

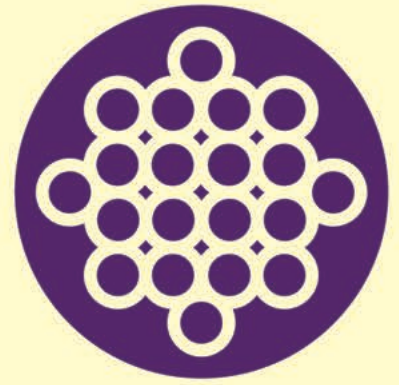
Life with r(20) syndrome

Zara's Story: The Early Years



ring 20

research & support uk



Ring20 Research and Support UK CIO are a charity providing information and support for individuals, families and healthcare professionals who are affected by, or who come into contact with Ring Chromosome 20 Syndrome [r(20)] - a rare epilepsy syndrome.

Based in the UK we connect families from across the world and our support services may be accessed by anyone affected by r(20) syndrome.

We actively advocate for research to try to improve outcomes for all those living with r(20) syndrome.

Our aim is to improve understanding of the impact of r(20) on daily life, the challenges and how to overcome them. We recommend sharing this comic with families, friends, carers, schools/colleges, work colleagues, social workers, sports coaches/group activity leaders - indeed anyone involved in the support and care for a person living with r(20).

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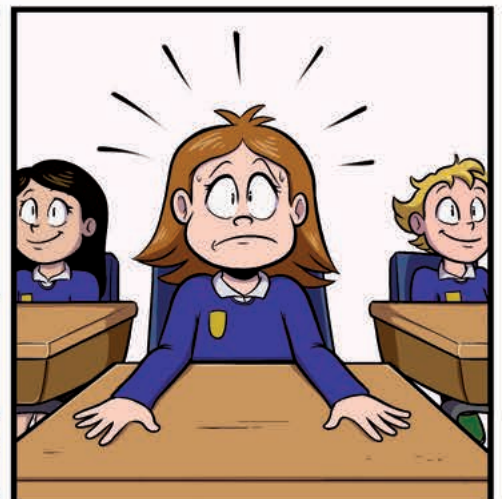
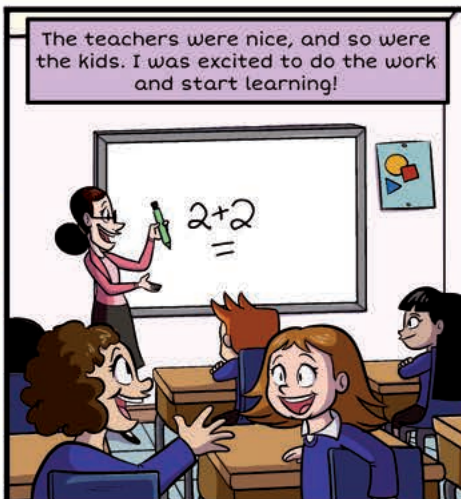
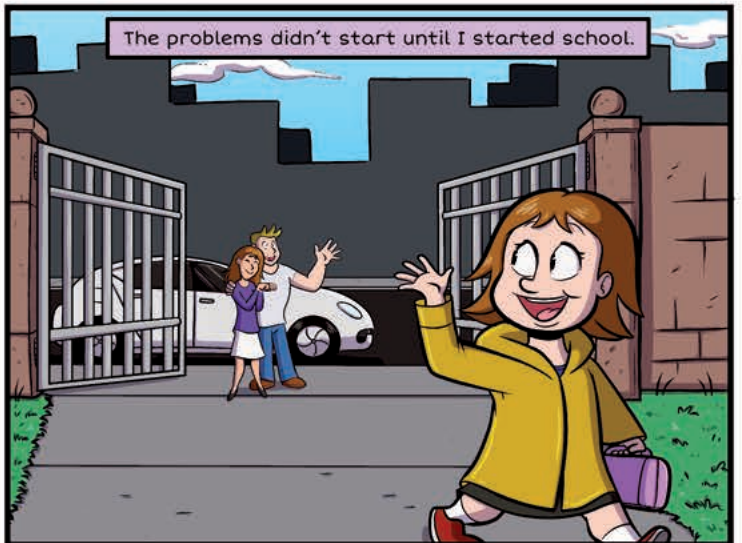
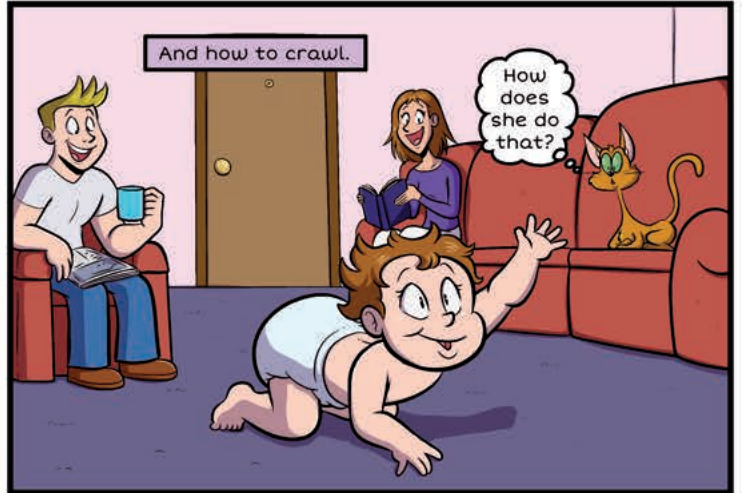
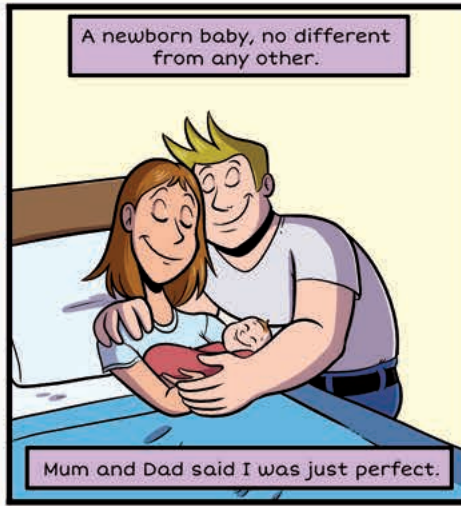
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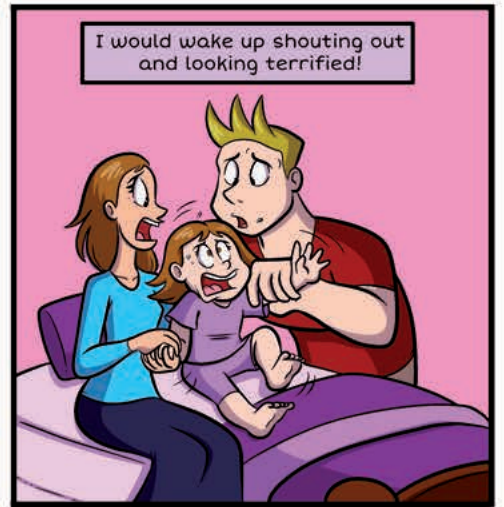
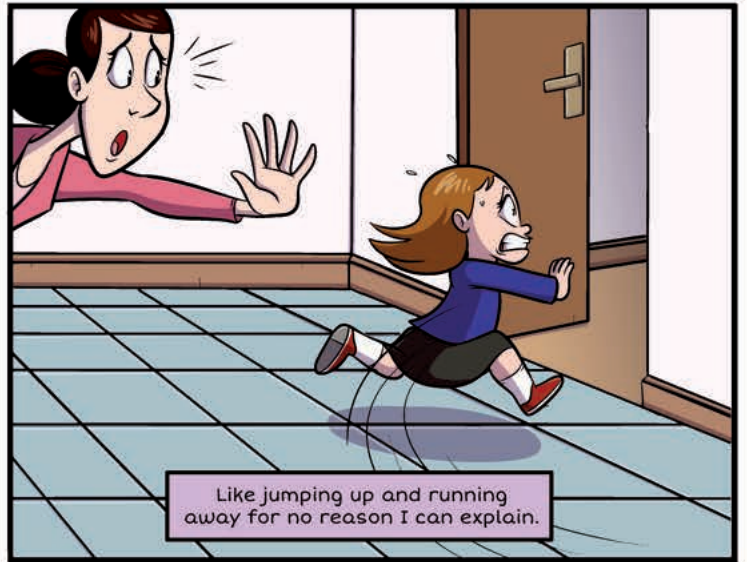
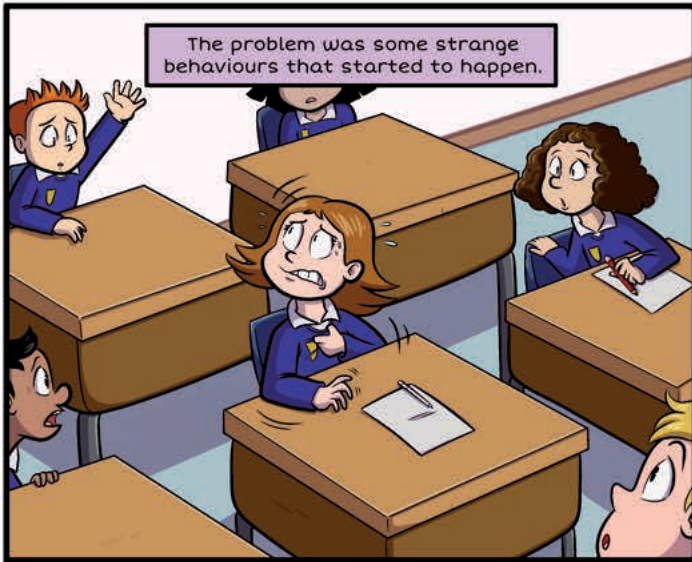


