

Epilepsy and Related Syndromes - Useful Organisations

What is a Syndrome?

There are many different types of epilepsy classifications. Nowadays, most people are given a specific diagnosis or name for their epilepsy, rather than just told that 'you have epilepsy'. Some types of epilepsy are classified as syndromes and are defined based on a unique combination of symptoms.

An epilepsy syndrome is a type of epilepsy that depends upon:

- family history
- what age the seizures started
- seizure type[s]
- cause, and other linked diseases or conditions
- how the syndrome progresses over time
- presence or absence of brain abnormalities
- EEG findings
- brain scan findings [such as CT, MRI, PET]
- response to medication.

A diagnosis of a particular epilepsy syndrome is useful in deciding the possible treatment options, what course the condition may take, and the possible genetic risk of passing it on to offspring.

Useful Resources

Below is a list of syndromes associated with seizures and epilepsy, and related support organisations or groups. This list is not exhaustive, and there are some organisations that are not based in Australia but contactable online.

Epilepsy Syndromes Causing Disability	
<p>Epileptic Encephalopathy with Continuous Spike and Wave Syndrome during Sleep (CSWS) ^{1,2,3} Also sometimes termed Electrical Status Epilepticus in Sleep (ESES)</p>	<p>This is a rare epilepsy syndrome or epileptic encephalopathy and often presents between the ages of 3-7 years. The child shows deterioration in a wide range of developmental abilities, including speech, language, motor skills, learning, memory, and behaviour. Development before onset may be normal. EEG during Non-REM sleep shows continuous epileptiform activity (with no clinical signs of seizures). A number of different seizure types can be seen, and can occur during wakefulness. This syndrome can disappear around the age of 11 years, approximately 3-4 years after onset. This results in cognitive and behavioural improvements as well. Seizures usually disappear around puberty but neuropsychological consequences may persist. There are some similar features to Landau Kleffner Syndrome.</p> <p>https://www.facebook.com/groups/EpilepsyLKS/ Raising a child with LKS and CSWS</p> <p>https://www.facebook.com/groups/149591171771780/ Raising a child with CSWS</p>

1 http://crf.med.harvard.edu/publications/JMPeters_JClinNeuro2011.pdf

2 <http://link.springer.com/article/10.1684%2Fepd.2012.0482#page-2>

3 <http://www.acnr.co.uk/2012/12/continuous-spike-wave-discharges-in-slow-wave-sleep-dont-let-it-catch-you-napping/>

<p>Dravet Syndrome Also called Severe Myoclonic Epilepsy of Infancy (SMEI)</p>	<p>This is a severe form of epilepsy where seizures are poorly controlled. It appears during the first year of life with frequent fever related seizures. Later, other types of seizures appear, including myoclonus (muscle spasms). Status epilepticus also may occur. Children with Dravet syndrome usually experience poor development of language and motor skills, hyperactivity, and difficulty relating to others.</p> <p>http://www.dravet.org.uk/ Dravet Syndrome UK</p> <p>http://dravetdata.com/ DravetData.com Go to recommended links for support.</p> <p>http://www.dravetfoundation.org/ Dravet Foundation USA</p> <p>https://www.facebook.com/DravetSyndromeFoundation</p>
<p>Landau Kleffner Syndrome (LKS)⁴ Also called Infantile Acquired Aphasia, acquired Epileptic Aphasia or Aphasia with Convulsive Disorder</p>	<p>This is a rare childhood condition. LKS occurs most frequently in normally developing children between 3 and 7 years of age. A major feature of LKS is the gradual or sudden loss of the ability to understand and use spoken language. Not all children have seizures, and they are usually associated with sleep when they happen. Behavioural disorders such as hyperactivity, aggressiveness and depression can also accompany this disorder. Typically, children with LKS develop normally, but then lose their speech and language skills.</p> <p>Most children outgrow the seizures, and epileptiform activity on the EEG usually returns to normal by age 15. Long term outcomes vary greatly.</p> <p>https://www.facebook.com/groups/EpilepsyLKS/ Raising a child with LKS and CSWS</p> <p>https://www.facebook.com/groups/66923310016/</p> <p>https://www.facebook.com/FamiliesOfLandauKleffnerSyndrome Friends of LKS (FOLKS)</p> <p>http://www.mdjunction.com/landau-kleffner-syndrome MD Junction - People Helping People</p>
<p>Lennox Gastaut Syndrome (LGS) Is challenging behaviour a listed defining characteristic?</p>	<p>Any condition that produces major brain abnormalities is a potential cause of Lennox-Gastaut syndrome, which has three defining characteristics: multiple seizure types, a distinctive brain-wave pattern and intellectual impairment that can range from slight to profound. There are multiple seizure types, tonic seizures being the most common. Seizures are frequent and usually difficult to control.</p> <p>http://lgsfoundation.org/ The LGS Foundation provides information, support, services, and forums.</p> <p>https://www.facebook.com/groups/59073585620/ LGS Facebook group</p>

