

Clinical Genetics

AB119. Computer-aided facial recognition of Chinese individuals with 22q11.2 deletion-algorithm training using NIH atlas of human malformation syndromes from diverse population

Gary Tsz Kin Mok, Brian Hon-Yin Chung

Department of Paediatrics & Adolescent Medicine, Li Ka Shing Faculty of Medicine, the University of Hong Kong, Hong Kong

Background: 22q11.2 deletion syndrome (22q11.2DS) is a common genetic disorder with an estimated frequency of 1/4,000. It is a multi-systemic disorder with high phenotypic variability. Our previous work showed substantial under-diagnosis of 22q11.2DS as 1 in 10 adult patients with conotruncal defects were found to have 22q11.2DS. The National Institute of Health (NIH) has created an atlas of human malformation syndrome from diverse populations to provide an easy tool to assist clinician in diagnosing the syndromic across various populations. In this study, we seek to determine whether training the computer-aided facial recognition technology using images from ethnicity-matched patients from the NIH Atlas can improve the detection performance of this technology.

Methods: Clinical photographs of 16 Chinese subjects with

molecularly confirmed 22q11.2DS, from the NIH atlas and its related publication were used for training the facial recognition technology. The system automatically localizes hundreds of facial fiducial points and takes measurements. The final classification is based on these measurements, as well as an estimated probability of subjects having 22q11.2DS based on the entire facial image. Clinical photographs of 7 patients with molecularly confirmed 22q11.2DS were obtained with informed consent and used for testing the performance in recognizing facial profiles of the Chinese subjects before and after training.

Results: All 7 test cases were improved in ranking and scoring after the software training. In 4 cases, 22q11.2DS did not appear as one possible syndrome match before the training; however, it appeared within the first 10 syndrome matches after training.

Conclusions: The present pilot data shows that this technology can be trained to recognize patients with 22q11.2DS. It also highlights the need to collect clinical photographs of patients from diverse populations to be used as resources for training the software which can lead to improvement of the performance of computer-aided facial recognition technology.

Keywords: 22q11.2 Deletion Syndrome; computer-aided facial recognition technology; Chinese

doi: 10.21037/atm.2017.s119

Cite this abstract as: Mok GT, Chung BH. Computer-aided facial recognition of Chinese individuals with 22q11.2 deletion-algorithm training using NIH atlas of human malformation syndromes from diverse population. *Ann Transl Med* 2017;5(Suppl 2):AB119. doi: 10.21037/atm.2017.s119