Issue 4

July 2016

Registered Charity Number 1165651

Newsletter

ring 20
research & support uk

Onwards and upwards...

Special points of interest:

- Checkout the stupendous efforts of our fundraisers (page 4/5)
- Have you joined the r(20) community? Get involved in the discussions— see RareConnect (page 8)
- Latest research news (page 9)

Hello again

Has it really been 6 months since our last newsletter?

This time last year I said a lot had happened; if only I'd had a crystal ball to see how far we've come just 12 months on.

With 4 conferences so far under our belt, plus awareness raising at the new Birmingham Rare Disease Centre, fantastic fundraising events in Wales, IT challenges changing webhosting provider for our website/ emails and launching a European online community forum in 6 languages—we didn't stop there...oh no...

We've attended meetings and are currently liaising with various medical professionals regarding a number of potential research opportunities for r(20).

We launched our r(20) Activists poster campaign.

And we've submitted grant funding applications—the first of which has been successfully accepted!

We couldn't do it all without the invaluable help and support from our team of volunteers—we just don't have the time or resources being a small charity run by just 4 Trustees (with day jobs and families).

To Tamzin, Jon, Dale and Dawn we are extremely grateful for your input and efforts and we hope that you will continue to provide ongoing support, on which we are reliant to function.

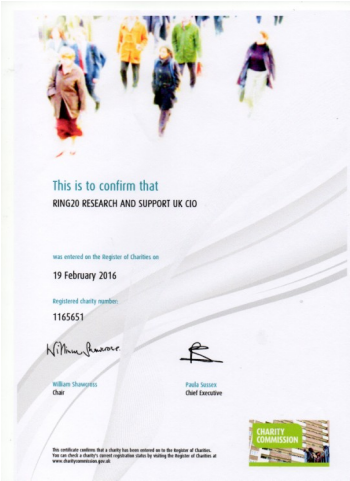
If you have some time to spare and think you could help us out too, do get in touch.

There is so much more that we'd like to do, but are unable to progress faster without more volunteers.

I look forward to hearing from you...

Yours

Allison Watson Co-Founder / Secretary


This is to confirm that
RING20 RESEARCH AND SUPPORT UK CIO
was entered on the Register of Charities on
19 February 2016
Registered charity number:
1165651
William Shawcross
Chair
Paula Sussex
Chief Executive
CHARITY COMMISSION

INSIDE THIS ISSUE:

Charities Commission Registration / AGM

As of 19th Feb 2016 we changed our legal status to Ring20 Research and Support UK CIO and became a registered charity with the Charity Commission for England and Wales.

Registered Charity Number 1165651

Registration will open new opportunities for us increasing our eligibility for such things as raffle prizes and donations, through to matched funding and importantly grant funding applications.

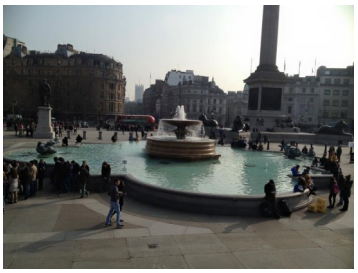
Please save the date for our forthcoming AGM, to be held In Sept—date to be confirmed in due course..

Voting forms will be sent out prior to the meeting, so watch out for these in your inbox. It would be great to meet with some of you in person on the day .

Get to meet with all the Trustees so we can hear your views on what's important to you, in terms of the support group service we provide going forwards.

