Evaluating the Benefit and Efficiency of Using Computer-Aided Facial Dysmorphology Novel Analysis in the Clinic

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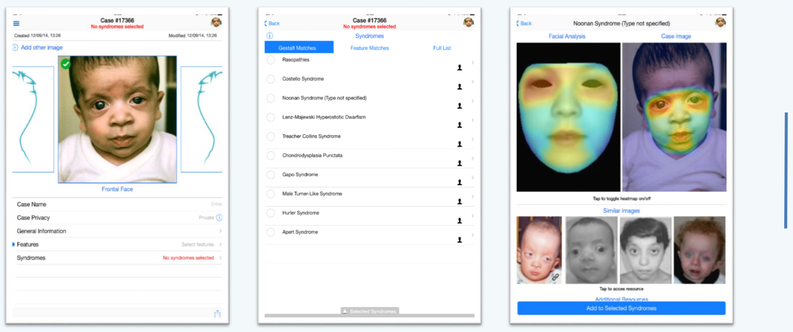
INTRODUCTION

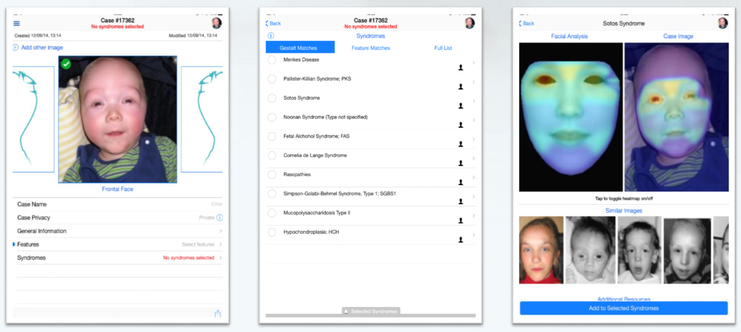
We demonstrated previously that computer-aided facial analysis technology (developed by FDNA) is accurate in distinguishing targeted selected syndromes by processing 2D facial images. The computer-generated analyses were able to produce results comparable with those of human experts in dysmorphology. In this study, we evaluate the performance of the FDNA technology by submitting for analysis a set of images of molecularly diagnosed individuals.

FACE2GENE

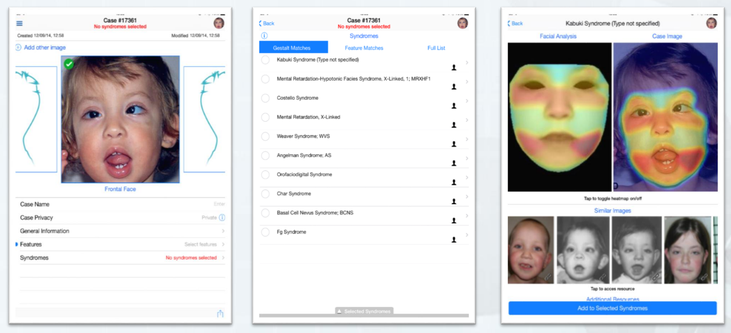
Figure 1. *Sample analysis applied to public images collected from the web (NOT the images used in the study)*



Source: "Clinical manifestations of Noonan syndrome", http://openi.nlm.nih.gov/



Source: "What is Sotos Syndrome", http://sotossyndrome.org/sotos-syndrome



Source: http://en.wikipedia.org/wiki/Kabuki_syndrome

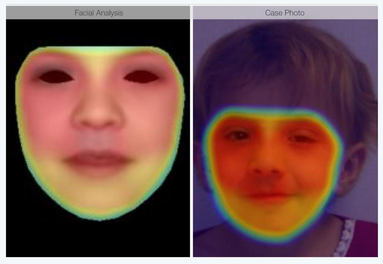
METHODS

Forty six images of dysmorphic children were submitted through the Face2Gene mobile or online interfaces developed by FDNA. No clinical information other than the facial image and molecular diagnosis was specified. Cases with syndromes not currently supported by the technology (15) were excluded, resulting in a test group of 31 cases. A match was considered positive where the diagnosed syndrome was listed among the top twenty diagnoses suggested by the technology using FDNA's proprietary "gestalt analysis".

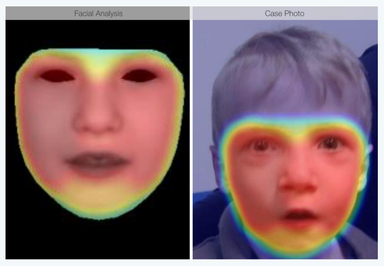
RESULTS

The Facial Dysmorphology Novel Analysis technology analyzed 31 cases which had a molecular diagnosis indication. In 26/31 (84%) patients there was a positive match between the confirmed diagnoses and one of the first 20 syndromes suggested by the technology. In 19/31 (61%) patients, a positive match was identified within the first 5 syndromes.

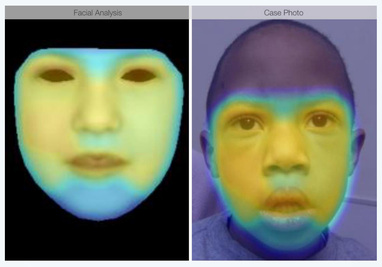
Figure 2. *Facial analysis of selected patients from this study showing the molecularly-confirmed diagnosis and FDNA match ranking using the "gestalt analysis" of this technology. Heat maps displaying the most distinguishable regions of the syndrome are drawn over the patient's facial images and compared to the matched syndrome's masks as suggested by the FDNA analysis.*



18674
Noonan syndrome
Rank: 1

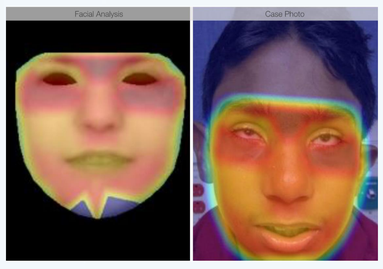


18760
22q11.2 deletion
Rank: 1

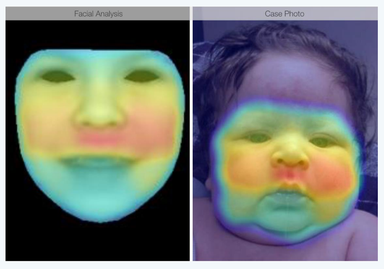


18746
22q11.2 deletion
Rank: 3

18683
Stickler syndrome
Rank: 11

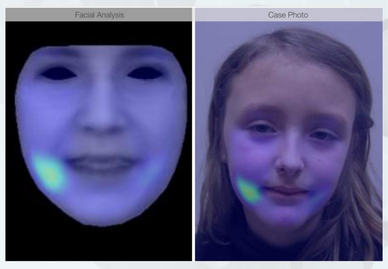


18754
CHARGE syndrome
Rank: 1



18862
Williams syndrome
Rank: 2

18943
CHARGE syndrome
Rank: 4



18755
Marfan syndrome
Rank: 30

CONCLUSIONS

The technology is able to distinguish dysmorphic syndromes by processing 2D facial images with 84% accuracy when checked against a diagnosis which is molecularly confirmed.
Routine use of the FDNA technology in the clinic could assist clinicians in creating a more accurate differential diagnosis and shorten the time for achieving molecular confirmation of rare disease patients.

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