Juvenile Myoclonic Epilepsy: A Case Report

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Abstract: Juvenile myoclonic epilepsy is one type of epileptic syndrome typically manifesting in adolescence, presented with myoclonic seizure in all affected individuals. We reported a 12-year-old girl with her recurrent episodes of myoclonic seizure. Physical examination, vital signs, and general status were normal. An increased muscle tone was found in neurological examination. An electroencephalogram was performed and showed polyspike-and-wave patterns consistent with JME. An abnormality was found in brain MRI. The patient was managed with antiseizure medication. Subsequently, the patient showed an improvement. This case report highlights the importance of early diagnosis, proper management, and prognosis of children with juvenile myoclonic epilepsy.

1 INTRODUCTION

Juvenile myoclonic epilepsy (JME) is a common form of idiopathic generalized epilepsy syndrome. Typically emerging during adolescence, JME accounts for 5-10% of all epilepsy cases and approximately 26% of idiopathic generalized epilepsies. The condition often presents between the ages of 12-18 years, with a slight female predominance. Genetic disorder is considered the causes of JME, with several genes implicated in its etiology, though the exact genetic mechanisms remain complex and multifactorial. A family history of epilepsy may play a role in the occurrence of JME.

JME is characterized by the onset of myoclonic jerks, generalized tonic-clonic seizures, and less frequently, absence seizure. Myoclonic jerks, often occurring shortly after waking, may progress to generalized tonic-clonic seizures if left untreated.

Despite its relatively high prevalence among young individuals, JME is frequently underdiagnosed or misdiagnosed, partly due to the subtlety of myoclonic jerks or the lack of awareness among healthcare providers. This condition requires long-term management and is generally well-controlled with antiepileptic drugs (AEDs). However, adherence to treatment and lifestyle modifications are critical factors in reducing seizure frequency and improving quality of life. Understanding the unique characteristics, clinical presentation, and management strategies of JME is essential for optimizing patient outcomes and developing tailored therapeutic approaches.

2 CASE ILLUSTRATION

A 12-year-old girl presented with her first episode of seizure four months prior to hospital admission. The semiology included rhythmic jerking movements in both hands, accompanied by upward gaze with seizure duration roughly 3 minutes, after which the patient limp and fell. She experienced recurrent episodes of seizure on daily basis. No history of seizure in the family. She was diagnosed with myoclonic seizure. Valproic acid 7 ml twice a day and phenobarbital 30 mg three times a day were given. Unfortunately, the seizure did not improve. Three months later, she came to Pediatric Neurology Clinic with worsening continuous involuntary jerking movements in all extremities, making her unable to engage in daily activities.

Physical examination showed normal vital sign and general condition. Neurological assessment revealed an increase of muscle tone. Laboratory tests showed normal result. EEG examination was performed and showed showed epileptiform activity characterized by 4-5 Hz polyspike-and-wave patterns, consistent with JME (Figure 1). A brain MRI identified a subacute infarction in the bilateral lentiform nucleus (Figure 2).

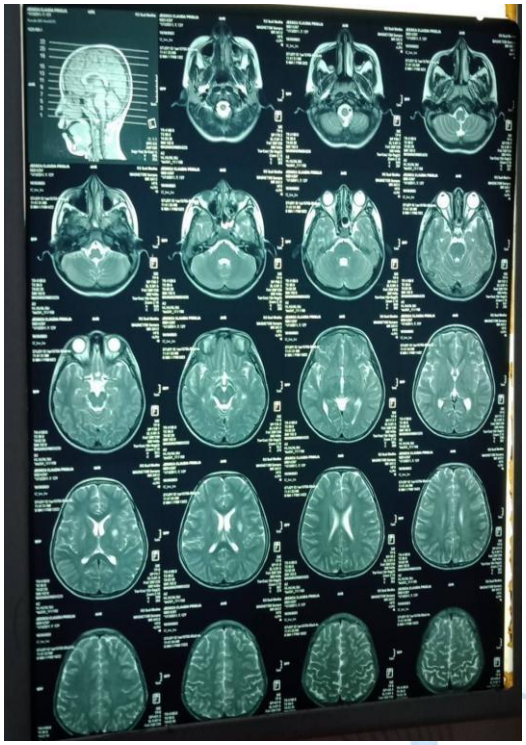

Figure 2. Brain MRI of the patient showing an abnormality

Clobazam 0,1 mg/kg/day titrating into 0,3 mg/kg/day was added to the medication. She responded well to the treatment. Currently, the patient's seizure frequency has reduced to at least 3-5 times a day.

