Epilepsy pathogenesis

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The pathogenesis of epilepsy is complex and not completely understood, but it is believed to involve various factors, including genetics and environmental factors.

Genetics can contribute to the development of epilepsy. There are several genes that have been identified as being associated with the disorder, including genes that encode ion channels, neurotransmitter receptors, and other proteins involved in synaptic function. Mutations in these genes can alter the function of neurons and increase the likelihood of seizures.

Environmental factors can also play a role in the development of epilepsy. Brain injuries, infections, and other conditions that affect the brain can increase the risk of developing the disorder. Additionally, exposure to toxins, such as lead or mercury, can increase the risk of seizures.

The pathogenesis of epilepsy is also related to the mechanisms that regulate the excitability and inhibition of neurons in the brain. Excitatory and inhibitory neurotransmitters, such as glutamate and GABA, respectively, play a crucial role in this regulation. Alterations in the balance between excitatory and inhibitory neurotransmitters, such as increased glutamatergic activity or decreased GABAergic activity, can lead to excessive neuronal activity and seizures.

In addition to these factors, inflammation, oxidative stress, and other mechanisms can also contribute to the pathogenesis of epilepsy. Overall, the pathogenesis of epilepsy is multifactorial and involves complex interactions between genetic, environmental, and neurological factors.

Brain angiogenesis inhibitor 3 (ADGRB3/BAI3) belongs to the family of Adhesion G protein-coupled receptors. It is most highly expressed in the brain where it plays a role in synaptic function. Genome-wide association studies have implicated ADGRB3 in disorders such as schizophrenia and epilepsy.

Khan et al. investigated the cellular role of TAK1 in experimental epilepsy. C57Bl6 and transgenic mice with inducible and microglia-specific deletion of Tak1 (Cx3cr1CreER: Tak1fl/fl) were subjected to

the unilateral intracortical kainate mouse model of temporal lobe epilepsy (TLE). Immunohistochemical staining was performed to quantify different cell populations. The epileptic activity was monitored by continuous telemetric electroencephalogram (EEG) recordings over a period of 4 weeks. The results show that TAK1 was activated predominantly in microglia at an early stage of kainate-induced epileptogenesis. Tak1 deletion in microglia resulted in reduced hippocampal reactive microgliosis and a significant decrease in chronic epileptic activity. Overall, the data suggest that TAK1-dependent microglial activation contributes to the chronic epilepsy pathogenesis [1].

Genetics

Molecular cytogenetics and cytogenomic studies have made a contribution to genetics of epilepsy. Current genomic research is generally focused on the molecular genetic aspects (i.e. gene hunting, detecting mutations in known epilepsy-associated genes, searching monogenic causes of epilepsy). Nonetheless, chromosomal abnormalities and copy number variants (CNVs) represent an important part of genetic defects causing epilepsy. Moreover, somatic chromosomal mosaicism and genome/chromosome instability seem to be a possible mechanism for a wide spectrum of epileptic conditions. This idea becomes even more attracting taking into account the potential of molecular neurocytogenetic (neurocytogenomic) studies of the epileptic brain. Unfortunately, analyses of chromosome numbers and structure in the affected brain or epileptogenic brain foci are rarely performed. Therefore, one may conclude that cytogenomic area of genomic epileptology is poorly researched. Accordingly, molecular cytogenetic and cytogenomic studies of the clinical cohorts and molecular neurocytogenetic analyses of the epileptic brain appear to be required.

Iourov et al. performed a theoretical analysis to define the targets of the aforementioned studies and to highlight future directions for molecular cytogenetic and cytogenomic research of epileptic disorders in the widest sense. To succeed, they formed a consortium, which is planned to perform at least a part of suggested research. Taking into account the nature of the communication, “cytogenomic epileptology” has been introduced to cover the research efforts in this field of medical genomics and epileptology. Additionally, initial results of studying cytogenomic variations in the Russian neurodevelopmental cohort are reviewed with special attention to epilepsy. In total, they concluded that (i) epilepsy-associated cytogenomic variations require more profound research; (ii) ontological analyses of epilepsy genes affected by chromosomal rearrangements and/or CNVs with unraveling pathways implicating epilepsy-associated genes are beneficial for epileptology; (iii) molecular neurocytogenetic (neurocytogenomic) analysis of postoperative samples are warranted in patients suffering from epileptic disorders [2].

1)
Khan D, Bedner P, Müller J, Lülsberg F, Henning L, Prinz M, Steinhäuser C, Muhammad S. TGF-β Activated Kinase 1 (TAK1) Is Activated in Microglia After Experimental Epilepsy and Contributes to Epileptogenesis. Mol Neurobiol. 2023 Mar 2. doi: 10.1007/s12035-023-03290-2. Epub ahead of print. PMID: 36862288.

2)
Iourov IY, Gerasimov AP, Zelenova MA, Ivanova NE, Kurinnaia OS, Zabrodskaya YM, Demidova IA, Barantsevich ER, Vasin KS, Kolotii AD, Ushanov VV, Sitovskaya DA, Lobzhanidze TB, Iuditskaia ME, Iakushev NS, Zhumatov MM, Vorsanova SG, Samochernyh KA. Cytogenomic epileptology. Mol Cytogenet. 2023 Jan 5;16(1):1. doi: 10.1186/s13039-022-00634-w. PMID: 36600272.

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