metrics

Syndrome	GestaltMatcher-Grad-CAM	GestaltMatcher-SIDU	ResNet50- Grad-CAM	ResNet50- SIDU
Cornelia de Lange	0.76	0.56	0.30	0.89
22q11.2 deletion	0.78	0.86	0.20	0.67
Down	0.34	0.20	0.34	0.43
Noonan	0.45	0.36	0.56	0.67
Prader-Willi	0.67	0.89	0.56	0.78
Wolf-Hirschhorn	0.56	0.76	0.65	0.10
Williams-Beuren	0.45	0.65	0.78	0.34
Average	**0.58**	**0.62**	**0.48**	**0.55**

Table 3: Comparison table for IoU score. The IoU values are calculated between the eye-tracker heat maps and different XAI heat maps considered.

Revisiting the research questions...

RQ1. What are the image saliency methods available to identify the AOI in a given image to make predictions?
SIDU approach was able to emphasize the crucial regions more accurately than the approach Grad-CAM.
SIDU approach highlights the regions spread over some portion of the face, whereas, Grad-CAM approach highlights small regions in the face.

RQ2. What are the important AOI in a face to make predictions related to genetic disorders?
There is **no genetic disorder specific AOI** identified by the visual explanation method.
SIDU based approach tends to highlight AOI in the images more specifically than the Grad-CAM based approach.

Revisiting the research questions...

RQ3. Do the saliency maps of clinicians and eye-tracker saliency maps align?
There is a huge difference in the features observed by a model and a human while looking at features in an image.

RQ 4 Do the clinicians and non-clinicians look at the same facial features?
Clinicians tend to look at other features **excluding the obvious ones**.
Whereas **non-clinicians** always tend to look at the **eye, nose,** and **mouth** region.

Contributions

- Conducted literature review for rare-disorder identification and classification and also explanation methods.
- Implemented selected classification methods and explanations generated.
- Compared saliency maps from XAI to eye-tracker maps.
- Presented the ideas as a poster at Arbeitsgemeinschaft für Gen-Diagnostik e.V. (AGDev).
- Contributed to recently published paper *"Comparison of clinical geneticist and computer visual attention in assessing genetic conditions"*in PLOS genetics.

Future work

- Improvement to dataset
- Extending eye-tracking survey
- Find approach to quantify results

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Motivation and background

- Perform eye tracking analysis and to understand how clinicians diagnose patients.
- Comparing with explainable AI to understand difference between human and AI.

Datasets used

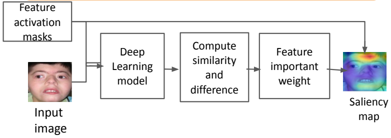
- **GestaltMatcher DataBase** (7 syndromes)
- **Custom dataset** (11 classes -10 syndromes, 1 unaffected

Deep Learning classifiers used

Deep Learning classifiers are used to classify the images into various syndromes. Deep Learning classifiers used in this work,

- **GestaltMatcher ArcFace**
- **ResNet50**

Explainable AI Methods



The regions responsible for prediction are highlighted using explainable AI method (**GradCam**, **SIDU**). The figure above represents the saliency map generation using SIDU.

Eye-tracking data using Tobii-Pro

A Tobii-eye tracker device is used for collecting data from clinicians and non-clinicians provided with syndromic and non-syndromic faces. Heatmaps are generated from the eyetracker based on the time spent on each facial feature.

Comparison to eye-tracking data

The eye tracking heatmaps are compared with the explainable AI heatmaps using KL-Divergence and got 0.31 for ResNet gradcam, 0.51 for ResNet SIDU approach.

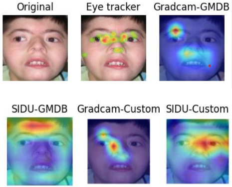


Figure 2: Comparison of eye tracker heatmaps with Gradcam and SIDU.

Conclusion and results

- AI and human look at different features in an image
- Difficult to compare AI and human for different syndromes

References
[1] Satya M. Muddamsetty, Visual explanation of black-box model: Similarity Difference and Uniqueness (SIDU) method, Pattern Recognition,2022

Thank you