Clinical Genetics

AB052. Application of facial dysmorphology analysis technology (Face2gene) in Korean rare genetic diseases

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Background: Since 2011, FDNA (facial dysmorphology novel analysis) has started to develop a technology called the Face2gene which is a suite of phenotyping applications that facilitate comprehensive and precise genetic evaluations. The Face2gene has been collected and studied mainly in USA and Europe, and it is limited to application to Asian people. Therefore, this study was conducted to evaluate the usefulness of the Face2gene in Korean patients who were already molecularly diagnosed.

Methods: The study was carried out at four university hospitals. After obtaining the consent of the patient's parents, the photographs were taken and analyzed by the Face2gene. The inclusion criteria were molecularly diagnosed children and syndrome characterized by facial dysmorphology.

Results: A total of 46 patients were recruited and reviewed. The patients were affected by 12 syndromes. In 43/46 (93.4%) patients, the confirmed diagnoses were listed in the top 30th. In 13/46 (28.2%) patients, the diagnoses were listed for the top 1st. Down syndrome had the highest positive matching rate (33%) within the top 1st ranking.

Conclusions: The Face2gene system has a high possibility to match the molecularly confirmed syndrome by only analyzing facial gestalts of the patients. The Face2gene system can be used as an assistant tool in the developmental medicine and genetic clinic.

Keywords: Dysmorphology; rare disease; genetic medicine

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