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Juvenile Myoclonic Epilepsy

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Introduction

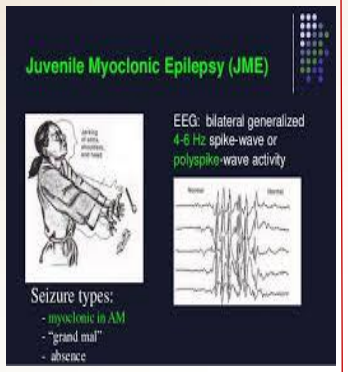
Juvenile myoclonic epilepsy (JME) is one of the most common epilepsies of childhood (Korff, Nordli, & Eichler, 2014). JME typically occurs in otherwise healthy teenagers, and is characterized by one or more of the following seizure types: myoclonic jerks, generalized tonic clonic seizures, and absence seizures (Korff, Nordli, & Eichler, 2014). JME is thought to have genetically mediated factors (Park, Shahid, & Jammoul, 2015). JME is considered to be gender equal (Korff, Nordli, & Eichler, 2014). Most JME patients are diagnosed between 12 and 18 years of age (Korff, Nordli, & Eichler, 2014). Triggers linked to onset of seizure activity include: sleep deprivation, fatigue, alcohol, mental stress, and flashing lights (Park, Shahid, & Jammoul, 2015). Although the frequency of seizures typically decreases as the individual progresses through adulthood, JME is usually a life-long disease (Korff, Nordli, & Eichler, 2014). The majority of patients with JME respond to antiepileptic treatment, and are able to live free from seizure, as long as they maintain treatment (Afra & Adamolekun, 2011). However, antiepileptic medication can have adverse effects, possibly leading to poor tolerance, and compliance with treatment (Afra & Adamolekun, 2011). Education of JME patients and their families, by their healthcare provider, on disease and treatment (including non-pharmacological treatment such as life-style modification) is essential for disease management, and improved patient outcomes.

Case Study Presentation

A 15 year old white male is admitted to the pediatric intensive care unit from the emergency department, accompanied by his mother. The juvenile is unable to maintain a wakeful state, therefore, initial information is obtained from his mother. The mother reports her son had stayed at a friend's house overnight, she received a phone call from the friend's mother this morning stating her son had a seizure. The juvenile was brought to the emergency room by ambulance. The mother reports the information she received was that her son and his friend had been up all night playing video games. While playing, the juvenile fell from the edge of the bed into the floor and began convulsing, followed by an episode of vomiting. The episode was reported to have lasted about two minutes. The mother reports the juvenile has no known history of seizures, no prior medical conditions. Diagnostic evaluation completed in the ER includes MRI and laboratory testing. MRI results indicate no abnormalities. Laboratory testing reveals complete blood count, and comprehensive metabolic panel within normal limits; elevated lactate, and creatine kinase; drug and alcohol screening are negative. Vital signs are within normal limits. The juvenile has abrasions to his forehead, and bite marks to his tongue. After waking, the juvenile states he does not recall the episode. However, he reports that throughout the night he noticed he had been having some jerking episodes of both his hands that would cause him to drop his video game controller, he also reports that following this jerking episodes he briefly forgot what he was doing. EEG results are abnormal, with findings consistent with juvenile myoclonic epilepsy.

Signs and Symptoms of JME

Juvenile myoclonic epilepsy is characterized by myoclonic jerks that usually occur after awakening, and are described as shock-like or arrhythmic movements of both arms (Sirven & Fisher, 2013). Patient's with JME may present with myoclonus, generalized tonic-clonic seizure, or absence seizure (Korff, Nordli, & Eichler, 2014). However, most patient's present following a generalized tonic-clonic seizure, and further assessment, and testing leads to diagnosis of JME (Korff, Nordli, & Eichler, 2014). Interictal EEG pattern in JME is classically, "4 to 6 Hz bilateral polyspike and slow wave discharges with frontal predominance over a normal background activity (Korff, Nordli, & Eichler, 2014, p.3). However, other abnormalities are less commonly noted including, "2.5 to 4.5 Hz bilateral spike-waves, single spikes, and irregular spike-wave complexes" (Korff, Nordli, & Eichler, 2014, p.3). JME typically manifests during adolescence, and persists into adulthood (NIH, 2012).



