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Infantile Spasms

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Introduction

In the Neurology Department at Nationwide Children's Hospital , we see many patients who have infantile spasms, also known as West Syndrome. In an attempt to improve care provided to my patients and better gain an understanding of the underlying pathophysiology of this epilepsy syndrome, I chose to pursue this topic.

Infantile Spams is an epilepsy syndrome that usually appears in children between the ages of four and eight months. The spasms generally occur upon waking or right after feeding, and can occur in clusters of a hundred or more at a time (Go et al., 2012). Children with infantile spasms present with a classic, abnormal electroencephalogram. The pattern that determines if it is indeed infantile spasms is called hypsarrythmia, which is a disorganized pattern (Iype et al., 2016). Children with infantile spams will also regress in milestones and can have mental retardation.

Many studies have been done to determine the best treatment options for infantile spams. The standard treatment consists of either high dose oral steroids, vigabatrin, or adrenocorticotropin hormone (ACTH). The quicker the initiation of treatment, the better the outcome for the child (Beatty, Wrede, & Blume, 2017). For this project research will be done to find articles that support the most effective treatment for infantile spamS.

Underlying Pathophysiological Process.

The exact pathophysiology of infantile spasms is unknownbut there are many theories that share a common pathway in the etiology (Wheless et al., 2014). Infantile Spasms is a rare disorder and occurs in about 1.4 to 4.5 per 10,000 live births (Glaze, 2017). It is equally common in boys and girls and most cases are random, while some have a genetic component. Some patients who have infantile spasms will have another neurological disorder known as Tuberous Sclerosis, it is also seen frequently in patients who have hypoxic-ischemic encephalopathy, as well as Down syndrome (Inoue et al., 2014). A fairly common cause of infantile spasms is dneonatal hypoxic-ischemic injury, meaning the whole brain is deprived of oxygen at birth. This accounts for nearly ten percent of all infantile spasms diagnoses (Gano et al., 2013).

TABLE 42.1 Causes of Secondary Generalized Epilepsy Syndromes (Infantile Spasms and LGS)

Idiopathic, Cryptogenic
Symptomatic
Perinatal factors: hypoxic-ischemic encephalopathy, hypoglycemia, and hypocalcemia
Infection: intrauterine infection (toxoplasmosis, rubella, and cytomegalovirus, herpes), meningoencephalitis
Cerebral malformation: holoprosencephaly, lissencephaly, polymicrogyria, Aicardi's syndrome
Vascular: infarction, hemorrhage, porencephaly
Neurocutaneous syndromes: tuberous sclerosis complex, Sturge–Weber syndrome, and others (e.g., neurofibromatosis type 1)
Metabolic disease: nonketotic hyperglycinemia, pyridoxine deficiency, aminoacidopathy (phenylketonuria, maple syrup urine disease)
Chromosomal disorders: Down's syndrome, Angelman's syndrome (happy puppet syndrome: abnormality in chromosome 15q11–13, seizures, developmental delay, dysmorphic features, and paroxysms of inappropriate laughter)

Abbreviation: LGS, Lennox–Gastaut syndrome.

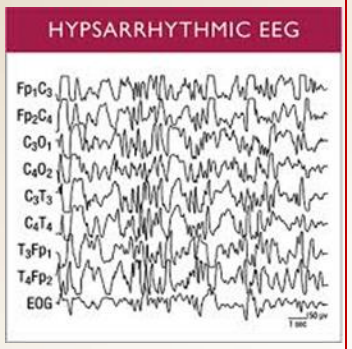
Significance of Pathophysiology

While the incidence of infantile spasms are rare, the pathophysiology is devastating. Most children have mental retardation or significant delay. It is common for infantile spasms to progress into other epilepsy syndromes. The most common being Lennox–Gastaut syndrome (LGS). LGS is diagnosed between the ages of one and eight years old. Seizures begin to change from spasms to tonic-clonic, myoclonic and absence seizures. These seizures usually occur frequently and have a high chance of being refractory to treatment (Go et al., 2012). If they do respond to treatment, many side effects are usually going to be present as they are on multiple antiepileptic drugs. Children with LGS have a poor prognosis and frequently die from complications related to their seizures, such as head trauma or sudden unexplained death in epilepsy patients (SUDEP) (Wheless et al., 2014).

