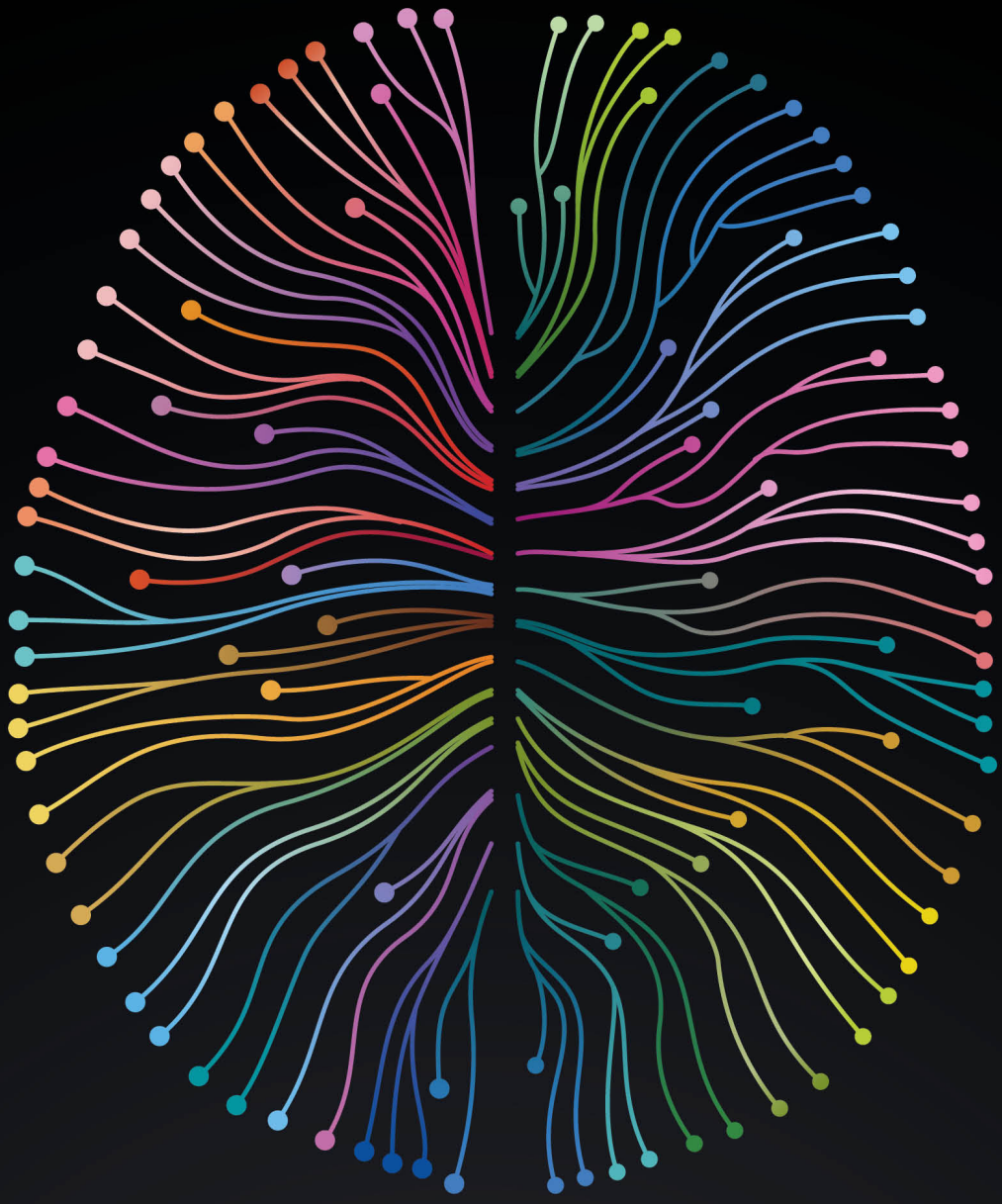
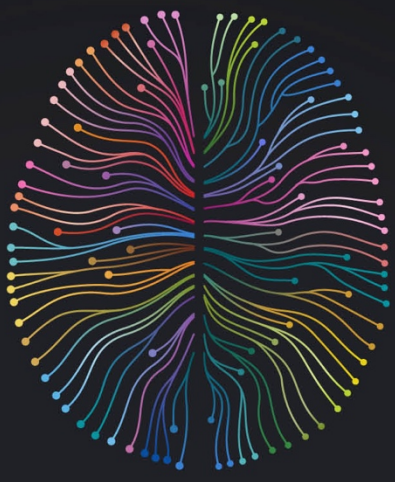
DEB PAL KELLY PRETZ CAROLINE MUIR NICOLA JOLLEFF



WHAT EVERY PARENT SHOULD KNOW ABOUT ROLANDIC EPILEPSY



WHAT EVERY PARENT SHOULD KNOW ABOUT ROLANDIC EPILEPSY

Deb Pal
Kelly Pretz
Caroline Muir
Nicola Jolleff

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CONTENTS

About the authors

Deb Pal

Deb Pal is Professor of Paediatric Epilepsy at King's College London and Consultant Paediatric Neurologist at Evelina Children's and King's College Hospitals. He trained in paediatric neurology at Great Ormond Street Children's Hospital, London UK. He studied medicine at Cambridge University, has a PhD in Neuroscience, and spent ten years at Columbia University, New York researching the genetics of complex diseases. His research has spanned the public health aspects of childhood epilepsy in developing countries and the genetic basis of common childhood epilepsies. Since 2005, he has been head of the world's leading research group on rolandic epilepsy.

Kelly Pretz

Kelly Elizabeth Pretz is a fifteen-year-old from the USA diagnosed with Rolandic epilepsy at the age of ten. She is an active high school freshman. She plays the flute in her school's marching band as well as the concert band. She is a senior girl scout who earned the prestigious silver award with a platform of epilepsy. Kelly spends at least one day a week volunteering at the National Epilepsy Foundation where she mentors other kids with epilepsy. She has also started an epilepsy foundation of her own and began an awareness campaign called the "Purple Mission". This mission has enabled Kelly to involve her school as well as the surrounding communities in the support of epilepsy awareness. Kelly has a book called' "Rain at Midnight." It is a story about a girl who must deal with epilepsy. Kelly feels that having epilepsy has given her purpose and motivation. Kelly says, "If you say I can't do something, then I will prove you wrong."

