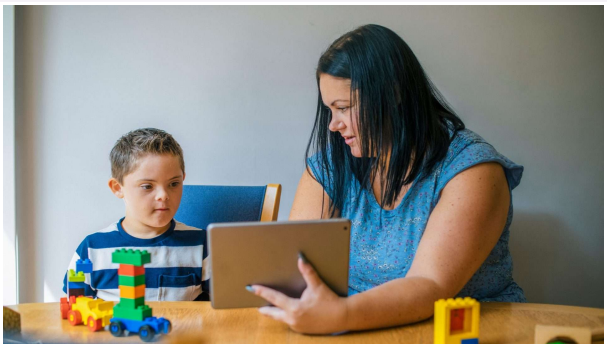
Developmental Epileptic Encephalopathy



Developmental and Epileptic Encephalopathies (DEEs) are a group of rare and severe epilepsies. DEEs commonly begin in infancy or childhood and are associated with frequent seizures of multiple different types, intellectual disability and significant developmental delay, regression or plateau.

UNDERSTANDING DEES

Other conditions, such as movement disorders, autism spectrum disorder (ASD), and behavioural, feeding and sleep problems, are common. Frequent epileptic activity, which further disrupts development, over and above that expected from the underlying cause alone, is the hallmark feature of DEEs. These conditions can be challenging to diagnose and manage because of their complexity, as seizures tend to be resistant to anti-seizure medication (ASM). Early diagnosis and treatment are essential in improving long-term developmental outcomes. (1)

CHALLENGES OF DEES

Every person's DEE experience and challenges are different. Apart from high seizure frequency, other common challenges can include:

- Intellectual disability
- Impaired communication skills (e.g. speech, facial expressions, gestures and body language)
- Behaviour issues
- Loss of motor skills
- Psychiatric issues (e.g., autism spectrum disorder, depression, anxiety, psychosis)
- Sleep issues

- Gastrointestinal issues (e.g. loss of appetite and weight loss, abdominal pain, cramping, nausea or vomiting, constipation, assisted feeding)
- Visual impairment
- Respiratory issues
- Orthopedic issues
- Sudden Unexpected Death in Epilepsy (SUDEP)

WHAT CAUSES DEES

DEEs are mostly genetic conditions where changes in the genetic code (DNA) disrupt normal brain function and brain development. These gene changes are often new in the person with DEE (*de novo*). They can also be inherited from healthy parents (recessive). Other non-genetic causes of DEEs include:

- Head or brain injuries
- Metabolic factors (can have a genetic basis)
- Unknown factors

SEIZURE TYPES

People with DEE can have a wide range of seizure types. Every person's seizure experience is different, and not all seizures are alike. However, the patterns of seizures tend to be consistent.

Some people with a DEE will meet the criteria for specific epilepsy syndromes. This refers to a cluster of symptoms and features that usually occur together. Some common epilepsy syndromes that are DEEs include:

- Early infantile epileptic encephalopathy (EIDEE), including Ohtahara syndrome and early myoclonic encephalopathy (EME)Infantile epileptic spasms syndrome (IESS), including West syndrome
- Dravet syndrome
- Lennox Gastaut syndrome (LGS)
- Febrile infection-related epilepsy syndrome (FIRES)
- Epilepsy of infancy with migrating focal seizures (EIMFS)
- Epilepsy with myoclonic-atonic seizures (EMAS), otherwise known as myoclonic-atonic

epilepsy (MAE) or Doose syndrome.

- Landau-kleffner syndrome (LKS)
- Developmental and epileptic encephalopathy with spike and wave activation in sleep (DEE-SWAS)

If a person has DEE, the diagnosis and evaluation may include:

- Genetic testing via blood, saliva samples or a skin biopsy
- Sleep and wake electroencephalogram (EEG) and or Video electroencephalogram monitoring (VEM)
- Magnetic resonance imaging (MRI)
- Positron emission tomography (PET)
- Determining the seizure types and frequency
- Identifying other associated conditions that impact development, learning, and everyday living
- Choosing the best treatment options for each person

TREATMENT FOR DEE

While ASMs are always the first line of treatment for epilepsy, gaining seizure control for DEEs can be difficult. The type of treatment a person receives will vary significantly based on their unique condition. Understanding and accurately diagnosing the underlying causes can better help inform treatment options and improve developmental progress.(1) Only some people with a DEE can achieve seizure control. Therefore the goals around seizure management may be different for each person. It is essential that you discuss ASM options with your neurologist and find what best works for you.

Treatment and management options for DEEs may include:

- Anti-seizure medication (usually the first line of treatment)
- Surgical treatment
- Dietary management (Ketogenic Dietary Therapies)
- Allied health (such as occupational therapies, physiotherapies, and speech therapies)
- Managing individual seizure triggers and risks (around heights, water or fire)

DEE RESOURCES

The National Epilepsy Support Services (NESS) is an Australia-wide resource that provides support and information for people living with epilepsy and DEE and those who assist them. NESS supports people of all ages and at all stages of life.

The NESS is available Mon – Sat, 9:00am – 7:00pm (AEST) to provide support and information across Australia. Phone: 1300 761 487. Email: support@epilepsysmart.org.au

Genetic Epilepsy Team Australia (GETA) is a group of parents whose children have rare genetic epilepsies. Join us to help shape a better healthcare system for our children by supporting the development of treatments and ultimately finding a cure for genetic epilepsy.

Website: www.geneticepilepsyteam.com.au

Facebook: @geneticepilepsy

Twitter: @geneticepilepsy

Email: hello@geneticepilepsyteam.com.au

Genes4Epilepsy is a website developed by the Epilepsy Research Centre which contains information and resources about genetic epilepsies, including informational videos about a range of topics related to DEEs.

Website: www.genes4epilepsy.org

References

1. Scheffer IE, Liao J. Deciphering the concepts behind “Epileptic encephalopathy” and “Developmental and epileptic encephalopathy”. Eur J Paediatr Neurol. 2020;24:11-4.

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