Epilepsy Action Australia

FACT SHEET:

Epilepsy and Related Syndromes

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
- The age 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 an 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	
Epileptic Encephalopathy with Continuous Spike and Wave Syndrome during Sleep (CSWS), sometimes termed Electrical Status Epilepticus in Sleep (ESES)	A rare epilepsy syndrome or epileptic encephalopathy which 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 outward signs of seizures). Different seizure types can be seen, and this can occur during wakefulness. This syndrome can disappear around puberty, which may result in some improvements, but cognitive and behavioural symptoms may persist. This syndrome has similar features to Landau Kleffner Syndrome.
	Raising a child with LKS and CSWS - https://www.facebook.com/groups/EpilepsyLKS/
	Raising a child with CSWS - https://www.facebook.com/groups/149591171771780/

Epilepsy Syndromes Causing Disability	
Dravet Syndrome Also called Severe Myoclonic Epilepsy of Infancy (SMEI)	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.
	Dravet Syndrome Australia - https://www.facebook.com/groups/dravetsyndrome/
	Dravet Syndrome UK - http://www.dravet.org.uk/
	Dravet Foundation USA - http://www.dravetfoundation.org/
	DravetData.com – http://dravetdata.com/
Landau Kleffner Syndrome (LKS)	This is a rare childhood condition and occurs most frequently in normally developing children between 3 and 7 years of age. Typically, children with LKS lose their speech and language skills. Not all children have seizures, and they are usually associated with sleep when they happen. Most children outgrow the seizures and by about age 15. Behavioural disorders such as hyperactivity, aggressiveness and depression can also accompany LKS. Long term outcomes vary greatly.
	Raising a child with LKS and CSWS - https://www.facebook.com/groups/EpilepsyLKS/
	Landau Kleffner Support Group - https://www.facebook.com/groups/66923310016/
	Friends of LKS (FOLKS) - https://www.facebook.com/FamiliesOfLandauKleffnerSyndrome
Lennox Gastaut Syndrome (LGS)	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 can be many seizure types which are usually frequent and difficult to control.
	The LGS Foundation - http://lgsfoundation.org/
	LGS Facebook group - https://www.facebook.com/groups/59073585620/

Epilepsy Syndromes Causing Disability	
Mitochondrial Disease Examples include: 1. MELAS: Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis And Stroke Like Episodes. 2. MERRF: Myoclonic Epilepsy with Ragged Red Muscle Fibres.	Mitochondrial disease is an inherited disease that can be present at birth or develop later in life. It causes debilitating physical, developmental, and cognitive disabilities including poor growth; loss of muscle coordination; muscle weakness and pain; seizures; vision and/or hearing loss; gastrointestinal issues; learning disabilities and organ failure.
	Mito Foundation - https://www.mito.org.au/
	United Mitochondrial Disease Foundation - http://www.umdf.org/
	Mitoaction Facebook Group- https://www.facebook.com/mitoaction?_rdr=p
Myoclonic Astatic Epilepsy (MAE) Also known as Doose syndrome	This is an epilepsy syndrome of early childhood usually characterised by difficult to control generalised onset seizures. Children will have several 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.
	Doose Syndrome Epilepsy Alliance - http://www.doosesyndrome.org/
	Doose Syndrome MAE Facebook group - https://www.facebook.com/groups/552067731663874/
	Myoclonic Astatic Epilepsy Facebook group - https://www.facebook.com/pages/Myoclonic-Astatic-Epilepsy/190872344263079
Ohtahara Syndrome Also called Early Infantile Epileptic Encephalopathy (EIEE) with burst suppression	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.
	Aarons Ohtahara - https://sites.google.com/a/ohtahara.org/ohtahara2/home
	Ohtahara Facebook Group - https://www.facebook.com/AaronsOhtaharaFoundation/timeline/

Epilepsy Syndromes Causing Disability	
Progressive Myoclonic Epilepsies Examples include: 1. Myoclonic epilepsy with ragged red muscle fibres 2. Severe myoclonic epilepsy of infancy (Dravet syndrome) 3. Unverricht-Lundborg disease (also called Baltic myoclonus) 4. Lafora disease 5. Mitochondrial encephalopathies	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. Also see rare disease links or general links within this document and in the 'General help and support' section below.
	Rare Voices Australia - https://www.rarevoices.org.au/
	Steve Waugh Foundation - https://www.stevewaughfoundation.com.au/
	National Organisation for Rare Diseases (NORD), USA - http://www.rarediseases.org/
Rasmussen's Syndrome (Encephalitis) (RE)	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 and characterised by frequent seizures (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.
	Encephalitis Society - https://www.encephalitis.info/rasmussens-encephalitis
	The Hemispherectomy Foundation - http://hemifoundation.homestead.com/welcome.html
Ring Chromosome 20 Syndrome	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 medication. Episodes of non-convulsive status epilepticus are common and most people with this syndrome also have some degree of intellectual disability and behavioural difficulties.
	The Ring 20 Research and Support UK - https://ring20researchsupport.co.uk/
	Ring 20 Facebook Group - https://www.facebook.com/Ring20ResearchandSupportUK