ring 20 research & support uk

Our Volunteers



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

easyfundraising.org.uk

Dale—Global Leadership symposium

Hey my name is Dale ward I suffer with (R20) I got diagnosed when I was 8, when I first got R20 I was having 32 seizures a day later on in my teenage years due to bad medication I had eating problems as I struggled to gain an appetite and lost a lot of weight rapidly. I am now 23 and have been living in supported living with supporting staff 24 hours around the clock for nearly two years, they have supported me achieve a lot of goals the past couple of years these include visiting Longleat, attending and completing college courses but my biggest achievements are organising and running my fundraising events for R20 these include pub charity events, 24hour Gamethon and my family have even climbed Snowdon. My family have been fantastic working with me to organise a run these events. I am now on the edge of completing my Level 2 Health & Social Care in the next step of my pursuit to have a long career in the care sector, support while doing some voluntary work but my ultimate goal is to become a counsellor, which at this moment of time is a high likelihood. These days I'm much more confident, have much more self-believe that I can accomplish my goals and got everything there to do well. I'm down to earth and have a distinct likability factor which goes far in the line of work I am looking to go into. I still have 4-5 seizures a day and can last up to 30-40 minutes or 15-20minutes depending on the day and how I'm feeling, I have no control over them except I know when I'm going to have a bad day if I have a seizure in the morning as they usually occur in the evening and night. I am on medication at the moment which is keeping me relatively stable seizure wise and am not having any side effects (touch wood). I want to give back to people who once helped me and I want to do the same for others, I have met some incredible people living in supported living. People with various disabilities such as autism, mental health problems and alcoholism and they are all fantastic, the support staff have been amazing helping me through tough times I always have someone I can talk to and always there to help me through a seizure. I am living independently and anyone out there who feel there in need of help don't hesitate to ask for a social worker , there is a lot of things said about social workers such as 'child catchers' but there not they can help you if you need it don't be afraid to ask. They will search for the best possible solution for you and who you are looking out for. I have to say my life has changed for the better the R20 foundation have been great, I have travelled to conferences all over the UK such as Cardiff, Birmingham and even London. They were all fantastic experiences and I wish to have more of them but without funds it gets more and more difficult to attend these conferences and raise awareness which the charity has been doing fantastically so far but we don't want it to stop here. I will work to achieve my goals while working with R20 as I am also volunteer for R20 and represented them in London for the Rare Genetics UK Symposium while there I met some fantastic people where I learned more about other rare genetic disorders and what they may have in common with Ring Chromosome 20. I have stayed in contact with some of these people so we can work together to find a solution to our common cause. I always had this believe which kept me going that there is always someone worse off than you no matter how bad life seems there always someone worse and meeting some of these fantastic people proved that. I personally am quite old fashioned in terms I have my own sayings and this is a couple of them "things must always get worse to get better" and a wise old man once told me which proved true "good things comes to those who wait".

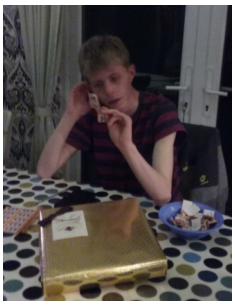
Thank you for listening and reading my passage I hope I helped in any way I could.

Dale Ward

Dawn—BCH—Rare disease Day and Undiagnosed Disease Awareness Day

Dawn hosted a stand for us at 2 events at Birmingham Children's Hospital (BCH), namely Rare disease Day (on 29th February) and Undiagnosed Disease Day (on 29th April). Dawn did a great job talking to everyone to raise awareness of r(20).

Here she is with Larissa Kerecuk—Rare Disease Lead at BCH.



Update from Dawn:

Callum did a raffle of Thornton's chocolates to raise money for Ring 20 Research and Support UK. We managed to raise £53. It was an easy thing to do as life has been very busy at the moment.

Callum is now 16 and staying on at school for 2 more years.

Dawn Gray

Our Volunteers…(continued)

Tamzin—social media update/ Jon—Website+

During the six months since the last newsletter came out, my husband Jonathan and I (Tamzin) have been continuing to help look after the social media and web site side of things. We don't have much free time but we are happy to do what we can to support the Trustees as we understand when dealing with a very rare condition the pool of volunteers is likely to be rather small.

On the social media side, we have seen a steady increase of new followers on both Facebook and Twitter. I continue to try to keep people up to date on new research and treatments as well as highlighting fundraising activities. In February, it was an interesting experience to take part in the Twitter debate on Rare disease day; communicating directly the Bath MP Ben Howlett who would be discussing the concerns of the rare disease community in parliament. It is also enjoyable to reach out to professional bodies via Twitter partially when someone is representing R20 at an event to encourage them to visit our stall and find out about us.