Caroline Muir

Caroline Muir, M.A.(Hons) Edin, M.A., London College of Printing, has an editorial background in general publishing. She took a career break to bring up her three children, one of whom has had rolandic epilepsy. She has worked for many years in the voluntary sector. She recently worked with the National Centre for Young People with Epilepsy to create a unique diary for children with this condition, DAY BY DAY BY ME. This is available free of charge through the charity Young Epilepsy.

Nicola Jolleff

Nicola is retired Principal Speech and Language Therapist at Guys and St Thomas NHS Foundation Trust.

1.What is Rolandic Epilepsy?

Deb Pal

Epilepsy

The medical definition of epilepsy is that there is a tendency to have seizures as a result of changes in the electrical activity of the brain. The word "repeated" is a key part of the definition, as a single seizure is not the same thing as epilepsy.

An epileptic seizure results from a sudden burst of electricity in the brain that results in an alteration in sensation, behaviour or consciousness. Seizures can take many forms because the brain is responsible for such a wide range of functions. The symptoms that occur during a seizure depend on where in the brain this abnormal burst of electrical activity occurs and where it spreads to. Seizures may last from a few seconds to a few minutes.

Seizures are often classified as focal or generalized depending on what your doctor concludes about where in the brain the seizures have arisen. Focal means that the seizures are limited to one side of the brain, and the symptoms will depend on where in the brain the seizures start from and spread to. In generalised seizures both sides of the brain, not just one area, is affected by abnormal electrical discharges and the child becomes unaware of their surroundings or may lose consciousness completely as in a tonic-clonic ("grand-mal") seizure. The terms focal and generalized are explained further in the Seizures section below.

Epilepsy in Children

Many types of epilepsy only occur in childhood, starting and often finishing before 16 years of age. The epilepsies of childhood include some of the simplest as well as the most difficult seizure types to treat. Quite often, there are effects on a child's development and behaviour, and it is important to be aware of these when considering the whole child with epilepsy.

Rolandic epilepsy (RE)

Rolandic epilepsy is the most common type of epilepsy - affecting about one-sixth of all children with epilepsy in the UK - that means over 10,000 people! So you are definitely not alone. We will use the simple term "rolandic epilepsy" (RE) throughout this book, although you will also find it labelled elsewhere as "benign rolandic epilepsy" or "childhood epilepsy with centrotemporal spikes" - a bit of a mouthful! Children with RE usually have their first seizure between the ages of 4 and 10 years, and twice as many boys are affected as girls - we don't know why this is so. However, the fact that the seizures disappear at teenage suggests that the seizures and other features (below) are a manifestation of the different way that brains mature in children with RE. Although the diagnosis is made after seizures have occurred, often seizures are not the first symptom of the condition - earlier in life there may have been concerns about speech delay, or about reading difficulties. Below is a description of some of the features that children with RE may encounter. That is not to say that these features are inevitable – about 1 in 5 children with RE may only experience the odd seizure and have no other complications.

Seizures

Seizures are usually related to sleep, whether during the day or night. Most children with RE have less than six seizures in their lifetime, but a small minority have several hundreds. Most children have a single type of seizure (known as focal), but about one quarter may have both focal and generalised seizures. The typical features of the focal seizures are
(a) contraction, twitching or weakness of one side of the face, sounds from the throat, and drooling with an inability to swallow.

(b) not being able to speak because the muscles of the vocal tract are paralysed.
(c) tingling or numbness of the tongue, lips, gum and cheek.
These seizures typically last for a matter of a few minutes.

Generalized seizures may occur more often in younger children and are assumed to spread as an electrical activation across the brain from a focal seizure. A generalized seizure may start off as a focal seizure (as above), but then the seizure may spread to other parts of the body from the face, eg to involve the arm and leg, and then become a generalized convulsion with loss of consciousness. In a generalized convulsion, the person goes stiff, may fall if standing, and then has repetitive and rhythmic jerking of the arms and legs for several minutes. During this seizure they will be unresponsive and unaware of their surroundings.

Speech sound disorder

Speech sound disorder is defined as problems in learning how to make the sounds that make up intelligible speech. We all remember how infants begin to make speech sounds, and how this matures to distinct and clear words over the first two to three years of life. Children with speech sound disorder do not make this transition at the appropriate time and may start words with the wrong sounds or leave sounds off words. Frequently, teachers or strangers cannot understand their speech, even by the age of four years old. Children with SSD may be late in speaking words and sentences and may already have been referred for speech therapy. We estimate that about 30% of children with RE have a speech sound disorder. Speech sound disorder is different to stuttering or aphasia.

Dyslexia

Children with RE frequently have problems in language and learning and we estimate about 40% will have problems in learning to read. Dyslexia or reading disability is one of the most common complications of RE. Dyslexia is defined as significant impairment in the development of reading skills that are not a result of mental retardation, hearing or visual impairment, having a different mother tongue, or inadequate schooling or home environment. Dyslexia is often not detected until entering primary school and becomes evident in the ease of learning reading skills in relation to peers. Dyslexia is remediable through educational methods and remediation is easiest and most effective when introduced early. Unfortunately, some children are not diagnosed until they have been at school for four or five years, by which time the impact of their dyslexia has spread into other school subjects that rely on fluent and accurate reading of the written word.

Attention problems

Attention comprises a number of capacities that also mature and develop over time, like our ability to speak clearly or to learn to read. The three main aspects of attention allow us to (a) respond to changes in our sensory world, eg the sound of a telephone; (b) to maintain focus over a period of time; and (c) to suppress distracting stimuli, eg classmates, while working on another task. The development and maturation of these three aspects of attention is impaired in most children with RE. Attention continues to develop until teenage and beyond. Problems in the development of attention are not the same thing as attention deficit hyperactivity disorder (ADHD or ADD), which refers to a persistent set of behavioural features that are seen in different settings. A small proportion of children with RE may also have features of ADHD.

