Research study

Proband information for PEDIA study
(Prioritization of Exome Data by Image Analysis)

We are inviting you to take part in a research study. Before you decide whether to participate or not it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and to decide whether or not you wish to be involved.

Due to certain clinical findings your doctor diagnosed you with a genetic disorder or you are carrier of a pathogenic mutation. The clinicians analyzed your genes and also took all your special features into account. Facial abnormalities („gestalt“) can help clinicians finding the right diagnosis.

Here, we would like to use software for an automated, reproducible and fine-grained biometric analysis of frontal face photographies. The software we develop imitates a neuronal network and it is trained on images of probands and their healthy family members to compute the so called “similarity score” to known syndromes. In addition we collect genetic and clinical information. Your diagnosis, the similarity score and the genetic information are used to train the neural network. Our study aims at evaluating whether the additional information of automated image analysis improves the diagnostic yield in molecular genetics. We aim to apply this technology in exome or genome data analysis.

Personal data such as name, date of birth, address and clinical data will be encrypted and stored in separate databases. The research group of Peter Krawitz, MD, is responsible for the data analysis of the pseudonymized clinical data. Only the person taking the consent and further individuals named by the participant have access to the personal data. Results of the analysis of the prioritization algorithms will not allow drawing conclusions regarding who participated in the study.

Participating in the study is free of charge and participants are not entitled to any compensation of any kind. All participants have the right to obtain information about how their data is used and informed consent can be withdrawn at any time. In this case all data will be removed from the databases.

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The study aims at improving the diagnostic procedure for genetic disorders and will shorten the diagnostic odyssey for future patients. However, participating in the study will not have an immediate advantage for the participant.

Additional information:

Explanation of data processing in the example of DeepGestalt.

Computer assisted image analysis applies methods of artificial intelligence. First, parts of the face (such as eyes, nose, mouth, etc.) are automatically identified. Then these parts are compared to known syndromic phenotype models (such as Down Syndrome, Mabry Syndrome, etc.). These comparisons result in the “similarity score”. A detailed explanation of the method is described in the manuscript entitiled: “DeepGestalt-Identifying Rare Genetic Syndromes Using Deep Learning“ (https://arxiv.org/pdf/ 1801.07637.pdf).

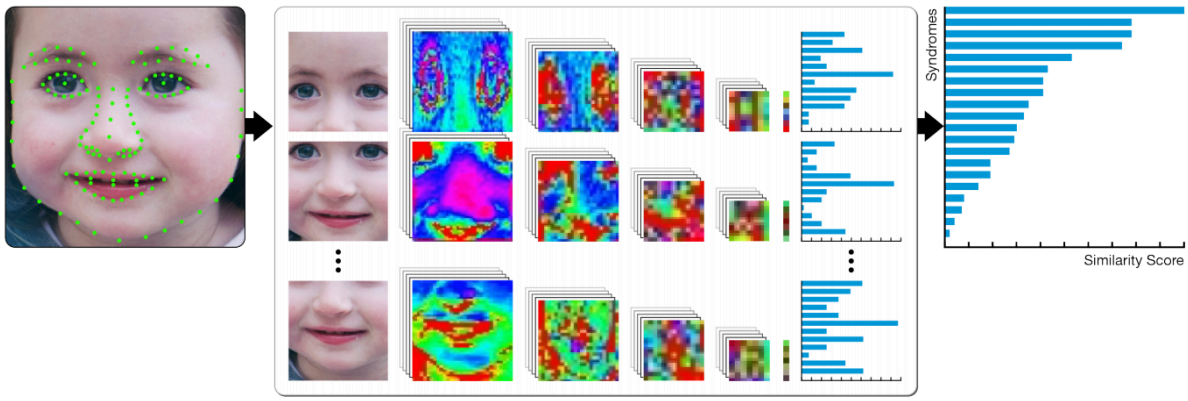


Figure 1 - Results of the facial analysis by Deep Gestalt are stored as similarity scores

We use the similarity scores obtained from many probands to train the neural network to improve prediction of the gene affected in patients with rare syndromes.

