COMPUTER-AIDED FACIAL ANALYSIS AS A TOOL TO IDENTIFY PATIENTS WITH SILVER-RUSSELL AND PRADER-WILLI SYNDROME

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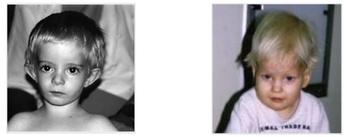
INTRODUCTION

Genetic syndromes often show suggestive facial features that provide clues for the diagnosis. Considering the high number of genetic syndromes and the possible overlap of some features, memorizing facial gestalt is a challenging task for clinicians.

DeepGestalt technology, and its app Face2Gene, can help in the diagnosis of genetic syndromes by analyzing the features detected in one or more facial images of affected individuals.

AIM

To assess the clinical utility of the Face2Gene app in diagnosing patients with Silver-Russell syndrome (SRS) and Prader-Willi syndrome (PWS).



On the left: SRS patient. On the right: PWS patient. Source: Face2 Gene database (FDNA, Inc., Boston, MA, USA)

METHODS

- 23 pediatric patients with clinically or genetically diagnosed SRS and 29 pediatric patients with genetically confirmed PWS were enrolled between December 2020 and April 2021.
- One frontal picture of each patient was acquired.
- Top-1, top-5 and top-10 sensitivities were analyzed.
- Correlation with the specific genetic diagnosis was investigated.
- When available, pictures of the same patient at different ages were compared.

CONCLUSIONS

Face2Gene app can be a useful tool to support clinicians in the diagnosis of SRS and PWS.

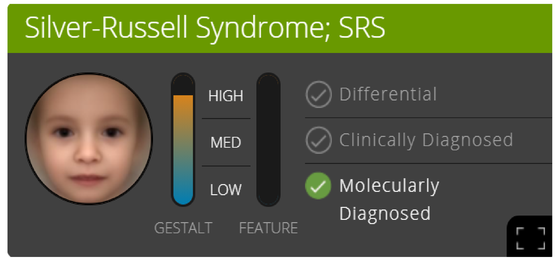
The sensitivity is higher in PWS patients and was comparable throughout all age ranges.

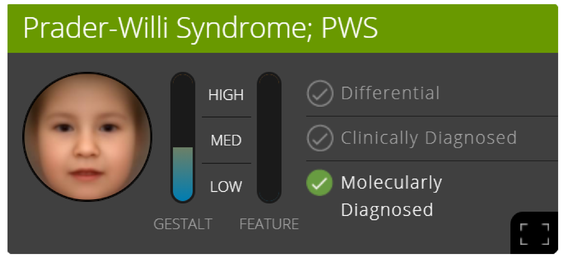
In SRS patients, the app performed better in the younger age group, probably due to more pronounced facial features at a younger age.

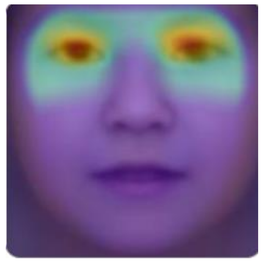
RESULTS

- In the SRS cohort Face2Gene showed a top-1, top-5 and top-10 sensitivity of 39%, 65% and 91% respectively.
- 2 patients (8.7%) were not correctly diagnosed by the app; this did not change after adding clinical features.
- 13 SRS patients (56.5%) had SRS suggested with high level of probability.
- SRS was the first syndrome suggested in 44% of genetically confirmed patients vs 28% of clinically diagnosed patients (p=0.49).
- Face2Gene performed better in younger patients: for all patients SRS was suggested as top-1, albeit with variable degree of probability.

- In the PWS cohort the top-1, top-5 and top-10 sensitivity were 76%, 97% and 100% respectively.
- 21% of PWS patients had PWS suggested with high level of probability.
- PWS as first suggestion was equally distributed throughout the age range.
- PWS was suggested as top-1 in 60% of patients genetically diagnosed with mUPD(7) and in 83% of patients presenting with paternal deletion of chromosome 15q11-13 (p=0.17).







On the top: SRS suggested with high probability in one of the analyzed patients, molecularly diagnosed.

On the bottom: PWS suggested with medium probability in one of the analyzed patients, molecularly diagnosed.

On the right the heat maps marking the areas of resemblance with a composite face resulting from the photos of affected individuals uploaded in the database. The intensity of the color scale goes from light blue to red, where the red areas are more suggestive for the analyzed syndrome. No pictures of the enrolled patients are shown.

REFERENCES

Gurovich Y. et.al Identifying facial phenotypes of genetic disorders using deep learning. Nat Med. 2019;25:60-64.

Azzi S. et al. A prospective study validating a clinical scoring system and demonstrating phenotypical-genotypical correlations in Silver-Russell syndrome J Med Genet 2015;52:446–453.

Goldstone AP. et al. Recommendations for the diagnosis and management of Prader-Willi syndrome. J Clin Endocrinol Metab. 2008;93:4183-97

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