

# GROWING UP

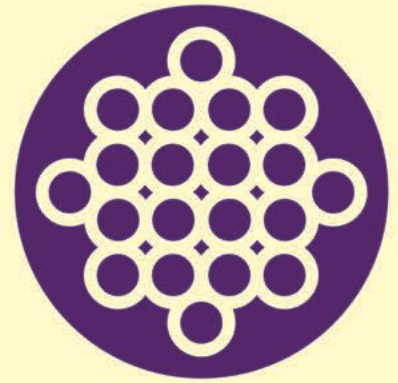
WITH

# r(20) SYNDROME



# 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).

Ring20 Research and Support UK CIO,  
26 Headley Chase,  
Warley Brentwood,  
Essex,  
CM14 5BN,  
England,

Registered Charity no.: 1165651 (England and Wales), SC048893 (Scotland)



+44 (0)7385 292797



[www.ring20researchsupport.co.uk](http://www.ring20researchsupport.co.uk)



[ring20@ring20researchsupport.co.uk](mailto:ring20@ring20researchsupport.co.uk)



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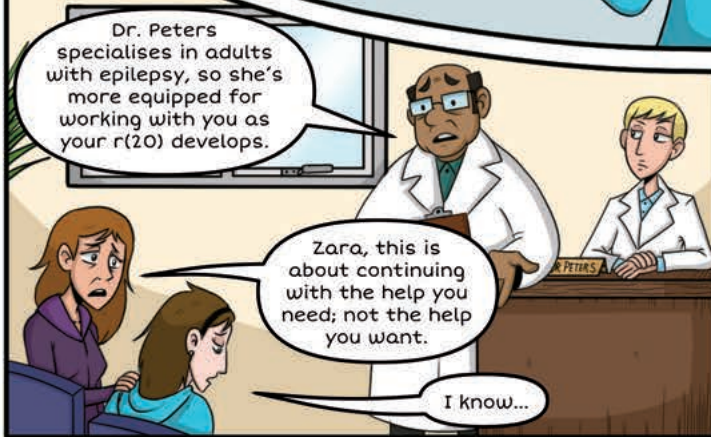
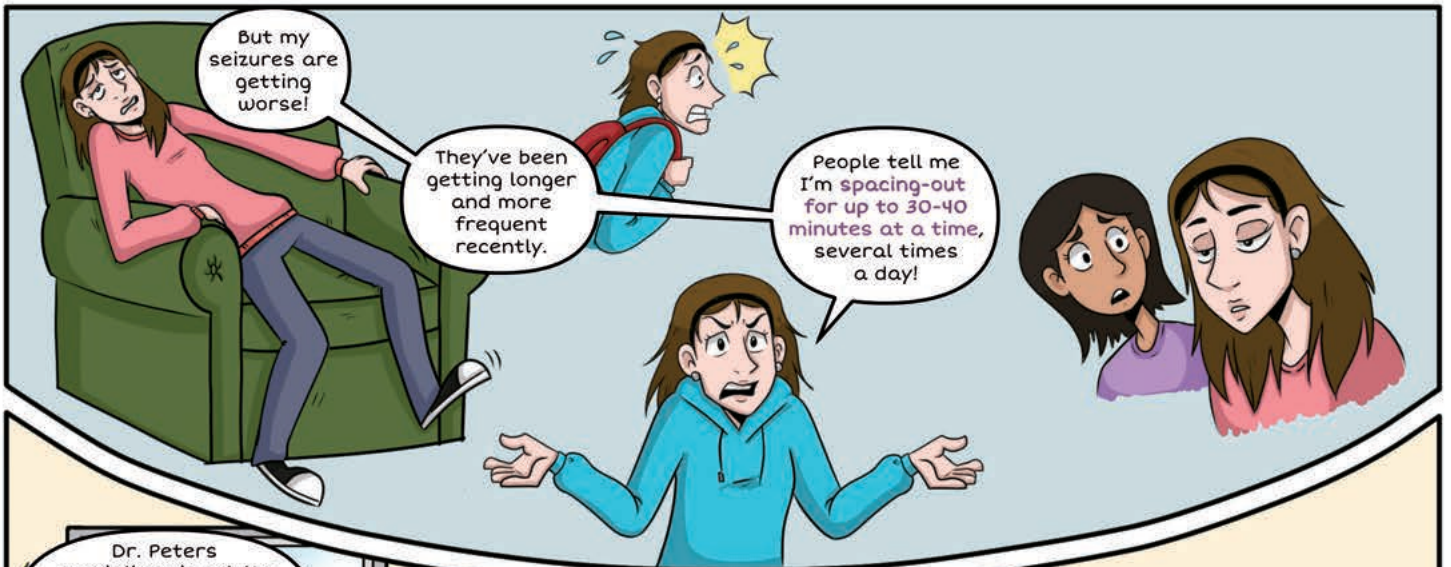
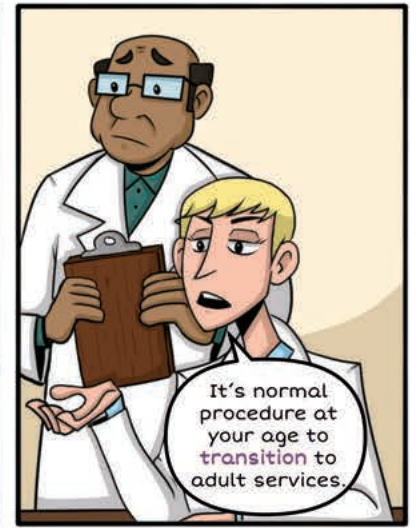
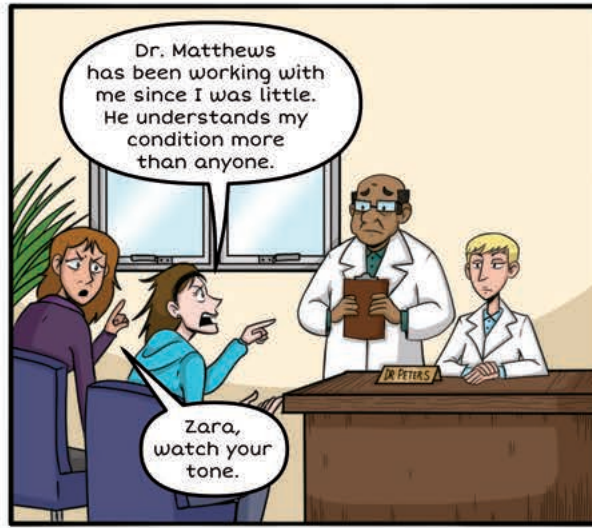
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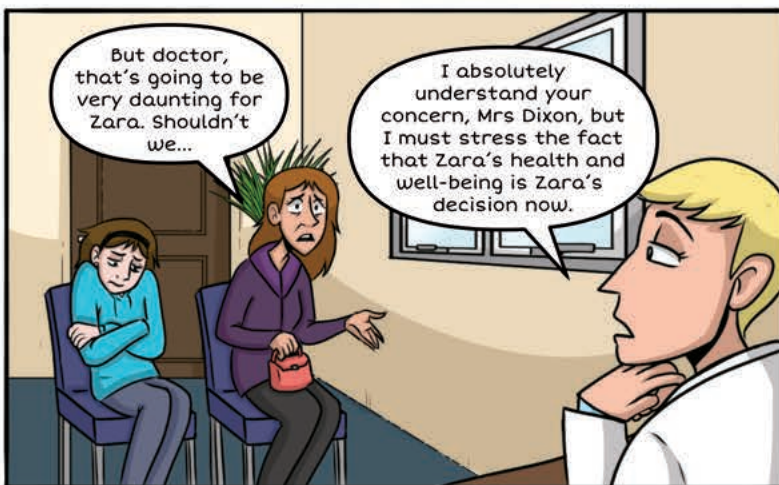
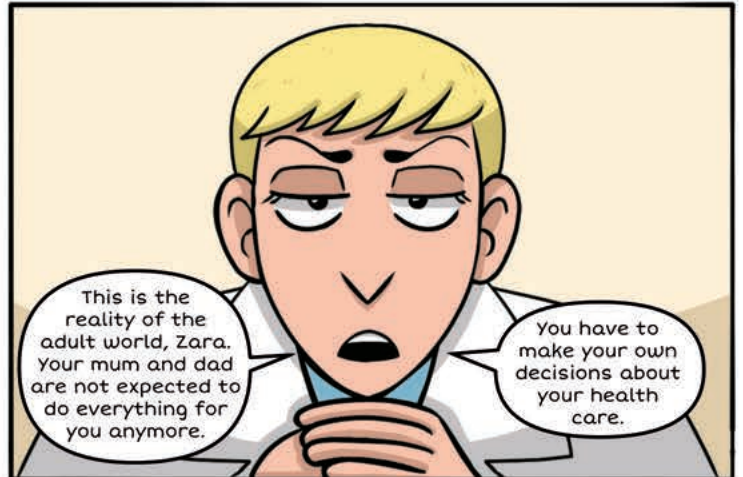
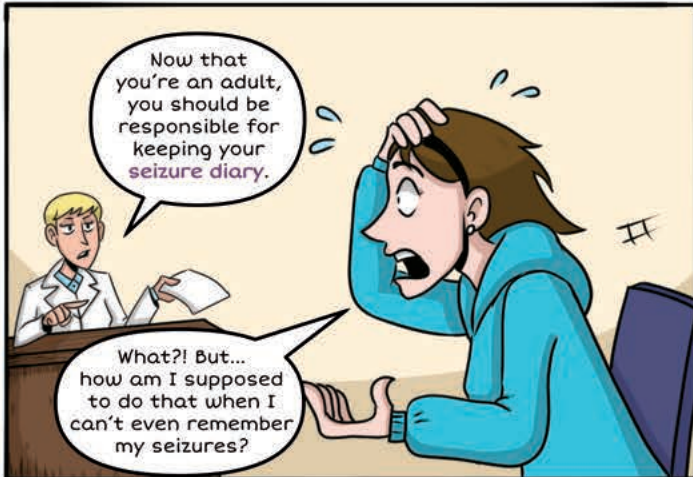


