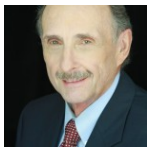
Disorder Directory: Learn from the Experts

EDUCATIONAL ARTICLES PLUS FAMILY STORIES AND RESOURCES

INDEX A B C D E F G H I J K L M N O P Q R S T U V W X Y Z

Infantile Spasms

ARTICLE FAMILY STORIES RESOURCES



DON SHIELDS, MD

Dr. Shields is Professor Emeritus of Neurology and Pediatrics, David Geffen School of Medicine at UCLA

Positions held in National and International and other medical organizations

- President, Child Neurology Foundation
- Vice President, Pediatric Epilepsy Foundation
- Councilor, Child Neurology Society, 1994-1996
- Nominating Committee, American Epilepsy Society, 1996
- Membership Committee, Professors of Child Neurology, 1995
- Chairman, Membership committee, Child Neurology Society, 1988-1993
- Membership committee, Child Neurology Society, 1986-1988
- Co-chairman Awards Committee, American Epilepsy Society, 1989-1992

Awards for scientific accomplishment

- UCLA School of Medicine Sherman Mellinkoff Faculty Award for dedication to the art of medicine and to the finest in doctor-patient relationships 2010
- Who's Who in the World 2007, to present
- Who's Who in America, 2006, to present

Areas of contributions to the literature and research

- Childhood epilepsy, primarily difficult to control epilepsies such as infantile spasms and Lennox-Gastaut Syndrome
- Surgery to treat epilepsy in very young children
- Consequences of epilepsy on development

SUMMARY

Infantile spasms (IS) is a rare seizure disorder that occurs in young children, usually under one year of age. The average age of onset is around four months, but some children may experience spasms as early as one month. A few children may begin as late as two years. Only about 2,500 children in the US are diagnosed each year with IS. It often has a very subtle appearance so it is difficult for parents to recognize that it is a serious problem. When most people think of a seizure disorder, they may think of someone falling to the ground and having all-over body convulsions. It is very obvious when that happens that there is something wrong. Many people seeing a seizure for the first time are quite scared; while others may think that the person is going to die.

A young child having infantile spasms, on the other hand, may just have little head drops that do not appear to be anything serious. However, it is a much more serious seizure disorder than the generalized convulsion. Not only is it difficult for the parent to realize that this is a seizure disorder, it is also challenging for pediatricians. Infantile spasms are so uncommon that most pediatricians will see only one or two IS cases during all the years of practice. Also IS often looks similar to common disorders such as a normal startle reflex, colic, or reflux. It is very important to recognize that a child has IS as soon as it begins because:

- there are medications that may control the spasms
- the longer the spasms last before they are treated and controlled, the poorer the child may do developmentally

Unfortunately, children who develop IS are at great risk for developmental disability and autism, but some children will do well if they are treated early. Because the spells may be subtle, the diagnosis may be delayed for weeks or months

DESCRIPTION/ SYMPTOMS

WHAT DO INFANTILE SPASMS LOOK LIKE?

Infantile spasms were first described by the English physician, Dr. W.J. West in 1841. His description is as accurate today as it was then. It is a remarkable report because Dr. West was describing his own son and he was asking for help. The following is a quote from Dr. West's report describing his son:

> *"The child is now a year old; it was a remarkably fine, healthy child when born, and continued to thrive until he was four months old. It was at this time that I first observed slight bobbings of the head forward, which I then regarded as a trick, but were, in fact, the first indications of disease;*

Like Dr. West's son, many children with IS appear to be normal until the spasms begin. It seems that he was not concerned when he first saw the spasms because they did not appear to be serious. Just like Dr. West, parents today often tell us that they were not concerned at first. But, as the spasms become more obvious they realize that this is something serious. Dr. West then described what happened to his son over the next few months.