Courtesy of Pediatric Neurology Review, Atlantic Health.org

Underlying Pathophysiology

An epileptic seizure results from neurologic dysfunction with abnormal neuronal firing, manifesting as changes in motor control, abnormal sensory perception, abnormal behavior, and/or abnormal autonomic function (Stafstrom & Rho, 2014). Epilepsy results from cellular level biochemical processes that promote aberrant electrical activity causing neuronal hyperexcitability, and hypersynchrony (Stafstrom & Rho, 2014). Juvenile myolonic epilepsy is an idiopathic generalized epilepsy syndrome (Carroll, 2013). JME seizures are linked with cortical hypersensitivity (Korff, Nordli, & Eichler, 2014). Routine brain analysis of patients with JME is typically normal (Carroll, 2013). However, advanced imaging techniques have suggested "involvement of frontal thalamocortical circuits and dysfunction in the dopaminergic and serotoninergic neurotransmission systems" (Korff, Nordli, & Eichler, 2014, p. 2). The underlying cause of JME remains unknown (Korff, Nordli, & Eichler, 2014). Several research studies have shown that genetic factors play a role in JME (Mefford et. al., 2010). At least 15 chromosomal loci are suspected in playing a role in JME including: EFHC1, GABRA1, CLCN2 (Korff, Nordli, & Eichler, 2014), CPA6 (Sapio, et. al., 2015), and BRD2 (Velisek, et. al., 2011). Some studies have found a slightly higher rate of JME among females, but most studies have found JME to be gender equal (Korff, Nordli, & Eichler, 2014).

Courtesy of Optimum Wellness Labs

Significance of Pathophysiology

The generalized tonic-clonic seizure (GTCS) of JME that often leads to the patient seeking medical attention is similar to GTCS of other causes (Wilfong, 2014). Also, neuroimaging often fails to find any abnormalities in patients with JME (Korff, Nordli, & Eichler, 2014). Given the unclear etiology of JME, the lack of abnormal structural findings, and the number of causes that can lead to GTCS, a careful history is very important for accurate diagnosis of JME (Wilfong, 2014). The clinician should obtain information related to: the setting in which the episode occurred including time of day, recent illness or new medication, behavior immediately prior to the event, activity engaged in immediately prior to event, was the event accompanied by other conditions such as headache, were any focal symptoms present such as twitching, a physical description of the event related to body movements and duration of event, behavior after the event including confusion or focal findings (Wilfong, 2014). A family history should also be obtained (Wilfong, 2014). Although some patients with JME will have normal EEG findings, EEG should be performed to help confirm diagnosis of JME (Wilfong, 2014).

Implications for Nursing Care

Accurate diagnosis of JME, differentiation from other seizure disorder, is imperative for accurate treatment (Korff, Nordli, & Eichler, 2014). Some antiepileptic drugs are ineffective, or contraindicated in JME (Schachter, 2015). With appropriate antiepileptic drug therapy, most patients respond quickly, and completely but will still require lifelong treatment (Korff, Nordli, & Eichler, 2014). Patients should be educated that they need to continue drug treatment even after prolonged periods of remission from seizure (Korff, Nordli, & Eichler, 2014). Patients should be educated to avoid seizure triggers (Korff, Nordli, & Eichler, 2014). Patients should also be educated on lifestyle modifications related to potential safety concerns surrounding swimming/bathing, driving, and other potentially dangerous activities (Korff, Nordli, & Eichler, 2014). Consistent seizure control to decrease, or prevent seizure episodes is imperative to prevent seizure comorbidity, and disability (Kessler, Lane, Shahly, & Stang, 2012).

Conclusion

Although the etiology of JME remains unknown, it can be successfully diagnosed by thorough assessment of patient history, presentation, and testing. With appropriate, and consistent antiepileptic drug treatment, and lifestyle modification, most patients are able to remain seizure free. Many of the potential seizure triggers in JME are particularly common for teenagers to experience such as lack of sleep, stress, and flashing lights. Juveniles newly diagnosed with JME, and their families, need to be well educated on the disease, disease treatment, and safety considerations to promote the best outcomes for the juvenile. .

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Courtesy of Noran Neurological Clinic

Additional Resources

Epilepsy Foundation www.epilepsy.com/

NICHQ.Org | Epilepsy Resources for Patients and Families www.nichq.org/.../epilepsy/resources/epilepsy-resources-for-patients

CDC - Epilepsy - Toolkit - Resource Guide www.cdc.gov/epilepsy

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