<https://bit.ly/ring20YouTube>

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 follow her progression into adulthood. If you haven't already, we recommend you also read: Life with r(20) syndrome Zara's Story: The Younger Years.







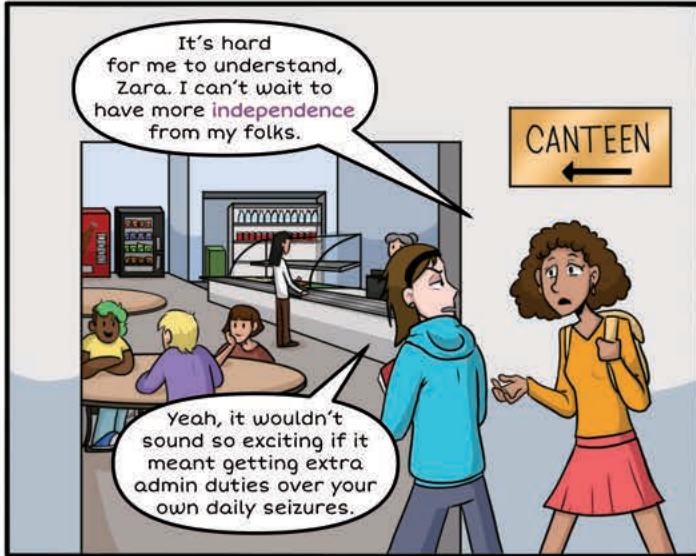
Growing up... sucks.



r(20) sucks.



My whole life right now just... sucks!



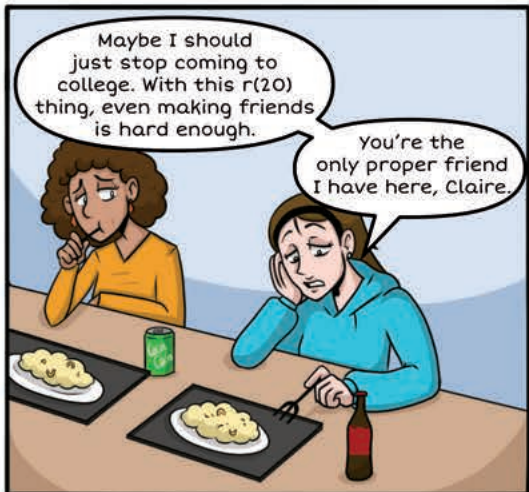
It's hard for me to understand, Zara. I can't wait to have more independence from my folks.

