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Dementia 2018: Paroxysmal Kinesigenic Dyskinesia with genetic diagnosis of Wilson's disease

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Paroxysmal kinesigenic dyskinesia (PKD), a rare paroxysmal movement disorder often misdiagnosed as epilepsy, is characterized by recurrent, brief dyskinesia attacks from seconds to 5 minutes triggered by sudden voluntary movement like dystonia , tremor ,myoclonic jerks .Ion channelopathy has been suggested, since the disease responds well to moderate dosage of carbamazepine/oxcarbamazepine.Secondary causes of PKD which may well be associated with wilsons disease and other concurrent movement disorders should be sorted out if no evidence of ion channelopathy or genetic mutation is present. A 22 year male patient presented to our OPD with voluntary movement of Right hand with minimal dystonia present in resting as well as moving state , depression caused because of not able to perform daily activities .The patient was diagnosed initially with PKD because it lasted for few seconds to 2 minutes. Routine labs were performed including blood ceruloplasmin ,urine and serum copper which was consistent with diagnosis of WD. The ATP 7B gene mutation was positive in this case with no hepatic involvement .PKD gene testing was negative.Patient was started on traditional dosage of D -Penicillamine and being continued long term.For PKD we gave 50 mg bid dose of carbamazepine which was later increased to 100 mg bid with complete resolution of dyskinesia and depression. We think PKD might be secondary to WD in our case or some unknown ion channelopathy might be present which is not yet reported till date.Response to CMZ and penicillamine was very obvious. Myoclonus of PKD can be easily confused with myoclonic epilepsy and use of antiepileptic drug may be inappropriate in this setting. So careful monitoring of symptoms as well as associations with other diseases should be considered while evaluating this type of rare treatable cases. Inappropriate treatment can easily exacerbate the symptoms and can degrade the quality of life in young patients.

Paroxysmal movement disorders (PMDs) are rare neurological diseases typically manifesting with intermittent attacks of abnormal involuntary movements. The term "paroxysmal" indicates a well-defined onset and termination of clinical manifestations. Two main categories of PMDs are recognized based on phenomenology: Paroxysmal dyskinesias (PxDs) are characterized by transient episodes hyperkinetic movement disorders, while attacks of cerebellar dysfunction are the hallmark of episodic ataxias (EAs). From an etiological point of view, both primary (genetic) and secondary (acquired) causes of PMDs are recognized. Some aspects of clinical history may help to distinguish primary from secondary PMDs: Most primary forms occur as sporadic or familial cases with autosomal dominant inheritance, and most often onset of manifestations is set in childhood or adolescence, and interictal neurological examination is unremarkable; secondary forms occur sporadically, more usually begin after the second decade of life, and clinical examination is frequently abnormal also outside of attacks.

A further category that may manifest as PMDs are functional (psychogenic) movement disorders (FMDs). Patients with FMDs may show tremor, dystonia, myoclonus, parkinsonism, speech and gait disturbances, or other movement disorders whose patterns are usually incongruent with that observed in organic diseases, although sometimes diagnosis may be challenging. Diagnosis of FMDs is based on positive clinical features (e.g., variability, inconsistency, suggestibility, distractibility, and suppressibility) during physical examination and should be considered in presence of some clues such as intra-individual variability of phenomenology, duration and frequency of attacks, and/or precipitation of the disorder by physical or emotional life events. Other supporting information can be helpful (i.e., neurophysiologic and imaging studies).

Recognition and diagnosis of PMDs are based on personal and familial medical history, physical examination, detailed reconstruction of ictal phenomenology (possibly including video-recording of at least one attack), brain magnetic resonance imaging (MRI), and genetic analysis. Neurophysiological (i.e., standard electroencephalogram or long-term monitoring) or laboratory tests are reserved for cases in which an epileptic origin of the attack cannot be excluded, or brain MRI reveals alterations that are compatible with genetic-metabolic or secondary causes. Genetic knowledge of PMDs has been largely incremented by the advent of next generation sequencing (NGS) methodologies, which allowed to increase both molecular diagnosis and identification of ultra-rare or new genes. The wide number of genes involved in the pathogenesis of PMDs reflects a high complexity of molecular bases of neurotransmission in cerebellar and basal ganglia circuits. This comprehensive review is focused on clinical and genetic features of PMDs according to current nosology. As this review is mainly targeted on genetic causes of PMDs, functional PMDs will not be discussed further

Foot Note: This work is partly presented at 13th Annual Conference on Dementia and Alzheimers Disease, December 13-15, 2018 Abu Dhabi, UAE