Pediatric Epilepsy Syndromes

Monisha Goyal MD discusses pediatric epilepsy syndromes. She examines the difference between benign & catastrophic epilepsy syndromes and she shares the role of genetic testing in for these conditions. She offers the latest treatment options available and when it is important to refer to the experts at UAB Medicine.

Featured Speaker:

Monisha Goyal, MD

Monisha Goyal, MD Specialties include Clinical Neurophysiology, Epilepsy Neurology, Pediatric Neurology and Pediatrics.

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Release Date: June 7, 2021
Expiration Date: June 6, 2024

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Welcome to UAB Med Cast, a continuing education podcast for medical professionals. Bringing knowledge to your world. Here's Melanie Cole.

Melanie: Welcome to UAB Med Cast. I'm Melanie Cole and, today we're discussing pediatric epilepsy syndromes. Joining me is Dr. Monisha Goyal. She's a pediatric epileptologist at UAB Medicine. Dr. Goyal, it's pleasure to have you join us today. Tell us a little bit about pediatric epilepsy syndromes, this umbrella and tell us a little bit about how they've evolved over the years.

Dr Monisha Goyal: So epilepsy is a common neurologic disorder affecting 1 to 1.5% of the world's population. And it's mostly diagnosed in children and in the adults, but more in the elderly. And so a pediatric epilepsy syndrome is defined the cluster of signs and symptoms typically occurring together. Examples include age of seizure onset, types of seizures, precipitating factors, et cetera. The grouping of these signs and symptoms into an identifiable epilepsy syndrome helps determine appropriate therapy. It helps determine prognosis and including genetic risk for other family members.

So pediatric epilepsy syndromes can be grouped or classified in many ways. Most commonly, this is done by age of presentation, such as neonatal epilepsy syndromes, syndromes presenting in infancy, childhood, adolescence, et cetera. However, one of the distinguishing features of pediatric epilepsy is the remarkable dichotomy between the so-called benign and the catastrophic epilepsy syndrome. And that's what I sort of want to emphasize and discuss further.

Melanie: Well, then tell us a little bit more about that. Expand on the difference between benign and catastrophic epilepsy syndromes for us.

Dr Monisha Goyal: So the so-called benign syndromes have no structural brain abnormality. They're sensitive to seizure medications, have a higher emission rate and have a low association with comorbidities such as cognitive delay or behavioral issues. These children, epilepsy may have a negative impact on their quality of life, but this is more an irritation and it's usually temporary. It's not a disaster.

However, for children with catastrophic epilepsy syndromes, seizures are part and parcel of their everyday life. They cause significant morbidity and mortality, including global developmental delay and intellectual disability.

Melanie: What an interesting topic we're discussing here today. So what are some of the common benign and catastrophic syndromes that you see?

Dr Monisha Goyal: So I'm going to discuss a few benign epilepsy syndromes and then we'll go into the catastrophic ones. The benign epilepsy syndromes that I'm going to discuss include benign familial neonatal convulsions or BFMC, childhood absence, benign rolandic epilepsy and juvenile myoclonic epilepsy. Again, these are just some of the examples, but I thought I'd choose a few of the more notable ones.

So, benign familial neonatal convulsions. this is an autosomal dominant condition. It presents with focal or generalized seizures, sometimes with apneas or autonomic symptoms. And it generally begins between two and eight days of life. Seizures do resolve in 60 to 70% of patients by one and a half months of age. Neurodevelopment is usually normal, but there is an increased risk of subsequent epilepsy in about 15% of the patients. And treatment with phenobarb is usually initiated due to the flurry of seizures, which can be up to 30 per day.

However, treatment may not affect seizure recurrence or cessation. And typically, this syndrome has a favorable outcome with spontaneous resolution. There are patients who do develop epilepsy after the syndrome and these type of seizures usually go into the epilepsy syndrome of benign epilepsy with centro-temporal spikes or benign rolandic epilepsy that we'll discuss in just in a few minutes.

Another example of a benign syndrome is childhood absence. And childhood absence usually begins between four to eight years of age and the main seizure type are absence seizures or brief staring spells. Children are usually not aware or responsive and abruptly return to normal. The staring can sometimes be accompanied by automatisms or automatic movements thought to be of sub-cortical origin. These include lip-smacking or pill-rolling type movements. So you can have some motor manifestations along with staring.

And the seizure duration is 10 to 20 seconds and these usually occur multiple times a day. The classic EEG pattern that most of us are familiar with is 3 Hertz generalized spike-and-wave pattern during a seizure. Development is usually normal in these kids. though they may have a higher rate of attention problems. And two out of three children with childhood absence do respond to treatment and seizures usually disappear by late childhood or mid-adolescence.

And then another common example of a benign epilepsy syndrome is benign rolandic epilepsy. This has many names. It goes by benign centro-temporal epilepsy or epilepsy with centro-temporal spikes. This is the most common focal epilepsy of childhood and the onset is usually around five to seven years of age. Children complain of unilateral sensory motor symptoms, including paresthesias of the mouth, the tongue, lips, their face may pull down. So they have a tonic deviation and have clonic movements of the lower face.

And occasionally, the seizure will spread to the ipsilateral lower body and, in some instances, generalized. Seizures can begin on either side and speech arrest may also occur due to loss of coordination of muscles needed for speech articulation. The seizures generally tend to occur from sleep. Benign rolandic epilepsy remits by 16 or so years of age, and many children require no treatment whatsoever.