4 http://www.medicinenet.com/landau-kleffner_syndrome/article.htm

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

<p>Mitochondrial Disease</p> <p>Examples include:</p> <ol style="list-style-type: none"> 1. MELAS: Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke Like Episodes. 2. MERRF: Myoclonic Epilepsy with Ragged Red Muscle Fibres. 	<p>Mitochondrial disease is an inherited chronic illness that can be present at birth or develop later in life. It causes debilitating physical, developmental, and cognitive disabilities with symptoms including poor growth; loss of muscle coordination; muscle weakness and pain; seizures; vision and/or hearing loss; gastrointestinal issues; learning disabilities; and organ failure.⁵</p> <p>http://www.amdf.org.au/ Australian Mitochondrial Disease Foundation. The AMDF's foremost purpose is to find a cure, or at the very least some form of meaningful treatment. To this end it is committed to supporting and funding research projects directed towards understanding more about mitochondrial medicine.</p> <p>http://www.umdf.org/ United Mitochondrial Disease Foundation.</p> <p>https://www.facebook.com/mitoaction?_rdr=p Mitoaction Facebook Group</p>
<p>Myoclonic Astatic Epilepsy (MAE)</p> <p>Also known as Doose syndrome</p>	<p>This is an epilepsy syndrome of early childhood usually characterised by difficult to control generalised seizures (tonic-clonic, myoclonic, atonic, tonic, absence). Children will have a number of seizure types, sometimes associated with falls. Myoclonic Astatic refers to the most common seizure type which is a myoclonic seizure (jerk) followed immediately by an atonic seizure (drop or head nod). Outcome is variable.</p> <p>http://www.doosesyndrome.org/ Doose Syndrome Epilepsy Alliance Information and global parent support group online.</p> <p>https://www.facebook.com/groups/552067731663874/ Doose Syndrome MAE Facebook group</p> <p>https://www.facebook.com/pages/Myoclonic-Astatic-Epilepsy/190872344263079 https://www.facebook.com/groups/DooseSyndrome/ Doose Foundation (for parents of children living with Doose syndrome (Myoclonic Astatic Epilepsy) or variants) MAE Facebook group</p>
<p>Ohtahara Syndrome</p> <p>Also called Early Infantile Epileptic Encephalopathy (EIEE) with burst suppression</p>	<p>This is a rare progressive epileptic encephalopathy. The syndrome is outwardly characterised by tonic spasms and focal seizures. It is a debilitating progressive neurological disorder, involving poorly controlled seizures and intellectual impairment. No single cause has been identified, although in many cases structural brain damage is present.</p> <p>https://sites.google.com/a/ohtahara.org/ohtahara2/home Aarons Ohtahara A site with parent resources and blogs.</p> <p>https://www.facebook.com/AaronsOhtaharaFoundation/timeline/ Ohtahara Facebook Group</p>
<p>Progressive Myoclonic Epilepsies</p> <p>Examples include:</p> <ol style="list-style-type: none"> 1. Myoclonic epilepsy with ragged red muscle fibres 2. Severe myoclonic epilepsy of infancy (Dravet syndrome) 	<p>These are rare epilepsies and frequently result from hereditary metabolic disorders. They feature a combination of myoclonic and tonic-clonic seizures, unsteadiness, muscle rigidity, and often intellectual disability.</p>

