Is a Computer-Based Facial Dysmorphology Novel Analysis Ready for the Clinic?
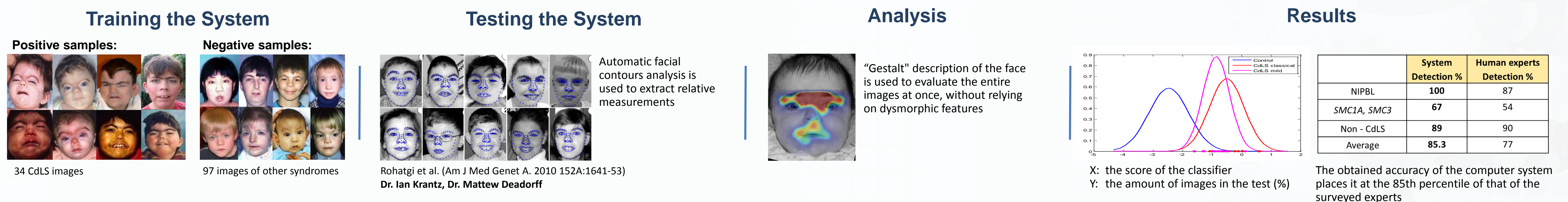
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Previously, we were able to demonstrate that the facial dysmorphology novel analysis technology was successful in recognizing the facial dysmorphology associated with targeted selected syndromes by processing 2D facial images. In this study we investigated the performance of the Facial Dysmorphology Novel Analysis technology by analyzing a random set of images of dysmorphic individuals affected with a random variety of syndromes.

PREVIOUS WORK – SPECIFIC SYNDROMES

Example of performance -- Computer-aided facial recognition of Cornelia de Lange syndrome: a comparison to the recognition by human experts

Training the System

Positive samples:

Negative samples:

34 CdLS images

97 images of other syndromes

Testing the System

Automatic facial contours analysis is used to extract relative measurements

Rohatgi et al. (Am J Med Genet A. 2010 152A:1641-53)
Dr. Ian Krantz, Dr. Mattew Deadorff

Analysis

"Gestalt" description of the face is used to evaluate the entire images at once, without relying on dysmorphic features

Results

X: the score of the classifier
Y: the amount of images in the test (%)

	System Detection %	Human experts Detection %
NIPBL	100	87
SMC1A, SMC3	67	54
Non - CdLS	89	90
Average	85.3	77

