Abstracts *(Oral Presentation)*

Comparison of the Accuracy in Diagnosis of 22q11.2 Deletion Syndrome and Williams Syndrome by Facial Photos Between De-identified Facial Program Features and Clinicians

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Abstract

Introduction: Since there are more than 6,000 genetic syndromes, the recognition facial pattern may present a challenge for general clinicians. 22q11.2 deletion syndrome (22q11.2 DS), aka DiGeorge syndrome, and Williams syndrome (WS) are two genetic syndromes, with a not too low prevalence. However, underdiagnosis is unsurprising, given that genetic testing is only for patients who are suspected of any genetic syndromes. These two syndromes are different, but they share some common phenotypes, such as global developmental delay, cardiac defects and some subtle facial dysmorphisms. Therefore, if there is any tool to help clinicians recognize any genetic syndrome, the diagnosis will be more accurate.

Objectives: We would like to evaluate the accuracy of the Face2Gene application in suspected 22q11.2 DS and WS, to compare the accuracy between the app and clinicians, and if the accuracy is best when combining clinicians' decision and the application.

Methods: Our participants include a total of 64 two-dimensional children's frontal facial photos, divided into 3 groups of 22q11.2 DS (16 photos), WS (16 photos) and unaffected (32 photos). We also have nine clinicians, divided into three group of interns, pediatric residents and pediatricians. The result demonstrates the accuracy in the app is higher than in clinicians, especially when comparing between two diseases. However, the accuracy of clinicians is higher when comparing between 22q11.2 DS and unaffected control. The accuracy is higher when clinicians use the app combined with their own decision.

Conclusions: The application will be a great tool to help clinicians to recognize facial features of genetic syndromes, before ordering specific tests to confirm the definite diagnosis.

Keywords: 22q11.2 Deletion syndrome, Williams syndrome, Accuracy, Face2Gene
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