Successful treatment of two cases of rare Movement Disorders

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Introduction

Movement disorders can manifest in numerous ways, with symptoms ranging from subtle to disabling. The first step to diagnosis depends critically on an accurate description of the abnormal movement. The neurological examination needs to be tailored to the clinical history in many cases. Paroxysmal movement disorders are another potential pitfall. These can be misdiagnosed as epilepsy despite the absence of any disturbance of awareness. Patients have very little understanding of the condition. We present two patients with rare movement disorders. Right medical intervention, administered at the right time, is the key for effective recovery.

Case 1

The first patient, a girl child from Bangladesh aged five, was initially brought to the hospital after two years of ongoing treatment for Myoclonic Epilepsy, a type of seizure with sudden muscle spasms. She was treated with multiple anti-epileptic medications by her previous doctors as the girl continued to have these clinical events in spite of treatment for epilepsy. After careful and thorough observation, we concluded that the girl had been misdiagnosed as Epilepsy. She was actually exhibiting a rare movement disorder called Hyperekplexia, which is also called exaggerated startle reaction in which affected individuals have increased muscle tone (hypertonia) as a response to unexpected stimuli, especially loud noises.

The child underwent a test called video-EEG to demonstrate that there was no epileptic basis for these abnormal clinical events. This study captures the brain activity with synchronised video recording, enabling us to do a time locked capture of these abnormal movements. We can use this special study of the brain to distinguish seizures from other non-seizure problems. The family of the patient was then informed of the correct diagnosis and reassured that this was not a seizure disorder. The patient received the most appropriate medication for the condition-Clonazepam, with a gratifying outcome.

This is a rare condition with some genetic basis. Many patients do have a benign course. with lesser events as, they grow up.

Case 2

The second patient, a young adult aged twenty, was undergoing a similar management for seizures for almost 15 years, for an involuntary movement that occurred especially when he woke from sleep. These movements were quite dramatic, with flailing movements of the limbs but with preserved consciousness. The patient was again on multiple drugs for epilepsy. Careful history taking and special testing using Video-EEG proved that he was not suffering with seizures but that it was a rare disorder of the nervous system called Paroxysmal Non-Kinesogenic Dyskinesia (PNKD) that is distinguished by periods of involuntary movement like irregular shaking and writhing movements of the limbs. The patient and family were counselled and we could remove many of the unwanted anti-epileptic medications that he was taking.

Discussion

Both these syndromes are very rare and cannot be easily identified. PNKD-Paroxysmal Non-Kinesogenic Dyskinesia is so rare that its prevalence rate is 1 in 5 million people while Hyperekplexia occurs for 1 in 50000 people. When these patients presented to us, we had to thoroughly study their case history as things were not adding up. On proper clinical examination and after witnessing some of the episodes, it was clear that these clinical events were not seizures. We immediately organized Video-EEG (Electroencephalography - a test that detects abnormalities in the brain waves, or in the electrical activity of the brain) to capture episodes and proved that these episodes did not have an epileptic basis. It is very unfortunate that both these young patients were on the inappropriate medications for such long periods of time. If undiagnosed, both these syndromes might have done much more damage to their already troubled lives. We are happy that we were able to correctly identify the condition and steer the patient on the right direction to recovery. Both patients have been responding well to the treatment and are doing well.