Yeah, it wouldn't sound so exciting if it meant getting extra admin duties over your own daily seizures.



AND my studies are suffering because of them.

There's no way I'm making it through college this year.



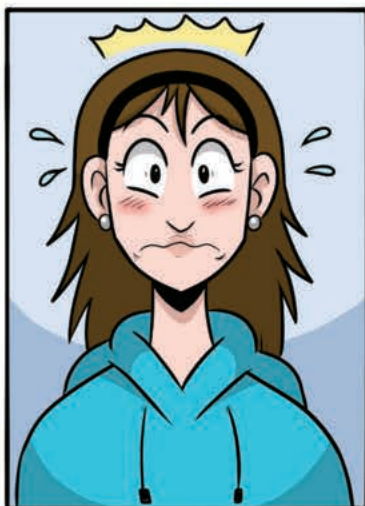
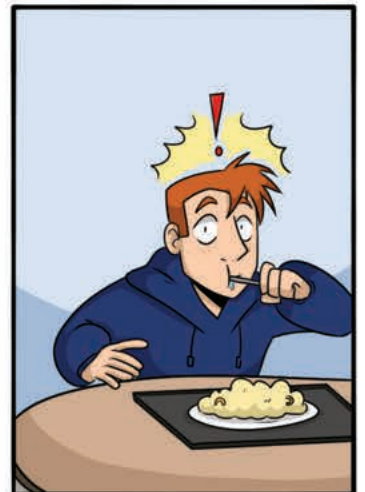
Maybe I should just stop coming to college. With this r(20) thing, even making friends is hard enough.

You're the only proper friend I have here, Claire.



Don't blame college, girl. You don't give people a chance.

Example: It's common knowledge that Ryan probably likes you. A LOT.



You think you're so funny, Claire. But you're fooling yourself.

Hey, I'm just saying. I've seen the way he looks at you.

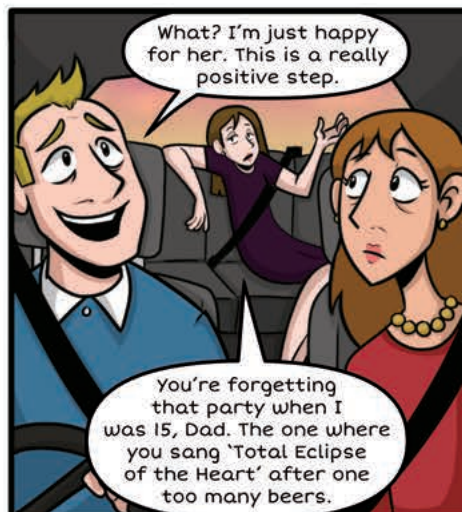
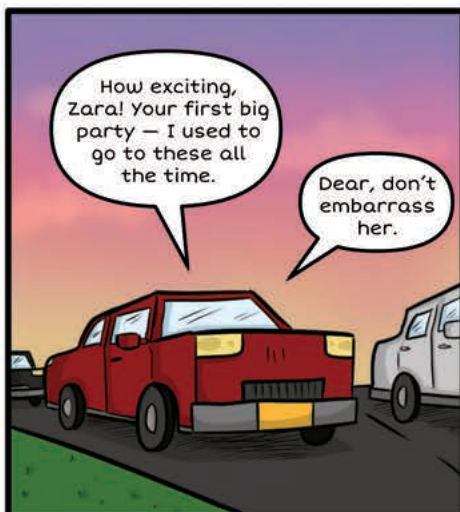
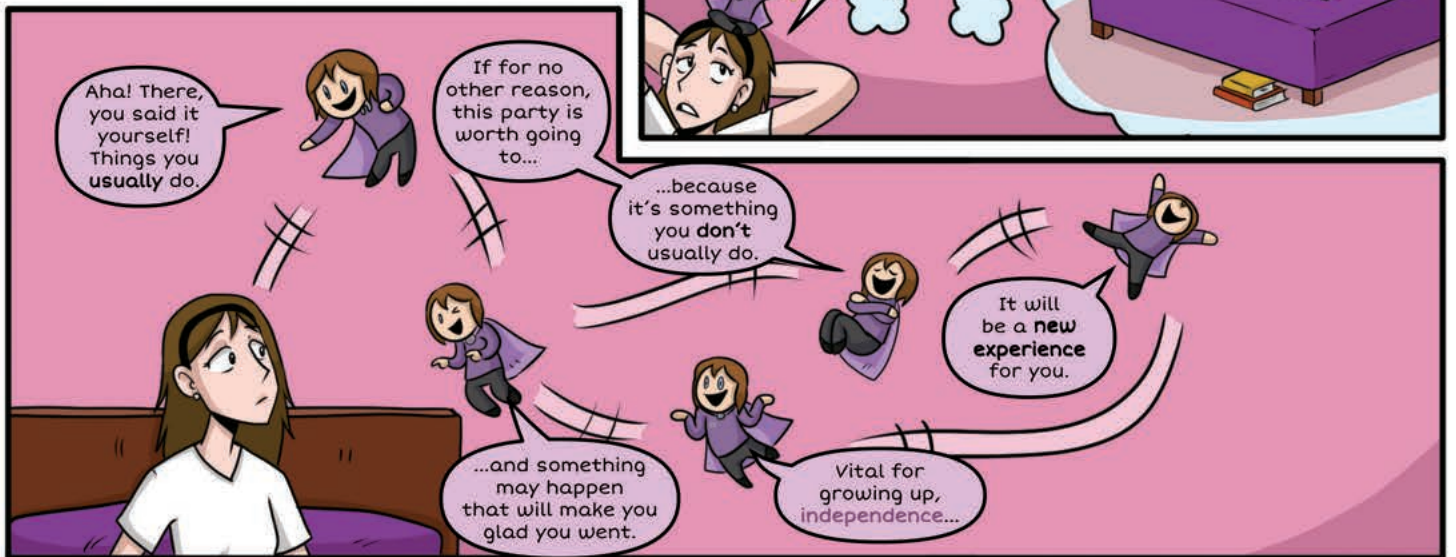
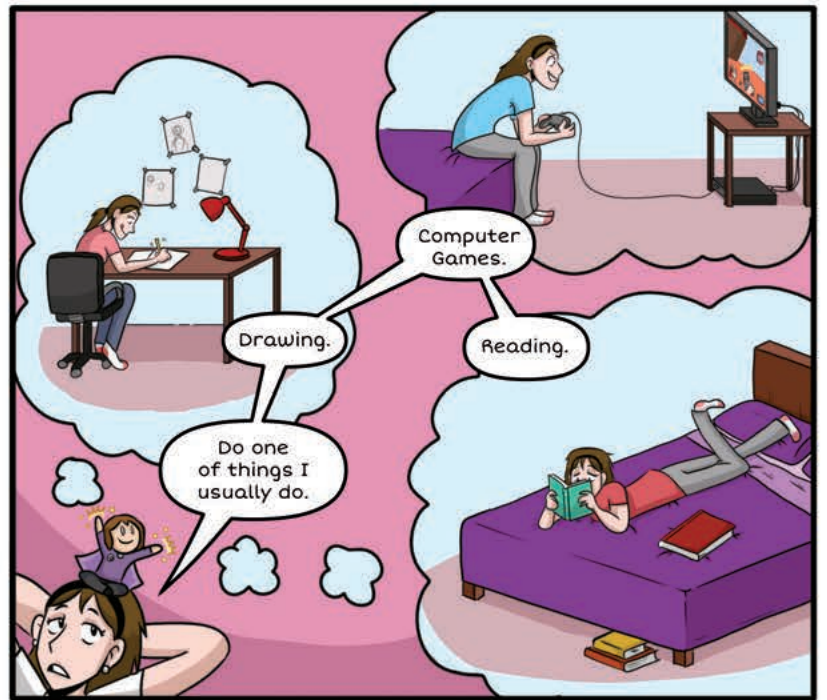