> *"for these bobbings increased in frequency, and at length became so frequent and powerful, as to cause a complete heaving of the head forward toward his knees, and then immediately relaxing into the upright position: these bowings and relax things would be repeated alternately at intervals of a few seconds, and repeated from 10 to 20 or more times in each attack, which*

> *attack would not continue for more than two or 3 minutes; he sometimes has two, three, or more attacks in the day; they come on whether sitting or lying; just before they come on, he is all alive and in motion, making a strange noise, and then all of a sudden down goes his head and upwards his knees; he then appears frightened and screams out;*

The spasms usually become more obvious as they did in this case. Sometimes they remain subtle, maybe just a brief head nod but nothing else. Dr. West's son had what's called "flexor spasms" where the child's head goes forward and the legs come up. The arms usually flare out from the body as the head and body come forward. Sometimes spasms occur with the head going back and the legs straightening out. These are called "extensor spasms." Each spasm episode lasts just a second or two but they often come in clusters. Although the spasms may happen as single jerk event, clusters are more common and often occur on awakening in the morning or after a nap.

By the time Dr. West wrote about his son, he had been having the spasms for several months and the effect on his development was obvious, as Dr. West wrote:

> *"at one time he looked pale and exhausted, but lately he has regained his good looks, and independent of this affection, is a fine growing child, but he neither possesses the intellectual vivacity or the power of moving his limbs, a child his age; he never cries at the time of the attacks, or smiles or takes any notice, but looks placid and pitiful, yet his hearing and vision are good; he has no power of holding himself upright or using his limbs, and his head falls without support"*

Unfortunately for Dr. West's son, there were no effective treatments available in 1841 so he had the worst possible outcome. In addition to not developing mentally, he also seemed to be very weak and was not even able to control his head when he was seated.

To help make clear the appearance of infantile spasms, click here to watch a video of a child having infantile spasms.

HOW ARE INFANTILE SPASMS DIAGNOSED

Most children who have infantile spasms will have a very abnormal electroencephalogram (EEG) pattern called hypsarhythymia or modified hypsarhythmia. Hypsarhythmia is a very high-voltage, disorganized pattern of EEG abnormality. The hypsarrhythmia pattern is not present during the entire EEG in many cases. It is most likely to be observed during sleep. A less chaotic pattern, called modified hypsarhythmia, actually may be more common than hypsarhythmia. Hypsarhythmia or modified hypsarhythmia is seen in about 2/3 of cases. Some children may have an EEG pattern that is less dramatic than hypsarhythmia or modified hypsarhythmia and in those cases, it is very important to see what the EEG does when the child has a cluster of spasms. At the time of a spasm, the EEG usually flattens out for a very brief time. This is called an "electrodecremental response." Thus, the diagnosis of infantile spasms can be confirmed by observing either hypsarhythmia, modified hypsarhythmia or an electrodecremental response. Many patients will have all three.

The diagnosis of infantile spasms can be confirmed by observing the patterns of hypsarhythmia, modified hypsarhythmia or an electrodecremental response on an EEG.

If the EEG is normal, the diagnosis of infantile spasms should be reconsidered because there are benign disorders that may appear clinically similar to

infantile spasms (for example, benign infantile myoclonus or benign familial infantile convulsions).

WHAT CAUSES INFANTILE SPASMS

The seizures' appearance and EEG are so distinct that the clinical diagnosis of infantile spasms can be made with certainty in the most cases. Once it's recognized, the diagnosis of infantile spasms is usually easy, but determining what caused the spasms may be difficult. Determining the cause of infantile spasms is very important because it affects treatment and prognosis. After careful evaluation, the underlying cause can be identified in more than 70% of cases. There are dozens of disorders that are known to cause infantile spasms. When neurologists are able to identify the cause, it is labeled "symptomatic." It is essential that an appropriate diagnostic evaluation be performed in every child.

A diagnosis is important because it leads to specific treatment that may improve the long-term developmental outcome. In fact, some children with infantile spasms may ultimately lead normal lives, but only if they are diagnosed and treated correctly. Examples of specific therapy include:

- Pyridoxine-dependent seizures are treated with pyridoxine (vitamin B6)
- Tuberous sclerosis associated seizures are most likely to respond to vigabatrin

