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

Chad Haldeman-Englert, MD, FACMG,1 Lior Wolf, PhD2,3
1 Department of Pediatrics, Section on Medical Genetics, Wake Forest School of Medicine, Winston-Salem, NC
2 Tel Aviv University, Tel Aviv, Israel
3 FDNA Ltd., Herzliya, Israel

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.

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.

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.