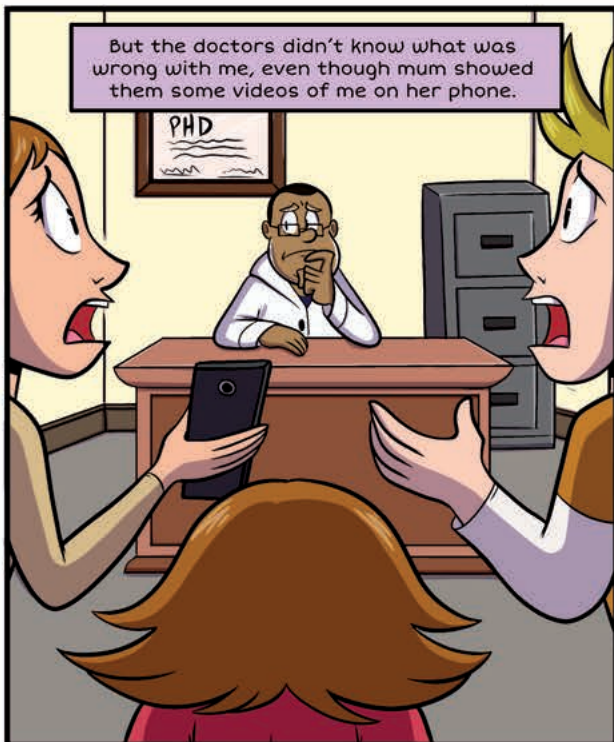
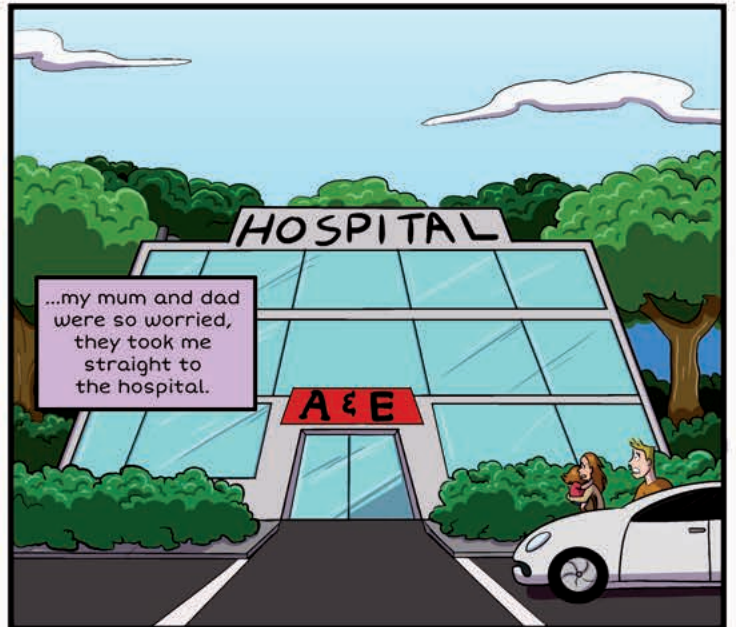
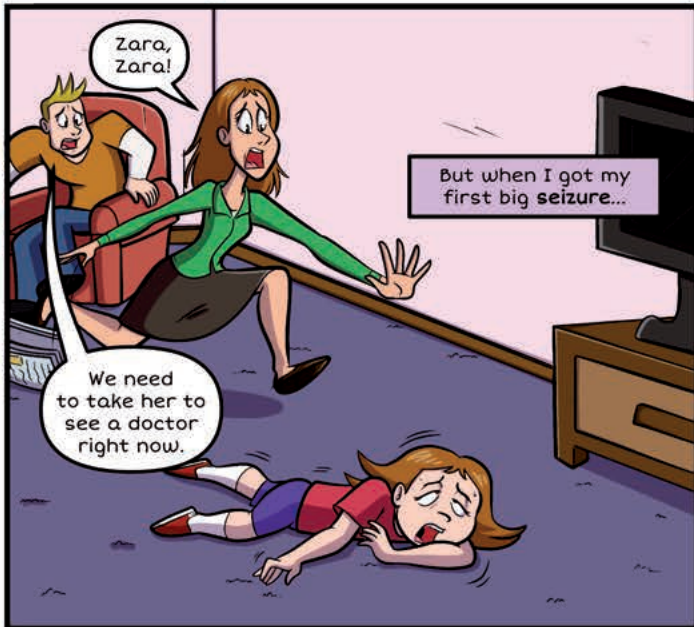
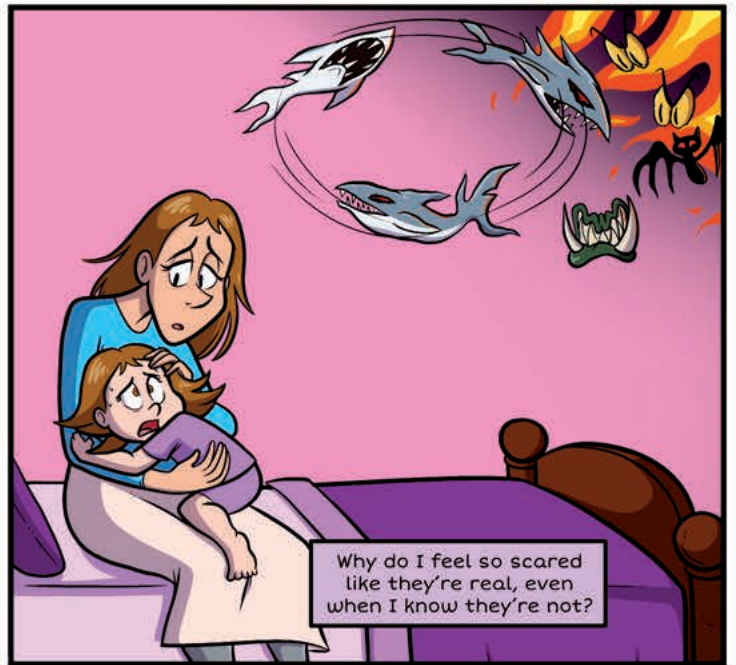
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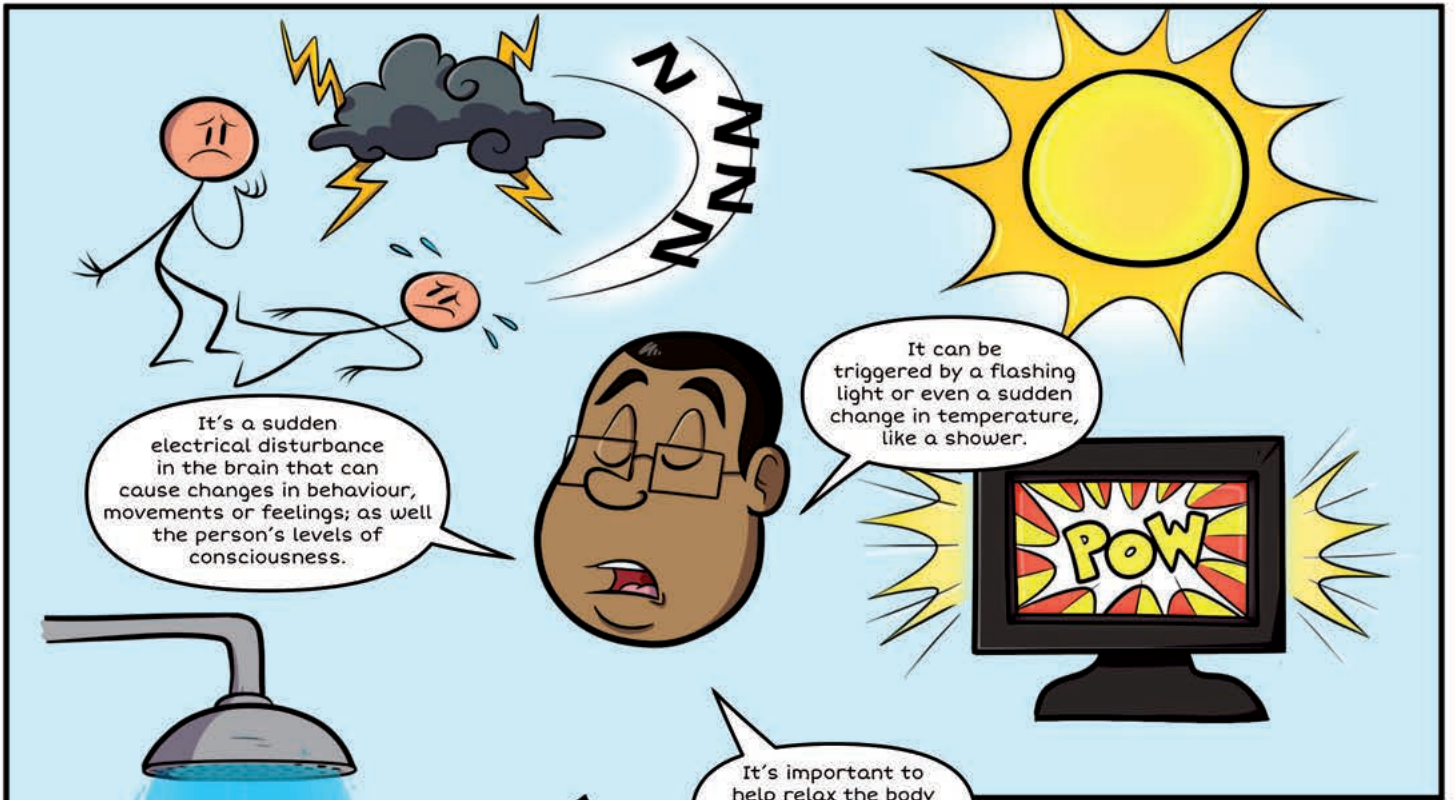
The experience that Zara has in this story is not what every person living with r(20) goes through. It aims to address some of the complications that r(20) can bring to the already difficult experiences of growing up, and help people who don't have r(20) but have experienced growing pains understand what it's like.

