




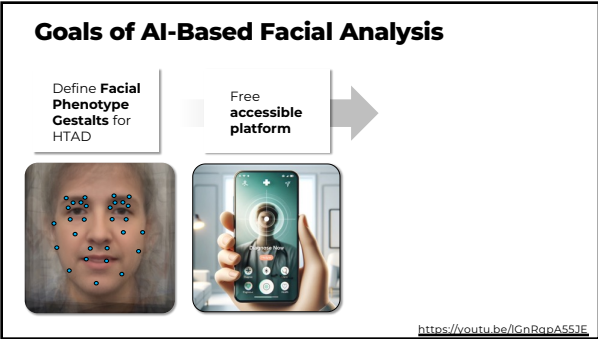
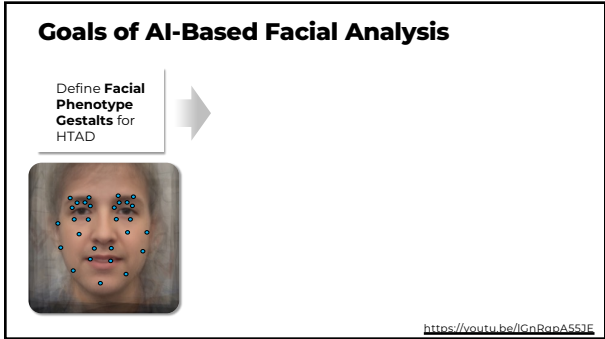
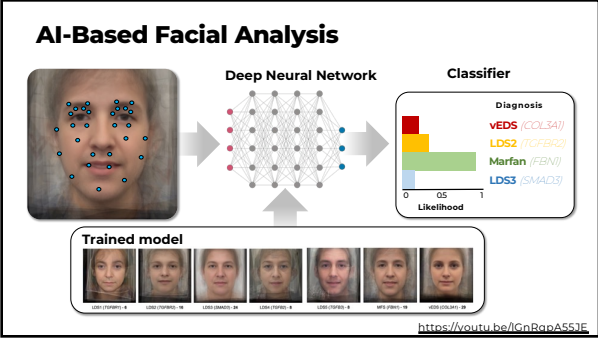
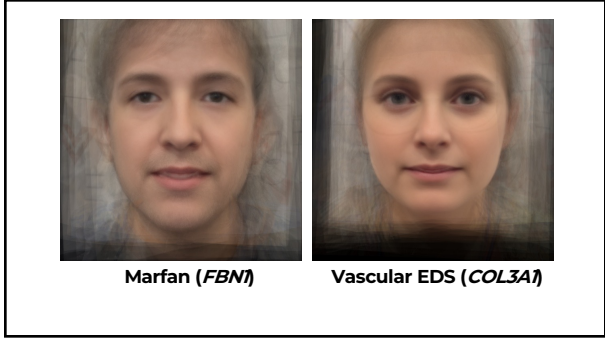


AI Based Facial Analysis Identifies Phenotypic Features Of Heritable Aortic Diseases

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McGovern Medical School
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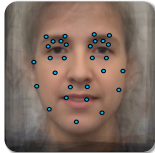






No Disclosures




Goals of AI-Based Facial Analysis

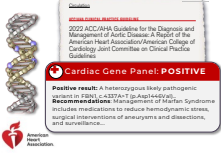
Define **Facial Phenotype Gestalts** for HTAD