Migraine

Migraine is a particular type of recurrent headache characterised by a throbbing pain, made worse by exertion and sometimes bright lights and loud noises. Children may also feel sick. The headache usually lasts at least an hour and usually relieved by sleep. Ice-pick headaches, in which the pain may be described as sudden excruciating sharp pains, are a variant of migraine. About 15% of children with RE suffer migraines. They are often triggered by insufficient sleep, skipping meals, or other loss of daily routine

Coordination

Developmental coordination disorder is a medical term for "the clumsy child". Children with DCD have problems with motor coordination of arms and legs serious enough that it interferes with home and school life. Often, children with DCD also have speech and language impairments, reading disability, autism spectrum disorder or ADHD. DCD may occur in association with RE.

Cause

RE has a genetic cause. About one in ten children have a brother, sister or other family member affected with RE. The characteristic brainwave or electroencephalographic (EEG) feature of RE, known as CentroTemporal Spikes, is passed on from parent to child with a 50% probability. However, just inheriting the EEG feature does not give you epilepsy. Our current understanding is that RE results from a combination of gene variants acting together, one of which is the variant in the gene PAX6 that causes the EEG feature. Our studies also show that the EEG abnormality and speech sound disorder are probably caused by the same genetic influence. Reading disability also appears to be genetic and, along with speech sound disorder and the EEG, also runs in families. Not surprisingly, the tendency for migraine also runs in RE families.

While RE may be genetic, this does not mean that the condition cannot be treated. For example, speech sound disorder can be improved with the help of a speech therapist, dyslexia can be remedied using conventional methods, and changes in lifestyle can reduce the frequency of migraine attacks. Nearly all the symptoms can be ameliorated in some way.

Prognosis

RE is a developmental condition, meaning that it evolves as the child matures. Certain symptoms completely resolve like the seizures and the EEG abnormality. 100% of children "grow out" of their seizures by the age of sixteen years old. The EEG usually goes back to normal at around the same time, indicating that the tendency for recurrent seizures has disappeared. A small risk of later seizures remains, about 1-2% over a lifetime. Speech also improves over time, becoming better articulated, but there may be some residual signs even in adulthood. Dyslexia can be persistent, especially if not acted upon in early school life. The prognosis for motor coordination and attention impairment or ADHD is not well known.

History

Most people assume that rolandic epilepsy is named after the anatomical landmark in the brain close to where the seizures are presumed to arise from, the fissure of Luigi Rolando (1773-1831). However, the real origin of the name may be from the Bavarian physician Martinus Rulandus who gave one of the first known descriptions of the condition in 1597. Rulandus taught medicine at several places and was physician to the Duke of Bavaria. He left behind a series of medical case books in one of which he describes a typical seizure in a ten-year-old boy that we would now undoubtedly classify as a rolandic seizure. This was treated by Rulandus with a drink of lime tree blossom and purgation with brandy and aloe!

The electroencephalograph or EEG (brainwave test) was invented in Germany in the 1940s. A series of electrodes is used to pick up the tiny electrical signals beneath the scalp. There then began an era in which the EEG was applied to normal people and to people with epilepsy to explore the diagnostic usefulness of this new technology. Within a few years, the French neurophysiologist Yvette Gastaut noticed that a number of children exhibited spikes in the pre-rolandic area, and soon afterwards, another French duo, Nayrac and Beaussart, reported that the seizures described by Rulandus were associated with this particular EEG pattern – thus was born the syndrome of rolandic epilepsy. It has remained defined mostly by the characteristic seizure and EEG pattern until the 1990s, when a German paediatrician, Ute Staden who was dual trained as a speech therapist, studied 20 children with RE and noticed speech and language impairments were very common among this group.

Diagnosis & Assessment

Deb Pal

Eye-witness accounts

In many cases, children who are having a focal rolandic seizure do not lose consciousness and retain awareness throughout their seizure, able to give a good account of what they experienced. The most prominent feature is an inability to speak, drooling of saliva from the mouth, and guttural sounds. Often the seizure occurs out of sleep either in the night, or in the day for example while taking a nap. There may be distortion or jerking of one side of the mouth or face, numbness or tingling of the same area; during the course of the seizure there may be jerking of the arm or leg on one side. About one-quarter of children will have a generalised convulsion at some time, which may start as a focal seizure.

EEG

If the features of the seizure are typical, then confirming the diagnosis of RE rests on the findings in the electroencephalograph (EEG or brainwave test). The EEG typically shows a normal background rhythm that is symmetrical and reacts in the normal way when the technician asks your child to open his eyes. Similarly, when s/he drifts off to sleep, there should be the normal EEG features that indicate the different stages of sleep. Superimposed on this normal rhythm are "spikes" recorded over the rolandic areas on either side of the head near the temples. These are known as rolandic or centrotemporal spikes, giving this type of epilepsy its unique name. These spikes may be seen on one side of the head or on both sides. The side on which they are seen may change from one EEG to the next. They typically increase in frequency as the child falls asleep. Spikes may also be seen over other areas of the brain on the same EEG recording. Interestingly, these same spikes are also found in about 2% of the population, although less than 10% of such children have RE. Thus to make the diagnosis of RE, both the seizures and the characteristic EEG must be present. As children approach late teens, these spikes disappear, and this transition heralds the end of the vulnerable period for seizures.

Brain Imaging

Because the diagnosis can be reliably made from the seizure description and the EEG, neuroimaging is not usually performed in European countries. However, when it is performed, the results are usually normal or reveal incidental findings not relevant to the epilepsy.

Blood tests

There are no routine blood tests performed to make a diagnosis of RE. However, where the features seem unusual, doctors may request tests to look for rare underlying causes. These may be detected by genetic testing or by MRI scan of the brain.

3. Treatment

Deb Pal & Nicola Jolleff

Self-management

There are many things you can do yourself for your child with RE. As you will have discovered from the above, RE is about much more than just seizures. Your child may have problems with speech and language, with reading, attention or coordination. All of these problems may be improved with the correct assessment and intervention.

An adequate amount of good quality sleep is very important for children with RE. This is because sleep deprivation may trigger seizures. Antiepileptic drugs may appear to work less well if the child is not sleeping properly. Children with RE may appear sleepy during the day if they are having frequent night-time seizures. These seizures disrupt normal sleep patterns and put the child at further risk of seizures. The relationship between sleep and epilepsy can be a vicious cycle, and ensuring good quality sleep may protect against seizures, while treating seizures can improve sleep quality too.