⁵ <http://www.mitoaction.org/>

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

<p>Progressive Myoclonic Epilepsies cont.</p> <ol style="list-style-type: none"> 3. Unverricht-Lundborg disease (also called Baltic myoclonus) 4. Lafora disease 5. Mitochondrial encephalopathies 	<p>There are no specific support organisations. See rare diseases link or general links below.</p> <p>http://www.rarediseases.org/ National Organisation for Rare Diseases. NORD, USA.</p>
<p>Rasmussen's Syndrome (Encephalitis) (RE)</p>	<p>Rasmussen's encephalitis is a rare, chronic inflammatory neurological disease that usually affects only one hemisphere of the brain. It usually occurs in children under the age of 10. It is characterised by frequent seizures (of many types, but predominantly focal) affecting one side of the body, loss of motor skills and speech, paralysis on one side of the body, inflammation of the brain (encephalitis) and intellectual disability.</p> <p>https://www.encephalitis.info/rasmussens-encephalitis</p> <p>https://www.facebook.com/REChildrensProject/ The RE Children's Project. Aims to increase awareness regarding RE for the primary purpose of supporting scientific research directed towards a cure.</p> <p>http://hemifoundation.homestead.com/welcome.html The Hemispherectomy Foundation provides emotional, financial, and educational support to individuals and their families who have undergone, or will undergo, a hemispherectomy or similar brain surgery.</p>
<p>Ring Chromosome 20 Syndrome</p>	<p>This is a rare condition that affects the normal development and function of the brain. The most common feature being recurrent focal seizures in childhood which respond poorly to medications. Episodes of non-convulsive status epilepticus are common and most people with ring chromosome 20 syndrome also have some degree of intellectual disability and behavioural difficulties.</p> <p>https://www.facebook.com/Ring20ResearchandSupportUK http://www.ring20researchsupport.co.uk/ The Ring 20 Research and Support UK has been set up to support families, individuals and professionals who are affected by, or who come into contact with Ring Chromosome 20 Syndrome [r20].</p>
<p>West Syndrome (Infantile Spasms)</p>	<p>This is a severe epilepsy syndrome seen in infants and is sometimes called infantile spasms. The spasms are seizures that are like 'jack-knife' movements where the whole body bends in half and the infant lunges forward. They tend to occur in clusters. There is a characteristic EEG in between seizures and some degree of intellectual disability. Most children go on to have other kinds of seizures in later childhood including the epilepsy syndrome Lennox-Gastaut syndrome.</p> <p>https://www.facebook.com/groups/122838701073070/ There are a number of support groups on facebook for West Syndrome.</p> <p>http://www.infantilespasms.com/forum/ Infantile Spasm Forum/Community</p> <p>https://www.facebook.com/infantilespasms Facebook page</p>

