Issue 3

December 2015

Newsletter

ring 20 research & support uk

Its official—we're getting registered!

Special points of interest:

- See what our volunteers have to say in this issue (on our volunteers and Events pages)

AGM

In July we held our first AGM. It was a shame that no family members were able to join us, but the key focus of the meeting for the Trustees was preparing our registration application for the Charities Commission. There was a lot of paperwork to be completed and thanks to a lot of hard work from Chris, we ploughed our way through this over the summer. We will be changing our legal status, to officially become **Ring20 Research and Support UK CIO**—this name change doesn't affect yourselves, but becoming a CIO offers the Trustees greater protection, going forward. So in due course we hope to receive a registered charity number that can be quoted on all correspondence—and importantly will make applications for funding/grants much easier and open more opportunities for us in the future as a recognized charitable organization.

Families Informal Meet

In the summer, a few r(20) families decided to meet up at Stratford –upon –Avon. Luckily we had good weather and picnicked by the river, chatting whilst the youngsters played ball games. It was lovely to meet up with each other and share stories and just 'hang-out'. We hope to do this again and maybe a few more of you may be able to join us?

A few words of thanks...

I do hope many of you have a strong support network; living with r(20) isn't always easy. I would like to take a moment to thank many of my family and friends who continue to support me and my charity work donating their time, skills and hard-earned cash—I couldn't have achieved as much this year without them. It is encouraging that we have new families taking up the fundraising baton and I hope more of you will be inspired by their efforts and take up the challenge yourselves in 2016?

May I take this opportunity to thank you all for your ongoing support and wish you all a very Merry Christmas and Happy New Year!

Yours

Allison Watson Co-Founder / Secretary

INSIDE THIS ISSUE:

Genetics Disorders Media Campaign

Did you see us in the Independent Newspaper on Friday 18th September, on the middle page spread of the Genetics Disorder supplement, as part of Jeans for Genes day?

The feature included one of our own member patient stories in the 'mums and dads panel'

But the Independent was only the start—we also now have our own page in the Genetics section with more information here:

http://www.healthawareness.co.uk/genetics/stem-cell-research-brings-insight-into-chromosome-disorder?utm_source=propeller

This page features

- An interview with Nancy Spinner on her ongoing genetic research
- 2 patient case studies
- Rachel's story
- Diana's story
- A video on r(20) - as kindly provided by the Italian Collaborative Research Group
- Branding, contact info and links to our website

Our Volunteers

Following our call for volunteers in the last issue, we have been privileged that 2 of our family members have stepped forward. We really value their contribution, which is vital if we are to grow as a charity. If you think you could help too, we are continually looking for more volunteers, so do get in touch...

Here's what they have to say:

Tamzin

My son Kieran was diagnosed with Ring20 7 years ago. Whilst supporting him with the control of his seizures and trying to help him work around his learning difficulties, I still felt quite helpless in the face of his diagnosis. With such a rare condition, who would be working on a treatment and what would the future hold for my son?
When I read in one of the Ring20 Research and Support's newsletters that volunteers would be welcome, it got me thinking. Maybe I could help in this small way by raising awareness and hopefully help gain some attention for this syndrome. So, I contacted Allison (one of the trustees) explaining that I would like to try looking after the social media for the charity. I am really glad I did as Allison was very supportive and I have so far really enjoyed posting regularly on Facebook am currently working on an awareness/fund raising day at Kieran's school which is really exciting.

Dale

Hi, my name's Dale Ward, I have had R20 since I was 8 years old and am now 22 years old and living an independent life. At age 8 I had my first seizure and despite having rubbish memory I remember it like it was yesterday, I was watching my Dad play football over the park with my cousin, when I started tugging my shirt and phasing out. My cousin was calling my name with no response and after the football game had finished he told my Dad what had happened. My Dad asked me about it, but we thought nothing of it and moved on. We went home and my Dad told my Mum what happened, but I seemed fine. Later that night I came downstairs with Lucy and Adam (my cousins) and I was tugging my shirt again, my Mum called my name but I didn't answer after three or four times of calling my name with no response my Mum started to panic and called the ambulance, later that night I was diagnosed with epilepsy. As time went on I was on full fat milkshakes—I was that close to being classed anorexic. I was told to fry everything in the chip pan and eat all fatty foods, I was in this state for about 5-6 years from about 13 years old to about 19 years but I gradually got better and now I'm starting the gym. Also as a teenager I had no social life whatsoever, only from school to playing out on the street but later on playing on the street wasn't an option. I had no life outside of school, I was stuck inside just playing on my game all day, until I started going to my local football team around the park and watching them every Saturday, then going with them to have a drink with them over the pub (even if it's a just a coke). It was something to look forward to and my Mum knew I was safe with them as I had family playing for them as well, cousins, my uncle, stepdad and close friends. Going with them also gave me a drilling down on what life is about. What I love most about this club is that they treated me as one of their own and that feeling of acceptance was the best feeling ever, now still to this day I watch Marshfield play 'week in week out' . I started to put the weight back on and started to feel healthier.