Visualization of data interpretation in the example of Face2Gene.

Clinicians can apply our results on the platform Face2Gene to evaluate patient information and prediction.

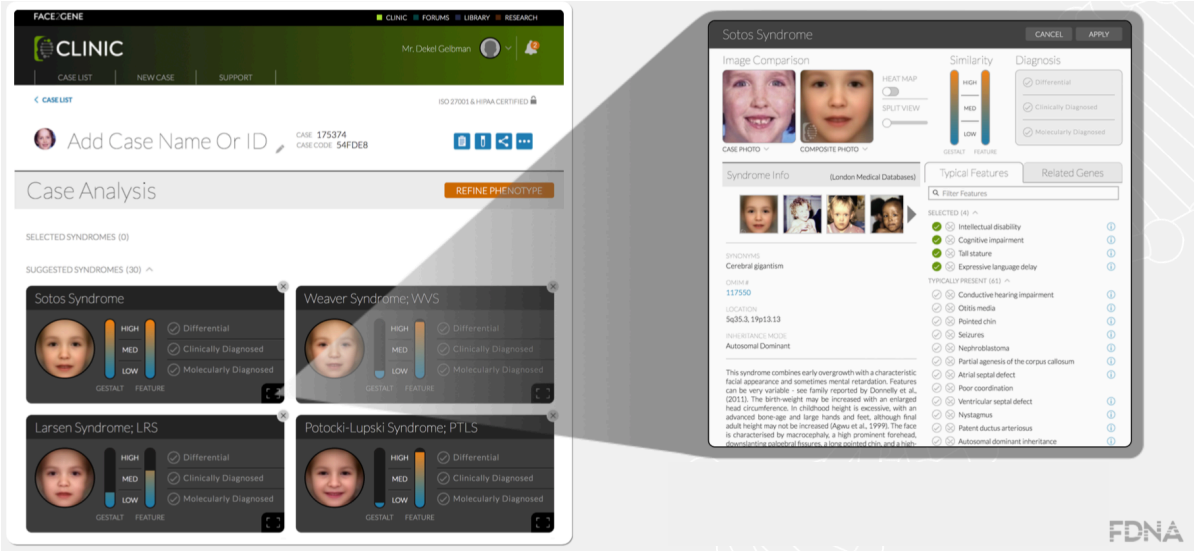
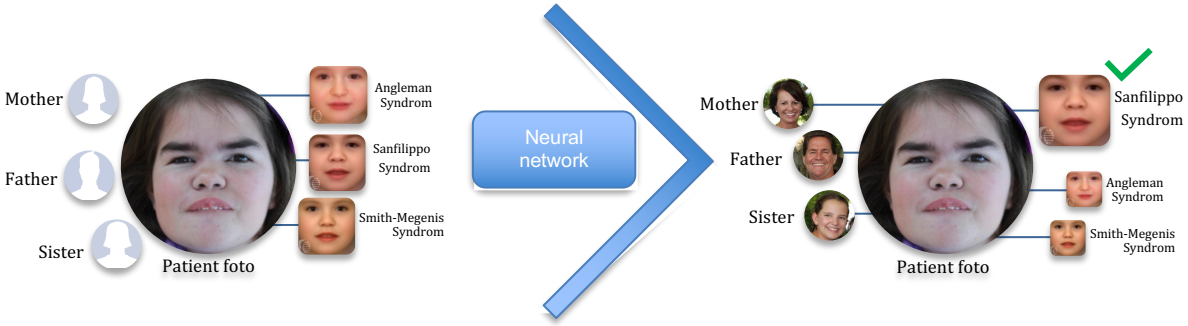


Figure 2 - A clinician can compare patient information with predicted syndromes.

A schematic example for usage of family images for improvement of syndrome prediction.

We want to increase the prediction of the syndrome in affected probands by training a neural network with frontal face photos of healthy family members. Thereby we want to reduce familial similarity and increase syndromic features.



If you have further questions about the research study, please do not hesitate to contact us via email, telephone or mail.

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