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Genetics of paroxysmal dyskinesia and the implications in pathogenesis of epilepsy and paroxysmal neurological diseases

Yo-Tsen Liu Taipei Veterans General Hospital, Taiwan

Paroxysmal dyskinesia (PD) is a clinically and genetically heterogeneous group of diseases characterized by episodically recurrent involuntary movements and classified into three groups: paroxysmal kinesigenic dyskinesia (PKD), paroxysmal non-kinesigenic dyskinesia (PNKD) and paroxysmal exercise-induced dyskinesia (PED). The breakthrough of genetics of PD was the identification of PRRT2 mutations in 2011. Following this, it has been well recognized that there is increasing genetic and clinical overlaps between PD and other paroxysmal neurologic disorders, including epilepsy, hemiplegic migraine, episodic ataxia and migraine. Advancement in genetics of PD has contributed to well-addressed genotypic-phenotypic correlations of the above diseases, which help greatly in improving the efficiency of genetic diagnosis and thus benefit more treatable patients. The rapidly-accumulated genetic knowledge has also shed light on understandings of the normal functions of the related genes and the underlying pathogenesis how these mutated genes to cause diseases. Furthermore, the reversibility of PD may be the key to unravel the mechanisms of other irreversible neurodegenerative diseases and inspire potential targets of treatment in the future.

Biography

Yo-Tsen Liu earned her MD at National Taiwan University in 2001. After that, she completed her Neurological Residency Training and became a Neurology Consultant at Taipei Veterans General Hospital (TVGH) in 2005. After winning "Studying Abroad Scholarship" supported by Taiwan's Ministry of Education, she studied her PhD at Institute of Neurology, University College London, London, UK in 2010-2014. She is now a Neurology Consultant at Department of Neurology of TVGH and Associate Professor at National Yang-Ming University, Taiwan. Her academic interests are the applications of next-generation sequencing in diverse neurological diseases, including paroxysmal dyskinesia, epilepsy and neurodegenerative diseases.

ytliu2@vghtpe.gov.tw

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