Well you can tell him to stop wasting his time. I'm not interested.

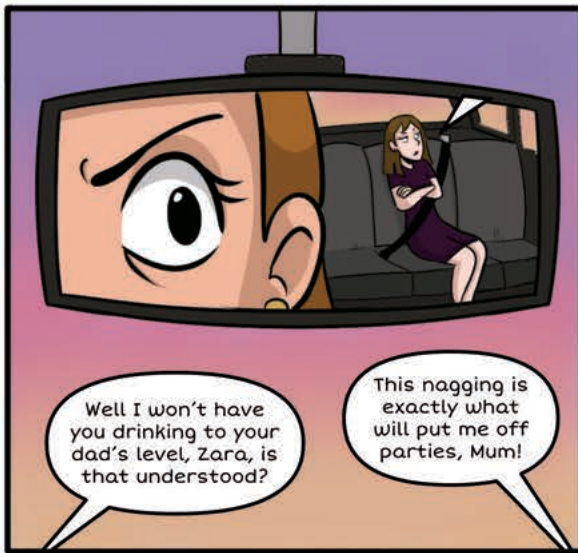
Besides, if I ever did meet a boy I liked, my awkwardness would single-handedly kill any chemistry between us.











Well I won't have you drinking to your dad's level, Zara, is that understood?

This nagging is exactly what will put me off parties, Mum!



Mum did have a point. Alcohol has a much faster effect on me because of my meds.



But once I was at the party, it seemed to be a lot better than I worried about, and I was keen to make a good impression.

I felt good. To begin with...

Said without a care. I've got to consult a genetic counsellor before even considering the idea in case I pass on my r(20) genes to my child.

My boyfriend was so funny last night!



He did that again? God, my Andy is just the worst for that.

Relationships? Sex? Urrgh! When am I going to feel ready for those sorts of things.



Things are going so well between Gavin and I. Think we may even have children together.



Would you do those drugs if you were offered? Andy says they're pretty good, apparently.

Pff, yeah right. And risk having a seizure in public?



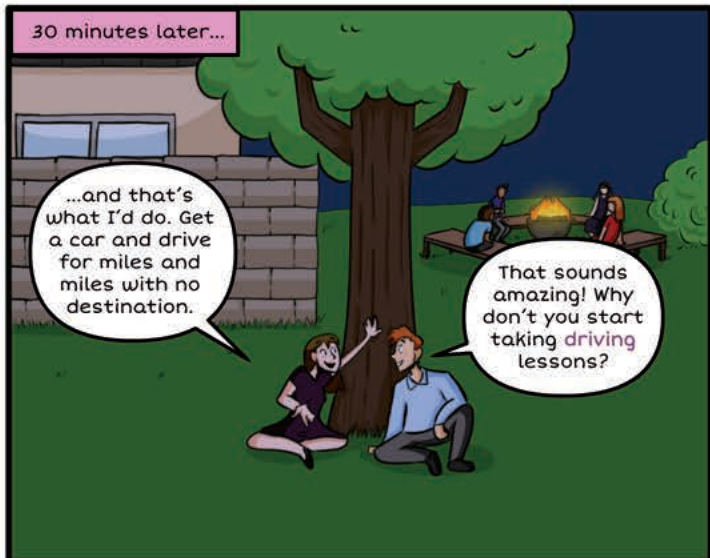
I dunno. What do you think, Zara?

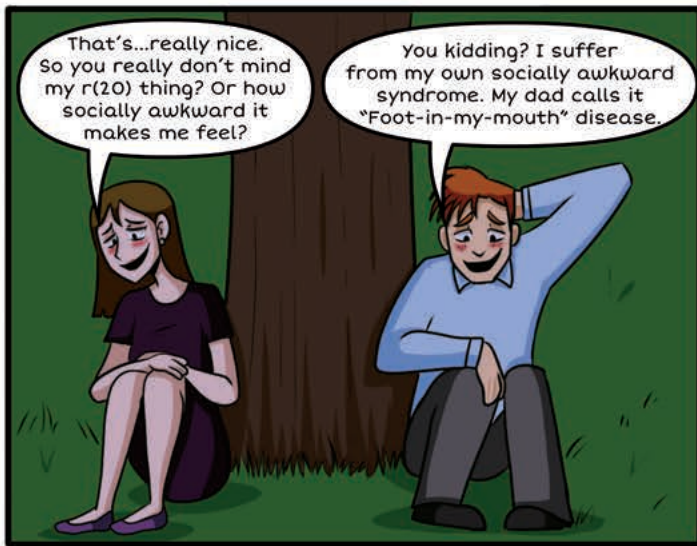
Huh?!