Due to our web site hosting company having a strategic change of business, we were given notice of the web site's hosting. This meant finding another provider and the migration of the site. Whilst this means no difference to you whilst browsing the site, it did mean quite a few hours work behind the scenes. One big advantage of the new hosting company 34SP.com is that they offer free hosting to registered charities. This helps us save more money for other purposes.

As part of our charity's new "charitable incorporated organisation (CIO)" status, Jonathan has updated the website to meet the new requirements. We've also added a new "Personal Stories" section. We're slowly publishing family stories here and we are hoping more people will share their experiences of Ring20 as this is a great way to connect as a community. Ongoing Jonathan continues to provide IT support for the site, provide web traffic statistics for the trustees and maintains the Ring20 maps.

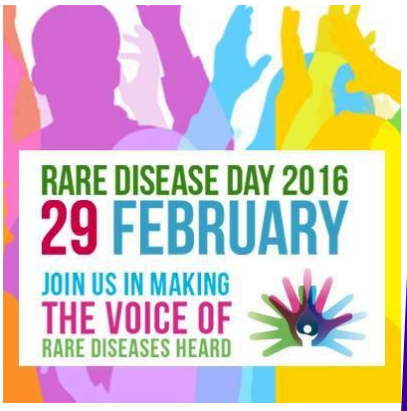
Tamzin Dunn

Our Website - http://ring20researchsupport.co.uk/

R20 Map - https://www.google.com/maps/d/view?mid=1qp4LBEeah3UaDQV4j-bEU9DkFGY

Ben Howlett: http://www.ben4bath.co.uk/
34SP: https://www.34sp.com/

CIO Status: https://www.gov.uk/guidance/change-your-charity-structure



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 2 cheques for £200 each from Cellular Solutions (Leanne Jackson's workplace) to help us with our vital work.

cellularsolutions

IFDS International Financial Data Services United Kingdom

IFDS have granted us £150 (and also £150 to Jeans for Genes) in respect of our families conference.

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

Matched funding has recently secured us a:

- £500 donation from Lloyds Bank in support of the Gamethon
- £1,000 from Barclays in respect of the Snowdon Climb.

Jeans for Genes Grant application

We are really pleased that our grant funding application to Jeans for Genes to host an r(20) families weekend workshop has been approved.

However payment of the full grant is subject to total monies raised on Jeans for Genes Day on Sept 23rd 2016. If we receive full funding, we will can start to organise this event for 2017—watch this space…

Jeans for Genes Day

GENETIC DISORDERS UK
Bringing together those affected by genetic disorders

BIG Lottery—Awards for All Grant Application

We have submitted a new application as a backup for the families conference, and for much needed IT equipment, training and new fundraising items and conference materials—so we can continue to provide important support to families.

AWARDS FOR ALL BIG LOTTERY FUND

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Fundraising

The Ward family and their friends have really been pulling out al the stops with two amazing fundraising events for us in the past few months—they're now thinking about what event they will do next!

We are incredibly grateful to all involved for their efforts and the huge sums of money raised through sponsorship and matched funding.

Read on and be inspired to do something equally amazing:

Snowdon Climb - May 2016 £1,003.00 (+ £1,000 matched funding from Barclays)

We managed to conquer Snowdon is the most horrendous weather. We met at 2am in Asda car park so we could plan for the 4 hour drive ahead in the pouring rain and follow each other up, so no sleep that night. We got to Snowdonia about 6.30am and parked at the bottom of Snowdon car park. The weather was grey, dark and foggy but no rain at this point, yeah we thought our luck was in!

