Special Issue

Genetics of Epileptic Encephalopathies: From Gene Discovery to Clinical Diagnosis and Management Implications of Genetic Diagnoses

Message from the Guest Editors

Epileptic encephalopathy is characterized by cognitive dysfunction associated with ongoing epileptiform activity and refractory seizures. Epileptic encephalopathy is heterogenous, and there are likely more than 1000 genetic disorders associated with it. In recent years, NGS technologies such as targeted panels and exome sequencing have increased the genetic diagnosis in patients with epilepsy. Molecular genetic diagnoses and the type of underlying genetic disease (e.g., SCN1A, SCN2A, and KCNQ2 associated epilepsies or some of the inherited metabolic disorders) can guide physicians in the management of epilepsy. This Special Issue will focus on the genetic basis of epileptic encephalopathies, recently discovered genetic epilepsies, and treatable inherited metabolic disorders. We extend an invitation for reviews on the current state of genetics in epileptic encephalopathies, as well as original research articles on the discovery of genetic variations or mutations that could be used to distinguish clinically relevant disease or predict therapeutic efficacies.

Guest Editors

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Deadline for manuscript submissions

closed (30 September 2022)

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Message from the Editor-in-Chief

Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving field. There is a need for good quality, open access journals in this area, and the Genes team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised. Why not consider Genes for your next genetics paper?

Editor-in-Chief

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