Say something funny and clever, Zara! ANYTHING!

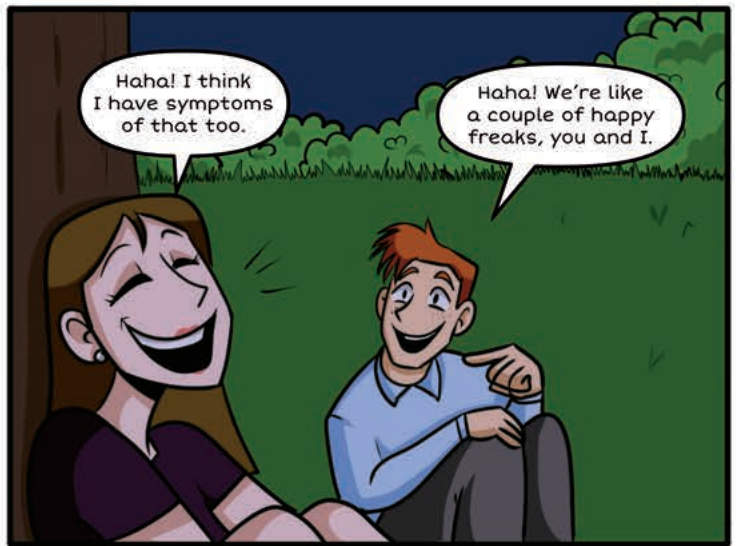
Oh, heh-heh... Not me. I think I'm on enough drugs already, eh?





That's...really nice. So you really don't mind my r(20) thing? Or how socially awkward it makes me feel?

You kidding? I suffer from my own socially awkward syndrome. My dad calls it "Foot-in-my-mouth" disease.



Haha! I think I have symptoms of that too.

Haha! We're like a couple of happy freaks, you and I.



Wh-WHAT did you just say? My r(20) makes me a freak, does it?!

Huh?? I only meant that as in we're both like social freaks together...

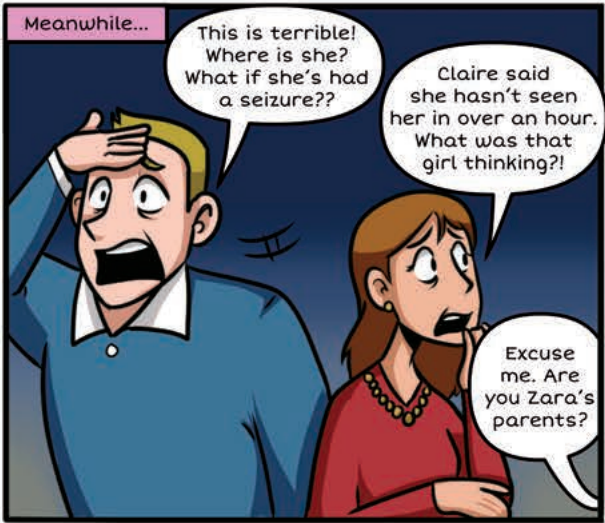


You...I never want to...Just go and find another "freak" to be awkward to!

Zara, wait! I didn't mean it like that...I was trying to be funny...!



Foot-in-my-DAMN-mouth disease!



Meanwhile...

This is terrible! Where is she? What if she's had a seizure??

Claire said she hasn't seen her in over an hour. What was that girl thinking?!

Excuse me. Are you Zara's parents?



And there you have it. What I feared would happen at this party, happened.

I hate this. I just hate it all. I hate boys, I hate parties, I hate r(20)!



Oh no...\*Huff\*...Not now. \*Huff\*...Please not now....

But just before they could start to get any worse...