Got ourselves layered up, hoody's, water proofs gloves, hats the works. Then started our climb, the route we did was the most scenic so even though the cloud and fog was thick we could see the lakes and waterfalls that were immediately around us, beautiful! This route is a steady incline to start with some harder sections until we meet the part where you really have to climb the mountain. Hands, feet and all sorts are needed to make it up, and this is when the heavens decided to open. Not just torrential rain but hail, the wind was blowing a gale with the hail bouncing off our faces but we kept on going strong. At this point there are no longer paths, you just grab a rock and climb but the whole thing had turned into waterfalls where there was so much down pour. We had a few slips and a few falls but thankfully and luckily no one was hurt and every one kept in good spirits. We finally make it to the top where we could go in the dry café and take off our wet clothes. We waited for Dale's train to pull in and there he was, it was an incredible feeling for us all to see his smiling face beaming with pride for us all. It was so amazing to see him at the top, we took him to the summit with us which he loved. It was an emotional and amazing day,

Claire said "the hardest, amazing, emotional day ever, such an incredible experience" The weather made it harder but made it more worthwhile. It was a great day and for such a great cause, loved every minute of it.'

it took 3.5 hours to get to the top and another 3.5 to get to the bottom then the 4 hour drive home. Long day lol.

Jemma Ward

Gamethon Cardiff - Jan 2016 £928.09 (+ £500 matched funding from Lloyds)

I was at a charity event last autumn for Ring20 which had been arranged by Dale Ward where I first had the idea to set up and host a Gamethon to raise money to support r(20) research. I had known Dale for years through my wife and since meeting him I had wanted to help raise money for research into his condition.

Sadly I'm no athlete, too many rugby injuries meant marathons, mountain climbs, and long distance events were out of the picture, but there was one thing I knew I could do better than most and that was sit and play computer games for prolonged periods. It was then I thought I could easily get a group of people together to play games for 24hours in aid of a good cause, and I'm good at games so how hard could it be? It turns out very hard!

Rhys Jenkins and Michael Westerberg were there to help me the next morning setting up the Facebook page and getting the word out. Our friend Paul Aboy (IX Duality) was already a YouTube wizz and he set up the live feeds, graphic overlays for the event, and plugged us on his YouTube page; its fair to say without the help of these three the event would not have been watched and funded as much as it was.

With everything in place for promoting next to sort was a venue, and Lawrence Facey came forward and offered us his house to run everything from. He also spoke to his neighbours and asked them for access to their internet just so we could get 12 Xboxes, 2 PC's and 6 live streams running at once (once again Paul Aboy was brilliant here). Luckily they agreed and after I had rounded up some friends , got enough consoles, TV's (a lot of partners left staring at walls that day) some amazing prizes for raffles and games everything was good to go, hard part over I thought… I was wrong.

The night before the event one of the neighbours decided that we could no longer use their internet access, it was now not clear if there was enough access to get everything to work. Then the morning of the event we had the realisation that getting 12 men, 12 40"plus TV's, 12 Xbox one consoles and 8 tables into one room was going to be a real struggle, never mind the lack of plugs. Some how we managed it and we found a slight side effect to all of this electronics in one place, extreme heat, the room was sweltering, and we were going to be sat in 1 spot for 24 hours—suddenly it started to dawn that this was going to be tough…

We started at 11am, and regularly had around 20 viewers across our channels (hundreds throughout the day) all being able to see our overlays, and t-shirts promoting the charity, this was a great feeling knowing so many people were getting made aware of the charity.

Fundraising...(continued)

People watched us play an entire FIFA league, everyone on at all times, 22 games each, 264 games of FIFA in total and 6 hours of football. Luckily I was crowned champion and we were all feeling the strain on our eyes after that, sadly there was 18 more hours to go!

After 12 hours we held the prize draw and had Dale Ward arrive to draw prizes and play some games with us. Michael Westerberg allowed Dale to shave his head as we passed the £1000 target that he said we would not pass, it was now 11pm, I had been up since 3am with my new-born and getting the event ready, it was really starting to get difficult now.

