Highlighting phenotypic differences of Down Syndrome and Turner (45X) Syndrome Patients from different ethnic groups using computer-aided facial dysmorphology analysis

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Introduction:

As computer-aided facial dysmorphology analysis technology becomes more frequent in the clinic, the issue of phenotypic syndrome variation based on ethnicity has been highlighted. In this study, we compare the ability of this technology to differentiate individuals with Down or Turner Syndrome and individuals without these syndromes in 3 ethnic population groups: Congolese African, Caucasian and Latin-American.

Methods:

47 frontal images of Latin-American (LAT) children affected with cytogenetically diagnosed Down Syndrome (DS) and 17 frontal images of Latin-American children affected with cytogenetically diagnosed Turner Syndrome (TS) were compared using the Facial Dysmorphology Novel Analysis (FDNA) technology (Fig.2) with 4 groups of facial images: Latin-American and Caucasian (CAU) children affected with other syndromes, Latin-American and Caucasian children not affected (Normal) by any syndrome. 19 frontal images of Congolese African (CON) children affected with cytogenetically diagnosed Down Syndrome and 51 frontal images of Caucasian children affected with DS (Table1) were compared with non-affected children from their own ethnic group.

The mean area under the curve (AUC) was used as a means of comparison between the cohorts (Table2.) The separation quality between different populations was evaluated by measuring the area under the curve (AUC) of the ROC (Receiver Operating Characteristic) curve plotting the true positive rate as function of the false positive rate (Fig.1)

Figure 1

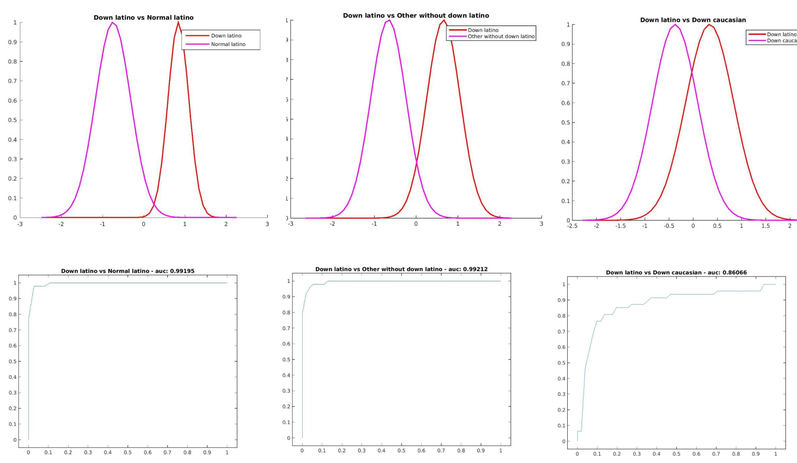

Figure 1: *Histograms and ROC curves describing level of separation between of the cohorts in this study, illustrating higher recognitions capabilities between non-affected and syndromic individuals, than between the different ethnic groups studied.*

Figure 2

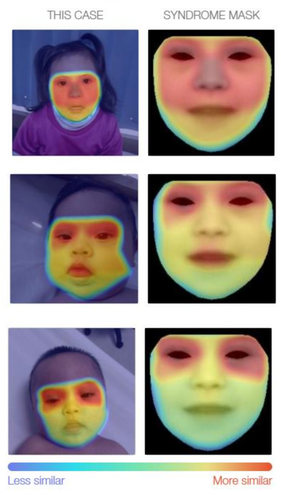

Figure 2: *Resulting heat-maps and DS masks of selected patients from this study. Heat-maps display the level of similitude between the patient photo and syndrome mask. (Courtesy of FDNA)*

Table 1

Cohort	No. of images
Down LAT	47
Normal LAT	37
Other syndromes LAT	54
Down CAU	51
Down CON	19
Normal CON	54
Turner LAT	17
Turner CAU	13
Normal CAU	38

Table 2

Comparisons conducted	Mean AUC obtained
Down LAT vs Normal LAT	0.99
Down LAT vs Other syndromes LAT	0.99
Down LAT vs Down CAU	0.86
Down CON vs Normal CON	0.96
Turner LAT vs Normal LAT	0.975
Turner LAT vs Other syndromes LAT	0.943
Turner CAU vs Normal CAU	0.99

Table 1 and 2: *Description of test cohorts and comparisons conducted*

Results:

The mean AUC of the comparison between DS Latin-American with not-affected LAT was 99% compared to the comparison between LAT DS with CAU DS (86%) illustrating **higher recognition cap abilities of not-affected vs. syndromic individuals, than between the different ethnic groups**. The mean AUC of the comparison between LAT DS and non-affected LAT was 97% compared to the mean AUC of the comparison of LAT DS with LATs having other syndromes (94%) illustrating the **higher recognition capabilities of syndromic versus non-affected as opposed to recognition between different syndromes within one ethnicity**. A parallel comparison between syndromic and non-affected individuals in the Caucasian group yielded similar results. A slightly lower mean AUC of 96% resulted from the comparison between African Congolese DS and African Congolese non-affected (96%) than the parallel comparisons with different ethnic groups listed above (99% for both Latin-American and Caucasian), due probably to the smaller sample of images of African children in the database.

Conclusions:

Our preliminary results show that the deep phenotyping provided by computer-aided facial recognition technologies is preserved across three ethnic groups, despite the presence of ethnic-specific features. The challenge of recognizing between different syndromes remains similar across the ethnicities tested. Larger study samples and additional comparative studies are needed.