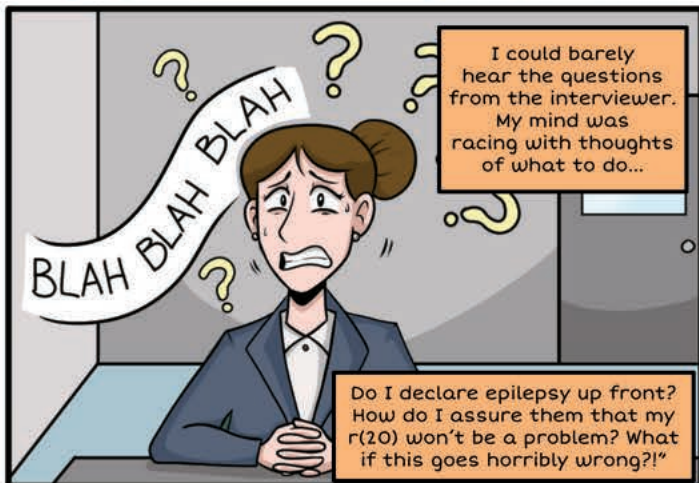
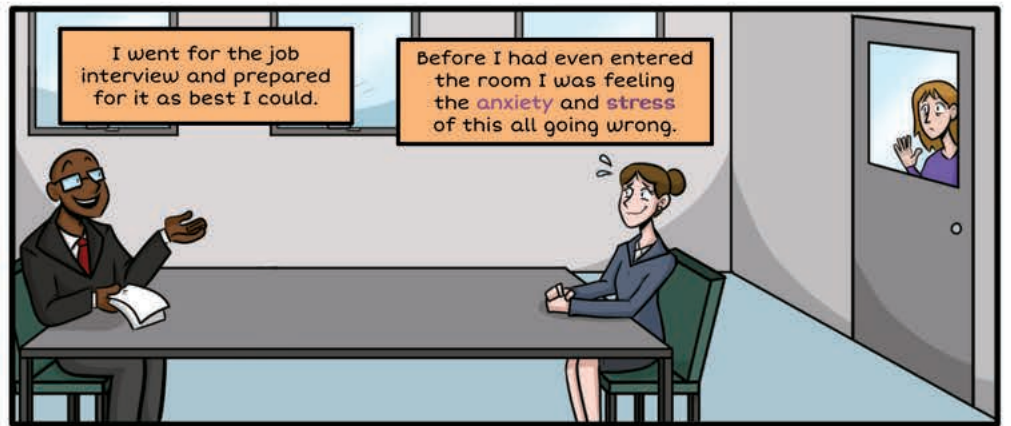
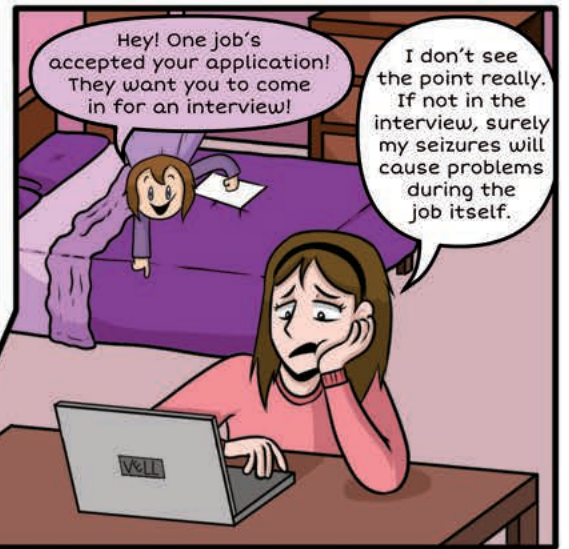
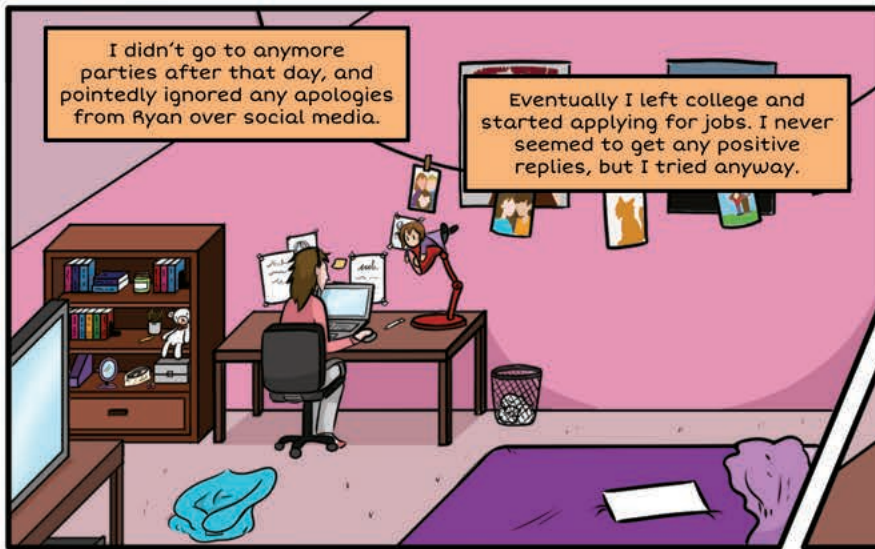


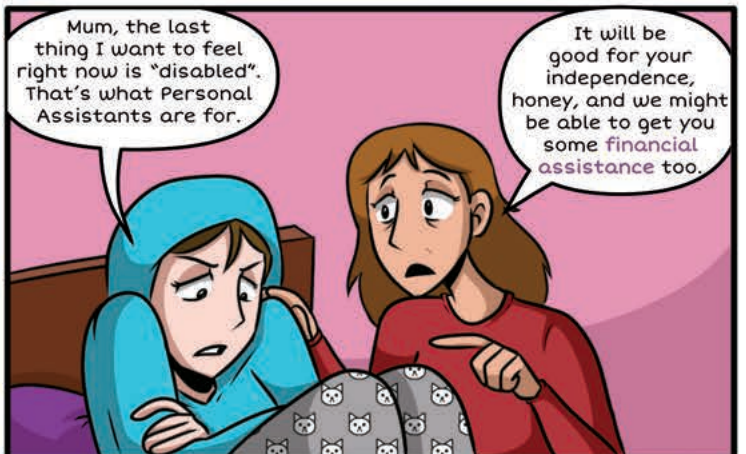
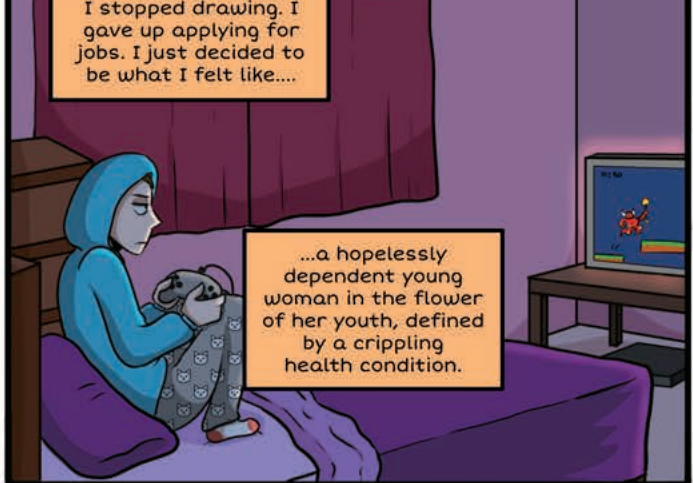
My parents came to take me home.

There she is, Mr & Mrs Dixon. I'm sorry I lost her.