Free accessible platform



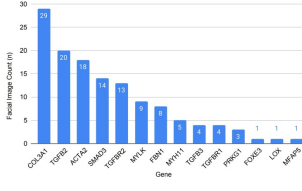
Early diagnosis and management



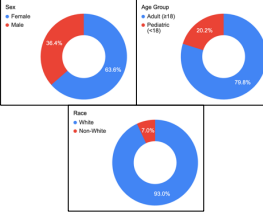
<https://youtu.be/GnRopAS5JE>

Images Collected to Date = 130*

MAC AORTATM
Montalcino Aortic Consortium



Gene	Count
COL3A1	28
TGFBR1	20
ACTA1	18
SMAD3	14
TGFBR2	13
PRK	9
MYH11	8
TGFBI	5
TRIM3	4
FOXP2	3
COL	1
MAP3K4	1



Sex

- Female: 76.4%
- Male: 23.6%

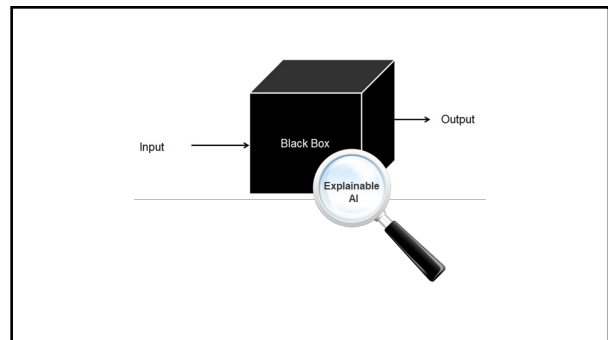
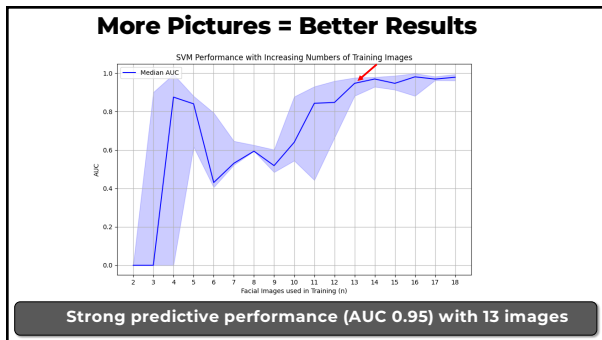
Age Group

- Adult (18-): 80.2%
- Pediatric (<18): 19.8%

Race


- White: 79%
- Non-White: 21%

*Not all images are usable (glasses, expression, etc.)




Local interpretable model-agnostic explanations (LIME)

- Creates perturbed versions of the input image
- Analyzes how these changes affect the model's predictions
- Highlights which parts of the image most influence the prediction using a heatmap
- Deep blue areas** = distinguishing features compared to controls



VEDS vs Controls

vEDS vs Controls



Blue areas distinguish vEDS

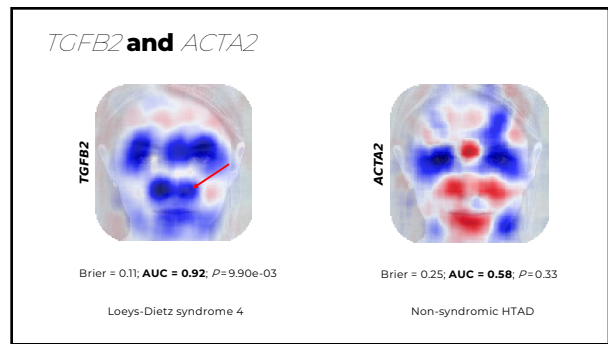
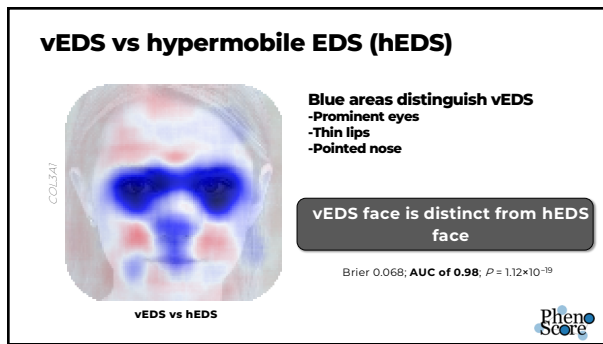
- Prominent eyes
- Thin lips
- Pointed nose


vEDS face is distinct from average face

Brier 0.068; **AUC of 0.97**; $P = 2.93 \times 10^{-11}$

VEDS vs Controls

PhenoScore



- ### Conclusions
- 
- AI-based facial recognition offers a promising approach to early HTAD diagnosis
 - Non-invasive, cost-effective, rapid
 - Useful for non-geneticists and regions with low access to genetic testing
 - Limited number of training images needed (13 for vEDS)
 - Excellent accuracy, with an AUC = 0.97 in distinguishing vEDS from general population and hEDS
 - Effectively identifies known vEDS-specific facial features

Use Cases

Dilated Aorta

Aortic Dissection

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