Signs and Symptoms

Infantile Spasms are typically classified one of two ways. They are either symptomatic or cryptogenic. If a patient has symptomatic Infantile Spasms that means there is either an identified underlying cause and/or there is a developmental delay present at the time of the onset of spams. Cryptogenic Infantile Spasms have no known underlying cause and there is not a developmental delay at the onset of spasms (Glaze, 2017). The patients have a normal exam and imaging, yet still have hypsarrhythmia on the electroencephalogram (EEG). Cryptogenic accounts for 40 to 50 percent of all cases of infantile spasms.

When infantile spasms are suspected, quick actions should be taken to confirm the diagnosis so treatment can be promptly started. The parents are almost always the first to notice symptoms and will usually take their child to the family practitioner or pediatrician for concerns. Symptoms that are usually noticed are contractions (spasms) of the extremities, neck and trunk of the baby, with brief periods of unresponsiveness (Samson, 2012). The spasms are almost always symmetrical, involving both sides of the body. These spasms can occur anywhere from twenty to one hundred times per day (Go et al., 2012). It is very helpful in making a diagnosis if the family is able to video tape the spasms to show the provider. Another symptom that usually brings the parents in is regression in developmental milestones. This most often occurs after the onset of spasms. As Advance Practice Registered Nurses (APRNs) it is prudent that we know what signs to look for to make an accurate diagnosis and referral to a neurologist so as to provide the best outcome for the patient. In addition to the spasms and developmental regression, an EEG is done to see what the brain waves look like. If the patient has infantile spasms, their EEG will have the unmistakable pattern of hypsarrhythmia which confirms diagnosis. Hypsarrhythmia is a description of high voltage spikes and slow waves. These spikes and waves are not synchronized and give a chaotic appearance to the EEG (Hancock, Osborne, & Edwards, 2013).



Treatment Options

Once diagnosis has been confirmed the neurologist and family decide on the best treatment option for the patient. The standard treatment options most frequently utilized are vigabatrin, high dose steroids, or ACTH. Each treatment has the potential for both a positive outcome and/or harmful side effects. With treatment the goal is to control the infantile spasms so that they are either greatly reduced or completely absent. Infantile Spasms generally last up to around three years of age and long term prognosis is poor. (Hancock, Osborne & Edwards, 2013). Most of these patients will develop Epilepsy and 70 percent will have severe mental and psychomotor retardation (Samson, 2012).

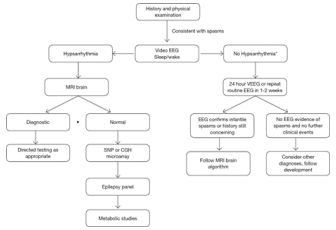


Table 1. Management of infantile spasms

Implications for Nursing Care

As APRNs, we have to be prepared to not only make a swift diagnosis and to initiate treatment , bur to also have tough conversations with families. A diagnosis of Infantile spasms is life changing for a family. Not only is their child having seizures but they are going to progressively get worse as time goes on. Linking these families with appropriate resources is a must. Education is key in maintaining high quality of life for these patients, as parents are often taken aback by the challenges that they face in raising a child with multiple disabilities. The more resources and help a family can get, the better overall outcome for the whole family. Monitoring of the patient for efficacy and side effects must be done diligently. The side effects for each medication option can have devastating effects including permanent vision loss, irritability, hyperglycemia, hypertension, sodium and water retention, weight gain, gastric ulcers and bleeding, immune system dysfunction, infection, cardiomyopathy, congestive heart failure, and diabetic ketoacidosis (Samson, 2012).

Conclusion

In conclusion, infantile spasms must be recognized and diagnosed quickly in order to have the best outcome for the patient. Diagnosis is made based upon three symptoms/signs: spasms, developmental regression, and an EEG with hypsarrhythmia. ACTH, vigabatrin, and high dose steroids are the first line of treatment. Spasms typically resolve by three years of age, however, further neurological devastation usually transpires. Most patients will go on to develop epilepsy and mental and psychomotor retardation. Support and education must be provided to the patient and family to optimize quality of life and outcomes

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Additional Resources

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