We hope that you enjoy reading Zara's story and will go on to read part 2 as Zara grows up and comes to terms with growing into an adult.









It's a sudden electrical disturbance in the brain that can cause changes in behaviour, movements or feelings; as well as the person's levels of consciousness.

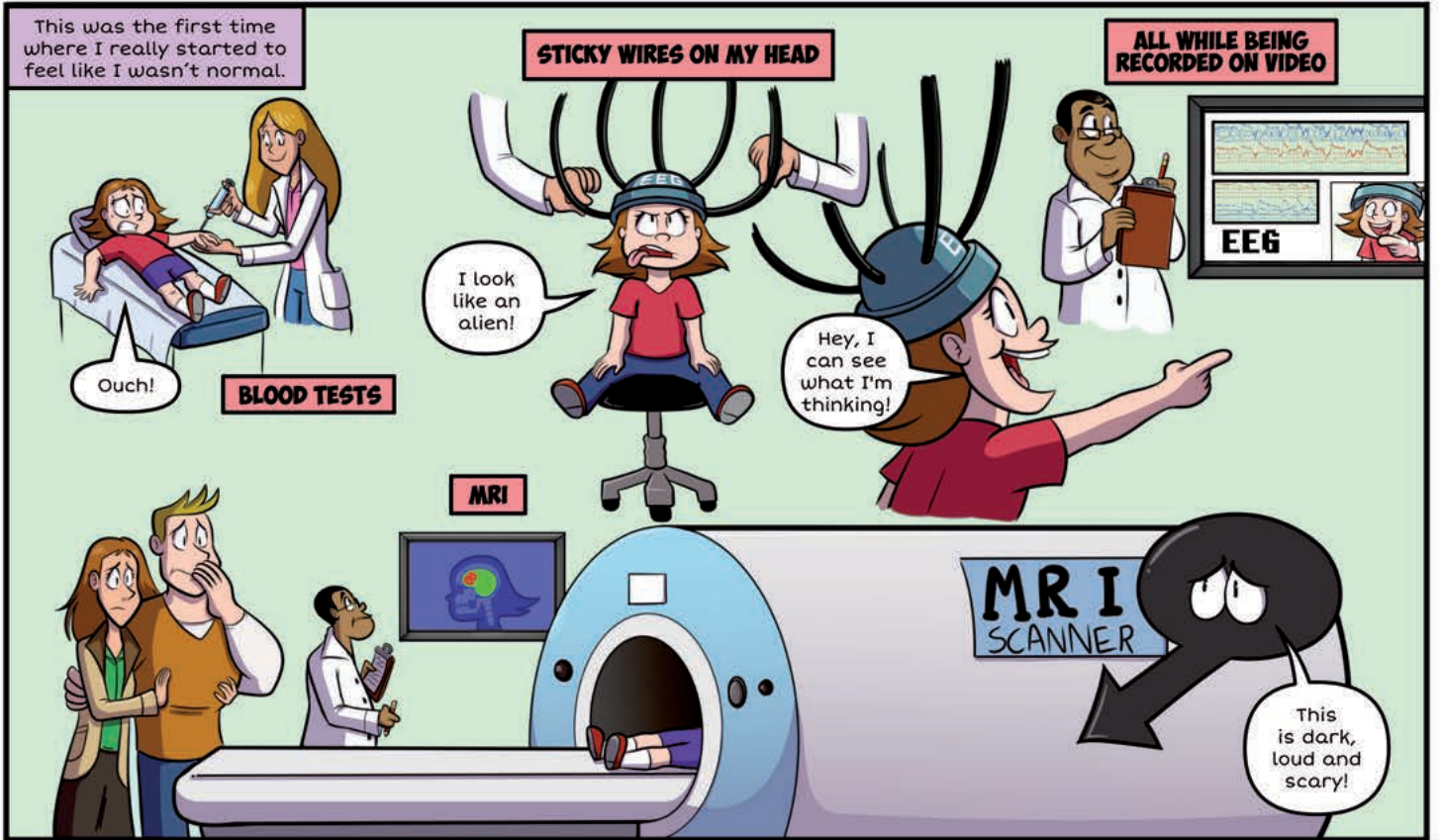
It can be triggered by a flashing light or even a sudden change in temperature, like a shower.