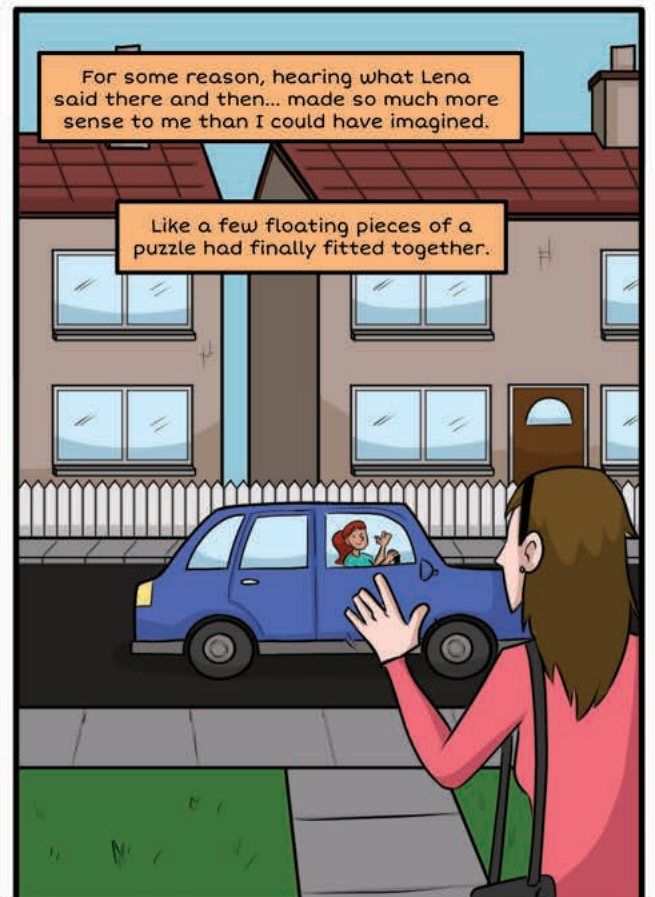
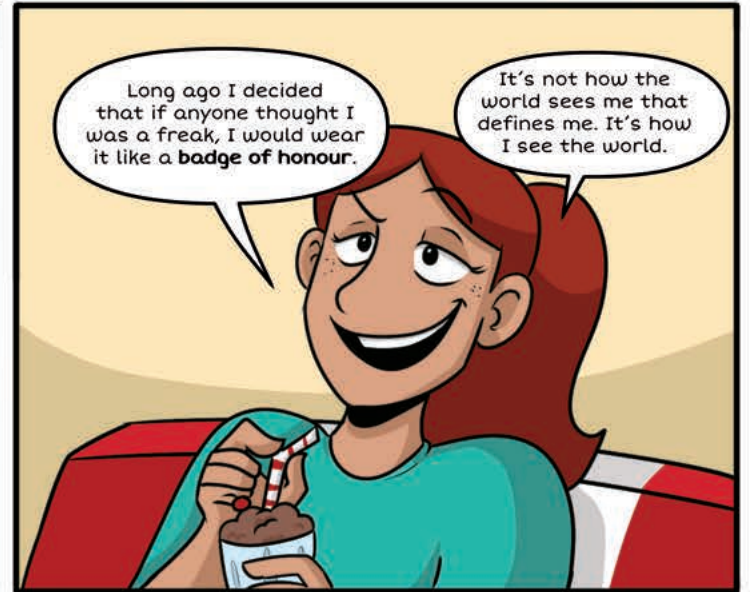
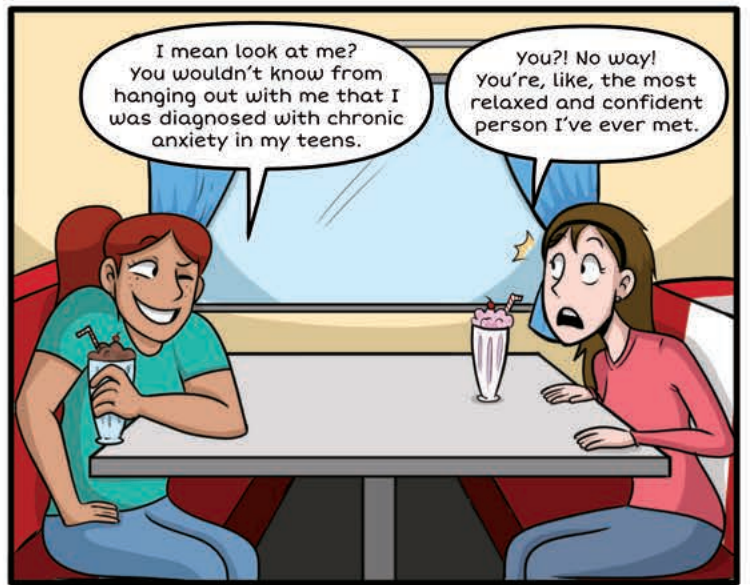
Zara! Are you okay?

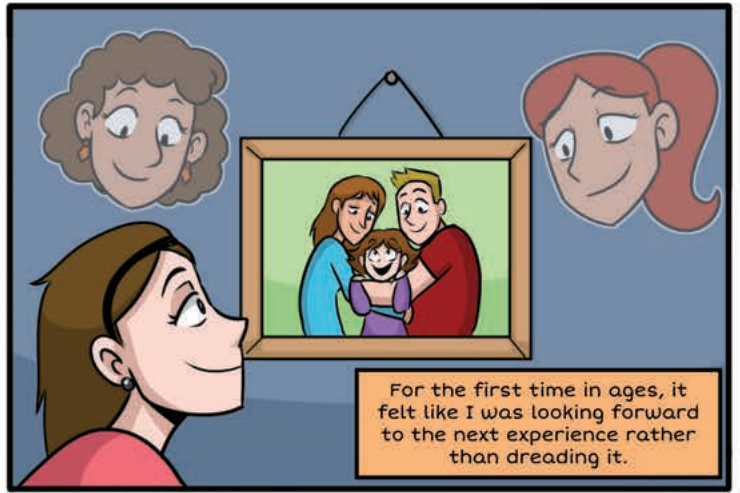
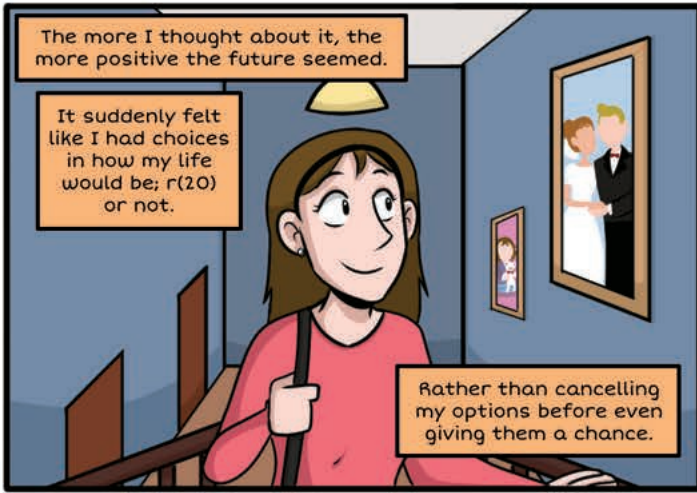
This nice boy helped us find you.