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

Disabilities associated with seizures/epilepsy	
Acquired brain injury (ABI)	Acquired brain injury (ABI) is damage to the brain that occurs after birth but is not due to an inherited disorder or degenerative disease. Damage may be caused either by a traumatic or non-traumatic injury to the brain.
	http://www.braininjuryaustralia.org.au/ Brain Injury Australia. Click "Services" for a list of state organisations that can help people with brain injury.
	http://families4families.org.au/ Families 4 Families. Assist adults who have ABI and their families by providing information and education, social opportunities, online resources and other family focussed supports.
Angelman Syndrome	This is a genetic syndrome characterised by intellectual and physical disability, abnormally small head, poor coordination, frequent jerky limb movements and flapping of the arms and hands, poor muscle tone, hyperactivity, seizures, absence of speech, frequent smiling and outbursts of laughter, and a great propensity for protruding the tongue (tongue thrusting).
	http://www.angelmansyndrome.org/ Angelman Syndrome Association. This is the umbrella Organisation for children with Angelman Syndrome and their families in Australia. There are support groups in each state and territory. Angelman Syndrome Australia Facebook Group https://www.facebook.com/angelmanaustralia/
Autism Spectrum Disorder (ASD)	A disorder characterised by deficits in social interaction and communication, and a limited range of activities and interests, and often with the presence of repetitive, stereotyped behaviours.
	https://raisingchildren.net.au/autism/learning-about-asd/about-asd/asd-overview Raising Children Network. Getting help for ASD
	https://www.facebook.com/AutismAwarenessAustralia Autism Awareness Facebook Group There are a number of local facebook groups throughout Australia.
	https://www.autismspectrum.org.au/aap#Other%20states Autism services in each state
Batten disease⁶	Batten disease is a rare but fatal, inherited disorder of the nervous system.. Symptoms usually appear between the ages of 5 and 10 years, where the child will begin to develop vision problems or seizures. In some cases these signs are subtle, appearing as personality and behaviour changes, slow learning, clumsiness, or stumbling. Over time, affected children suffer intellectual impairment, worsening seizures, and progressive loss of sight and motor skills.
	http://www.battens.org.au/ Batten Disease Support and Research Association provides a parent communication network, information and emotional support to families of children affected by Batten disease [refer to Contacts section].

⁶ http://www.ninds.nih.gov/disorders/batten/detail_batten.htm

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

Batten disease cont.	<p>http://bdsra.org/ Batten Disease Support and Research Association Founded by parents, is dedicated to funding research for treatments and cures, providing family support services, advancing education and raising awareness</p>
Cerebral Palsy (CP)	<p>Cerebral palsy is an umbrella term that refers to a group of disorders affecting a person's ability to move. It is a physical disability, so affects movement and posture due to damage to the developing brain either during pregnancy or shortly after birth. It is a permanent life-long condition, but generally does not worsen over time.</p> <p>https://cpaustralia.com.au/ Cerebral Palsy Australia. The national peak body of organisations that work with people with cerebral palsy and people with similar disabilities and their carers.</p> <p>http://www.cpsn.info/ Cerebral Palsy Support Network Australia</p> <p>https://www.cerebralpalsy.org.au/ Cerebral Palsy Alliance</p>
Down Syndrome	<p>A genetic disorder, associated with the presence of an extra chromosome 21, characterised by mild to severe mental impairment, weak muscle tone, shorter stature, and a flattened facial profile.</p> <p>http://www.downsyndrome.org.au/index.html Down Syndrome Australia is made up of State and Territory associations providing support, information and resources. The associations come together to represent and progress the needs, interests and aspirations of people with Down syndrome and those that support them.</p>
Fragile X Syndrome (FXS)	<p>This is a genetic condition causing intellectual disability, behavioural and learning disability and various physical characteristics. People with FXS who have seizures are most likely to have focal seizures. It is also the most common single gene cause of autism worldwide.</p> <p>http://fragilex.org.au/ The Fragile X Association provides support to families living with Fragile X. This includes specialised clinics, mobile workshops, counselling and casework, fundraising and increasing public awareness of Fragile X. There is also a facebook group on this site.</p>
Neurofibromatosis⁷	<p>There are two types of neurofibromatosis. It is a genetic disorder of the nervous system that primarily affects the development and growth of neural (nerve) cell tissues, causes tumours to grow on nerves, and may produce other abnormalities. Signs and symptoms may include neurofibromas or lumps anywhere on the body, learning difficulties, scoliosis and hearing loss. One type is seen in childhood and the other develops late adolescence or even in the 40-50's. Some people live almost unaffected by it; rarely, others can be severely affected.</p> <p>Childrens Tumour Foundation https://www.ctf.org.au/ https://www.facebook.com/pages/Neurofibromatosis-Support/345030432837</p>

7 <http://www.betterhealth.vic.gov.au/bhcv2/bhcarticles.nsf/pages/Neurofibromatosis>

