

Understanding Sleep-Related Hypermotor Epilepsy (SHE) in Children

Overview

Sleep-Related Hypermotor Epilepsy (SHE), formerly known as **Autosomal Dominant Nocturnal Frontal Lobe Epilepsy (ADNFLE)** is a rare and complex form of epilepsy characterized by mild to severe seizures that occur predominantly during sleep, and often occur in clusters. These seizures are often mistaken for night terrors or nightmares due to their intense nature and the sudden, vigorous movements they provoke. However, SHE is a distinct neurological condition that requires specific diagnostic and treatment approaches. It is known to be caused by gene mutations from several genes including KCNT1.

Symptoms

Children with SHE often display a variety of symptoms, including:

- **Sudden, vigorous movements during sleep:** This can include thrashing, facial grimacing, vocalizations, rocking, and abrupt awakening or wandering. Seizures may be preceded by an aura such as choking, breathlessness, shivering and others.
- **Cognitive and behavioral challenges:** Most people with SHE are intellectually normal, however, untreated seizures or sleep disruption from frequent seizures can lead to behavioral challenges.
 - **Autistic-like behaviors:** Some people with SHE may experience psychiatric disorders, behavioral disorders or intellectual disability. Some may receive an autism diagnosis, or exhibit repetitive behaviors, social challenges, or heightened sensory sensitivities, potentially exacerbated by medications.
 - **Impact of Sleep Disruption:** Poor sleep can cause irritability, mood swings, and difficulties with concentration and memory.
 - **Neurological Differences:** Brain abnormalities associated with SHE may result in hyperactivity, impulsivity, and challenges with executive function, and may appear as defiance or intentional misbehavior.
 - **Medication Side Effects:** Anti-seizure medications are essential for controlling seizures but can also cause side effects such as dizziness, weakness, and changes in behavior, including mood swings, depression, and anxiety.
- **Speech Delay and Fine Motor weakness:** Research suggests that epilepsy, particularly when seizures involve the frontal lobe, can lead to broader developmental delays, including in speech and fine motor skills. This is because the frontal lobe is integrally involved in coordinating and planning movements, as well as in executing complex motor activities. ([BioMed Central](#))

Diagnosis

The Importance of Nighttime EEG Monitoring

Diagnosing Sleep-Related Hypermotor Epilepsy (SHE) requires careful and often repeated evaluations due to the complex nature of the condition. A nighttime Electroencephalogram (EEG) is a critical tool for identifying the electrical brain activity associated with nocturnal seizures. However, it's important to understand that a single EEG may not always capture all seizure activity, especially in cases where seizures are deep-seated within the brain or occur infrequently.

Key Points to Consider:

- **One EEG May Not Be Enough:** Seizures originating deep within the brain, particularly in the frontal lobe and extra-frontal areas, can sometimes be missed on a standard surface EEG due to the skull and scalp's insulating properties. Additionally, the semiology of these seizures—how they present physically—can be subtle or complex, making them harder to detect. This is why repeated or extended EEG monitoring, possibly including video EEG, is often necessary to capture these events.
- **Continuous Monitoring:** Given the episodic nature of SHE, continuous or overnight monitoring increases the likelihood of capturing seizure activity that may be missed in shorter, routine EEG sessions. This comprehensive approach is essential for making an accurate diagnosis and guiding effective treatment and safety strategies.

Neuroimaging: MRI scans can help identify structural abnormalities in the brain, particularly in the frontal lobe, which is often implicated in SHE.

Genetic Testing: Genetic mutations, such as those in the KCNT1 gene, are strongly associated with SHE. The KCNT1 mutation is one of the more serious genetic factors, with higher penetrance, meaning it is more likely to cause the disorder if present. It is important to note that there are many KCNT1 Variants of Uncertain Significance that have not been widely seen but present with likely pathogenic clinical symptoms and should be discussed with a genetic counselor. Additionally, other genetic variants, as well as psychological and environmental factors can also result in behavioral disorders, making it crucial to consider a comprehensive approach when assessing these conditions.