# 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

## TRANSITION

Typically between the age of 16 -20 years, a young person with r(20) syndrome will be transferred from children's medical services into adult services, where their epilepsy will be managed by an adult neurologist. The transfer of records and care may take place over a short period of time when the young person sees both their old and new doctor for a few appointments. This period is referred to as transition.

## SPACING OUT FOR 30-40 MINS

People with r(20) syndrome may display seizures where they appear vacant or confused, unable to verbally respond properly and these episodes can last for many minutes, sometimes hours (and rarely, days). Unlike absence seizures, which typically last a few seconds, where these prolonged periods of confusion do not abate and the person's consciousness is impaired, they may be described as non-convulsive status epilepticus (NCSE).

## SEIZURE DIARY

As the person with r(20) syndrome gets older, their medical team may ask them to keep their own seizure diary. For some people this may be difficult for them e.g. where they are unaware of their seizures themselves and/or where they are unable to track their own seizures. It is still important to maintain a record of seizures to see if they change over time or with any changes in treatment, so if the young person is unable to do this themselves then it is OK to ask for other adults/carers to help.

## SUDEP

When someone is believed to have died during or after a seizure where no other cause of death can be found. In young adults, who experience uncontrolled tonic clonic seizures at night their risk of SUDEP is increased and therefore it is recommended that appropriate precautions are taken to minimise this risk e.g. continuing to take medications regularly and on time and having a seizure monitoring device or alarm to alert someone if you have a seizure so they can time your seizure and call for additional help/ambulance if necessary.

## SEIZURE ALARM

For any person with uncontrolled epilepsy a seizure alarm can be useful to alert caregivers in the event of a seizure, so they can come to your aid. Seizure monitoring devices come in all different forms, there are alarms that can be placed under a sheet on the bed to alert if someone has a tonic clonic seizure at night, video cameras/sound projecting systems similar to a baby monitoring device as well as wrist based devices that can alert when someone has a motor seizure during the day or night-time. Your medical team or epilepsy nurse may be able to provide some helpful advice on whether you could benefit from an alarm and what type of device might suit you.

## INDEPENDENCE

As people with r(20) syndrome grow into adults they may wish to seek independence just like any other person of their age. This may mean they will want to travel, work and/or socialise without their parents, seek their own accommodation, go on to form relationships and have a family of their own. Every person's situation is different, so speak with your parents/carers, medical team and social worker to achieve the right balance for what is safe and appropriate for your needs.

## ALCOHOL

For any person taking anti-seizure medication, it is important to understand the effects that alcohol can have on you and your seizures. It is therefore important to read further information from your local epilepsy organisation on the impact of drinking alcohol, so you are informed of the risks and what you can do to minimise them.

## SEX AND RELATIONSHIPS

Just because you have seizures doesn't mean you won't enter into relationships, but you may worry about how a partner might view your epilepsy - when do you tell someone you have r(20)? Finding a partner that understands your condition can really help. And when you are ready for a sexual relationship ensure you take appropriate precautions and discuss this with your GP as some contraceptive pills can interact with your anti-seizure medication. For females of child bearing age prescribed Sodium Valproate, new guidance has been issued and your medical team should talk to you about this.

## GENETIC/GENOMIC COUNSELLOR

Genetic counsellors (or Genomic Counsellors as they may be referred to) work directly with patients and families offering genetic/genomic information and support allowing them to make health decisions. When you receive your r(20) diagnosis you should be referred to a genetic counsellor for information on the condition and what this means to you. Later in life, if and when you would like to start a family of your own, again you should seek support from a genetic counsellor as there are (a few) cases where r(20) syndrome has been passed on from parent to child.

## DRUGS

Similar to alcohol, taking illegal drugs can impact on your seizures and overall health. Ensure you stay informed of the risks if you are offered illegal drugs such as cannabis, ecstasy, speed, cocaine and other recreational drugs which can all increase the chance of having a seizure.

## DRIVING

If you continue to experience seizures you will not be able to drive. In many countries you will need to prove that you have been seizure free for 2 years before applying or reapplying for a driving licence. However there may be support for help with the costs of travelling on public transport, so do check these out to see if you are eligible.

## TRIGGERS - STRESS AND ANXIETY

Seizure triggers can be many and varied. In teen and adult years anxiety or stress may be triggers not experienced (or as prominent) in childhood, so it is best to try to find ways to combat these and avoid stressful situations if you can as you get older. Tiredness, lack of sleep or not taking anti-seizure medications regularly each day can all trigger seizures. Other triggers commonly reported by r(20) families, which may be not so common in the general epilepsy population, include taking bath/showers, significant changes in temperature (going from a hot room to cold outside or vice versa) and exercising.

## PERSONAL ASSISTANT

Adults with a complex disability such as r(20) syndrome may be eligible for funding someone to help them access the community safely and/or work with them on life skills and increase independence – sometimes referred to as for a Personal Assistant. Speak to your adult social care team about whether you are eligible to apply.

## FINANCIAL ASSISTANCE

Your symptoms may make it difficult to do all the day-to-day tasks that someone without r(20) may perform unaided. If this is the case, you may be able to apply for financial assistance to help you pay for the additional support you need. Usually you will undergo an assessment of your needs first, filling out complicated forms and potentially having to argue your case at tribunal if your request is initially refused, which can be very daunting. Ensure you/your family get help with this process – an independent local charitable body that helps disabled people claim benefits may be best placed to support you and these services are often available free of charge.



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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