Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB045. Studying the probabilities of Down syndrome recognition in Thai children using de-identified computer-aided facial features analysis

Nattariya Vorravanpreecha, Tanayoot Lertboonnum, Rungrote Rodjanadit, Pak Sriplienchan, Kitiwan Rojnueangnit

Department of Pediatrics, Faculty of Medicine, Thammasat University, Pathumthani, Thailand

Background: Craniofacial dysmorphism plays a major part in the evaluation of many genetic syndromes. Facial pattern recognition has been typically used by clinicians before sending the patients' blood for specific diagnostic tests. However, the number of genetic syndromes is enormous, and the specific facial features are difficult to memorize for all syndromes. Therefore facial dysmorphology novel analysis (FDNA) technology, innovative software, was developed to help clinicians recognize probable genetic syndromes from patients' facial gestalts by ranking. Ethnic differences might have a major effect on patients' facial features. The FDNA database mainly consists of Caucasian patients; therefore, the probability recognition for Asian patients may be limited. The aim of this project is to test the software's recognition probability (sensitivity) on Thai Down Syndrome (DS) children compared to Thai non-Down syndrome (non-DS) children (specificity).

Methods: Thirty Thai children with DS, genetically confirmed diagnosis, aged between 1 month and 15 years, were recruited in the study group. The control group were 140 Thai non-DS children, either normal or having other genetic syndromes. Frontal, and when possible side, photographs were obtained from all participants. Images were uploaded to FDNA software, at least 2 pictures for each participant. Positive probability recognition for DS was if DS was reported in the first 10 ranks.

Results: Our study showed all 30 Thai-DS children were recognized as DS in the first 10th ranks, and 27 cases were recognized as DS in the first 5th ranks. Therefore the sensitivity of this app was 100% or 90% if cut point was the first 10th or 5th ranks, respectively. In group of Thai non-DS, 12 were recognized as DS, so the specificity was 89.2% or 91.36% if the cut the first 10th or 5th ranks, respectively.

Conclusions: This application was proved to be effective in recognition of Thai-DS children. However, the specificity result was probably due to the limitation of Face2Gene's database, which has being compose of mostly Caucasians. Therefore, misdiagnosis by ethnicity can occur.

Keywords: Down syndrome (DS); craniofacial dysmorphism; facial dysmorphology novel analysis (FDNA)

doi: 10.21037/atm.2017.s045

Cite this abstract as: Vorravanpreecha N, Lertboonnum T, Rodjanadit R, Sriplienchan P, Rojnueangnit K. Studying the probabilities of Down syndrome recognition in Thai children using de-identified computer-aided facial features analysis. Ann Transl Med 2017;5(Suppl 2):AB045. doi: 10.21037/atm.2017.s045