It's important to help relax the body during a seizure. Calming music can help, for example.



So what can we do?

I recommend Zara's gets some tests. That way we can find out what's wrong with her.



This was the first time where I really started to feel like I wasn't normal.

STICKY WIRES ON MY HEAD

ALL WHILE BEING RECORDED ON VIDEO

BLOOD TESTS

I look like an alien!

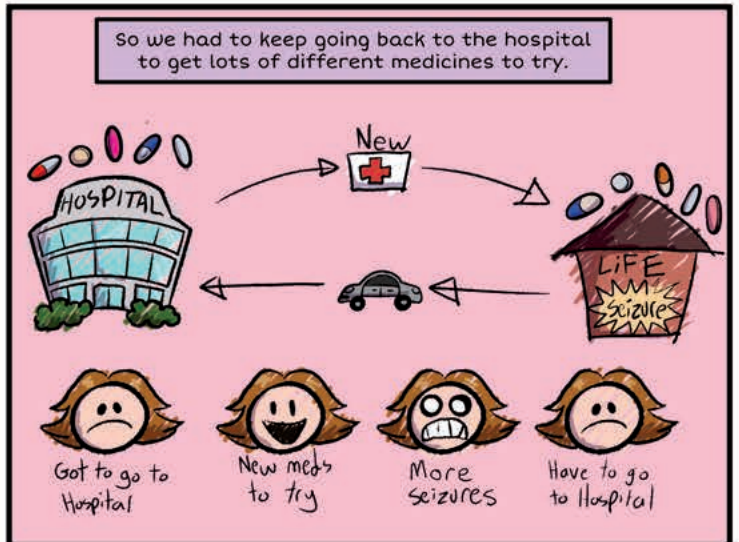
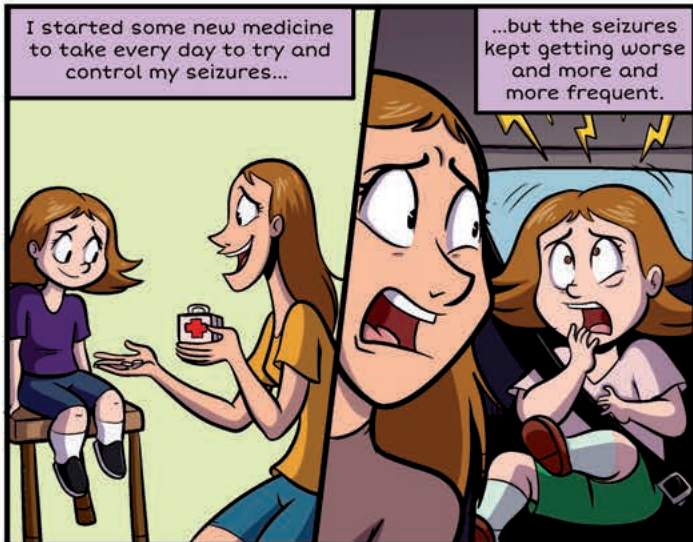
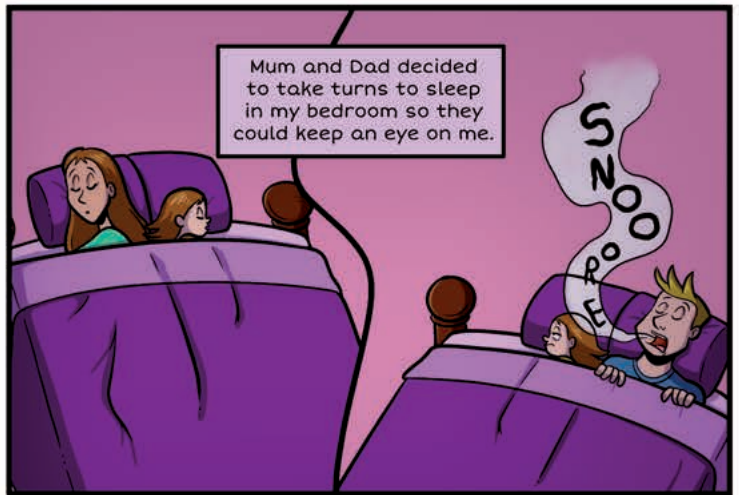
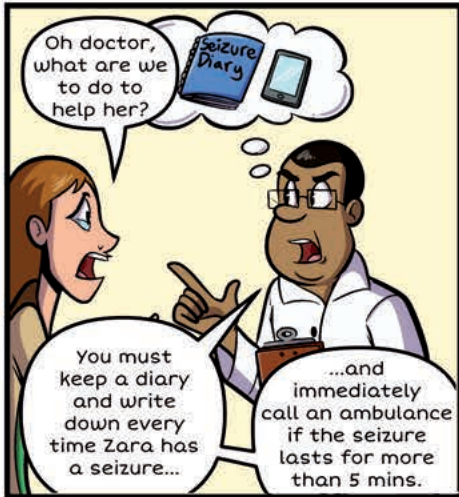
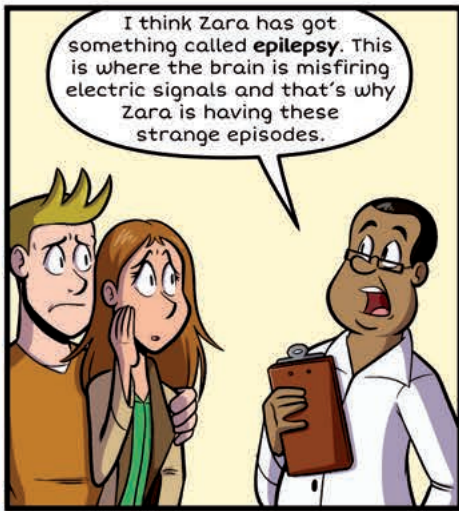
Hey, I can see what I'm thinking!