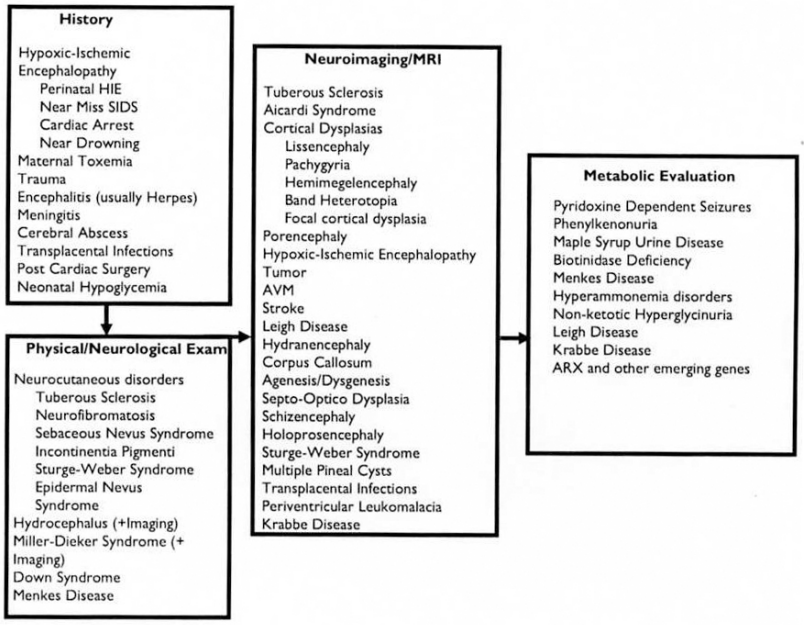
Another important reason for establishing a causal diagnosis is that some of the disorders are genetic, which may carry a significant risk of recurrence in another sibling. In those cases, genetic counseling is essential if recurrence is to be avoided.

In some cases, the cause cannot be identified; such cases are called "cryptogenic." Cryptogenic means that the cause is hidden. It does not mean that there is not a cause; it just means that we don't know all the causes yet and don't have a way to find them all.

Doctors follow a standard evaluation process to try to identify the underlying cause of the infantile spasms. This process, as noted in the figure below, includes different steps a neurologist will perform to identify disorders that are associated with IS by:

- First, taking a history
- Second, a careful physical and neurologic examination
- Third, doing a magnetic resonance imaging (MRI) scan.
- There are many metabolic and genetic disorders that have been associated with infantile spasms, but they are very rare.

Table 1



HISTORY OF THE CHILD

The first step is a careful history. Sometimes the underlying cause of infantile spasms is determined by taking a careful history of the patient. The history includes all information related to the pregnancy, labor and delivery as well as all of the events, including developmental milestones, which have occurred prior to the time of evaluation for the infantile spasms. A child may be developing normally, but then development stops, or even reverses. If that happens, it is an important clue. Some children may have seizures that occurred prior to the infantile spasms. Sometimes the seizures begin on one side of the body and that is also an important clue to understanding possible causes. Although there are many disorders associated with infantile spasms that may be identified by taking a careful history, two stand out as particularly important:

1. A history of brain injury from lack of oxygen
2. Brain injury from infection such as meningitis or encephalitis.

A child may be developing normally, but then development stops, or even reverses. If that happens, it is an important clue.

PHYSICAL AND NEUROLOGIC EXAMINATION

The second step is a careful physical examination and neurologic examination of the child. Many patients may have been developmentally delayed prior to the onset of spasms. Several underlying disorders can be identified by careful examination, such as:

- Cerebral palsy: Some children may have signs of cerebral palsy. These symptoms would be a clue of a brain problem that occurred during development or at the time of birth, such as stroke causing brain damage.
- Down syndrome: If Down syndrome had not been identified prior to presentation with infantile spasms it will be apparent on the physical examination.
- Tuberous sclerosis: An examination of the skin, especially the characteristic of “ash leaf” spots, is evidence of tuberous sclerosis. Ash leaf spots are small areas of skin that appear to be white compared to the rest

of the skin because they lacked normal pigment. Tuberous sclerosis is one of the more common associated disorders. Tuberous sclerosis also has very characteristic changes on the MRI brain scan that are diagnostic.

Several other disorders can be diagnosed by skin examination including neurofibromatosis and incontinentia pigmenti.

NEUROIMAGING

The third step is an MRI scan. Neuroimaging has been an important advancement in the last several decades in the diagnosis of the underlying etiologies or causes of infantile spasms. Both the MRI scan and computed tomography (CT) scan have been used to detect brain abnormalities in children with infantile spasms. However, the MRI scan is much more sensitive and more likely to discover an abnormality of the brain than the CT scan. A large list of neurologic abnormalities can be seen on MRI scan (including developmental brain abnormalities) and evidence of brain injury events (such as brain injury from lack of oxygen, trauma, brain tumor, or injury from past infection).