Another example of a benign epilepsy syndrome is JME or juvenile myoclonic epilepsy. And this is a syndrome with onset around adolescence in otherwise healthy individuals. The most common seizure type is a myoclonic jerk. And these are quick jerks of the arms and legs, which is the hallmark of this disease. And these usually occur upon awakening. This may be the only symptom in about 15 to 20% of cases. Other seizure types in this syndrome include generalized tonic-clonic seizures. These usually appear a few months after onset of myoclonic jerks and some folks will also have absence seizures.

Seizures in this syndrome are triggered just like many others syndrome. Triggers include lack of sleep, fatigue, stress, alcohol, flickering lights or menses. And in most, seizures are well-controlled with a single drug such as lamotrigine or valproic acid. However, in some other cases, you may need two or three drugs. Although patients usually require life-long treatment with anti-seizure medications, their overall prognosis is usually very good.

So those were examples of benign syndromes. I'm going to go ahead and focus now on catastrophic epilepsies. Examples that I'm going to discuss are infantile spasms, Lennox-Gastaut syndrome, Dravet syndrome, and progressive myoclonic epilepsy. So they're sort of age-based.

The first syndrome is infantile spasms. The infantile spasms, essentially this is an epileptic spasm. It's a seizure type of infancy and childhood. And this syndrome that the infantile spasm encompasses is called West syndrome. Epileptic spasms are more commonly called infantile spasms since they typically are seen in the first year of life.

So West syndrome or infantile spasms is characterized by epileptic spasms, developmental delay, and an interictal EEG pattern called hypsarrhythmia, which means the EEG pattern seen in between seizures or spasms. The onset is usually in the first year of life, typically between four to eight months.

The seizures often look like sudden bending forward of the body with stiffening of the arms and legs. These lasts about one to two seconds. Some children arch their backs as they extend their arms and legs. Spasms tend to occur in multiple clusters per day and most children, but not all will have hypsarrhythmia. Spasms usually resolve, but they may be replaced by other seizure types.

Etiologies of the syndrome are diverse. They include birth injuries, such as hypoxic ischemic encephalopathy, metabolic and other genetic etiologies. And in some children, no cause is found. The prognosis in this syndrome is largely dependent on the underlying cause of this syndrome. Intellectual prognosis is generally poor. Many of these

children have neurological impairment prior to the onset of the spasms and that dictates the future prognosis in terms of seizure control and development.

Children who have rapid initiation of treatment who have normal development prior to infantile spasms and have no identifiable cause, these children may do well. Infantile spasms usually resolve by mid-childhood. But more than half of the children with infantile spasms will later develop other seizure types, such as Lennox-Gastaut syndrome. This is a electroclinical syndrome with onset between one to eight years of age. And this syndrome includes many seizure types, including tonic, atonic, atypical absence. The EEG has interictal widespread 1.5 to 2.5 Hertz slow spike-and-wave discharges. And these children have severe intellectual disability.

Treatment includes multiple drugs, ketogenic diet, and palliative measures such as corpus callosotomy or vagal nerve stimulator. However, the outcome for LGS remains disappointing in regards to both seizures and cognitive outcome.

Melanie: Well, thank you for that comprehensive answer, Dr. Goyal. So tell us a little bit about the role of genetic testing in these epilepsy syndromes you've been discussing. Who should get genetic testing and when should genetic testing be performed and on who? Does that mean siblings? Just briefly touch on genetic testing for us.

Dr Monisha Goyal: Historically, up to 70% of epilepsy etiology has been deemed idiopathic. But with currently available genetic testing, an etiology may be found in more than 30% of the population. So what was considered idiopathic may not be and that is because of the availability of genetic testing. So according to recent International League Against Epilepsy recommendations, all infants with seizures and all individuals of any age who have failed one anti-seizure medication, these folks should be referred to a tertiary care center and genetic counseling should be available to all epilepsy patients.

So though the cornerstone of epilepsy valuation includes EEG and MRI, all epilepsy that appears to be following a refractory pathway should have other testing, which includes genetic testing. So genetic testing includes many things such as chromosomal analysis, gene panels.

When should one get this type of testing? I think anybody with global developmental delay who has seizures should get a chromosomal microarray. This is indicated for all children with developmental delay. Metabolic testing should be taken at the onset of epilepsy in infants without structural or syndromic cause of seizures and then genetic testing follows the metabolic testing.

Timely genetic diagnosis may reduce overall cost, limit invasive tests and improve prognostic accuracy. Usually, the genetic testing is done on the patients and, depending on the results, family testing can also be done.

Melanie: Dr. Goyal, as we wrap up, do you have any final thoughts for other providers and what you would like them to know about pediatric epilepsy syndromes and when you feel it's important they refer to the specialists at UAB Medicine?

Dr Monisha Goyal: Anytime a patient is not responding to seizure medications, and this usually involves the first or second try. If they have failed one or two medications and continue to have seizures, they should be referred to a tertiary care center such as our center. Also any infants with seizures, because they may have other underlying causes, should be referred to a tertiary care center for further evaluation.

Melanie: Thank you so much, Dr. Goyal. What an informative episode this was. Thank you for joining us. And a physician can refer a patient to UAB Medicine by calling the MIST line at 1-800-UAB-MIST.

That concludes this episode of UAB Med Cast. For more information on resources available at UAB Medicine, you can always visit our website at UABMedicine.org/physician. Please also remember to download, subscribe, rate, and review this podcast and all the other UAB Medicine podcasts. I'm Melanie Cole.