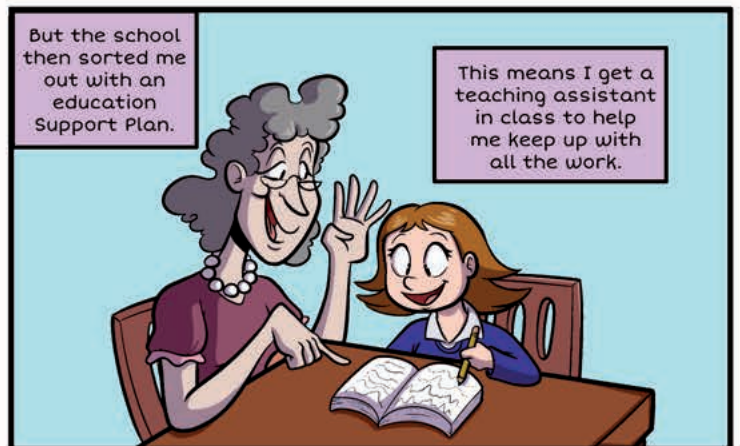
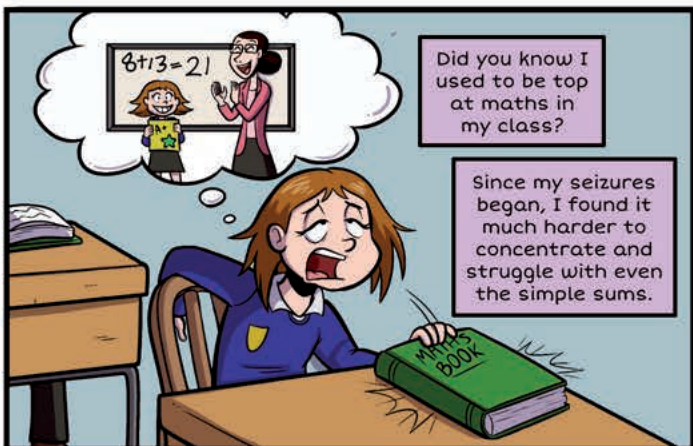
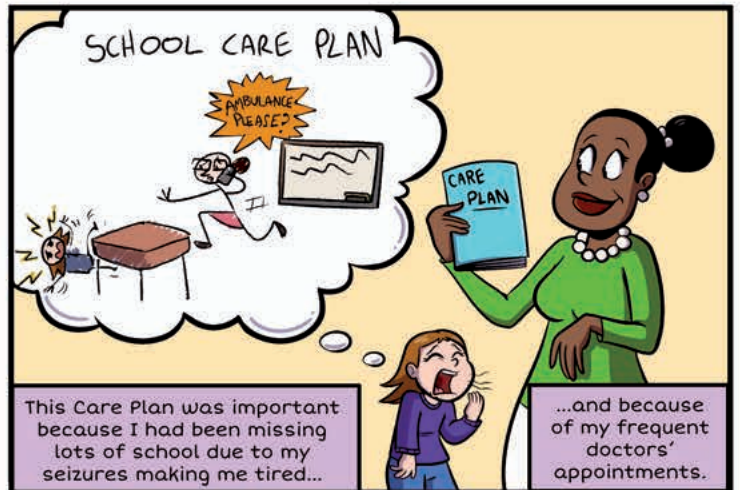
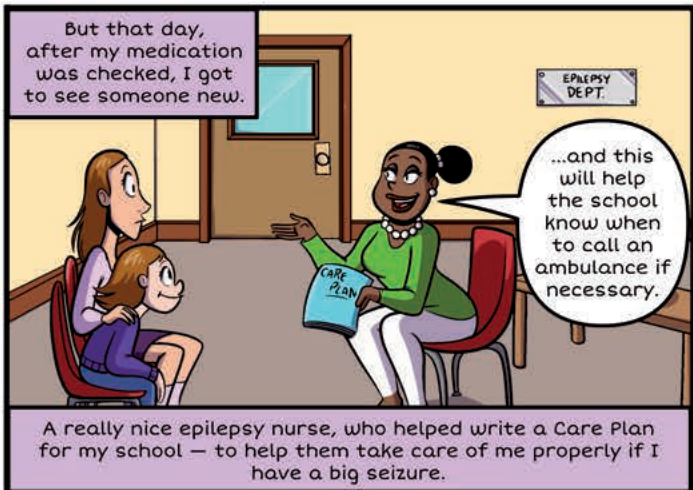
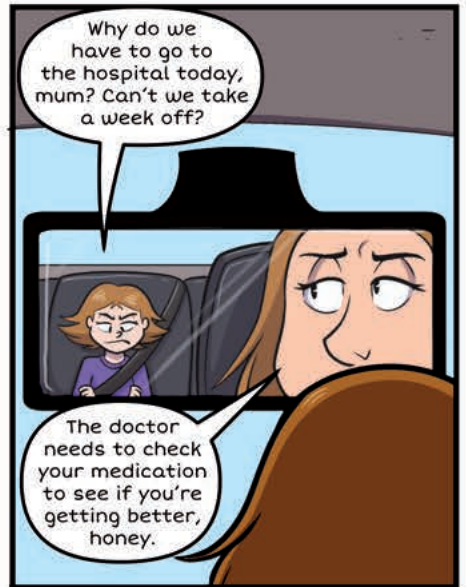
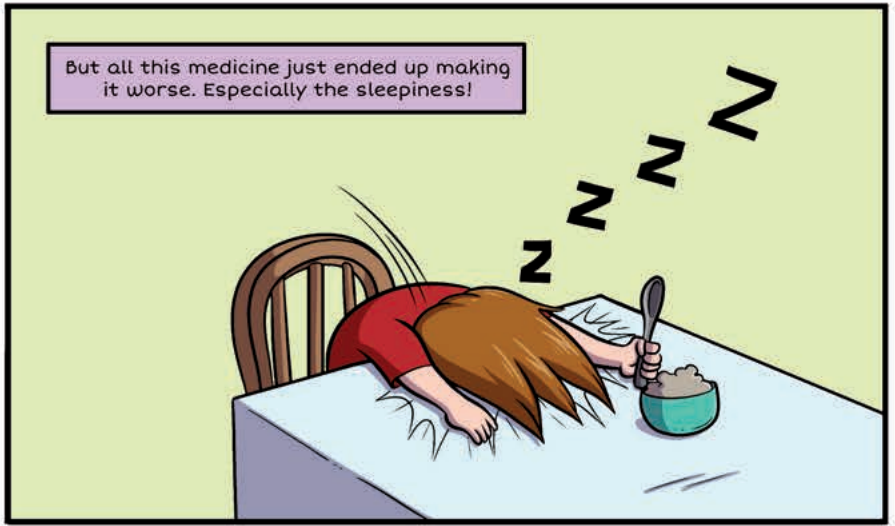
MRI

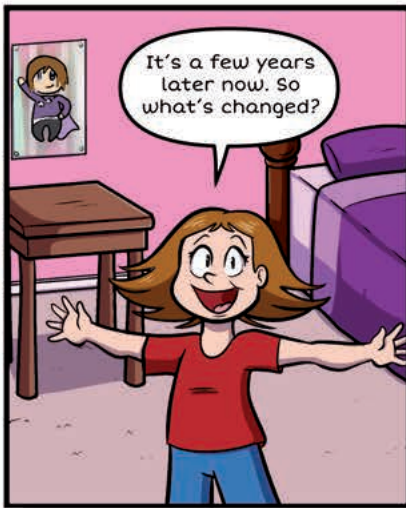
MRI SCANNER

This is dark, loud and scary!

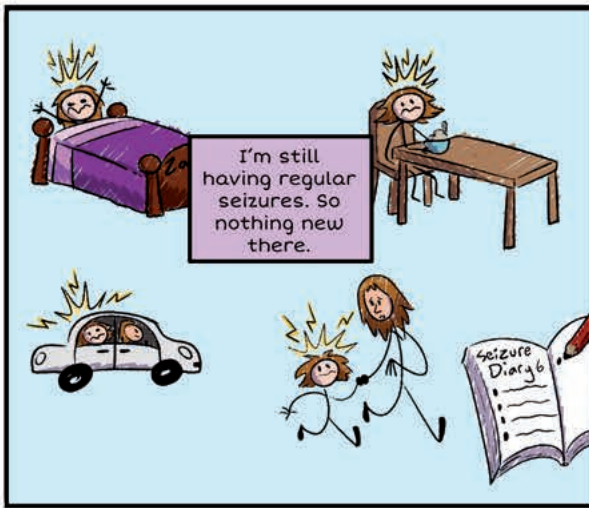
Ouch!