Once the doctor has completed a careful history, physical and neurologic examination, and performed an MRI scan, almost all of the underlying disorders that we know of which are associated with infantile spasms will have been identified.

METABOLIC AND GENETIC STUDIES

Finally, if the history, physical examination, neurologic examination, and MRI scan do not reveal the underlying cause, then the genetic/metabolic evaluation may be considered. More than 50 genetic/metabolic diseases have been associated with infantile spasms. It is not always necessary to do the complete metabolic/genetic workup on every child. If the history or exam suggests the possibility of a metabolic or genetic disorder then the treating physician will undertake the evaluation. A rare cause of infantile spasms is a disorder called “pyridoxine (vitamin B6) dependent seizures.” It is sometimes useful to administer a trial of intravenous pyridoxine. If that is unsuccessful, blood and urine tests for metabolic or genetic diseases may be performed. A spinal tap (lumbar puncture) may be necessary since some disorders can be detected only in spinal fluid. If, at the conclusion of the history, EEG, MRI, and metabolic testing, no etiology has been identified, the cause of infantile spasms is then called “cryptogenic .”

More than 50 genetic/metabolic diseases have been associated with infantile spasms

THERAPEUTIC INTERVENTIONS

The goal of therapy is to achieve complete control of the spasms. This is very important because if the spasms cannot be controlled, the child is unlikely to develop typically. Unfortunately, even a 50%-90% reduction in the number of seizures does not provide for typical development. For example, if a child’s infantile spasms disorder was causing 100 spasms a day before treatment, but then only has one spasm per day after treatment, his therapeutic intervention is still considered unsuccessful. Unfortunately, for many patients, the underlying disorder does not allow for normal development even if the spasms are completely controlled (for example: brain damage from lack of oxygen or infection). Nevertheless, seizure control should still sought because it greatly improves the patient’s and the parents’ lives.

For other patients, there may be an opportunity to greatly improve the developmental outcome. The patients with the best prognosis are those with a cryptogenic etiology and possibly some patients with tuberous sclerosis or a localized cortical dysplasia. In either case, the treatment that gives the child the best chance to achieve complete control of spasms is the treatment of choice. On rare occasions, the testing will reveal a metabolic or genetic disorder that has a specific therapy which would then be the treatment of choice.

ANTICONVULSANT MEDICATIONS

Medical treatment options are somewhat different for infantile spasms than for other seizure types. There are only two drugs that are approved by the FDA for the treatment of infantile spasms:

- Adrenocorticotropic hormone (ACTH)
- Vigabatrin

Medications that are used to treat older children or adults with epilepsy, such as phenobarbital, carbamazepine, or phenytoin, are rarely helpful. There is some evidence that one or two of the newer drugs or the ketogenic diet may be effective in some patients. So, even patients who fail treatment with ACTH and with vigabatrin may occasionally be controlled with other therapies. Therefore, it is very important to keep trying if the first treatments don't seem to be successful.

FIRST LINE MEDICAL THERAPIES

Adrenocorticotropic Hormone (ACTH)

ACTH is the oldest of the approved medications for infantile spasms. It was the first drug that ever was shown to be successful in treating infantile spasms. In 1958, Sorel reported administering ACTH to seven patients, four of whom responded within a few days and only one of whom had no response at all. ACTH was used for more than 50 years before it was approved by the FDA in 2010 for the treatment of infantile spasms. One drawback from the use of ACTH is that it must be given by injection once or twice each day. One or both parents are taught how to give the injections so that the shots can be given at home.

ACTH is a very powerful medication and has many of side effects. The majority of children will become very hungry and gain weight. Most become very irritable. More serious side effects include:

- High blood pressure
- Decreased glucose levels in the blood
- Stomach ulcer
- Growth retardation
- Heart problems
- Immunosuppression (difficulty fighting infection)

In one study, the risk of serious side effects with ACTH was 43%. ACTH is more likely to be effective at higher doses than lower doses. In addition, the more serious complications occur when the children have been on the medication for an extended period of time. Thus, by treating at high doses and by keeping the duration as short as possible, the child has the best chance for controlling the spasms and the risk of serious complications can be decreased. Children being treated with ACTH must see their doctors frequently for regular blood pressure measurements and blood tests to minimize the risks.