To treat or not to treat?

Antiepileptic drug treatment is usually effective at preventing further seizures, although many doctors believe that treatment is not necessary if the seizures only occur at night, or the child never loses consciousness. As a general guide, treatment should be considered in people when the seizures are disrupting everyday life. It is important to bear in mind first that antiepileptic drugs have little or no proven effect on any other feature of the condition than the seizures, and second that antiepileptic drugs do not cure the condition, they merely prevent seizures from occurring. These drugs are therefore prescribed over the vulnerable years when seizures are likely to occur, and then withdrawn when it is believed that seizures are not likely to recur. This vulnerable period is about three years, shorter for some, longer for others. The decision on whether to treat with antiepileptic drugs is frequently a balance between the potential benefits to the child and the potential risks of using antiepileptic medications.

Who should treat?

Antiepileptic drugs are specialised medicines and each one is used for a different purpose and is metabolised differently in the body. Only registered medical practitioners should prescribe these drugs, and a specialist in paediatrics or paediatric neurology should assess the child and start treatment after discussion with the child and family.

Evidence-based treatments

As rolandic epilepsy is a common condition, there is considerable variation in the choice of drugs to treat it with. In France alone there are five preferred treatments according to which region of France you live in! To date there have been no well-designed head-to-head studies to determine which drugs are best in terms of effectiveness and side effects. Some of the drugs in use have been around for 50 years, others are comparatively modern. Among the popular and effective treatments are carbamazepine, oxcarbazepine, sodium valproate, levetiracetam, and sulthiame. They have alternative trade names according to manufacturer. Most physicians advise sticking to the same manufacturer where possible. This is because the "strength" of the drug may vary slightly between manufacturers despite the dose (eg 500mg) being the same. This slight difference may not cause a problem in children whose seizures are well controlled on a relatively low dose but may be critical in those who have had difficulty achieving seizure control with more than one medication. In some difficult cases when seizures become frequent, children may be treated with steroids (eg Prednisolone) or benzodiazepine class (eg clobazam) of drugs for a short course.

Common side effects

Carbamazepine is the generic (non-brand name) and this drug may be branded as Tegretol. Carbamazepine is widely used and well tolerated by most people. The most common mild side effects are:

- Dizziness
- Sleepiness
- Unsteadiness
- Upset stomach
- Blurred or double vision
- Headache

If you notice any of these problems, contact your doctor or epilepsy nurse. Do not stop taking the drug and do not change the dose of drug without medical guidance. More serious side effects include:

- Allergic reactions – about 5-10% of people taking carbamazepine may develop a red rash within the first month of taking it. If this happens let your doctor or nurse know. A very small number of people may have a serious allergic skin reaction (known as Stevens-Johnson syndrome). People of south Asian descent are at particular risk of this and may be tested beforehand to see if they have the genetic risk for this reaction. A similar test will be available for people of European ancestry in the future.
- Blood disorders – these are very rare, affecting 1 in 30,000 people. The first symptoms may be fever, sore throat and mouth, nosebleeds, tiny red spots in the skin. If you notice these, call your doctor right away.
- Liver problems – again very rare. Symptoms include yellow eyes or skin, loss of appetite, vomiting and pale or dark bowel movements. If you notice these, call your doctor right away.

Sodium valproate is the generic name of a drug that may be branded as Epilim. Sodium valproate was widely used and well tolerated by most people but is now not recommended for females because of its teratogenic (birth malformation) side effects. The most common mild side effects are:

- Tiredness
- Dizziness
- Upset stomach
- Vomiting
- Tremor
- Hair loss
- Weight gain
- Irritability

If you notice any of these problems, contact your doctor or epilepsy nurse. Do not stop taking the drug and do not change the dose of drug without medical guidance. More serious side effects include:

- Liver or pancreas problems – rare, the symptoms may include weakness, swollen face, vomiting, yellow eyes and skin, abdominal pain.
- Blood clotting problem – easy bruising, nosebleeds.

If you notice these symptoms, call your doctor right away but do not stop taking the medicine.

Levetiracetam is the generic name of a drug better known as Keppra. Keppra is one of the newer drugs but very safe. Some mild side effects that have been reported include:

- Sleepiness
- Loss of strength and energy
- Dizziness
- Anxiety
- Irritability
- Other mood changes

Very few people have serious reactions to Levetiracetam, but some symptoms to look out for are:

- Depression
- Severe anxiety, agitation or confusion.

If you notice these symptoms, call your doctor right away.

Getting medical help

Your GP is usually your first point of contact, and s/he will have referred you to a specialist in epilepsy. Specialists may be paediatricians with special training and experience in epilepsy, or less commonly paediatric neurologists in larger children's hospitals. Many children's departments also have epilepsy nurses who are experts in the day-to-day management of children's epilepsy. They are much more accessible than doctors and can provide practical information and help on living with epilepsy and dealing with the common complications that arise. With experience of talking to the various members of your medical team you will find out who is best to contact for what problem or question. As a general guide, here are some suggestions:

Epilepsy nurse – information about your child's epilepsy; links to voluntary organizations; instruction in using rescue medications; access to community services; liaison with school

Paediatric specialist – makes the diagnosis of epilepsy; formulates a treatment plan; arranges tests; regular reviews.
GP – prescribing antiepileptic drugs recommended by the specialist; monitoring for side-effects; general health and medical management.

The ketogenic diet & surgical treatments

The **ketogenic diet** is a high-fat, low carbohydrate and controlled protein diet used in the management of children with treatment resistant epilepsy. It has no established place in the management of rolandic epilepsy.

Vagus nerve stimulation, a device like a pacemaker for the nervous system, may be used in children who are resistant to medication, who are not suitable for surgery, and for whom the ketogenic diet has been tried unsuccessfully. It is not used for the treatment of RE.

Only a small fraction of children with treatment resistant epilepsy are considered suitable candidates for **epilepsy surgery.** Because there is no surgical lesion to operate on in RE, surgery is not an option.