Eventually one day my seizures where hitting a bad patch so we asked for help and went to a social worker and that's when they recommended us to St. Helens House in Newport. It was a supported living accommodation where it is the first step of moving into your own place. I have a support worker present 24/7, there is a number of flats in the house in which there is number of residents with various disabilities. I have my own flat including bedroom, ensuite and a small kitchen with no oven. The oven is in the big kitchen which is open for all to use, along with the lounge and dinner table. Now I want to become a support worker myself and do for someone what my family, friends and support workers have done for me, while doing my bit to help raise funds and awareness of r(20). Going to various conferences has helped me in learning more about the genetic side of the condition, I really enjoyed my time in London and also the conference in Cardiff to learn about more rare diseases to help me in achieving my goals and helping others as I am now in Health and Social Care level 2 and looking towards the future to have a successful career in care.

Having a pre-xmas cleanup? Sell unwanted items and raise funds for us on eBay here:
http://www.ebay.co.uk/egw/ebay-for-charity/charity-profile/?NP_ID=71688

Don't forget to sign-up to *easyfundraising* and earn funds for us on your online shopping! Join here:
http://www.easyfundraising.org.uk/causes/ring20researchsupport

Fundraising

We've had fantastic support from our members fundraising for us since the last issue. Here are some of the highlights:

Dale's Fundraiser in Cardiff £750

Dale organised a fundraising evening at his local pub, inviting lots of family friends and colleagues to come along and join in the fun. Thy held raffles, tombola & karaoke,.

Leanne's Epic Bike ride £682.33

Leanne Lewis is the sister of one of our members and decided to take on a sponsored bike ride in France this summer 60km bike ride in France, cycling from Orléans to Poitiers.

Half Marathon £814.13

Allison successfully completed the Royal Parks Half Marathon, London in 2 hrs 17 mins.

Basildon Car Parking £197.76

Allison and David plus some family and friends manned the Basildon council car park for the day.

Billericay SummerFest £80.21

Sue, David and Allison hosted a fundraising stall selling Sue's hand made cards, tombola, and Guess the name of the Giraffe.

Christmas Card/ Cake sale & Xmas raffle at Virgin Clearview ~£250

Held on 30th Nov to get us in the Christmas spirit!

Brewers Club £267.87 so far this year...

The Brewers club in Romford continue to support upon a monthly basis.

<u>Upcoming events</u>

Dale's college fundraiser... Dale has put our name forward for a bag pack with his college where he will be helping organising the fundraising. Good luck Dale!

Walton High School, Milton Keynes Whole school fundraiser and r(20) awareness day—

Find us on Facebook

https://www.facebook.com/Ring20ResearchandSupportUK

Please Like our Facebook page to keep up-to-date on all that's happening!

Grants and Sponsorship

Corporate Sponsorship

We have received a cheque for £75 from Cellular Solutions (Leanne's workplace) to help us with our vital work.

cellularsolutions

IFDS charities' committee have also donated an additional £100 in respect of Allison's half marathon.

Does your workplace offer any corporate sponsorship for charities such as ours?

Jeans for Genes Grant application

We have recently submitted a grant funding application to Jeans for Genes to host a families workshop. If we are successful , we will look to book a date for this event in 2017, probably in the North of England.

JEANS FOR GENES DAY GENETIC DISORDERS UK

Bringing together those affected by genetic disorders

Our next grant funding application will be for a families workshop in 2016, through BIG Lottery—Awards for All.

