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Is the Genetic Diagnosis of Epilepsy Useful in Clinical Practice?

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The genetic diagnosis of epilepsy has become feasible and should help the clinical practice of epilepsy. Epilepsy syndromes where genetic diagnosis is applicable are still limited in number though the list of such epilepsy syndromes including progressive myoclonic epilepsies (PME), Dravet syndrome and other familial idiopathic epilepsies is now growing. For the diagnosis of epilepsy, genotyping ensures clinical diagnosis without further invasive examinations and may predict emerging symptoms. Genetic diagnosis considerably helps genetic counseling and prognostication. Genotyping also may distinguish epilepsies from non-epileptic conditions such as febrile seizures and alternatively identify underlying neurometabolic disorders, some of which are totally curable. Genetic information seems useful for the treatment of epilepsy as well. Based upon the molecular pathomechanisms, some antiepileptic drugs can be avoided before such drugs aggravate symptoms. For example, some sodium channel blockers should be avoided in the treatment for Dravet syndrome resulting from mutations of genes encoding sodium channels. Furthermore, early and intensive measures based on the genotyping may improve the prognosis of intractable epilepsy. However, there is no sound evidence, so far, whether or not such early interventions according to the genetic diagnosis improve the prognosis of epilepsy. Nevertheless, accumulating data on the early treatment based on the genetic diagnosis should clarify the effect of early interventions on the prognosis of epilepsy. Furthermore, novel treatments based on the molecular pathomechanisms such as the "read-through" drugs to ameliorate premature stop codons would emphasize the usefulness of the genetic diagnosis in clinical practice.

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