Three *potential* models for the **automated detection** of Mowat-Wilson and Noonan syndrome from facial features.

Machine learning for detecting congenital conditions

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1 Introduction

- A logistic regression model using manual delineation of features currently exists.
- This manual approach is subjective and resource intensive.
- In this study, three less labour intensive supervised training models were tested.

2 Methods

Anomaly detection (screening)

- CNN[1] Autoencoder trained on non-syndrome images.
- Density and reconstruction error distributions used to determine thresholds.

Logistic regression using facial coordinates

- Automated detection of facial features (*Python dlib library*).
- Standardised coordinates used in logistic regression model.
- Separate models for Mowat-Wilson and Noonan syndrome.

Classification using a CNN

- CNN learns distinguishing features.
- Activation layers show which features are more prominent such as eyes, nose and mouth.

3 Results

- Automated delineation and computer vision are viable alternatives to manual methods.
- CNN models are more accurate and stable.
- Anomaly detection shows much promise, as it only relies on images of non-syndrome subjects for training which are more readily available.

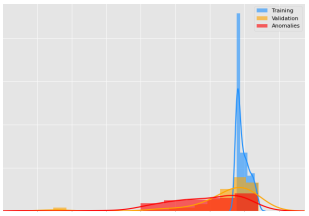
[1]Convolutional neural network

Additional information

	Mowat-Wilson	Noonan
Anomaly detection	78% (± 6%)[1]	
Logistic regression	73% (± 8%)	68% (± 11%)
CNN classifier	85% (± 2%)	82% (± 5%)

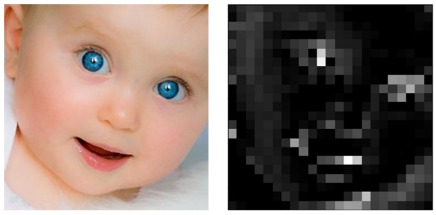
[1]Only used syndrome images

Density distribution for anomaly detection



Automated detection of facial features

Activation map showing detection of eyes/nose/mouth



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