Given the poor prognosis of Lennox-Gastaut Syndrome with regards to seizure control and cognitive outcome, it is very important to apply that term with caution.

How do we go about diagnosing it and what kind of investigations do we have to do once we diagnosis it? Well, the diagnosis really relies on that classic triad of many seizure types, mental retardation, and the classic EEG features that we just went over, mainly the slow spike in wave. Some people consider the presence of ten-hertz fast activity as an essential for diagnosis. This can be either associated with atonic seizures or can occur with minimal clinical manifestations, such as apnea or perhaps truncal rigidity, that can be seen mainly during non-REM sleep.

As we said, the diagnosis relies heavily on the EEG. Although it sounds easy to diagnosis since you have all these features, the features may not be very clear early at onset. Why? The cause, the history, the seizure types, the EEG features, are not pathognomonic for Lennox-Gastaut Syndrome. They are shared by other epilepsy syndromes and can occur in other seizure types. In addition, the core seizure types may not be present at onset. You can initially have focal seizure or the patient may present with myoclonic seizures, which can complicate the picture.

The EEG features we talked about extensively, but I just want to emphasize that a sleep recording is almost essential, not just a sleep recording, but a video sleep recording. Since a diagnosis relies so heavily on finding that classic EEG feature and since tonic seizures tend to occur mostly during sleep, a sleep EEG is essential. The background is abnormal early at onset being slow and poorly organized. Then, you start seeing these diffused slow spikes in wave discharges occurring mainly while awake. Sometimes, it is difficult to decide if this is an ictal or an interictal recording because come of these patients are not very responsive at baseline, so it is sometimes hard to decide whether there is an alternation in the level of consciousness or not.

As we mentioned, you have to focus on these fast rhythms that are essential for diagnosis and are essential in differentiating Lennox-Gastaut Syndrome from other epilepsy syndromes in which slow spike in wave discharges can be seen.
Every child with Lennox-Gastaut Syndrome should have an MRI, but there are no classic features for Lennox-Gastaut Syndrome and MRIs. Why? The etiology is very different. The findings on the MRI are inconsistent. You can see focal or multifocal malformations of cortical development. You can see evidence of ischemic injury that has occurred in the perinatal period or in early childhood. You can find manifestations of tuberous sclerosis or sometimes, you can have some MRI features of metabolic disorders.

The remainder of the testing really depends on the clinical situation. If you’ve resolved the problem with an MRI, there is no need for further investigations. But if you have not, you have to pursue a more elaborate evaluation, which can include chromosomal testing, and then depending on the patient, you might have to more specifically request genetic tests depending on what is your probability or what do you think the child might have. Metabolic disorders are a rare cause of Lennox-Gastaut Syndrome. Then again, if you have not reached a diagnosis with your chromosomal testing or MRI, you might have to pursue an elaborate metabolic evaluation, which may include CSF studies looking for neurotransmitters, vitamin dependencies, markers of vitamin dependencies, amino acids, and lactate for mitochondrial disorders.

Also when you diagnosis a child with Lennox-Gastaut Syndrome, it is important to think about what are your differential diagnoses. Well, Doose Syndrome or myoclonic astatic epilepsy is one important differential. These kids typically have multiple seizure types. However, tonic seizures are not a prominent feature. They are typically somewhat younger. They are more likely to be normal behaviorally and cognitively at the time of diagnosis. They are more or less easier to control and have a better prognosis. Other syndromes that you need to consider are Dravet Syndrome or infantile myoclonic epilepsy. Then you have to think about early onset absence epilepsy and sometimes, even focal epilepsy, such as frontal lobe epilepsy can have tonic seizures as a main feature. However, these tonic seizures are frequently asymmetric in their presentation.

Now what are the neuropsychological features of Lennox-Gastaut Syndrome? As we have heard, it affects cognition and behavior. Most patients are impaired even before they present with seizures. About 20 to 60 percent have mental retardation present at their presentation and this becomes more problematic over time. By five years into have the syndrome, 75 to 95 percent of patients will have mental retardation, and different studies have shown that IQ actually drops by about 15 points if you follow these kids ten years.

This cognitive deterioration is more frequent the more frequent the seizures are. It is more frequent in patients who have frequent episodes of status epilepticus, and it is worse the earlier the inset of epilepsy. However, always remember that cognitive deterioration can occur even if seizures are well controlled. Now, it is not clear why this cognitive deterioration occurs. It is not clear whether it is related to progressive brain damage or whether it is related to failure of brain development. Sometimes, these cognitive skills may fluctuate depending on the level of seizure control. For example, if you have a child having very frequent atypical absences, they’re going to perform less well than if their absences are well controlled.