The obtained accuracy of the computer system places it at the 85th percentile of that of the surveyed experts
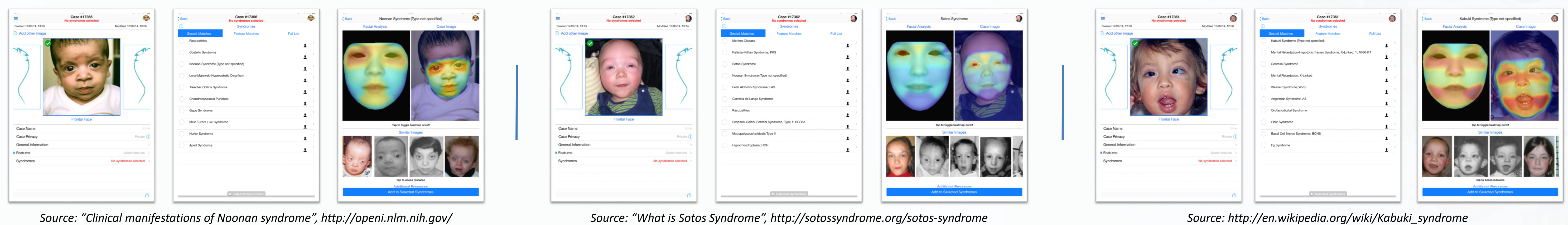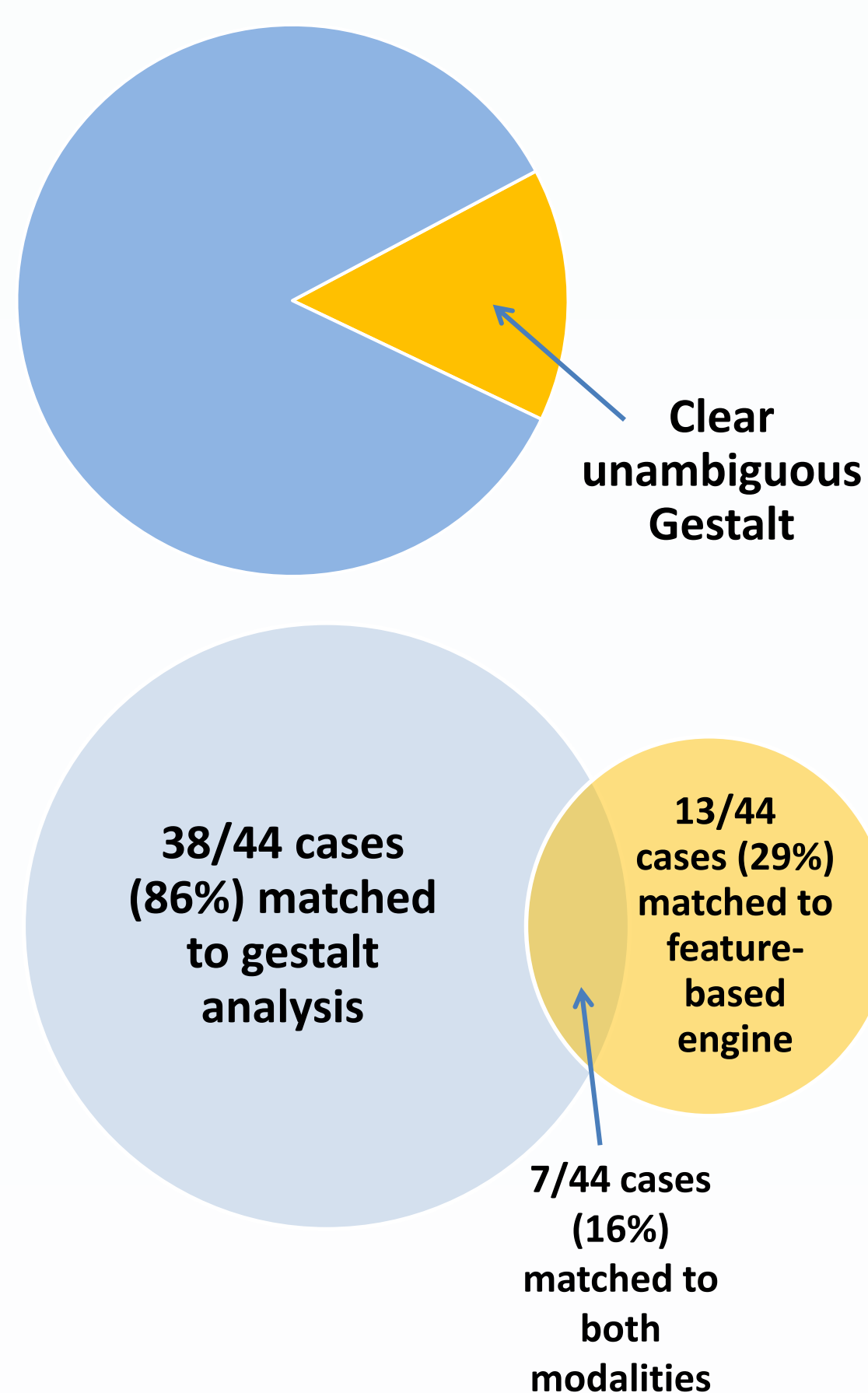
METHOD

The images in this study were submitted by more than a hundred clinical geneticists (users of the Face2Gene mobile application). Images of individuals suspected of being affected with rare chromosomal imbalances were excluded from this study. 350 images were chosen randomly and reviewed (without any additional information) independently by a single human geneticist experienced in dysmorphology (LBV). The results from the evaluation by the human geneticist were compared to gestalt and feature (based on a list of HPO terms representing facial features automatically detected by the system as the search criteria) search results returned by Face2Gene. A "Positive Match" means a syndrome determined by the human geneticist and listed among the ten highest ranked Face2Gene results.

FACE2GENE™

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

RESULTS

Clear unambiguous Gestalt

In 52/350 cases (15%), the human expert was able to clearly recognize and determine the presence of a specific genetic syndrome, based on gestalt only.

38/44 cases (86%) matched to gestalt analysis

13/44 cases (29%) matched to feature-based engine

7/44 cases (16%) matched to both modalities

In 44 of these cases (85%), there was a Positive Match between the system and the human geneticist. 38 cases of which, appeared in the gestalt search results, 13 cases appeared in the feature-based search results, and 7 cases appeared in both. Only 8/52 (15%) cases were recognized by the human expert, but not by the system.

It is unknown in how many cases the system recognized the "true" syndrome when the human expert did not, since the vast majority of the cases are submitted without molecular confirmation, other than 2 cases in which a molecular confirmation was indicated, and the system suggested the correct syndrome, while the expert did not.

CONCLUSIONS AND FUTURE WORK

30%–40% of genetic disorders manifest craniofacial abnormalities. Facial analysis software can successfully assist medical professionals of different specialties in the research and investigation of multiple genetic syndromes characterized by dysmorphic features. Possible future applications may include usage of facial analysis software to complement molecular studies, such as whole exome sequencing, by automatic phenotyping .

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