Complementary & alternative treatments

Complementary and alternative medicine (CAM) is increasing in popularity in Western countries, an estimated one-quarter of American parents using vitamins, diet, herbs, homeopathy, massage or aromatherapy. Many parents do not report their use of these CAM treatments to their doctor, but they should. This is because some products used in CAM may interact with antiepileptic medications in unexpected ways. Parents should also not discontinue their prescribed medications without discussing their plans with their child's physician.

Stress reduction through yoga or relaxation techniques forms of epilepsy. These include biotin, is a reasonable goal in epilepsy.

Cognitive behavioural therapy (CBT) has been tried to interrupt seizure onset and may help in the treatment of anxiety or depression. Neurofeedback is a technique where the person is able to see the activity of their brain in real time, for example through a simplified EEG monitor, and then influences brain activity through concentration or relaxation. Several forms of neurofeedback have been tried in patients with treatment resistant epilepsy and may work through enhanced awareness of early symptoms and identifying trigger factors, as well as real-time feedback of the EEG.

Craniosacral therapy is a based on the idea of wavelike movement of fluid between the head and the bottom of the spine, but there have been no clinical trials to assess its value.

Vitamins are sometimes used in the specific treatment of some rare pyridoxine and folate. These vitamins are found in normal healthy diets, and specific deficiencies occur only rarely in severe types of epilepsy. Other available supplements include gamma-aminobutyric acid (GABA), carnosine, taurine and carnitine. GABA, a chemical in the brain that reduces electrical excitability, is not absorbed well when taken by mouth. There is no evidence that carnosine or taurine work in epilepsy. Carnitine deficiency may occur with long-term use of the antiepileptic drug sodium valproate but is not recommended as a preventive treatment.

Some popular herbs should be avoided in people with epilepsy. Gingko biloba can decrease the effectiveness of certain antiepileptic drugs. Valerian binds to the same receptors as benzodiazepine drugs (eg diazepam, clobazam, clonazepam) and may cause sedation, as well as tremor, headache and heart problems. Primrose oil and borage may both lower the seizure threshold. Wormwood and sage, used for intestinal symptoms, may also promote seizures. Some herbs also affect the metabolism of antiepileptic drugs. There is little information on the risks or efficacy of homeopathy in childhood epilepsy, despite more than 500 homeopathic treatments for seizures.

Educational Assessment

Following diagnosis, a local Paediatrician and/or a Paediatric Neurologist will monitor a child with Rolandic Epilepsy regularly. Ideally the Paediatrician should refer to a multidisciplinary child development team at a Child Development Centre or a Children's Centre. In some areas there may not be an actual centre, but most local health services will be able to provide this service. The assessment should include a speech and language therapist and a psychologist to obtain a baseline profile of the child's language and cognitive abilities. Formal assessments will be important as a benchmark for medical, therapy or educational intervention.

Following assessment, therapy or educational support will be recommended according to the child's individual needs. Regular monitoring and re-assessment will be needed to inform about the benefit of any intervention.

A child should be referred to a specialist paediatric epilepsy service in particular circumstances. These include:

- Poor response to treatment
- Plateauing development
- Suspected variation in profile of skills, particularly language
- Concerns about educational placement

Educational Needs

In UK practice, routine assessment is achieved principally through the Education and Health Care Plan (EHCP) that has now superseded the Statement of Special Educational Needs (SSEN) in April 2018.

An EHCP is a document that lays out all the educational, health and social care needs that a child or young person may have until the age of 25 and pulls together the relevant providers to meet these needs. This contrasts with the previous SSEN that only considered educational needs.

In addition, young people and their families are more involved in the planning of an EHCP and a personal budget is available. An EHCP is a legal document following from the Children and Families Act 2014 in supporting vulnerable children and those with special educational needs (SEND) that aims to help the young person with epilepsy to attain the goals and ambitions that they and their families wish to achieve (http://www.legislation.gov.uk/ukpga/2014/6/part/3/enacted). The clinical nurse specialist (CNS) will need to know how the young person with epilepsy is affected by their condition at school, college or employment so as to prepare the health section of the EHCP.

Government Benefits

You may be eligible to claim Disability Living Allowance if you are watching over your child throughout the night and day. If you can claim this benefit, you may also be able to claim a Carers Allowance. Please see full details and application forms at www.direct.gov.uk

Travel Insurance

Very often, finding an insurance company, which covers epilepsy-related incidents, can be difficult. Most standard policies will not cover you. Competitive travel policies are available through Epilepsy Action. **www.epilepsy.org.uk**

4. Emotional aspects
Caroline Muir

Each parent will have his or her own experiences of processing diagnosis and day-to-day management of Rolandic epilepsy. It would be wrong to try to generalise about how one negotiates this emotional territory. I can only say that for me the early days of discovery and acceptance were bewildering. However, because of extremely professional medical personnel, from the GP to the consultants and specialist epilepsy nurses, my fears were managed at an early stage. I will just describe the processes I went through and hope these resonate with other parents in the same situation.

Discovery

The first time I was fully aware that my daughter Anna might have epilepsy was three in the morning, and unusually for her, she came into our room, mumbled something about a nightmare and got into our bed. She then had a generalised seizure lasting approximately 8-10 minutes. At that point she suddenly seemed barely recognisable. The colour drained from her face, her eyes rolled back, and she jerked and writhed vigorously. While we watched aghast, I called an ambulance, and my husband stroked her face. I could hear the servicemen come up the stairs as Anna began to breathe again, and the movements cease. The seizure we had witnessed had vanished as quickly as it had arrived, and there was an air of complete stillness and calm. I could barely believe what I had just seen. Then other incidents (Anna describing how she couldn't move her jaw, and a couple of reports of splitting headaches) suddenly took on a whole new significance. Anna was given oxygen and spoke a few words of surprise to see the ambulance men in the room. She had no knowledge of what had happened. It would take us all as a family some months to understand fully what was involved in this development.

