



FOR HEALTHCARE PROVIDERS: INFORMATION ABOUT INFANTILE SPASMS

WHAT ARE INFANTILE SPASMS (IS)?

IS are a rare but serious form of epilepsy that is characterized by sudden limb movements known as “spasms”. The spasms typically begin in the first year of life, when the baby is between 3 and 8 months of age, and in almost all cases have their onset prior to 3 years of age^{1,2}. It is common for IS to be misdiagnosed as normal startle reflexes or colic. The incidence of IS ranges from 2 to 3.5 cases per 10,000 live births^{1,3,4}.

IS are often seen in children with West syndrome, an epileptic encephalopathy that is accompanied by neurodevelopmental regression and cognitive impairment^{1,3}. The interictal electroencephalogram (EEG) often shows hypsarrhythmia, a high-amplitude irregular pattern of brain activity^{1,3,5}. If the spasms and hypsarrhythmia cannot be controlled, the child may continue to have severe chronic epilepsy and cognitive delay for life⁵. The spasms themselves are usually outgrown by age 5, but the cognitive delay continues, and the child often develops other forms of drug-resistant seizures, such as those of Lennox-Gastaut syndrome⁶. Research has found that 9-35% of children with IS also have autism spectrum disorder^{2,6}.

HOW DO YOU RECOGNIZE INFANTILE SPASMS?

IS are subtle and often go unnoticed since they tend to occur for less than 5 seconds⁷. Most of the seizures involve arm raising and/or extension, flexion at the waist, wide-eyed blinks or dropping of the head⁷. IS typically occur in clusters and often occur upon awakening or during early phases of sleep, though they can be present at other times⁸.

WHAT ARE THE UNDERLYING CAUSES OF INFANTILE SPASMS?

IS has a wide variety of causes. Some cases of IS can be attributed to chromosomal abnormalities and genetic mutations, including mutations in the *CDKL5*, *ARX*, and *DCS* genes². Many children with IS also have a genetic disorder called Tuberous Sclerosis complex, which arises from mutations in the *TSC1* and *TSC2* genes and results in brain malformations^{2,5}. Other known etiologies of IS include fragile X syndrome, focal cortical dysplasia, and Down syndrome^{5,7}. IS may also be acquired via infection, stroke, hypoxic-ischemic insults, or metabolic abnormalities^{2,5}. In the case of cryptogenic IS, there is no underlying or detectable cause³.

HOW ARE INFANTILE SPASMS DIAGNOSED?

If a parent suspects that their child has IS, it is suggested that the spasms be captured on video and shown to a clinician to determine whether the movements are truly spasms². An EEG is then ordered and the child is diagnosed with IS if an EEG shows hypsarrhythmia^{5,7}. The clinician may also suggest getting a magnetic resonance imaging (MRI) of the child's brain to identify potential etiologies². Pre-symptomatic EEG monitoring has been recommended in “at risk” children in an effort to predict the onset of IS and start early treatment.

TREATMENT OPTIONS

- ◆ Upon diagnosis of IS, vigabatrin, an anti-seizure drug, is typically prescribed as the first line of treatment in an effort to stop the seizures and eliminate hypsarrhythmia^{3,5}. If vigabatrin treatment is unsuccessful, then hormone therapy, including adrenocorticotropin (ACTH) injections or prednisolone (oral steroids), is used as a second line of treatment^{2,3}.
- ◆ Research indicates that combination therapy may also offer significant benefits to IS patients in controlling the seizures; in particular, vigabatrin combined with prednisolone^{1,2}.
- ◆ If the child has drug-resistant IS, then alternative treatment options, such as brain surgery or ketogenic diet therapy, should be considered².
- ◆ Early diagnosis and treatment are imperative since the seizures are often associated with poor long-term neurodevelopmental outcomes and cognitive impairment^{2,5,8}. Additionally, earlier treatment of IS may help reduce healthcare utilization⁶.

References

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