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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 like Carbamazepine/Oxcarbamazepine. Secondary causes of PKD which may well be associated with Wilson's 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. 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 Wilson Disease (WD). The ATP 7B gene mutation was positive and Wilson disease diagnosis was confirmed without any other phenotypic feature except dyskinesia/dystonia of right hand. Patient was started on traditional dosage of D-Penicillamine and being continued long term. In view of PKD we gave 50 mg bid dose of Carbamazepine which was later increased to 100 mg bid with complete resolution of symptoms. 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 can be easily confused with myoclonic epilepsy and use of anti-epileptic 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 and living in young patients.

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