Diagnosis

Ignorance And Fear

We learned our daughter had Rolandic Epilepsy after a series of tests – blood, EEG, and MRI, at Kings College Hospital, London. The MRI was a tough thing to guide my daughter through. It required a bit of acting on my part to appear confident and matter of fact about the procedure. I was grateful Anna was young enough to see it simply as an inconvenience: noisy, uncomfortable, and forcing her to miss the school she loves. For us parents the MRI test had much darker resonance; there is a high incidence of cancer in both our families. On reflection, it is a shame there is nobody able to offer an immediate response to such tests, but I learned to adopt the mask of patience, however churned up I was feeling. We waited for several weeks before I rang around the hospital and the GP to have the result confirmed. When we got the 'all clear' on the cancer issue, everything else seemed straightforward for a moment. The EEG indicated RE, and we were referred to a Paediatric Neurologist via our GP. I remember saying to my GP how relieved I was, since "nobody ever dies from epilepsy, do they?". She just looked me straight in the eye and said, "I'm afraid sometimes they do". I was shattered. This was the level of my ignorance about epilepsy. I realised I had some work to do to arm myself with the facts. The next thing I discovered was just how many different 'epilepsies' there were, and was told that RE was, in the words of the consultant "the one you want your child to have, since most children grow out of it". This was all very well, but it stood at odds with the conversation I had just had with the GP. Research told me just how many childhood pursuits were potentially hazardous because of epilepsy. All I could see for a week or so in my mind were worst-case scenarios.

Guilt

It was terrible to feel I had let my daughter down somehow, not reading 'the signs' of partial seizures. I had only the most rudimentary idea of what the condition involved in terms of symptoms; a patchwork of scraps of misinformation, and supposition. To get the better of this guilt I carried I began to find out what practical steps I could take to observe the correct safety precautions for Anna's new daily life. It was all about finding a balance between caring and flapping. I was also guilty that Anna's epilepsy could have been caused by an injury we parents could have averted in the first place by being more vigilant. Little did I know then that her epilepsy was much more likely to have a genetic cause. I was far too busy blaming myself to think straight.

5. Living with RE
Caroline Muir

Early Days

After we'd received Anna's diagnosis, we were told to keep our minds open and our eyes alert. The consultant thought that his diagnosis held considerable weight, but he could not be 100% certain. It would help, he said, if we could write down a detailed diary of Anna's physical state day by day. This is not easy with a child who seemed to have her worst seizures during the dead of night. I recorded details of partial seizures, the odd early morning generalized seizure, migraines, and events which we just weren't sure about – jaw stiffening, shaking, and shivery feelings, momentary pains around the eyes, things that didn't seem quite right. I found the simple act of writing these things down hard. I didn't want to remind myself what was staring me in the face. Our child had a condition that brought her experiences that neither she or we could control or even fully understand. These seizures might come when she was doing what other kids liked to do; ride a bike, climb a tree, have a sleepover. They would, with a bit of luck, disappear over time, if we made sure Anna took her medication and avoided triggers wherever humanly possible. In her case 'happy anticipation' could kick start a seizure. How in practical terms could we stop her from looking forward to Christmas or a trip to a theme park? These kinds of event seemed to bring about partial seizures. I realised I was being a perfectionist in trying to manage her condition. It was for the consultant to manage her medication, and for me to manage my feelings of dread and panic when I realised Anna had had a seizure. I learned somewhat slowly and painfully that it was OK to have partial seizures, and that with luck, nothing very bad need happen when they occurred. Moreover, the consultant explained that we weren't going for total eradication of seizures at the expense of very high doses of medication. This could be detrimental in terms of side effects. Rather, we were trying to find a happy medium between 'enough' medication and a manageable amount of partial seizures. What I had to do was worry less behind the scenes, or I would soon be an exclusively 'no fun' Mum to be around. At this point I made a conscious decision not to surf inexpertly through the information on the internet, and just be thankful that experts were monitoring my child's particular circumstances.

Sleep

What I couldn't do was sleep alongside Anna, as I found I just didn't sleep at all. At this point, the Muir Maxwell Trust was a Godsend. They provided an epilepsy alarm for Anna's bed, and this gave us parents a degree of faith that anything serious would be picked up. At the same time, the medication made Anna incredibly drowsy at first, and she seemed to sleep like a newborn baby. Early nights, reassurance, gentle encouragement to take daily medication on time - these were the priorities.

What To Tell The School

Getting staff at Anna's school to be vigilant required some practical steps, particularly after I discovered Anna had gone on a swimming trip with a supply teacher who had had no knowledge that Anna had epilepsy. I made up an A4 fact sheet with Anna's photo on it. This was placed in the staff room and school

office alongside profiles of children with other serious conditions. I also found a member of the office staff willing to come with me to our local hospital to be trained in how to administer emergency medication in the event of a prolonged generalised seizure. Then I made sure this went on all school trips and was kept somewhere easily obtainable, with clearly marked instructions. All children need watching during sports activities and in the playground.
I just had to remind myself that Anna was no different to the others as long as the medication was working correctly. (See also guidance notes for schools on how to support a child with epilepsy http://www.epilepsysociety.org.uk/whatwedo/schoolsawareness/programme)

Records

As to the diary I was supposed to be filling it, it was a hit and miss affair. First of all, I would write down Anna's symptoms on bits of paper, then lose them. Later the hospital provided me with a diary for adults with epilepsy. This was slim, and had tiny columns to fill in. Anna found it one day, read a few entries and put it in her mouth ready to take a bite out of it. That's how much she hated engaging with the daily business of her condition. After a fortnight of this, I came to her bedroom to take her pill up to her. She sat up in bed and her face crumpled. Tearfully, she told me how cheated she felt, to be only eight years old, feel tired all the time, and realise she would have to take these pills for as long as she could "ever imagine". She had had illnesses before, she explained, and they always went after two weeks, so why was she still forced to take these horrible pills? I realised I had not explained the basic facts about epilepsy in a way that Anna was able to fully absorb them. Just because I had explained them once, didn't mean she had taken them in. Nor had the doctors managed to make things clear to her, despite their patience and careful tailoring of language to her age and level understanding.
I would have to go over things with her, just as we did with times tables, until the important features of epilepsy had been absorbed.