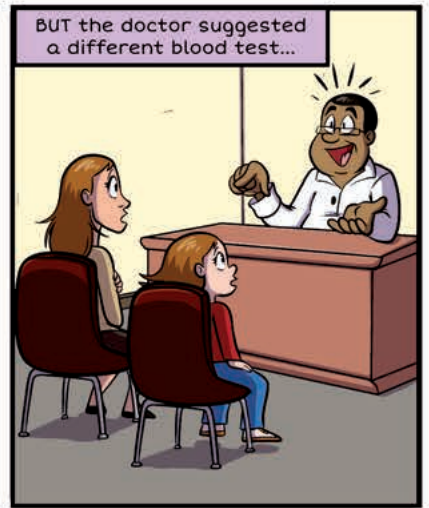




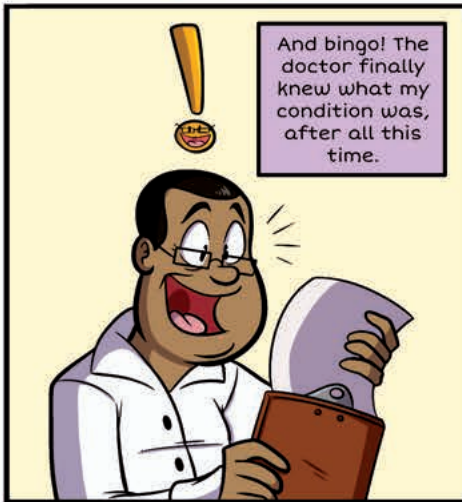
It's a few years later now. So what's changed?



I'm still having regular seizures. So nothing new there.



BUT the doctor suggested a different blood test...



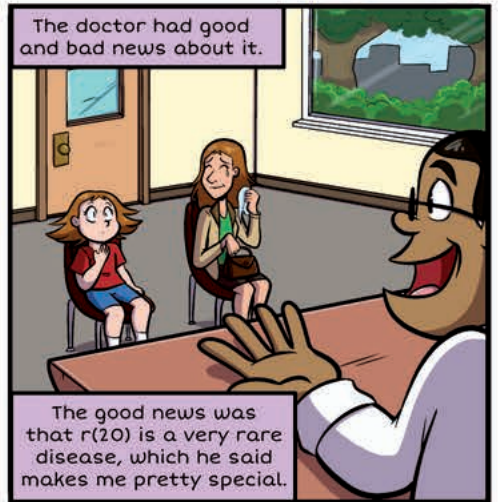
And bingo! The doctor finally knew what my condition was, after all this time.



r(20)

He told me I have something called "Ring Chromosome 20 Syndrome".

That's a bit of big mouthful, so let's call it "r(20)" for short.

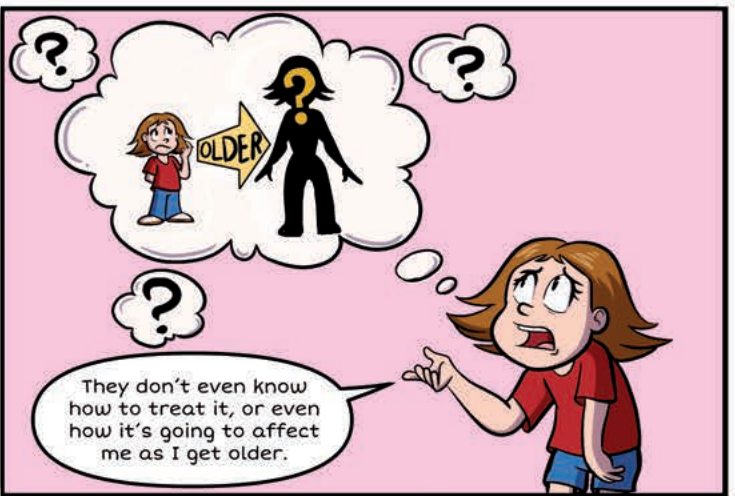


The doctor had good and bad news about it.

The good news was that r(20) is a very rare disease, which he said makes me pretty special.



The bad news was that there's not a lot known about r(20) and there is no cure.



They don't even know how to treat it, or even how it's going to affect me as I get older.



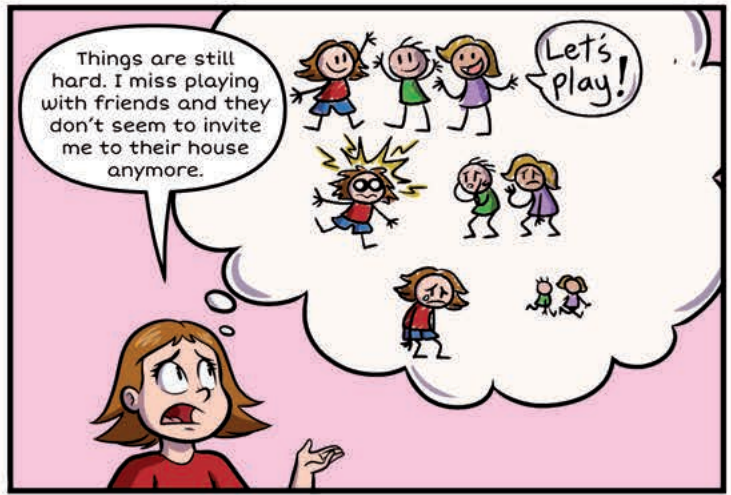
However, we found out there is a support group that we can join to learn more and speak to other families who have someone with r(20).

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R-20 Group



I've already made a great friend there. She's cool, and has r(20) just like me.



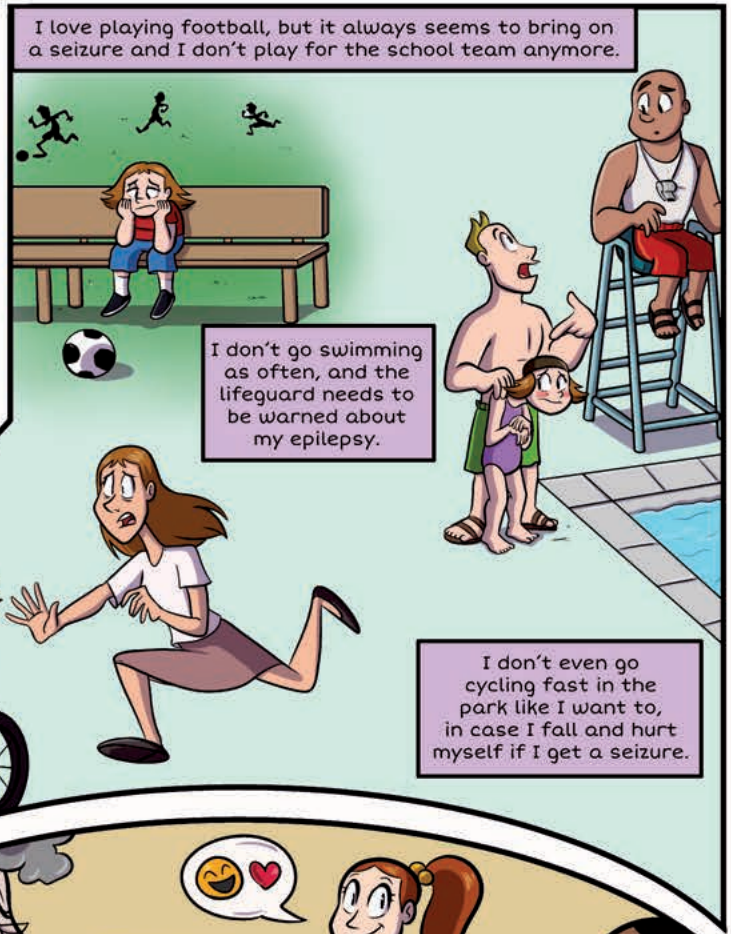
Things are still hard. I miss playing with friends and they don't seem to invite me to their house anymore.

Let's play!



If I get invited to a birthday party, mum and dad stay with me in case I have a seizure.

I don't feel the same as the other kids.



I love playing football, but it always seems to bring on a seizure and I don't play for the school team anymore.

