Clinical Genetics

AB085. Diagnosis of Prader-Willi syndrome using computer-aided facial dysmorphism analysis in Thai patients

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Background: Prader-Willi syndrome (PWS) is a genetic disorder with multisystemic involvement. Age-dependent subtle facial characteristics are noted, making it is difficult for physicians to recognize the syndrome. Computer-aided facial dysmorphism analysis, the Facial Dysmorphology Novel Analysis (FDNA), is an automated facial detection program associated with possible syndromes using 2D-photo. The FDNA was developed using pictorial data mainly from Caucasian individuals. This study intended to determine diagnostic yields of FDNA for Thai PWS patients and to compare the results before and after program training (PT).

Methods: Patients with molecularly confirmed PWS were enrolled. Frontal facial photos were obtained and randomly divided into 2 groups: (I) training group and (II) test group. Prior to training the program, all photos were analyzed by the FDNA (version 17.4.1). PWS ranks (PWSR) in 30 syndrome matching and scores of confidence level were reported. After PT, photos of each case from the test group were reanalyzed and the results were compared to the data obtained before PT.

Results: Twenty-five patients with 28 photos were included. Their age range was between 9 months and 25 years. Prior to PT, 29% (8/28) of photos was not listed in PWSR (3/9 in the test group and 5/19 in the training group). For those 25% (7/28) whose photos were ranked in the first place, their age was >2 years. After PT, 100% (9/9) of photos in the test group were redirected to the top-five ranking and 44% (4/9) in the first place. The PWSR and scores of confident levels were totally improved after PT. Age of PWS patient may affect the performance of the program and it is now an ongoing study.

Conclusions: Pictorial data of PWS patients from various ethnicities are important factor, leading to improvement of the program which results in a higher diagnostic yield.

Keywords: Prader-Willi syndrome (PWS); facial dysmorphism; computer-aided

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