Self-Consciousness

Anna had become increasingly unsure of herself in relation to her condition. She longed to keep it a secret, and hated it when I had to explain it to parents and carers during play dates and sleepovers. At one stage she thought children were "bullying" her because they were curious about it during playtime chats. It was pretty clear to me when she discussed this that these kids were just expressing their curiosity. I realised I had not equipped her to answer her own questions about epilepsy, let alone discuss it with confidence with her peers.

A few months later, with the help of the incredible people at the Education and Information department of the National Centre for Young People with Epilepsy, I began a project to create a diary for children with epilepsy between the ages of around 5-11 years. Within a year I sat down with Anna to fill this new diary in together. This diary was created to give children like her all the information they might need. But it would be couched in a book that looked "cool" and was filled with jokes, games, stickers and puzzles. Anna could carry this diary around with her, without worrying if anyone would tease her. She could stop looking for negative reactions where there weren't any, and instead enjoy the fact that since her seizures were now fully managed by the medication, she could do everything her friends did. Having a diary really helped us work with the consultant to get an accurate picture of seizure patterns, and it made me feel more confident about the consultant's decisions about medication levels.

Travelling And Holidays

When we go away, I always take Anna's emergency medication with us. This means that if she should have a prolonged generalized seizure, I am ready to try to help her. I have never had to use this treatment to date, but I have been grateful for the extra peace of mind it gives me. I have always put extra blister packs of Anna's regular medication in the car just in case we lose some pills along the way. I am always a bit more vigilant when Anna is swimming, but I haven't ever felt the need to stop her joining in the fun. It was a great relief to realise Anna does not have photosensitive epilepsy, and she loves fireworks and 3D cinema. But when we have pushed all our children physically, such as on a long hike, I can be pretty sure Anna will have a partial seizure. Having talked this through with the consultant, I have weighed up the value of exciting shared experiences against the relatively mild seizures that do not worry Anna. It seems a fair exchange. When Anna looks back on her childhood it will be these out of the ordinary family adventures she will remember, like climbing to the top of a mountain in the Lake District, not the shaky legs that came towards the end of the walk.

Sleepovers

It was not easy in the early days to relay basic information in a way that did not appear to alarm other parents, and then I realised that this was mostly my problem. I was anxious because at the back of my mind I had a picture of Anna having her first generalised seizure.

I felt bad just asking other parents to take this possibility on, and Anna felt bad that I had to bring the subject up. I just had to remind myself again and again that a) the medication worked and b) there was only the slimmest chance that any other parent might have deal with a medical emergency of this sort. On top of which her friend's parents were my friends too. And they would do whatever a parent had to do, given the worst-case scenario, a serious seizure. What helped, once again, was the fact sheet /medication pack I had made for Anna's emergency care. This meant parents could read it at their own convenience, before the event. We got to know a new level of generosity of spirit and were all the better equipped when my children's friends had 'serious condition issues' of their own.

Growing Out Of RE

As I write, my child is recently medication free. If I am honest, I am more watchful for signs of seizures than perhaps at any other time. I know that this too will pass, and I am grateful that for now, we don't have to begin and end each day with medication. As usual, my child takes the lead on the emotional front; in being more interested in the good things around her, not her past medical history. I keep the emergency medication around, I take each day as it comes, and I try not to be too fixed in my certainties. Anna is twelve, full of fun, and on to the next thing. We talk about her need to be completely honest with me, as much as before, if she experiences any partial seizure symptoms. We live in the needs of the day.

6. Growing out of RE
Kelly Pretz

Most kids would agree that growing up is not always easy. It can be a challenge for young kids to understand who they are. I was just beginning this process of figuring out who I was and where I was going five years ago. But on the 13th of December life changed dramatically. This was the day of my grand mal seizure. I was ten years old.

That night I was taken to the hospital and stayed overnight. I remember I couldn't fall asleep, no matter how much I wanted to. I was exhausted as I lay in the hospital bed with my mom next to me, not being able to sleep. The night seemed to go on forever but finally morning came, and we were allowed to go home. At this point there were no answers as to what had actually happened to me. I remember my doctor came to see me and suggested many different theories as to what I had just experienced. No one was really sure what had happened.

It was decided that I should have an EEG. I had no idea what to expect. My thoughts were racing with questions like: Why is this being done? Who's giving me the test? Is it going to hurt? But because I was 10, my real question was: "Were they saying 'egg'?"

The first EEG was by far the hardest out of all the EEGs that I have taken. It was so scary. I had to lie on an uncomfortable bed with weird sticky stuff in my hair that attached to my head with wires to a computer. I was told this was the way they could see my brain's activity. They were looking at the lines and if they went up and down like a mountain range that would mean something serious was going on. After 45 minutes they had enough on the monitor to see lines that looked like a model of Mount Everest.

I was taken to see the neurologist. He was smiling at me and asked me to talk about how I had been feeling. I talked about the times I felt like saliva in my mouth just welled up so quickly I had to run and spit it out. I talked about the weird feelings in my mouth and throat and neck. He asked me to talk about how they felt. All I could say was they were feelings that hurt sometimes and made me feel uncomfortable.

The neurologist said, "I know what this is". He said it was Rolandic epilepsy. My parents looked scared and so was I? I didn't know what this was but I knew it was not good. I somehow knew this was about to change my life.

Afraid of what my friends would say, I kept my condition a secret. No one wants to be different at school. Nobody wants to be known as the girl who has seizures. I decided to hide it and tell no one. My mother did tell the school nurse and the teachers I came into contact with. But I preferred to keep it to as few people as possible. So that meant not telling any of my friends. I was afraid I might lose friends if they heard about me. I figured they would be afraid.

So, life went on. My friends didn't know anything about my epilepsy. But it is hard to hide something this big. I was taking medication that made me tired and I really had no energy. I had to change certain things in my life. I couldn't do all the activities in gym. We have a rock wall and I was not supposed to be off the ground for fear I would have a seizure. I was not supposed to go to sleepovers because I needed to get my sleep. The list was short but it was a big change for someone like me.

The summer I turned 13 was the hardest. I was still having lots of feelings and I was put on more medication. I was more tired than ever. I had all these wonderful friends but because I was too tired to be with them I started to lose touch. It was awful and it was very upsetting. My friends thought I didn't want to be around them – which of course was not the truth – and I realized then they deserved to know what was really happening to me.

