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Genetic Risk Assessments Study on Epilepsy Individuals

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Epilepsy is a neurological disorder (central nervous system). Abnormal activity of brain causes seizures or unusual behavior, sensations, and sometimes loss of awareness. Epilepsy can affect males and females. Epilepsy is of two types of seizures. A generalized seizure affects the complete brain. Focal, or partial seizures, affect partial part of the brain. A mild seizure is difficult to recognize which lasts for few seconds during which you lack awareness.

Up to 40% of cases of rare neurological disorders with seizures are explained the cause of single variant rare genetic disease. To treat patients personalized therapies are in development with many of genes. In contrast to this the role of genetic factors plays a common form of epilepsy which has not guided in the disease management and treatment plans.

While GWAS (Genome-Wide Association Studies) identified few common variants in epilepsy genetically. They are challenging to quantify an individual patient's genetic risk based on variants (an indicator of the strength of an association). Moreover, analysing a single genetic risk variant does not take into consideration the effect other variants may have on overall epilepsy risk.

To address the clinical need, researchers calculated all known common genetic variants identified from large GWAS cohorts, includes more than 12,000 people with epilepsy and 24,000 healthy controls, the polygenic risk scores in more than 8,000 people with epilepsy and 622,000 population controls. By combining these effect sizes of thousands of common genetic variants, these scores are used to determine the risk in individual for epilepsy.

Polygenic risk scores are shown to predictive and diagnostic in a variety of medical conditions, including diabetes, myocardial infarction, and breast cancer. This is the first study to examine in the context of epilepsy. The research team presented the predictive power of the polygenic risk scores developed and performed well in this study.

We believe these findings will lay the groundwork for using genetic risk assessments in the clinic to diagnose common epilepsies and guide precision treatment earlier in the disease process," but additional research is necessary to find more accurate treatment for this epilepsy.

Epilepsy, affects about 1% of the U.S. population, and is typical to diagnose a patient after two unprovoked seizures. The two main epilepsy syndromes are generalized epilepsy (involves both cerebral hemispheres) and focal epilepsy (originates in one cerebral hemisphere), and they account for a combined 94% of incident epilepsies. This study analysis is the largest of epilepsy genetics to date.

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