The chance that ACTH will control the spasms at the highest doses may be as

high as 80+%. Some children may have a return of the spasms when the medication has been tapered and stopped. Whether the medication should be repeated a second time is not clear. If ACTH treatment has not been successful within two weeks, it should be rapidly tapered and stopped and another medication should be tried.

Vigabatrin

In 1991, it was reported that a new medication, vigabatrin, showed remarkable efficacy with infantile spasms. Sixty-eight patients who had failed other therapies were treated with vigabatrin as add-on therapy, and 29 of them (43%) showed complete resolution of the spasms. It was also noted that 12 of 14 patients who had tuberous sclerosis responded with complete control. There is some evidence that vigabatrin also may improve developmental outcome in patients with tuberous sclerosis.

It is important to understand how vigabatrin compares with ACTH. Vigevano et al. performed a study of children with newly diagnosed infantile spasms. The patients were given either ACTH or vigabatrin. Eleven of 23 vigabatrin patients responded (1 later relapsed) compared with 14 of 19 patients treated with ACTH (6 later relapsed). After relapses were taken into account, the long-term response rate was similar for both medications. In that study, ACTH was more effective for patients with perinatal hypoxic ischemic encephalopathy.

Vigabatrin generally is well tolerated in young children. Most side effects are not serious. There are reports of decreased muscle tone, sleepiness, or difficulty sleeping. However, there is one side effect that is potentially more serious – visual field constriction (tunnel vision). Blindness has not been reported and central vision appears to be unaffected. The visual loss is usually very subtle; most people who had it were not aware that it had occurred until special testing was performed. It was hard for people to realize that they had a problem, so it took more than a decade to recognize that it occurs. There have been many reports showing that peripheral visual fields are constricted in 15 to 50% of adult patients. It is not known if visual field constriction occurs in very young children because there is no effective method of testing for it. However, given the tragic nature of infantile spasms, even if it is proven to occur in infants, visual field constriction may be an acceptable side effect to trade for seizure control and an improved opportunity for normal development. Despite the visual field issue, many pediatric epileptologists consider vigabatrin to be the drug of choice for children with infantile spasms that are due to tuberous sclerosis and for other conditions as well.

Whether one chooses to treat initially with ACTH or vigabatrin is a decision made by the parents in consultation with their physician. There are circumstances where ACTH may be the best medication to choose and other circumstances where vigabatrin would be best. Most doctors recommend vigabatrin for patients who have infantile spasms associated with tuberous sclerosis.

Prednisolone

A possible alternative to the use of ACTH is an oral medication called prednisone or prednisolone. For many years it was not clear whether one was better than the other. Several studies showed the high-dose ACTH may be superior to what was then used as the dosage for prednisone. Some physicians are now using a much higher dose of prednisolone and finding that it is successful in treating many patients with infantile spasms. A recent study reported using 8 mg per kilogram of prednisolone for two weeks and if the medication was not successful , then trying ACTH. The main advantage of prednisolone is that it can be given orally whereas the ACTH requires daily

injections.

SECOND LINE MEDICAL THERAPIES

Pyridoxine

Pyridoxine (vitamin B6) dependency is a very rare cause of infantile spasms. A trial of 100-mg pyridoxine given intravenously should be administered if diagnosis remains in doubt after the history, examination, and MRI scan have been performed. An immediate normalization of the EEG suggests pyridoxine-dependent epilepsy.

Continued oral administration of high doses of pyridoxine also may be effective for some patients who do not have pyridoxine-dependent seizures. In Japan, high-dose pyridoxine is considered the initial drug of choice by many pediatric neurologists, with reports that approximately 15% respond. While this response rate is lower than either ACTH or vigabatrin, pyridoxine is their first choice based on the safety profile. Side effects include loss of appetite, irritability, and vomiting—all of which are relatively common but modest compared with those associated with ACTH or vigabatrin. Pyridoxine has not found support outside of Japan and a few other epilepsy centers. But, given the low risk associated with its use, it seems reasonable to give patients a 1 to 2 week trial of 100 to 400-mg pyridoxine before starting other medications. If these “standard” medications fail, other therapies must be considered, including other antiepileptic drugs (AEDs).