I made the decision to start with my best friend. We were playing volleyball in her backyard when I brought up the subject of my absences that looked like I was avoiding her and the others. I told her I had epilepsy. I held my breath, wondering how she would react.

"What's epilepsy?" she asked curiously.

So I told her, and I explained how I had been experiencing and taking medication for it. I told her I had to see the doctor a lot that summer which was why I was never around to be there for her.

'Well, you're still you." She told me and then told me it was my turn to serve the ball.

That moment, telling someone who I knew cared, made me feel like I could do anything. That simple conversation allowed me to be confident.

Confident to prove myself.

Summer came and went and soon it was time for my freshman year of High School.

I decided to make a choice. I would enter high school and not allow the epilepsy to be a negative in my life. Instead I would turn it into a positive. That was when my Purple Mission began. My dream is to bring awareness throughout my community and beyond about epilepsy. If one in ten people suffer from some sort of seizure activity I thought there are a lot of us not saying what is happening? This needs to change!

On my search for a change I stumbled across the Epilepsy Foundation of New Jersey where I met people making a difference. I knew that I belonged there. I met with two amazing women who welcomed me with open arms. Since my first meeting with them, I have been volunteering at the organization ever since. There I help with promotions and planning events and doing whatever I can to make a difference in everyone.

Once I was in High School I started an event to bring epilepsy awareness. This year I sold hand beaded bracelets at my lunchtime to the students and teachers. Each year I plan on doing some event at my school and in my community to continue to raise awareness. The monies I raise go directly to the foundation. Things were going well. Soon, though I would be getting even better news…

About two months ago I went back to see my neurologist. It was time to take another EEG. My test results were normal. For the first time in five years they were normal. I had just turned 15. It was time to start getting off the medication I had been taking for what seemed like forever. I am still in the process of getting off the medication. I was happy and a little nervous to start the process. Of course without the medication I could possibly have another grand mal seizure. But my doctor says, no. He believes this is almost over for me. What a wonderful thought to be done. I am lucky!

Actually, I really will never be done. I believe someone has to speak out about epilepsy and help to take away the fears that people have about it. It is a condition that must be handled and understood. But it doesn't change who you are. I wrote a book about someone like me who keeps her condition a secret. It is going to be published in summer 2011. Just like the character in my book we both learned to be brave and to love what we are. In the end this was never a bad experience for me. I grew in ways I never knew were possible. I am proud of all that I have done. I hope I can help someone else just like me.

7. Research
Deb Pal

Medical research is important because without it there would be no new medicines or tests, no improved treatments, or better ways of providing health care. For example, some treatments developed in the past 20 years have revolutionised survival for major health problems. The clot-busting drug streptokinase can reduce sudden death after a heart attack by a quarter. Tamoxifen in breast cancer has led to a 20-30% improvement in survival.

Genetics

Rolandic epilepsy is a complex genetic disorder, which means that it is caused by a combination of variations in different genes. We inherit genes from our parents and sometimes the combination of genes that are passed on to an individual result in the symptoms that make up RE. We also notice that many relatives of children with RE have speech problems or dyslexia, and we know that both of these conditions are also genetic. Thus the same genes that cause speech problems and dyslexia may also be involved in RE.

Genetic research is important because by knowing the exact cause of RE, we can develop specific tests to diagnose it more quickly and accurately. Also, by knowing the cause, we can design new treatments that act specifically on the cause and hopefully have fewer side effects than conventional antiepileptic drugs.

Our team at King's College have found the parts of the genetic code that cause the main features of RE. With your help we can narrow down exactly what is different about the genes in people with RE compared with healthy people. If your child has a diagnosis of RE, you may be eligible to take part in our research study. Please visit http://childhoodepilepsy.org to see which studies you may be eligible to take part in.

Neuroimaging

Children with rolandic epilepsy often have problems with speech, language and attention, probably because the brain circuits involved overlap with the areas from which seizures arise. We do not know though, why these circuits are affected, and whether the brain tries to compensate for this interruption. If we knew, then we might be able to help these circuits to reform or adapt by some other means. In our neuroimaging programme, we are visualizing the brain circuits in children with rolandic epilepsy. For latest progress visit our website http://childhoodepilepsy.org.

Sleep

Children with rolandic epilepsy often have seizures during sleep and are usually exquisitely sensitive to loss or disruption in their normal sleep patterns. But do they have different sleep patterns from the rest of us that put them at risk of seizures? Can the quality of their sleep be improved to protect them from seizures? We are launching a new clinical trial to find out whether it is better to treat children with antiepileptic drugs or not and also whether improving their sleep has a beneficial effect. See http://castlestudy.org.uk for the latest news.

Finding out about clinical trials

Clinical trials are research studies that involve people with a medical condition or healthy volunteers and are designed to compare new or existing treatments. Treatments might include drugs, vaccines, surgery, psychological therapy or even disease prevention. Involving people in clinical trials allows us to see whether the new methods are effective and safe. Trials are very carefully planned and regulated to ensure that human safety is paramount.

You can keep up to date with new clinical trials that might be relevant to your child through www.clinicaltrials.gov

Our clinical trial of treatment vs no-treatment plus sleep is at http://castlestudy.org.uk

Taking part in a research study allows you more contact with your medical team than you normally would have, and there will be opportunities to gain information about your condition that may help you to manage it better. In many cases the research may not help you personally, but it may provide vital information that will help others in the future.

8. Resources

Our lab website
http://childhoodepilepsy.org

Epilepsy Therapy Project (US)
www.epilepsy.com
The most comprehensive source of information and support about epilepsy on the web.

Epilepsy Action (UK)
www.epilepsy.org.uk

Epilepsy Wales
www.epilepsy-wales.co.uk

Epilepsy Scotland
www.epilepsyscotland.og.uk

National Centre for Young People with Epilepsy **www.ncype.org.uk**

Irish Epilepsy Association (Brainwave)
www.epilepsy.ie

Epilepsy Research UK
www.epilepsyresearch.org.uk

Joint Epilepsy Council
www.jointepilepsycouncil.org.uk

The Association for Child and Adolescent Mental Health ACAMH
www.acamh.org

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