Epilepsy Syndromes Causing Disability	
West Syndrome (Infantile Spasms)	An epilepsy syndrome seen in infants and is sometimes called infantile spasms. The spasms are seizures that are like 'jack-knife' movements where the 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 will have other kinds of seizures in later childhood including the epilepsy syndrome Lennox-Gastaut syndrome.
	West Syndrome/Infantile Spasms Australia Facebook support group - https://www.facebook.com/groups/377789189087140 (There are many other support groups on Facebook for West Syndrome)
	Cure Epilepsy - https://www.cureepilepsy.org/event_type/infantile-spasms-awareness-week-2019/
	Infantile Spasms Facebook page - https://www.facebook.com/infantilespasms
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.
	Brain Injury Australia - http://www.braininjuryaustralia.org.au/
	Families 4 Families - http://families4families.org.au/
Angelman Syndrome	This genetic syndrome is 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 propensity for protruding the tongue (tongue thrusting).
	Angelman Syndrome Association Australia - https://www.angelmansyndrome.org/
	Angelman Syndrome Australia Facebook Group - https://www.facebook.com/angelmanaustralia/
Autism Spectrum Disorder (ASD)	Autism spectrum disorder (ASD), refers to a broad range of conditions characterised by challenges with social skills, repetitive behaviours, speech and nonverbal communication.

Disabilities associated with seizures/epilepsy	
Autism Spectrum Disorder continued	Autism Spectrum Australia - https://www.autismspectrum.org.au/
	Autism Awareness Facebook Group - https://www.facebook.com/AutismAwarenessAustralia
	Autism Parents Australia Facebook Group - https://www.facebook.com/groups/autismparentsaustralia/
Batten disease	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.
	Batten Disease Support and Research Association Australia - http://www.battens.org.au/
	Batten Disease Support and Research Association USA - http://bdsra.org/
Cerebral Palsy (CP)	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.
	Cerebral Palsy Australia - https://cpaustralia.com.au/
	Cerebral Palsy Support Network - https://cpsn.org.au/
	Cerebral Palsy Alliance - https://www.cerebralpalsy.org.au/
Down Syndrome	A genetic disorder associated with the presence of an extra chromosome 21, characterised by mild to severe intellectual impairment, weak muscle tone, shorter stature and a flattened facial profile.
	Down Syndrome Australia - http://www.downsyndrome.org.au/index.html
Fragile X Syndrome	This is a genetic condition causing intellectual disability, behavioural and various physical characteristics. People with FXS who have seizures are most likely to have focal seizures..
	Fragile X Association Australia - http://fragilex.org.au/

Disabilities associated with seizures/epilepsy	
Neurofibromatosis	Is a genetic disorder that primarily affects the development and growth of nerve tissues, causing 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. There are two types of neurofibromatosis, one type is seen in childhood and the other develops later. Some people live almost unaffected by it; rarely, others can be severely affected.
	Childrens Tumour Foundation - https://www.ctf.org.au/
	Neurofibromatosis Facebook Support Group - https://www.facebook.com/pages/Neurofibromatosis-Support/345030432837
Pallister Killan syndrome (PKS)	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.
	PKS Foundation of Australia - http://www.pks.org.au/
	PKS Kids Facebook Group - https://www.facebook.com/pkskids/
Prada–Willi Syndrome (PWS)	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.
	Prader Willi Syndrome Australia - http://www.pws.org.au/
	International Prader-Willi Syndrome Organisation - http://www.ipwso.org/
Rett Syndrome	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.
	Rett Syndrome Australia Facebook group - https://www.facebook.com/groups/15688775612/
	Rett Syndrome and Related Disorders - https://rett.telethonkids.org.au/
	Telethon Kids Institute Facebook page - https://www.facebook.com/TelethonKids/

Disabilities associated with seizures/epilepsy	
Sturge Weber Syndrome (SWS)	A neurological disorder where there is a port-wine stain birthmark on the forehead and upper eyelid of one side of the face, 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 a loss of nerve cells and calcification of underlying tissue in the cerebral cortex (grey matter) 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.
	Genetic Alliance Australia (SWS) – http://www.geneticalliance.org.au/conditions_detail.php?Sturge-Weber-Syndrome-502
	Sturge Weber Foundation (SWF) USA - http://www.sturge-weber.org/
	SWF Facebook group - https://www.facebook.com/pages/The-Sturge-Weber-Foundation/231991960556
Tuberous Sclerosis Complex (TSC)	A genetic disorder that causes non-malignant tumours to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. Characteristics seen include: seizures, developmental delay, intellectual disability and autism spectrum disorder. The severity of TSC can vary widely between individuals.
	Tuberous Sclerosis Australia - https://tsa.org.au/
	TS Alliance Tuberous Sclerosis Complex Discussion Group - https://www.facebook.com/groups/TS.Alliance.Online.Discussion.Group/
Wolff Hirschorn Syndrome (WHS)	A chromosome disorder 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 tend 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.
	WHS Facebook support group - https://www.facebook.com/groups/151241528555/
	National Organisation for Rare Disorders - https://rarediseases.org/rare-diseases/wolf-hirschhorn-syndrome/

General help and support
Association for Children with a Disability - http://acd.org.au/ **Better Start for Children with a Disability** - http://betterstart.net.au/ **Disability Online Australia** - http://www.disabilityonline.org.au/homepage **Epilepsy (Online) Support Group** - http://tesg.forumotion.com/ **Genetic Alliance Australia** - http://www.geneticalliance.org.au/ **Intractable Childhood Epilepsy Alliance (ICE)** - http://www.ice-epilepsy.org/ **National Organisation for Rare Diseases (NORD)** - http://www.rarediseases.org/ **RareShare** - http://www.rareshare.org/ **Rare Voices Australia (RVA)** - http://www.rarevoices.org.au/ **The Brain Foundation** - https://brainfoundation.org.au/ **GETA – Genetic Epilepsy Team Australia** https://www.facebook.com/genetic.epilepsy.team.australia/

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