Valproic Acid

Valproic acid probably has the best anecdotal or unscientific evidence of success in treatment, but there have been no prospective randomized studies of efficacy for infantile spasms. Doses range from 20 mg/kg/day to 100 mg/kg/day. Although none of the reported patients developed liver failure, it nevertheless is a risk in children less than 2 years of age and should be used with caution for children with infantile spasms. This is a difficult limitation to valporic acid’s use because almost all patients are less than 2 years of age. Thus, the risk/benefit ratio should be determined.

Clonazepam

One of the earliest non-steroid treatments for infantile spasms were with the benzodiazepines , including clonazepam and nitrazepam. Nitrazepam was never approved for use in the US, so clonazepam is the only one available that has evidence of efficacy. Clonazepam was first reported to be helpful in a few patients with IS in the 1960’s. It is rarely used today because there are so many better medications.

Several of the new anticonvulsants have some evidence of efficacy.

Zonisamide

Zonisamide has shown some promise as an effective therapy for infantile spasms, but there have been no controlled or comparison trials to date. The Japanese experience suggests that zonisamide may be effective in about a 1/3 of patients. A recent report indicated that 5 of 25 patients with infantile spasms had a complete clinical and electrographic response (EEG pattern) to zonisamide within 1 to 2 weeks, with doses ranging from 8 to 32 mg/kg/day. Zonisamide is generally well tolerated. If the 30% or greater efficacy rates hold up in controlled studies, zonisamide could become a first-line therapy. However, most doctors are not finding the success rate to be that high.

Topiramate

In one study, topiramate was shown to be effective in 4 of 11 intractable infantile spasm patients in doses up to 25 mg/kg/day. Another study reported

that topiramate reduced seizures in 43% of 14 infantile spasms patients, but 29% were made worse and none became seizure free.

Lamotrigine

Lamotrigine is another of the newer AEDs with some anecdotal or scientific evidence of efficacy for infantile spasms, although there are no prospective controlled trials. One report of 3 patients who did not have a successful response to vigabatrin and ACTH treatment responded to lamotrigine after 1 dose. The usual dose of lamotrigine is 6 to 10 mg/kg/day. The major side effect is rash, which is dependent to some extent on how rapidly the dose is increased. The usual recommendation is to increase the dose slowly over 2 months to the minimum expected therapeutic dose. Given the severe nature of infantile spasms and the need to achieve control as soon as possible, taking 2 months to get to a therapeutic level obviously decreases the value of lamotrigine as a therapeutic option. However, if the lowest dose is effective, then lamotrigine becomes a drug to try when standard therapies have failed.

Finally, there are three non-drug therapies that should be considered as options when other therapies have failed:

- Ketogenic diet
- High-dose intravenous immunoglobulin (IVIG)
- Surgery

Ketogenic Diet

The ketogenic diet is a decades-old therapy that is back in popularity. Two recent retrospective reports of 40 children with infantile spasms show diet may control spasms in 20 to 35% of patients who are intractable to other therapies. In the past, there had been a question as to whether children less than 1 year of age could achieve and maintain ketosis. The recent reports indicate that young children can indeed achieve ketosis and may benefit from the diet. Most of the children tolerated the diet well, but there were adverse events, including renal stones, gastritis, hyperlipidemia, and gastroesophageal reflux.

IVIG

High-dose intravenous immunoglobulin (IVIG) has been reported to be helpful in a variety of seizure disorders. One study reported that all six children in their study who had cryptogenic infantile spasms achieved complete remission, but only one of five symptomatic patients responded. Intravenous immunoglobulin doses ranged from 100 to 200 mg/kg/dose given every 2 to 3 weeks to 400 mg/kg/day for five consecutive days. Although there is little data, intravenous immunoglobulin could be considered a possible therapeutic option in patients whose other medical therapies have not been successful. However, the actual efficacy is unclear and the most appropriate dosing and duration have not been defined.

SURGERY

The final nondrug therapy is removing an abnormal part of the brain (cortical resection). This option should be considered for patients who:

- Have failed other therapies including ACTH and Vigabatrin or both
- Have evidence of structural brain abnormalities in a defined area, such as developmental brain abnormalities, brain damage, or tuberous sclerosis.