Treatment Options

Treatment for SHE typically involves a combination of:

- **Antiepileptic Drugs (AEDs):** Medications like carbamazepine, oxcarbazepine, valproate, lamotrigine, topiramate, levetiracetam, and Epidiolex (CBD) are commonly used to control seizures. The choice of medication may depend on the child's specific symptoms and seizure types.
- **Supportive Therapies:** Speech and Physical therapy can address delays. Counseling and support for both the child and family can help address the emotional and social challenges associated with living with SHE. Family members should be educated on seizure first aid, and emergency plans should be in place at home and school.

Discuss the Risk of Sudden Unexpected Death in Epilepsy (SUDEP)

While rare, people with epilepsy, including SHE, are at risk for SUDEP. This risk underscores the importance of consistent monitoring, appropriate treatment, and safety measures to mitigate the risk. It's crucial to have a personalized Seizure Action Plan designed by a clinician. This plan should include medication management, emergency protocols, and safety measures like night-time monitoring.

Nighttime Safety Monitoring: Tools like night cameras and pulse oximeters can detect nocturnal seizures and alert caregivers for quick intervention. Some devices may be covered by insurance, so it's worth discussing options with your healthcare provider. Regularly review and adjust safety measures as your child grows.

Safe Sleep Environment: To minimize injury risks during seizures, consider special safety beds with railings, seizure-safe pillows, lightweight bedding, and door alarms. Padding sharp furniture edges and using night cameras can also enhance safety.

Regular reviews and adjustments to safety measures should be conducted as the child grows and their condition evolves.

Further Reading and References

For clinicians seeking more detailed information, the following references provide in-depth insights into the diagnosis, treatment, and management of SHE:

1. **"Neuropsychological and Behavioral Profile in Sleep-Related Hypermotor Epilepsy (SHE) and Disorders of Arousal (DOA): A Multimodal Analysis"**
 - URL: <https://www.mdpi.com/2077-0383/12/1/374>
 - This study explores the cognitive and psychiatric profiles of patients with SHE, emphasizing the impact of sleep instability on cognitive functions.
2. **"Sleep and Epilepsy: A Focused Review of Pathophysiology, Clinical Syndromes, Co-morbidities, and Therapy"**
 - URL: <https://link.springer.com/article/10.1007/s13311-021-01002-y>
 - This review provides an overview of the relationship between sleep, epilepsy, and cognitive and behavioral comorbidities, highlighting the effects of sleep-related seizures.
3. **"Sleep-Related Hypermotor Epilepsy: A Video-Polysomnographic and Genetic Appraisal of 40 Patients and Delineation of the Epileptic Syndrome"**
 - DOI: 10.1093/brain/awl033
 - This article discusses the genetic underpinnings and clinical features of SHE, providing valuable insights into its diagnosis and management.
4. **"Characteristics of Sleep-Related Hypermotor Epilepsy Pertinent to Dentistry: A Case Report and Review of the Literature"**
5. **"Sleep related hyper motor epilepsy (SHE): a unique syndrome with heterogeneous genetic etiologies"**, Bisulli, F., Licchetta, L. & Tinuper, P. Sleep related hyper motor epilepsy (SHE): a unique syndrome with heterogeneous genetic etiologies. *Sleep Science Practice* 3, 3 (2019). <https://doi.org/10.1186/s41606-019-0035-5>

If you suspect your child may be having seizures, it is crucial to seek a thorough medical evaluation and genetic testing. Early diagnosis and intervention can significantly improve the quality of life for children with this condition. Consult with a pediatric neurologist, discuss genetic testing, and explore all available treatment options to ensure the best possible outcomes. If your child has had a seizure, they may be eligible for free genetic testing through the [Unlock](#) program.

For additional support or questions, contact us at: support@kcnt1epilepsy.org

Medical Disclaimer

The information provided in this brochure is for educational purposes only and is not intended as a substitute for professional medical advice, diagnosis, or treatment. Always seek the advice of your physician or other qualified healthcare provider with any questions you may have regarding a medical condition. Never disregard professional medical advice or delay seeking it because of something you have read in this brochure. If you think you may have a medical emergency, call your doctor or emergency services immediately. The inclusion of references and resources is intended to provide helpful information, but does not imply endorsement of any particular diagnosis, treatment, or therapy.

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