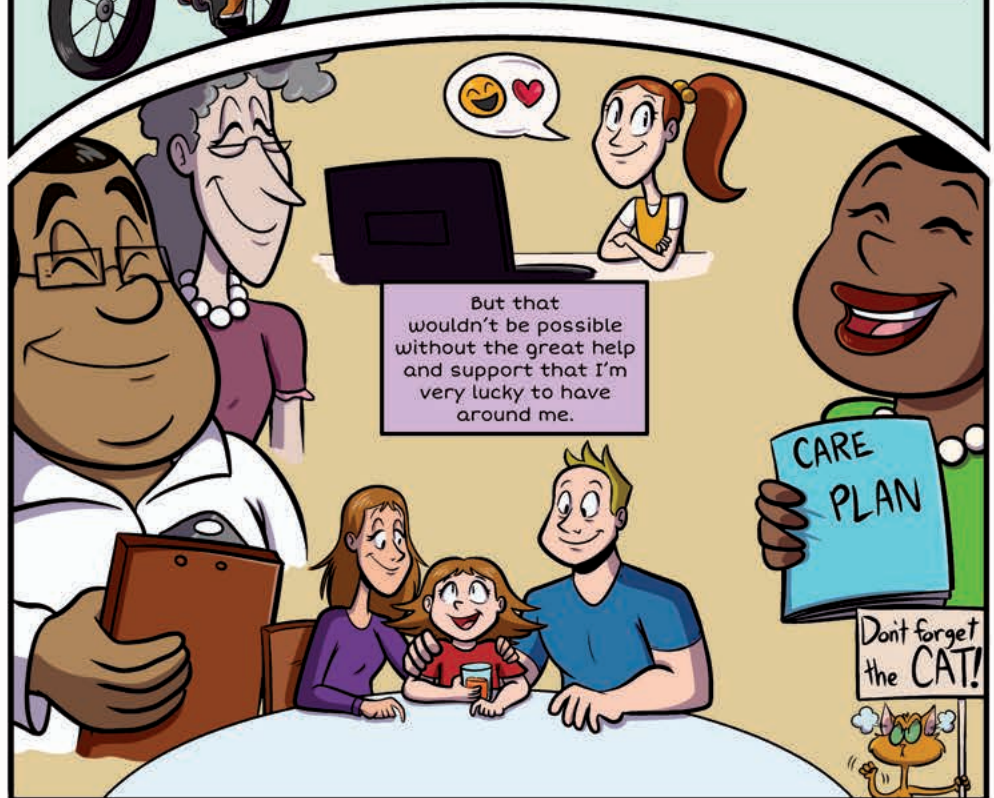
I don't go swimming as often, and the lifeguard needs to be warned about my epilepsy.

I don't even go cycling fast in the park like I want to, in case I fall and hurt myself if I get a seizure.



But you know, at least I'm still able to do these things.

At least I can still go swimming, cycling, and find ways around these difficulties.



But that wouldn't be possible without the great help and support that I'm very lucky to have around me.

CARE PLAN

Don't forget the CAT!



RING CHROMOSOME 20 SYNDROME

r(20) syndrome is a very rare condition in which one of the two copies of chromosome 20 has formed a ring rather than the typical linear chromosome structure. Why the tips of the chromosome join together to form the ring is not understood and how the formation of the ring affects the function of genes packaged on chromosome 20 is also not understood. The consequence of the ring formation is a difficult to control **epilepsy**, typically with onset in early childhood and associated with **intellectual disability** and **behaviour problems** which may be progressive. Sometimes the behavioural problems may present before the epilepsy. Seizures do not typically respond to treatment and there is currently no recommended treatment for r(20) syndrome.

We don't know how common r(20) syndrome is, though it is perceived to be underdiagnosed or misdiagnosed in some people with epilepsy.

There are two distinct forms of r(20) syndrome: non-mosaic where the ring is found in every cell in the body (the most severe form of the disease) and mosaic, where the ring is seen in only a percentage of the cells (this is the more common form of the disease).

Usually seizures begin in early childhood, typically around the time children start school, however symptoms may also present earlier in childhood, in adolescence or early adult life. Unlike many chromosomal disorders people with r(20) syndrome have a normal appearance and do not have intellectual disability before the epilepsy onset. There is usually a sudden onset of seizures without any clear trigger. Many people experience a change in behaviour before or around the seizure onset, manifested by poor attention and concentration, impulsivity and other behavioural problems.

Nocturnal focal seizures are often the first seizure type (sometimes mistaken as **night terrors**) and people describe frightening hallucinations or have a scared facial appearance. Prolonged periods of confusion (**non-convulsive status epilepticus** or **NCSE**) are commonly reported especially as the child gets older and these may last for minutes, hours (or rarely days). Other seizure types experienced might include tonic clonic seizures (either progressing from a focal seizure or alone), atonic seizures (drop attacks) and myoclonic seizures (jerks).

People with r(20) syndrome can go through periods where they have multiple very difficult to control seizures on a daily basis and this may be associated with significant intellectual and behavioural decline. These periods tend to be more of a problem in the first few years after presentation. Later in the course of epilepsy the seizures may become less frequent and may change over time with a more predictable pattern specific for that individual. Complete seizure freedom for prolonged periods (years) appears to be exceptional with most people having seizures in adult life.

Until the onset of seizures childhood development appears to be normal, however there often follows a rapid decline in intellectual function. Skills previously attained may be lost. This decline in ability varies from losing some skills but remaining in the normal range for intellectual function, to a significant loss in skills including difficulties with mobility and loss of speech and language functions. When this decline is associated with epilepsy it is called an **epileptic encephalopathy**. When very severe, r(20) syndrome may manifest in some people as a form of **childhood dementia**.

Support is likely to be required at school/college or even in the workplace. Behavioural issues such as bouts of aggression before and/or after a seizure are commonly reported. Individuals may also receive a diagnosis of **autism**. Predicting how severe these learning and behaviour problems will be in the future is not possible at the onset of the epilepsy.

Individuals with r(20) syndrome can benefit from:

- Safety advice especially re the 'confusional state' associated with episodes of NCSE
- A personalised rescue medication plan for prolonged or cluster seizures
- The side effects of medication particularly when changing treatment
- Genetic counselling
- Liaison with school or college for support during education
- Patient, carer & employer support requirements including neuropsychological evaluation, guidance, potential psychiatric support
- Sudden Unexpected Death in Epilepsy (SUDEP) risk management



Living with r(20) is neither simple nor easy. But here are a couple of boardgames to show you in a FUN way.

