Abstracts *(Poster Presentation)*

Genetic Diagnosis for Adult Patients at a Genetic Clinic

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Abstract

Introduction: Clinical utility of genetic testing has rapidly increased in the past decade to identify the definite diagnosis, etiology, and specific management. The majority of patients receiving genetic testing are children, while few adult patients have the tests. There are several barriers for genetic tests in adult patients; barriers may arise from either patients or clinicians who care for adult patients.

Objectives: The purpose of this study is to realize the detection rate and the benefits of genetic tests in adults.

Methods: A prospective study of 10 adult patients who were referred to a genetic clinic for a diagnosis during August 2020 - February 2021. Whole exome sequencing (WES) was pursued in all cases, and chromosomal microarray (CMA) was performed for 6 cases.

Results: The result is impressive; seven cases (70%) received the definite genetic diagnosis, four cases with known pathogenic variants in *KCNJ2, TGFBR1, SCN1A,* and *FBN1,* while the other three cases revealed novel likely pathogenic variants from in *GNB1, MID1,* and *DNAH9.* Two patients had abnormal CMA results.

Conclusions: This study demonstrates the success in genetic diagnosis in adult patients with intellectual disability and/or congenital anomalies. The advantage of the diagnosis is not only obtaining the diagnosis itself, but also relieving any doubt for the patient regarding any previous questionable diagnosis, guide for management, and recurrence risk in their children or family members. Therefore, this supports the value of genetic testing in adult patients.

Keywords: Adult genetics, Genetic clinic, Genetics test
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