Efficiency of Computer-Aided Facial Dysmorphology Analysis in the Medical Genetics Clinic

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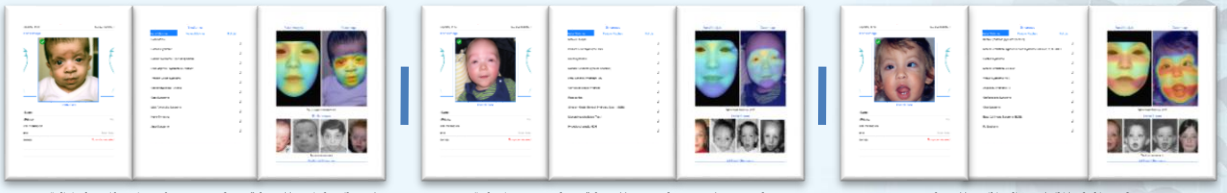
FELLOWSHIP AWARD ESHG 2015 Glasgow

INTRODUCTION

Facial Dysmorphology Novel Analysis (FDNA) technology has recently been introduced to clinicians through Face2Gene. The precision of syndrome detection when utilizing the automated facial recognition software was previously shown to be comparable to the accuracy of dysmorphology experts (Basel et al., ASHG 2014). In this study, the performance of FDNA technology was assessed by testing 2D facial images of patients with molecularly confirmed diagnoses and comparing the actual syndrome to the suggested syndromes.

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

Frontal facial images of patients were collected by the Department of Medical Genetics of Istanbul Medeniyet University. Images of cases with; a molecularly confirmed diagnosis, syndromes currently supported by FDNA, and well documented dysmorphic features were used in this study if the patient had given consent.

12 patients who were affected by 8 syndromes (Table. 1) were analyzed by FDNA technology. When the correct syndrome was listed in the top ten most likely syndromes, it was considered a positive match.

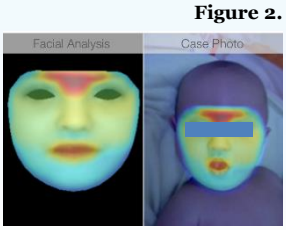
Table 1. *Syndromes included in this study and their OMIM entry*

Syndromes name	OMIM
Prader-Willi Syndrome; PWS	176270
Wolf-Hirschhorn Syndrome; WHS	194190
Rubinstein-Taybi Syndrome	180849
Chromosome 22q11.2 Deletion Syndrome	188400
Down Syndrome	190685
Achondroplasia; ACH	100800
Williams-Beuren Syndrome; WBS	194050
Trichorhinophalangeal Syndrome	190350

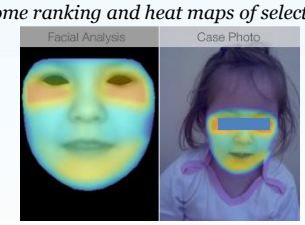
RESULTS

The Facial Dysmorphology Novel Analysis technology analyzed 12 cases. In 10/12 (83%) patients there was a positive match between the confirmed diagnoses and the list of syndromes retrieved by Face2Gene. The average rank of the correct syndrome was 3.8 (1 being the most similar and 10 being the least).

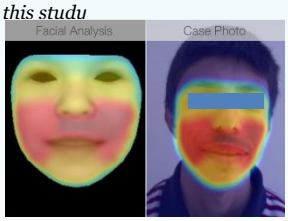
Figure 2. *Resulting syndrome ranking and heat maps of selected patients from this study*



Case #23311
Wolf Hirschhorn Syndrome WHS
Ranked 1st in the FDNA technology results



Case #23310
Prader-Willi Syndrome; PWS
Ranked 4th in the FDNA technology results



Case #23456
Williams-Beuren Syndrome; WBS
Ranked 1st in the FDNA technology results

Heat maps display the most distinguishable facial regions of the syndrome; red colours illustrate higher similarity while blue colours illustrate absence of similarity. The molecularly-confirmed diagnosis of the patient is listed along with the mask for that syndrome.

CONCLUSIONS

We believe that routine use of Facial Dysmorphology Novel Analysis in the clinic as a search and reference tool will assist clinicians, particularly medical geneticists, in the process of reaching a differential diagnosis and shorten the time and the cost for achieving molecular confirmation especially for rare disease patients.

This study was supported by the Research Fund of Istanbul Medeniyet University Project #: T-BEK-2015-709

FDNA Accessible Genetics