The last 12 hours were some of the hardest I have had to endure, slowly everyone deteriorated, the room got hotter and the strain on our eyes was becoming unbearable. We lost Matthew Miller to sleep around the 16 hour mark, Conor Sharpe went for some air at the 18 hour mark and was found in a bed 2 hours later, Rhys Jenkins made it to 21 hours before briefly succumbing to sleep, by the end I could barely function, my brain was numb, my eyes in pieces and speaking in long sentences a chore. It had been a marathon in every sense of the word and a lot harder than any of us had expected.

It took a week before I felt completely 100% again (a lot of us felt like this) i didn't touch FIFA for over a month, and the Xbox itself remained unpacked after the event for nearly 2 weeks. It had broken the inner gamer in me. In the end it was all worth it, we raised a total of £1428.09, and as hard and challenging as it was, we are already talking about arranging the sequel for next year!

Thanks for reading

Liam Facey

On YouTube if you search either IX Duality or The Ginna there are some excellent videos from the event.

Check out the time lapse video of the full 24hrs in a few minutes here:

https://www.youtube.com/watch?v=2JTbFdU1264

Be inspired!

Future events

Billericay Summerfext 2016 July 10th

Allison and family are hosting a fundraising tombola and cards stall in Essex.

Whole school fundraisers:

The following 2 schools have pledged to organise fundraising days on our behalf in the future:

Trinity School, Lewisham

Walton High School, Milton Keynes

Claire Sullivan SSE

Claire and some of her work colleagues are looking to give up a day of work to volunteer for us, supported by their employer SSE's 'Give something back' campaign..

Are you inspired to fundraise for r(20)?

If so, do get in touch with your ideas...

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Events

BPNA February Feb 2016, Sheffield

Following our medical advisor, Sophie's recommendation and introduction, Allison and David hosted a stand at the annual British Paediatric Neurology Association conference (BPNA) in Sheffield, held over 3 days. We arrived by train late the night before, ready for a prompt 9am start—travelling light to be able to carry all the stand essentials, plus David's ketogenic meals for 2 days! All the charity stands were located in a side hall and the first day was comparatively quiet compared to previous events, as many delegates grabbed a coffee in the main hall between heading off to the various talks. Still we had some interested visitors to our stand and come the next morning, almost a rush at morning tea break! Its great getting to speak with Paediatric Neurologists from all over the UK some who have/have not heard of r(20) and being able to share our information leaflets on diagnosis and signs and symptoms to recognise the syndrome. We hope this face to face engagement continues to raise awareness and will help more patients obtain a diagnosis and be advised of our support group.

The highlight of the meeting was meeting Dr Archana Desurkar from Sheffield Hospital who expressed a keen interest in r(20) having just diagnosed a new r(20) patient herself. She has kindly offered to help us with our research in identifying more r(20) cases around the UK.

Encounters with health professionals such as Archana, as well as liaison with other epilepsy related charities, helps to strengthen our support group and makes these events so worthwhile to attend.

Chair's Conference update – exciting news! RCPCH April 2016 Liverpool

Don had the opportunity of attending two events so far this year on behalf of our charity to raise awareness and learn more about building bridges and our capacity in order to achieve some of our core aims and objectives. The first was meeting was one that Professor Helen Cross and our medical adviser Sophia Varadker both agreed would enable us to reach the paediatric community.. This group of medical professionals are the group that get to see young children presenting with seizures. At this point we all know how important being able to rule out r(20) at an early stage is, or make being able to give an accurate diagnosis in order to avoid potentially unnecessary treatments. Until now we have never had the opportunity to reach out to this group.

Reading recent case histories from Callum's and Tilly's patient stories reinforces the importance of getting early knowledge and interventions in this area to as many institutions as possible in order to pick up the undiagnosed and identify future diagnoses. If we go some way towards achieving this through our presence at these conferences many more cases will be picked up and in turn grow the r(20) community and the capacity for research and action.

