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Coffee may cure rare genetic brain disorder

31 July 2019 | News | By Manbeena Chawla

Caffeine is the most commonly consumed drug in the world

A rare genetic brain disease causing a movement disorder has been reported from Mumbai where a 21-year-old patient has been diagnosed with '*Myoclonus-Dystonia'* with facial, neck and hand shaking and jerking due to rare genetic disorder called *ADCY5-related dyskinesia*. This is a rare disease with less than 400 cases reported in the world. The disorder is known to cause abnormal involuntary tremors/movements in the body and is usually seen in children, but this is the first time it has been reported in an Indian adult patient.

The patient suffered from involuntary jerky movements of his hands and head since the age of 13, but these jerks were initially mild. With time, this progressed to moderate severity over the last few years. "We were able to manage the trembling till the time they were mild. With time, the intensity increased and so did the frequency which hampered daily activities and even any work that needed concentration and attention", say the patients' parents.

"When the patient came to us, he would get movements in 'spells' intermittently in the early years. His whole body trembled when he was anxious or even concentrating on an activity such as riding his scooter. After examining him in the clinic and doing the initial tests, we were convinced he had a rare genetic disorder and advised genetic testing for certain genes, that later confirmed ADCY5 gene mutation. There was no family history of a similar illness indicating that some diseases may arise de-novo (on their own)", said Dr Pankaj Agarwal, Head of the Movement Disorders Clinic at Global Hospitals Mumbai, who made the diagnosis and is treating the patient.

As per Dr VL Ramprasad, COO, MedGenome Labs that performed the genetic testing, "ADCY5 mutation causes abnormal involuntary movements affecting the neck, arms and face. This mutation can also lead to episodic worsening triggered by anxiety, stress or inactivity -or characteristically periods before/after sleep. We have now published this case in MDCP (*Movement Disorders Clinical Practice*

) a leading and well-known international clinical neurology journal." This is the first reported adult-onset case of ADCY5 in India.

Further, recently in a serendipitous finding, French neurology researchers observed that caffeine helps manage *ADCY5-related Dyskinesia*. An 11-year-old boy in France of African origin with ADCY5 mutation was prescribed caffeine, which is also commonly used as medicine for ailments in in that patients' country of origin. Treatment started off with 1 cup of espresso produced a dramatic response that began in 45 minutes and lasted for 7 hours, the boy had nearly complete resolution of all dyskinesia (severe shaking) episodes. The parents accidentally gave him decaffeinated coffee once, resulting in immediate return of his dyskinesia episodes. After 4 days, the parents realized the problem and resumed treatment with caffeinated coffee, instantly bringing relief to the patient. This chance observation can now help similar patients with ADCY5 related Dyskinesia around the world to find a low-cost way to manage their symptoms. Caffeine is the most commonly consumed drug in the world, and its effects and safety, even in children is well known.