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

Pallister Killan syndrome (PKS)	<p>This is a rare chromosome abnormality. Affected individuals have unusual facial features, intellectual disability, seizures, patchy colour differences in the skin, and various other physical abnormalities.</p> <p>http://www.pks.org.au/ PKSFA provides help and support to individuals diagnosed with PKS and their families as well as raising awareness and providing information to the medical community.</p> <p>https://www.facebook.com/pages/Pallister-Killian-Syndrome-PKS/112628308772465</p>
Prada-Willi Syndrome (PWS)	<p>This is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviours, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity and, usually focal seizures.</p> <p>http://www.pws.org.au/ Prader Willi Syndrome Association: A self help group comprising people born with Prader Willi Syndrome, parents, friends and interested professionals.</p> <p>http://www.ipwso.org/ International Prader Willi Syndrome Association</p>
Rett Syndrome	<p>This is a neurodevelopmental disorder that affects girls almost exclusively. It is characterised by normal early growth and development followed by a slowing of development, loss of purposeful use of the hands, distinctive hand movements, slowed brain and head growth, problems with walking, seizures, and intellectual disability.</p> <p>https://www.facebook.com/groups/15688775612/ http://aussierett.org.au/ https://www.facebook.com/aussierett/ http://www.girlpower2cure.org/what-we-do/family-support.aspx Girl Power 2 Cure - Family Support</p>
Sturge Weber Syndrome (SWS)	<p>This is a neurological disorder where there is a port-wine stain birthmark on the forehead and upper eyelid of one side of the face. The birthmark is caused by an overabundance of capillaries around the trigeminal nerve just beneath the surface of the face. There are also abnormal blood vessels on the brain surface and the loss of nerve cells and calcification of underlying tissue in the cerebral cortex of the brain on the same side of the brain as the birthmark. Neurological symptoms include seizures that begin in infancy and may worsen with age.</p> <p>http://swscommunity.org/index.php http://www.sturge-weber.org/ Sturge Weber Foundation USA https://www.facebook.com/pages/The-Sturge-Weber-Foundation/231991960556 SWF facebook group</p>

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

Tuberous Sclerosis Complex (TSC)	<p>This is a genetic disorder that causes non-malignant tumours to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. The aspects of TSC that most strongly impact quality of life are generally associated with the brain: seizures, developmental delay, intellectual disability and autism spectrum disorder. The incidence and severity of TSC can vary widely between individuals.</p> <p>https://tsa.org.au/ Tuberous Sclerosis Australia</p> <p>http://www.inspire.com/groups/tuberous-sclerosis-alliance/ Tuberous Sclerosis Alliance Support Community. The Tuberous Sclerosis Alliance Support Community connects everyone affected by tuberous sclerosis complex for support and inspiration.</p> <p>https://www.facebook.com/groups/TS.Alliance.Online.Discussion.Group/</p> <p>http://www.dailystrength.org/c/Tuberous-Sclerosis-TSC/support-group TSC Online Support Group</p>
Wolff Hirschorn Syndrome (WHS)	<p>A chromosome disorder due to partial deletion of the short (p) arm of chromosome 4. It is therefore also called the 4p-syndrome. Features include midline defects with a scalp defect, wide spaced eyes, broad or beaked nose, oral facial clefts (cleft lip/palate); low simple ears; small and/or asymmetrical head; heart defects; and seizures [that tends to diminish with age]. There is severe to profound developmental and intellectual disability. There is usually very slow progress in development. Some people are prone to episodes of status epilepticus.</p> <p>http://www.awhssg.org.au/ Australian Wolff Hirschorn Syndrome Support Group is a voluntary not for profit self help organisation dedicated to educating parents about Wolf Hirschhorn Syndrome and networking them with other parents of affected children.</p> <p>http://www.4paussiekidz.org.au/about/wolf-hirschhorn-syndrome 4p Aussie Kidz</p> <p>https://www.facebook.com/groups/15124152855/ Facebook support group</p>
General Help and Support	
Association for Children with a Disability http://acd.org.au/	<p>Have knowledge of service systems and active members of families of children with any type of disability. Also Facebook group.</p>
Disability Online Australia http://www.disabilityonline.org.au/homepage	<p>Australia's disability hub.</p>
Epilepsy Parents Australia https://www.facebook.com/groups/epilepsyparentsaustralia/	<p>Active Facebook group</p>
Epilepsy (Online) Support Group http://tesg.forumotion.com/	<p>Online forums about a range of issues concerning epilepsy.</p>
Rare Voices Australia. (RVA) http://www.rarevoices.org.au/	<p>RVA is Australia's national organisation supporting all those who live with a rare disease. RVA provides information on rare diseases and services that may help. They work with governments, researchers, clinicians and industry to promote research, diagnosis, treatment and services for all rare diseases in Australia.</p>