One key insight Don discovered from talking with this group was that paediatricians in the UK do not get to see EEG's as part of their clinical evaluations, so they would not be able to pick up any irregularities in this data. Recent research from the French Epileptologist Arnaud Biraben's abstract points towards a distinctive EEG pattern in r(20)

Don was able to engage with a good number of paediatricians from all over Europe (some at the is event and

Key Connections –

British Paediatric Surveillance Unit (BPSU) – Don was fortunate to meet with the research director Richard Lynn and sat in on their presentation. They can survey the whole, or segments of the paediatric community with a quarterly survey that look at the following:

- The condition – natural history – when does it present?
- The test – is it adequate, consistent, adequate, accessible?
- The intervention – does it make a difference, is there any delay?
- The screening programme?
- Implementation procedure p when does treatment begin?

Being able to put r(20) on this map would have significant impact from an awareness and registry perspective. Also provide valuable reporting data for future potential diagnosis and treatment protocols, as well as research opportunities. Watch this space!

ECRD May 2106 Edinburgh

8th European Conference on Rare Diseases & Orphan Products
26 - 28 May 2016 | EICC, Edinburgh, Scotland, UK

Rare Connect Community – It was good to meet with Robert Pleticha who has been instrumental in helping us set up our own profile and connections across Europe with this incredibly powerful and professional platform. We have now managed to connect with personal stories and medical professionals through multiple translations and share with like minded advocates, researchers, families, individuals all across Europe. This community has the capacity to become an international resource hub and lead to many more families, institutions and medical profes-

Events…(continued)

sionals with a common interest in r(20) A good number of you have already contributed to this resource and eminent groups from France and Italy have recently connected check out the link here to see what's happening https://www.rareconnect.org/en

BPSU – Richard Lynn Scientific Director

Don had the chance to spend time in discussion with Richard Lynn from BPSU again and talk around the opportunities for potential r(20) questions to be included on the survey panel that is circulated amongst the entire UK paediatric community. It enabled Don to get a better understanding of the purpose of the unit, how it works and the considerations that the advisory board are looking for in terms of patient related outcome experiences and patient related outcome measures. For example, if we are seeking an independent study? or to have specific questions included as part of a general epilepsy panel related to rare disease, intractable epilepsy monitoring/testing.

OpenAPP - patient registry

Both Professor Cross and Richard Lynn suggested the requirement for a patient registry for r(20) which we do not yet have – and Don was able to meet with a new provider of such a tool who was exhibiting at the conference called **Open APP**.

Rare diseases are highly heterogeneous. International interoperable registries are particularly important for rare diseases. They bring together a small patient population which can be used to initially complete a natural history of the disease and then engage with academia and pharma companies. Patient Registries, ultimately, improve the quality of life of patients. Registries allow clinicians to discover the best care path for a disorder and then measure patient progress along that care path. Patient registries constitute key instruments for the development of natural history studies, the improvement of patient care, healthcare planning and quality of life outcome - *"If it can be measured, it can be managed."*

This tool has been developed by a team in Ireland and we now have the project details that we will be discussing at the next trustee meeting in early July before circulating to members shortly. The good news is its FREE development for up to 100 first cases! To sustain anything beyond that number will require funding which we will apply for in due course.

Birmingham Children's Hospital Rare Disease Centre

Had the opportunity to share with Dr Larissa Kerecuk who is Rare Disease Lead and Consultant Paediatric Nephrologist at Birmingham Children's Hospital. She has welcomed the opportunity of sharing these tremendous new facilities with our patient group for conferences, or meetings you can see the scope of the facility here http://www.bch.nhs.uk/news/article/14103-hospital-unveils-images-brand-new-%C2%A3375million-clinical-block

I was also able to obtain some valuable learning for our group by attending 3 days of programme presentations covering a wide variety of topics like:

- Research Ethics
- Establishment of European Reference Networks (ERN) model for rare diseases
- Social Innovation for better care for rare diseases
- Patients empowered in research & diagnosis
- Breaking through in diagnosis
- The impact of social media on globalisation: new knowledge and advocacy
- Boosting rare diseases in a global collaborative research environment
- Patients need accurate diagnosis

