Ion Channel Diseases of the Central Nervous System

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Key Words: Ion channels – Epilepsy – Ataxia – Schizophrenia — Alzheimer’s disease

ABSTRACT

In the last decade, advances in molecular genetics and cellular electrophysiology have increased our understanding of ion channel function. A number of diseases termed “channelopathies” have been discovered that are caused by ion channel dysfunction. Channelopathies can be caused by autoimmune, iatrogenic, toxic or genetic mechanisms. Mutations in genes encoding ion channel proteins that disrupt channel function are now the most commonly identified cause of channelopathies, perhaps because gene disruption is readily detected by the methods of molecular genetics. Ion channels are abundant in the central nervous system (CNS), but CNS channelopathies are rare; however, they overlap with some important neurological disorders, such as epilepsy, ataxia, migraine, schizophrenia, Alzheimer’s disease and other neurodegenerative diseases. It is possible that more CNS channelopathies will be discovered when additional ion channels are characterized and the complex mechanisms of brain function are better understood. At present, increased knowledge of the identity, structure and function of ion channels is facilitating diagnosis and treatment of many channelopathies.