Fact Sheet: Epilepsy and Related Syndromes - Useful Organisations

<p>Smile Foundation Australia (SMILE) http://www.smilefoundation.com.au/</p>	<p>The SMILE Foundation is a national charity that was established in 2007 to improve the quality of life of children with a rare disease or condition. SMILE provides financial assistance and case management services to affected families and supports research relating to rare childhood medical conditions.</p>
<p>Genetic Alliance Australia http://www.geneticalliance.org.au/</p>	<p>This site has a long list of contacts/support groups for rarer conditions, that are too rare to have their own support group. GAA endeavours to facilitate contact with another family/individual affected by the same or similar condition, and/or provide information about an overseas support group.</p>
<p>RareShare http://www.rareshare.org/</p>	<p>RareShare is a unique social hub building communities for patients, families, and healthcare professionals affected by rare disorders.</p>
<p>National Organisation for Rare Diseases (NORD) http://www.rarediseases.org/</p>	<p>NORD is a unique federation of voluntary health organisations dedicated to helping people with rare “orphan” diseases and assisting the organisations that serve them. NORD provides information, advocacy, research, and patient services to help all patients and families affected by rare diseases.</p>
<p>Intractable Childhood Epilepsy Alliance (ICE) http://www.ice-epilepsy.org/</p>	<p>ICE is dedicated to improving the lives of children affected by intractable epilepsy through evidence-based information, advocacy for appropriate medical treatment, promotion of antiepileptic drug development, data collection through patient registries, and funding of research that will lead to a cure for intractable childhood epilepsies.</p>
<p>Epilepsy Action Australia (EAA) http://www.epilepsy.org.au</p> <p>http://www.epilepsy.org.au/online-academy/</p>	<p>EAA has a number of online self management tools including:</p> <ul style="list-style-type: none"> • My Epilepsy Diary: a state of the art online epilepsy seizure diary adapted for the Australian community and accessed through the EAA website. It is a personal electronic diary to help keep track of your epilepsy - putting you in control of your information and helping you keep an accurate account of your seizures, triggers and medications. • Epilepsy Action Online Academy: provides a range of online, interactive epilepsy learning and education for people living with epilepsy, parents, carers, school children and health professionals. The Online Academy also offers useful resources including an online Seizure Management Plan and Emergency Medication Plan • Online Seizure Management Plan (SMP): this is a simple, online tool you can use to create an individualised SMP and Emergency Response Plan. This document provides essential information to anyone who may be in a position to assist someone having a seizure - such as family, friends, carers, teachers, colleagues or other involved persons. <p>Online services also include:</p> <ul style="list-style-type: none"> • Equip - an epilepsy resource for youth with epilepsy • rEaction - a resource to increase teens awareness of epilepsy (such as friends of someone with epilepsy) • Strong Foundations - for parents of children with epilepsy • Psychosocial Wellbeing for Adults resource • Yarning Epilepsy - a course for Indigenous Health Workers

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This information is given to provide accurate, general information about epilepsy. Medical information and knowledge changes rapidly and you should consult your doctor for more detailed information. This is not medical advice and you should not make any medication or treatment changes without consulting your doctor.