Not many years ago, infantile spasms were considered to be a generalized seizure disorder, and thus surgery was not possible. It has become clear in the last several years that in spite of the generalized nature of the seizures, an area of cortical abnormality can often be discovered. Removal of the abnormality may lead to control of seizures and, possibly, improved

developmental outcome. The majority of patients who have cortical resection have evidence of focal cortical abnormalities prior to surgical evaluation. For these patients, surgery should be considered early in the course rather than waiting for months or years. Selecting the appropriate candidates for surgery is usually more difficult in infantile spasms than for other types of epilepsy because of the generalized nature of the EEG abnormalities. The following should lead to a referral to a pediatric epilepsy surgery center for further evaluation:

- A careful review of the history (especially history of partial seizures that came first or accompanied infantile spasms)
- The presence of cortical disturbances on MRI scan
- Localized EEG abnormalities that suggest a localized cortical defect

PROGNOSIS

Dr.Riikonen from Finland has followed 214 infantile spasms patients for 20–35 years. She has collected the best long-term follow-up studies of these patients. In her series, nearly 1/3 of the patients died during the follow-up period; many in the first 3 years of life. Many of the 24 patients who died by age 3 died of complications of therapy with ACTH. During the time the study was done, patients were treated with high-dose ACTH for extended periods of time. This caused immunosuppression , which greatly increased the risk of infection such as pneumonia. The current approach to treatment of using ACTH or prednisolone for much shorter periods of time is associated with a much lower risk of death.

Of the 147 surviving patients, 25 (17%) had a favorable developmental outcome with an IQ of 85 or greater. Eleven others were somewhat lower with an IQ of 68–84. Thus, of the 214 patients diagnosed with infantile spasms, 31% died, 45% were developmentally disabled, but 24% had a reasonably favorable outcome.

The outcome is dependent on two major factors. First and foremost is the underlying etiology or cause. Some etiologies will lead to death or mental retardation, whether or not the patient developed infantile spasms. However, children with cryptogenic infantile spasms or infantile spasms that due to treatable causes, such as focal cortical dysplasia, may have a normal or near normal developmental outcome if seizures are controlled. Thus, the goal of therapy is to achieve control as soon as possible, especially for children who may have the potential for normal intellectual development.

The goal of infantile spasm therapy is to achieve seizure control as soon as possible

GLOSSARY

Adverse Event: An unexpected medical problem that happens during treatment with a drug or other therapy. Adverse events do not have to be caused by the drug or therapy, and they may be mild, moderate, or severe. Also called adverse effect.

AED's: Anti Epileptic Drugs

Autism: Autism is a developmental disability that interferes with a person's ability to communicate and socialize. In autism, the different areas of the brain fail to work together. Autism affects each person differently and to varying degrees of severity

Benign: A medical condition that is not considered to be severe or progressive.

Indicates a good outcome.

Benzodiazepines: Benzodiazepine medications are used to relieve anxiety, induce sleep, relax muscles, and relieve muscle spasms or seizures. Benzodiazepines work by slowing down the central nervous system.

Computed Tomography: A series of detailed pictures of areas inside the body taken from different angles. The pictures are created by a computer linked to an x-ray machine. Also called a CT or CAT scan, computed tomography scan, computerized axial tomography scan, and computerized tomography.

Controlled trial: The same as prospective randomized study. Comparison trial tests one medication verses another to determine which is most effective. Comparison trials are not necessarily randomized.

Cortical Dysplasia: A developmental abnormality of the brain.

Cortical Resection: Surgical removal of part of the brain.

Cryptogenic: The term literally means that the cause is “hidden.” It assumes that there is a cause but that we are not able to discover it with all of our testing.

Down Syndrome: A disorder caused by the presence of an extra chromosome 21 and characterized by mental retardation and distinguishing physical features.

Efficacy: Effectiveness. In medicine, the ability of an intervention (for example, a drug or surgery) to produce the desired beneficial effect.

Electrodecremental Response: An EEG change that occurs with infantile spasms where the EEG becomes “flat” for a very brief time when the child has a spasm.