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Events

ESHG – European Society for Human Genetics Conference 2015

Don attended this conference in June—here's his report:

As I write this update for this particular conference it struck me how quickly time has passed over the past decade and my personal involvement with r(20) syndrome . I mention this only in context of the number of meetings, conferences, family visits, conversations and engagement with many professionals around the world over the years. Certainly as part of this new team and the new organisation (Research Support UK) I sense a new chapter has begun. There is no doubt in my mind that this particular opportunity was very important in helping to establish new connections with a significant group of professionals we have previously not engaged with.

I know from past experience how costly outreach to professionals can be and our successful application to the Big Lottery Fund to enable this outreach has been a terrific shot in the arm for Ring 20 Research Support UK. We received funding to attend two conferences of which this was the largest.

There are so many factors involved in attending these events – many hours of preparation producing updated literature, stand materials, transportation and that's before the event actually begins. This particular event was held across four days and hosted over 2,000 delegates from over 40 different countries. In fact, I recorded speaking with delegates from 26 countries from all four corners of the globe, not just Europe.

I could spend lots of column inches telling you about the many conversations I shared (I did share some on twitter and face book)but wanted to try and give you the highlights here.

One of the interesting features of events like this is you do have time to learn from other charities trying to do much the same thing and what works for them. That said, the main highlights were talking to geneticists, clinicians and counsellors who were testing for rare disorders some of whom were familiar with r (20) and of course those who were not. Interestingly I talked with one of the lead research team who was involved in a abstract research paper that documents 5 cases of r(20) a number of years ago from Holland – D Lindhout Professor of Medical Genetics. He suggested some different avenues for research moving forward which I have passed on to our medical advisor Sophia Varadkar. I also connected with the geneticist from Iceland who recently diagnosed the first known set of twins with r (20) who took our literature to pass on to the family and colleagues.

Dr Jean Charles Hoda from Switzerland is setting up a foundation to help identify rare diseases and he had never heard of r(20) so I am following up to make sure we are represented in that endeavour. Geneticists from Austria and Slovakia have suspected cases and look to do more testing to rule out r(20). A team from Oxford had not heard of r(20) and were pleased to take away more information (and enquire if we were available to speak at their institution) as were a good number of other individuals from the UK and abroad. All those professionals that completed our stand questionnaire are being followed up be me personally, which will help us to further improve levels of diagnosis and awareness.

ILAE Scientific Meeting—UK Chapter

Allison and Dale attended this event in September—read on for their report :

Highlights from the day included talking to:

- Epilepsy Society—possible epilepsy genetics research initiatives
- Epilepsy Research UK—following up on a potential research opportunity,
- Sudep Awareness—engaging in their annual celebration day
- Nutricia—possibilities of attending a future r20 families event for keto cooking demonstration
- Cyberonics— to come and speak to our families at a future event about VNS therapy.

We also met with a number of medical professionals who support some of our member families, of whom they spoke very highly. We spoke to a genetics test team from Denmark who'd never heard of r (20) - but we've changed that now!

There is clearly still work to do in educating neurologists and geneticists that the newer genetic testing techniques won't pick up ring chromosomes, so many more patients may be going undiagnosed even though there is a greater move towards genetic screening in epilepsy.

Events...(continued)

We obtained suggested leads to follow up in terms of various research groups and consortia who might be interested in connecting with our group. We've been invited to speak to a medical team in Bolton, an epilepsy specialist nurse group in Dorset and host a stand at The Royal Free hospital training day next year to raise awareness of r(20) as well as forging stronger relationships with Young Epilepsy and discussing ways in which we can work together.

All in all a very successful event for networking and raising awareness of r(20) syndrome.

Rare disease Patient Network Launch

Genetic alliance UK/Rare Disease UK

Tues, 20th October 2015 Cardiff

Dale Ward & Dawn and Callum Gray attended on our behalf.

More information about the event here:

The rare disease network has been formed to engage patients, families and patient organisation representatives in the work that Rare Disease UK is undertaking in Wales, in particular to support the implementation of the Welsh Rare Disease Plan that was published earlier this year

Within the day, we will be covering topics such as an introduction to clinical trials, new sequencing technologies and what they mean for patients with rare conditions and also hearing from patients and carers about their experiences of living with rare conditions.

