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Genetics of Hypokinetic Diseases and Genetic Counseling

Zerrin Yılmaz Çelik

Başkent University Faculty of Medicine, Department of Medical Genetics, Ankara, Türkiye

Abstract

Hypokinetic diseases, a sub-branch of movement disorders, include a group of neurological conditions characterized by reduced voluntary movements. These diseases usually develop after a genetic disorder. These disorders can be transmitted from the family or may occur uniquely to the individual. A family history of hypokinetic disease may be a strong indicator of a genetic component. Individuals whose close relatives have been diagnosed with these diseases may have a higher risk due to shared genetic factors. Advances in genetic research have led to the investigation of gene therapy as a potential treatment for some hypokinetic diseases. Genetic information can enable the practice of precision medicine by tailoring treatment plans to an individual's genetic makeup. Current genetic diagnostic tests can help identify specific mutations or genetic variations associated with certain hypokinetic diseases. This can provide valuable information for both diagnosis and treatment planning. It is important to note that genetic disorders alone do not determine the development of hypokinetic diseases. To understand and manage these complex neurological disorders, it is necessary to consider both genetic and environmental factors. Environmental factors, such as exposure to toxins or lifestyle choices, can also play an important role in the onset and progression of diseases. Genetic counseling is important in terms of assessing genetic risk after family tree analysis, discussing potential consequences, and providing guidance for patients to make informed decisions about themselves. As a result, genetic approach to hypokinetic diseases; by examining the role of genetics in the pathogenesis of the disease, it aims to identify the relevant genetic factors, understand the personal/familial genetic and clinical risks of these factors and contribute to their management. In this presentation, it is aimed to discuss genetic approach and counseling by providing updated information about hypokinetic diseases and related genetic causes.

Epilepsies and Current Treatment Approaches from the Perspective of a Pediatric Neurologist

Rabia Tütüncü Toker

Bursa Uludağ University Faculty of Medicine, Department of Pediatrics, Division of Neurology, Bursa, Türkiye

Abstract

Although epilepsy is a historical disease, with the rapid progress of studies in the field of genetics, there is a need for new updates in terms of both definition, classification and treatment. The International League Against Epilepsy defined epilepsy as at least 2 untriggered seizures recurring at intervals longer than 24 hours; a diagnosis of epileptic syndrome, or one untriggered seizure accompanied by >60% probability of recurrence. Epilepsy; affects 5-10 per thousand of children, with the highest incidence in infancy. This rate varies between 8-14 per thousand in Türkiye. Epilepsy syndromes are defined and classified in newborns, infants and children in 2022 by considering certain criteria such as age of onset, family history, neurological examination, possibility of responding to medication, remission, comorbidities, seizure types, electroencephalography features, neuroimaging, genetics, and differential diagnosis. Epilepsy syndromes in newborns and infants were classified as self-limited epilepsies, developmental and epileptic encephalopathies and etiology-specific syndromes. Childhood epilepsies were grouped as self-limiting focal epilepsies of childhood, genetic generalized epilepsies and developmental and/or epileptic encephalopathies of childhood. Knowing the type and etiology of epilepsy and confirming the clinical suspicion of a specific syndrome avoids unnecessary additional tests. Contraindicated medications and ineffective treatment are avoided. Such as avoiding sodium channel blockers in patients with SCN1A-related epilepsy, or preferring these drugs first in channelopathy caused by KCNQ1 mutation in the neonatal period. It allows predicting the prognosis and allowing for information. It helps to personalize treatment based on the etiological gene.

Keywords: Epilepsy, epilepsy syndromes, gene, treatment