We always have to remember the behavioral problems that occur in patients with Lennox-Gastaut Syndrome, attention problems being very common. Aggression can also occur. Some of these kids have autistic features and less frequently, psychosis may be present.
When we approach a child with Lennox-Gastaut Syndrome, we have to think about it globally. A main thing is to reach an agreement with regards to treatment between the caregivers or the family and the treating physician. It is very important to outline this agreement so that you don’t set yourself up for failure. You have to focus on quality of life as your criteria for successful treatment more than seizure control.

However, having said that, the aims might be different depending on the stage of the disease and the particular child. For example, if you have just met a child that is newly diagnosed and is undergoing developmental deterioration, you’re going to try to address that child more aggressively than a child who has had Lennox-Gastaut Syndrome for a long period of time. You know their trajectory by now, and you know that quality of life issues are now more important to be addressed than seizure control.

Because the seizures are very resistant to treatment, seizure reduction, again, may not be your ultimate goal. Always remember that sometimes the quality of life is more impaired by the treatments that we try to give these patients rather than the seizures themselves.

When you try to treat a child with Lennox-Gastaut Syndrome, you have to think about their quality of life. The quality of life can be impacted in these kids by several factors. There is the social impact. The patients and their families have to deal with the stigma not only of this intractable epilepsy, but also of the cognitive issues and the behavioral issues. There are the sleep difficulties, which have a high impact on quality of life. Sleep is very hard in these patients. They may not sleep properly because of the medications, because sleep is interrupted by the frequent seizures, or sometimes, they simply have poor sleeping habits. This is a very common finding with patients with intractable epilepsy and behavioral problems.

There is also the physical impact of Lennox-Gastaut Syndrome. Injuries are very common, especially to the face and the mouth. Eventually as they grow older, gate disturbances are quite common. These frequently result in the patients being wheelchair bound. Sometimes, the wheelchair is an elective thing to reduce the effect or the impact of the falls on these patients, and they frequently end up wearing helmets. We have talked about the cognitive and the behavioral impact on these patients with progressive mental retardation and numerous behavioral problems that are difficult to deal with.

Of course, we should not forget the impact on the caregiver. The caregiver’s life is equally important as the child’s life. Seizures are unpredictable in these kids. The parents feel helpless. They cannot prevent the injuries. This results in parental anxiety. There are frequent reports of PTDS, depression and anxiety in these families. A lot of these families suffer from marital problems, job stress. There is the financial burden of having to deal with the seizures, the behavior and the cognitive problems. It is very difficult for these families to find resources within the community to help them care for these children.

Because of that, it is very important to have a team approach. It is very important for the neurologist or the Epileptologist to be the leader of that team, to have the treatment plan, the goals, and to identify all these important features that impact the quality of life, and based on that, recruit different resources. You might need mental healthcare providers with psychology or psychiatry that sometimes need to be involved. Then you have the sleep medicine, developmental pediatrics, social work. A dietician is frequently essential. You then come to a major thing in the child’s development or in the child’s life, which is transition into adult care.
About 80-percent of children with Lennox-Gastaut Syndrome will continue to experience seizures as adults. These seizures continue to be very difficult to control. Different studies have found that patients, who are able to control different aspects of their life and direct their care, have better quality of life. They also found that patients who end up living in group homes have a better functional status. They have a better level of seizure control and self-sufficiency. Also, their caregiver’s life is much better and they have better functional status.

However, most families are very hesitant to leave a pediatric practice. Most pediatric neurologists are very hesitant to give up their patients. Attachment happens at both sides of the aisle. There is always concern that if I leave my pediatric neurologist, I may not have enough time with an adult neurologist. Sometimes, it is very difficult to coordinate this transition and of course, there are insurance issues that come in the way.

Some people suggest a transition clinic staffed by an adult neurologist, a pediatric neurologist nursing from both sides, adult and pediatrics, so that the handing of care happens face-to-face with the patient present there. It is important when you transition these families into adult practice to educate them about all that’s going to happen. For example, you have to advise them to obtain guardianship before age 18. You have to advise them with regards to possible changes in health insurance. You have to try to refer them to supportive agencies for disabilities, and to tell them and help them organize their medical records and medications for these patients.

Things that need to be addressed as these kids became teenagers and young adults are productive care, bone health, being exposed to multiple anti-epileptic medications over time. Of course, there is the issue of safety at home and in the community, advising them to have personal and medical identification, alarms. There’s a high risk of abuse in patients with developmental disabilities and intractable epilepsy. You have to discuss with them the options for different living environments and vocational training and life skills.