Snakes & Ladders Medication & Treatments

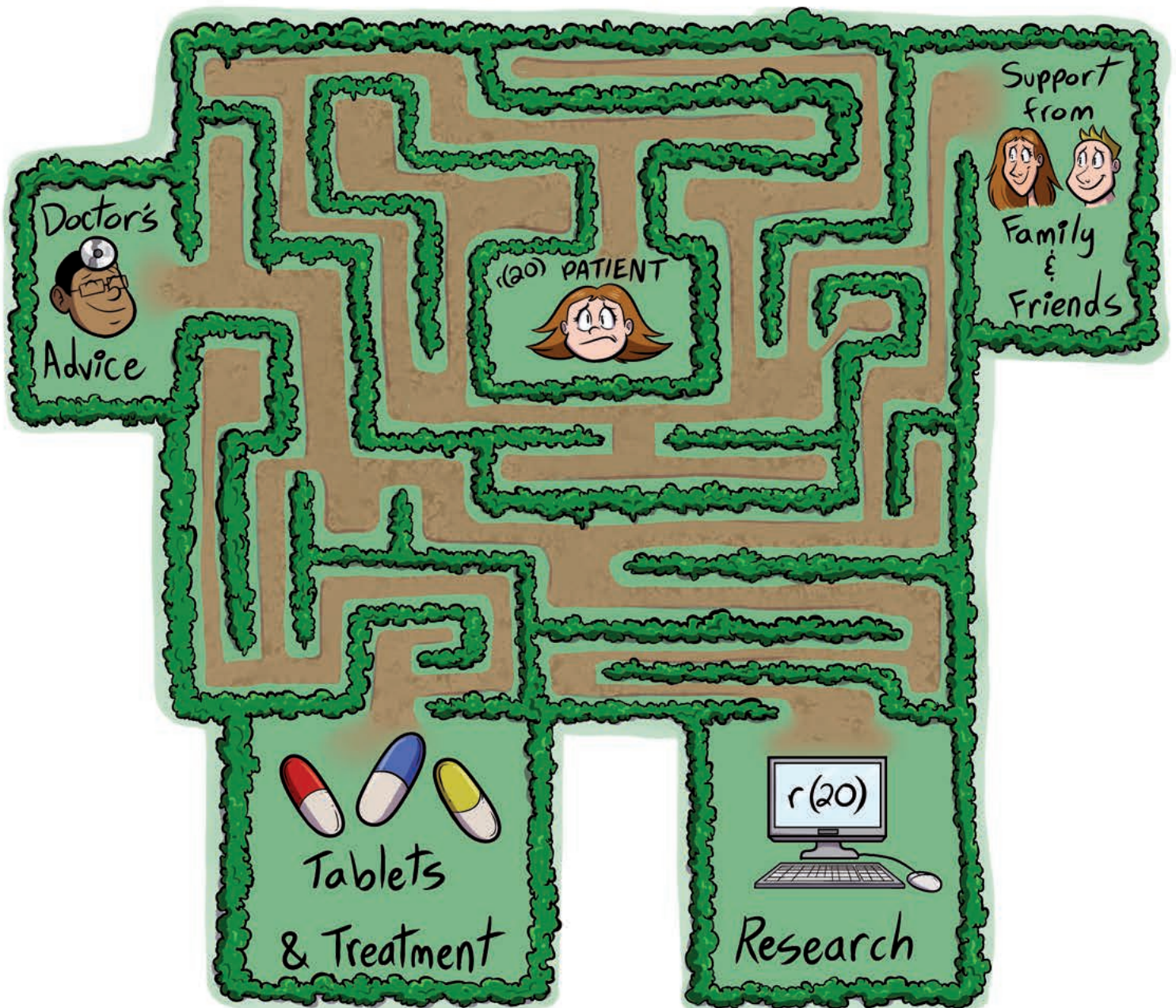
(A boardgame designed by a r(20) family about how nothing seems to work first time.)

Use 1 dice and play with up to 5 players.

Help the r(20) patient out of the



SPOILER: There is no way out. This is another puzzle designed by r(20) patients. Trying to live with r(20) is like navigating through a maze that you never really find a way out of.



However: within the maze, you find things that help you along the way and make that maze liveable.

See if you can find ALL 4 SUPPORTS.

NIGHT TERRORS

A night terror is a partial waking from sleep with behaviours such as screaming, kicking, panic, sleep walking, thrashing, or mumbling. They are harmless and each episode will end in deep sleep. However, in people with r(20) syndrome their night time seizures are often mistaken for night terrors causing a delay in diagnosis.

SEIZURE

A seizure is a sudden, uncontrolled electrical disturbance in the brain. It can cause changes in your behaviour, movements or feelings, and in levels of consciousness.

BLOOD TEST, EEG AND MRI

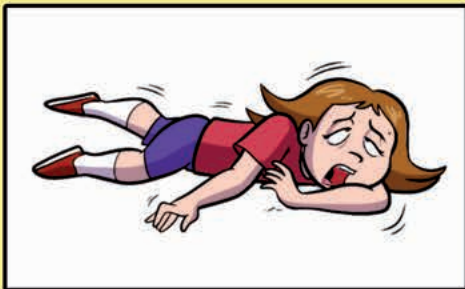
Doctors may perform tests to confirm if the person has epilepsy. Typical tests include an electroencephalogram (EEG) where you have wires glued to your head to read the electrical activity in your brain, a Magnetic Resonance Imaging (MRI) scan that uses strong magnetic fields and radio waves to produce detailed images of the inside of the brain and blood tests to check for genetic causes for the epilepsy.

EPILEPSY

If you have two or more seizures or a tendency to have recurrent seizures, you have epilepsy.

APPEARING VACANT OR DAYDREAMING

An atypical absence seizure or focal seizure with impaired awareness seizure may appear as if the person with r(20) syndrome is looking blank or not paying attention - as if they are daydreaming. However this is not the case and the person's brain is misfiring electrical signals such that they are unable to process information properly during this time.



FALLING ON THE FLOOR AND SHAKING

If someone is having a tonic clonic seizure their muscles stiffen and they may lose consciousness (the tonic phase) and then they may jerk or shake convulsively (the clonic phase).

KEEP A DIARY

It is a good idea for people with recurrent seizures to keep a record of their seizures in a diary. There are many apps available to help with this so you can keep a record on your mobile phone for example.

CALL AN AMBULANCE IF THE SEIZURE LASTS 5 MINS

Convulsive seizures or clusters of seizures lasting 5 mins or more are considered a medical emergency and you should call for an ambulance. Your epilepsy nurse should advise you of this when writing your Care Plan.



CARE PLAN

An epilepsy care plan should be drawn up with your medical team. It details your seizures and how you wish and need to be cared for when you have a seizure - and what to do in an emergency situation including the administration of any emergency medication you have been prescribed.

EDUCATION SUPPORT PLAN

A person with epilepsy who requires extra help at school or college with their learning and/or their seizures should have a support plan in place, detailing their learning, health and support needs in an educational environment.

ANGRY OR BADLY BEHAVED

Anger, aggression and poor behaviour may be exhibited by people with r(20) syndrome often before or around seizure onset.

SUDDEN UNEXPECTED DEATH IN EPILEPSY (SUDEP)

When someone is believed to have died during or after a seizure where no other cause of death can be found.



EPILEPTIC ENCEPHALOPATHY

Epileptic encephalopathy is when the epileptic activity and seizures contribute to more severe cognitive and behavioural impairments than would be expected from the underlying cause alone.

CHILDHOOD DEMENTIA

The loss of cognitive functioning—the ability to think, remember, problem solve or reason—to such an extent that it interferes with a person’s daily life and activities.

NON-CONVULSIVE STATUS EPILEPTICUS OR NCSE

A state of ongoing seizure or intermittent seizure activity with minimal or no motor movements and alteration of consciousness, often more simply described as ‘prolonged periods of confusion’.

TONIC CLONIC SEIZURE

The type of seizure most people recognise (previously called ‘grand mal’ seizures), where the person loses consciousness, their limbs become stiff, followed by jerks or shakes. Medical help must be sought for Tonic clonic seizures lasting 5 mins or more.

AUTISM

A developmental disorder of variable severity that is characterized by difficulty in social interaction and communication and by restricted or repetitive patterns of thought and behaviour.



Ring20 provide this comic as a FREE patient information resource to families receiving a diagnosis for the first time and those seeking insight into what to expect for their future with r(20) and how to prepare to meet their needs. If you would like to make a donation to help us continue our work supporting families please follow this link: <https://bit.ly/ring20donate>

MAKE A DONATION!



We would like to thank our r(20) families who attended our Birmingham workshop in 2019 for creating the content for this comic and our r(20) Youth Project participants who further developed those original ideas into the storyline you read here today. Special thanks go to Georgina Clunas (creator of Otsi), David Watson, Nicholas and Milan Colyer and Danielle Fenner for their valuable contributions and time devoted to this project. And of course, thank you to Rossie and the team at Dekko Comics for guiding us through the process and producing a wonderful, educational and fun comic!

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