Electroencephalogram: A recording of electrical activity in the brain. It is made by placing electrodes on the scalp (the skin covering the top of the head), and impulses are sent to a special machine. An EEG may be used to diagnose brain and sleep disorders. Also called EEG

Epileptologist: A doctor who specializes in the treatment of epilepsy
Etiology: The cause or origin of disease.

FDA: The Food and Drug Adminstration: The FDA is a government entity that requires drugs to be proven safe and effective. A drug cannot be sold in the US unless the FDA has approved it.

Generalized Convulsion: A sudden attack usually characterized by loss of consciousness and sustained or rhythmic contractions of some or all voluntary muscles. A condition in which muscles contract and relax quickly and cause uncontrolled shaking of the body.

Immunosupression: Suppression of the body’s immune system and its ability to fight infections and other diseases.

Immunosuppression may be deliberately induced with drugs, as in preparation for bone marrow or other organ transplantation, to prevent rejection of the donor tissue. It may also result from certain diseases such as AIDS or lymphoma or from anticancer drugs.

Intractable: Not able to be controlled

Intravenous: Into or within a vein. Intravenous usually refers to a way of giving a drug or other substance through a needle or tube inserted into a vein. Also called IV.

Ketogenic Diet: A diet sometimes used to treat severe seizures. Most of the calories in the diet come from fat and other parts of a normal diet are restricted. Must be done under a physicians direction.

Lumbar Puncture: A procedure in which a thin needle called a spinal needle is put into the lower part of the spinal column to collect cerebrospinal fluid or to give drugs. Also called spinal tap.

MRI: Magnetic Resonance Imaging. MRI is a diagnostic procedure that uses a combination of a large magnet, radiofrequencies, and a computer to produce detailed images of organs and structures within the body.

Neurofibromatosis: A rare genetic condition that causes brown spots and tumors on the skin, freckling in skin areas not exposed to the sun, tumors on the nerves, and developmental changes in the nervous system, muscles, bone, and skin. Also called NF1.

Peripheral Visual Fields:

Prognosis: The likely outcome or course of a disease; the chance of recovery or recurrence.

Prospective Randomized study: A study where the patients are randomly assigned to two different treatments to determine of one is better than the other.

Retrospective: A study of the results of a treatment by reviewing the records of patients.

Risk:Benefit ratio: This is a way to judge the risks (the likelihood that there will be a bad effect) verses the benefit (the likelihood that the treatment will help).

Tapered: Slowly decreased according to the directions of the treating doctor.

Therapeutic Dose: The amount of a medication needed to achieve the desired effect.

Tuberous Sclerosis: A genetic disorder in which benign (not cancer) tumors form in the kidneys, brain, eyes, heart, lungs, and skin. This disease can cause seizures, mental disabilities, and different types of skin lesions.

REFERENCES

Go CY, Mackay MT, Weiss SK, Stephens D, Adams-Webber T, Ashwal S, Snead OC 3rd; Child Neurology Society; American Academy of Neurology. Evidence-based guideline update: medical treatment of infantile spasms. Report of the Guideline Development Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. Neurology. 2012 Jun 12;78(24):1974-80.

Hussain SA, Shinnar S, Kwong G, Lerner JT, Matsumoto JH, et. al Treatment of infantile spasms with very high dose prednisolone before high dose adrenocorticotropic hormone. Epilepsia. 2014 Jan;55(1):103-7.

Kossoff EH. Infantile spasms. Neurologist. 2010 Mar;16(2):69-75.

Mackay MT, Weiss SK, Adams-Webber T, et.al. American Academy of Neurology; Child Neurology Society. Practice parameter: medical treatment of infantile spasms: report of the American Academy of Neurology and the Child Neurology Society. Neurology. 2004 May 25;62(10):1668-81.

Pellock JM. Balancing clinical benefits of vigabatrin with its associated risk of vision loss Acta Neurol Scand Suppl. 2011;(192):83-91.

Pellock JM, Hrachovy R, Shinnar S, Baram TZ, Bettis D, Dlugos DJ, Gaillard WD, Gibson PA, Holmes GL, Nordl DR, O'Dell C, Shields WD, Trevathan E, Wheless JW. Infantile spasms: a U.S. consensus report. Epilepsia. 2010 Oct;51(10):2175-89.

Shields WD. Infantile spasms: little seizures, BIG consequences. Epilepsy Curr. 2006 May-Jun;6(3):63-9.