3 DISCUSSION

Juvenile myoclonic epilepsy (JME) is recognized as a distinct epilepsy syndrome due to its characteristic clinical presentation, age of onset, and response to treatment. The majority of individuals with JME present with myoclonic jerks, predominantly in the upper limbs, that typically occur upon awakening. These jerks are often accompanied by generalized tonic-clonic seizures and, less frequently, absence seizures. The early identification of JME is crucial, as timely initiation of appropriate antiepileptic therapy can significantly reduce seizure frequency and improve patient outcomes.

Genetic predisposition plays a significant role in JME, with evidence supporting a polygenic inheritance pattern involving multiple genes, such as those encoding for ion channels and neurotransmitter receptors. Key genes associated with JME include KCNQ3, which affects potassium channels and may contribute to seizures by altering neuronal repolarization, and CLCN2, which regulates chloride channels involved in GABA-mediated inhibition. The inheritance pattern remains unclear, involving autosomal dominant, autosomal recessive, and multifactorial models. Most researchers believe that JME results from the interaction of multiple genes with genetic, epigenetic, or environmental factors, each having a modest impact on the condition's development.

The diagnosis of JME relies on clinical characteristic and typical EEG patterns. Key signs include myoclonic jerks, absence seizures (AS), and generalized tonic-clonic seizures (GTCS), alongside normal neurologic examination and head CT, a specific EEG pattern of generalized spikes or polyspikes especially with photic stimulation, and complete remission of seizures in 80% of patients during valproic acid therapy. Despite clear diagnostic features, JME is often misdiagnosed or delayed due to under-recognition of symptoms, like myoclonic jerks, or atypical EEG findings. Misdiagnosis can lead to inappropriate treatment, poorer outcomes, and complications such as brain damage or status epilepticus.

Myoclonic jerks in JME are usually sudden brief of single or repetitive, bilateral, symmetric, arrhythmic, involuntary movement, primarily in the upper extremities, but they can also affect other parts of the body. Consciousness remains intact, and most jerks occur upon waking. Only 3-5% of JME patients have just myoclonic jerks. GTCS develop in 80-97% of patients, often following myoclonic jerks. Absence seizures are less frequent, appearing in 12-54% of cases.

EEG is a key diagnostic tool of JME. Common interictal findings include bursts of generalized spike-and-wave or polyspike-and-wave patterns (3-5 Hz) that are often seen in the frontal regions. During myoclonic seizures, the EEG typically shows a pattern of polyspike-and-wave, with 5 to 20 high-frequency (10-16 Hz) spikes, often followed by slow waves at a frequency of 2.5-5 Hz. Brain imaging is generally normal in JME, but structural abnormalities such as increased cortical gray matter in the mesial frontal lobes, frontobasal regions, and the anterior portion of the thalamus may present.

Management of JME extends beyond pharmacotherapy; lifestyle modifications are also

critical. Many factors may trigger seizures in patients with JME, such as sleep deprivation, fatigue, photosensitivity, and stress. Consequently, patient education regarding these triggers, adherence to medication, and routine follow-ups play a vital role in managing the disorder effectively.

Continuous treatment is necessary to prevent frequent relapses, which can lead to status epilepticus. Although no randomized controlled trials have been conducted, clinical experience supports valproic acid as the first-choice AED, effectively controlling seizures in about 85% of cases. For those unresponsive to VPA, alternatives include lamotrigine, clobazam, levetiracetam, and topiramate. However, lamotrigine may worsens myoclonic seizures and GTCS, thus the use of lamotrigine may be use as combination with valproic acid. Clobazam has been used in the treatment of partial and generalized epilepsies. Some AEDs, like carbamazepine, oxcarbazepine and phenytoin, should be avoided as they can worsen myoclonic seizures.

4 CONCLUSIONS

JME is generally a manageable condition with proper diagnosis and treatment, but it poses challenges due to its genetic complexity, diagnostic subtleties, and the potential for a lifelong impact on patients. Future research should focus on elucidating the genetic underpinnings and pathophysiological mechanisms of JME, as well as developing personalized management approaches to improve patient outcomes.

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