We have a range of excellent guest speakers attending the day. We are excited that Professor Chris Oliver from Birmingham University will be our keynote speaker who will be drawing on his breadth of experience researching within the field of rare and genetic conditions particularly in relation to genetic syndromes associated with developmental delay.

Here's Dawn's report:

I just wanted to let you know that I took Callum to Cardiff to a Rare Disease Day. It was lovely to meet up again with Dale and share what it is like to have r20.

The day was short talks, which were all very interesting. One lady talked about her son and how she has now set up a website to give stories of rare diseases. She is happy for us to join her website, which will be good, especially as we are so ultra rare! Another talk mentioned on the medical side was that doctors and nurses are made more aware of rare diseases when training. We all felt this is important.

The day was 9.30 till 3.30 with a really nice lunch which Callum enjoyed. It was a great day and full of valuable information. I would recommend it to everyone.

Another thing I want to mention is Family Funding. Last year I applied for this through a friend's recommendation. Callum received a laptop which has been a great help to him in school and for homework. We did not pay anything just filled in the forms. A lady came to see us both and then it arrived. We do get Disability Living Allowance (DLA) and showed letters from the hospital about what Callum has. She had never heard of r20 so I told her about Ring20 Research & Support UK! You can apply every year even if it's a slide for the garden or an iPad.

Callum is going on camp in the summer with NCS. I had never heard of it. It's for 15 to 17 year olds to bring together people from different backgrounds and have fun canoeing climbing sailing walking cooking etc. It is government funded and I paid £50. Take a look for one in your own area and see what they have to offer if you are in this age bracket.

I am thinking of buying a box of chocolates and doing a raffle to raise money for R20. It is my busiest time of year so it won't take much effort or time to do. Even if it's a small amount it all helps.

I am so glad that I can be in touch with Ring20 Research & Support UK.

Research

At ILAE I took the opportunity to make contact with a number of medical professionals to discuss epilepsy research and r(20):

- Prof Tony Marson from Liverpool, is leading the recruitment of patients with epilepsy /learning difficulties for the 100,000 genomes project, however it is questionable as to whether ring chromosomes may be detected if present.
- We are privileged to have arranged a forthcoming meeting with Prof Helen Cross regarding potential research opportunities for r(20).
- We are hoping to follow-up with Prof Sameer Zuberi re: resurrecting his potential clinical research project

We would also like to follow-up with Prof Sanjay Sissodya and Dr Michael Johnson (amongst others) on genetic testing/research.

About Rare Diseases

In Europe, a rare disease is defined as one with an incidence of less than 1 in 2,000 people. While each of them is rare, there are more than 7,000 rare diseases, which together affect 24 million (six per cent) people in Europe and take 20 per cent of all health care costs. Improving the care and developing diagnostics and treatments for people with rare diseases are now international priorities.

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Your stories in print...

Read about those with r(20) who've been in the news recently...

Dolly Day

http://www.lancashiretelegraph.co.uk/news/13639455.Blackburn_teen_with_rare_condition_shows_off_amazing_collection_of_130_dolls/?ref=fbshr

Anya Belle-Brown (Canada)

http://epilepsyontario.org/student-attributes-surging-self-confidence-after-seizure-diagnosis-to-family-friends/

Helen's Heroes

http://m.columbiatribune.com/news/perspectives/helen-s-heroes-parents-work-to-make-a-normal-life/article_16c29bb8-6f40-55a9-b22e-9507b207ae6c.html#.Vgk_JXy0zYA.facebook

Merchandising

We now have a small selection of running vests available for anyone who wishes to take part in a sponsored sporting event to fundraise for us. Please get in touch if you're interested in running, walking, cycling swimming or anything else!

Following the success of last years calendars we've produced a limited number of new calendars for 2016 featuring some highlights celebrating this years events and fundraisers.

Calendars £10 plus P&P

Place your orders now via email or the website...when they're gone, they're gone!

We hope you have enjoyed reading our latest newsletter.

If you wish to receive future editions (we are anticipating producing these periodically, approx. 6 monthly), please ensure you signup to our mailing list on our website here:

http://www.ring20researchsupport.co.uk/

Any feedback on content and what you'd like included in future editions, please contact us to let us know. And of course, we would welcome any contributions of your own for inclusion.

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Supporting families, individuals and professionals affected by or who come into contact with Ring Chromosome 20 Syndrome

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