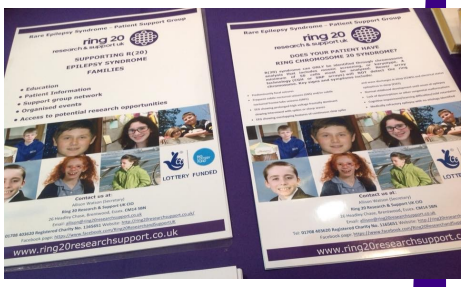
We can give all our members access to any of these presentations, so if you are interested in further details please contact Allison Watson.

ESNA June 2016 Manchester

Allison went along to the Epilepsy Specialist Nurse Association Annual conference in Manchester to meet with 80 ESN's from around the UK, to find out what they knew about r(20) and see if they had any patients in their care. It was quite a mixed bag in terms of those who had and had not heard of r(20).

All delegates were provided with our signs and symptoms list and r(20) leaflet about the support group in their delegate packs and I took time to chat to them about sharing this information with the colleagues. It was a privilege to hear Prof Sanjay Sisodiya speak about genetics and epilepsy and how understanding more about genetics causes will help to treat patients better in the future. There were also interesting presentations on women and childbearing with epilepsy and also driving regulations for those who have seizures.

One of the key reasons for attending was to promote our r(20) Activists campaign, so I gave out lots of our posters which should be appearing in an epilepsy clinic near you soon! However, there will be areas of the UK that I missed, or weren't represented at the conference, so please do try and take some posters along to your local hospital/clinic when you next attend for a routine appointment and help spread the word.



R(20) Activists Campaign

Have you signed up to help out with our r(20) Activists campaign as yet?

Posters have gone up in clinics:

- London GOSH, NHNN, Royal Free, Enfield
- NELFT Essex
- Birmingham—children's/adult hospital
- Milton Keynes
- Liverpool (Walton Centre)
- Oxford
- Gloucestershire/Worcester

Can you help distribute some posters to your local clinics to help raise awareness?

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RareConnect—join a wider r(20) community!

'What we learn from rare disorders often has profound consequences for our understanding of more common conditions.'

– Dr Francis Collins, Director of the National Institutes of Health

Have you checked out our new online community for r(20) on RareConnect yet?

If not you could be missing out!

Join the group here: https://www.rareconnect.org/en/community/ring-chromosome-20-syndrome

But we already have a website and a Facebook forum I hear you say, so why do we need another platform?

1. Not all our members are on Facebook (nor choose to be)
2. Facebook doesn't retain information forever and can delete historic discussions at whim.
3. Facebook re-orders posts, so you can't always find what you're looking for
4. *the best bits* - all content on RareConnect can be translated into 6 languages: English, Spanish, French, German, Italian and Portuguese by human translators and its FREE!

There are 5 key sections of the community:

Home

Where you can login and see what's new.

Understand

Here we have 4 patient stories already published. You can request to have your story added by selecting 'Tell my story'

Meet

This is where the discussion happens. Posts are grouped into logical topic areas., so you can easily follow a discussion or contribute a new question to the community yourself. All posts are moderated.

Posts will initially appear in their native language, however you can simply get an instant Google translate, or request a human translation into your own language within hours.

Learn

Key information about r(20) syndrome is published here—the benefit against our own website is the translation service—which will allow us to reach out to more r(20) patients/families around Europe and the world.

Members

Meet other members of the community. All members are vetted, for their interest and association with r(20) and we ask all members to signup to our member list here: http://eepurl.com/RtyQP

RareConnect is a platform for many different rare disease communities and the service is provided by EURORDIS—the European Organisation for Rare Diseases.

The community has already attracted new families to our r(20) community and with the use of Google Ads in multiple languages, searching for information r(20) just got easier!

We believe that once this gets going we'll find a lot more r(20) patients and families with whom we can provide mutual support and share information all over the world.

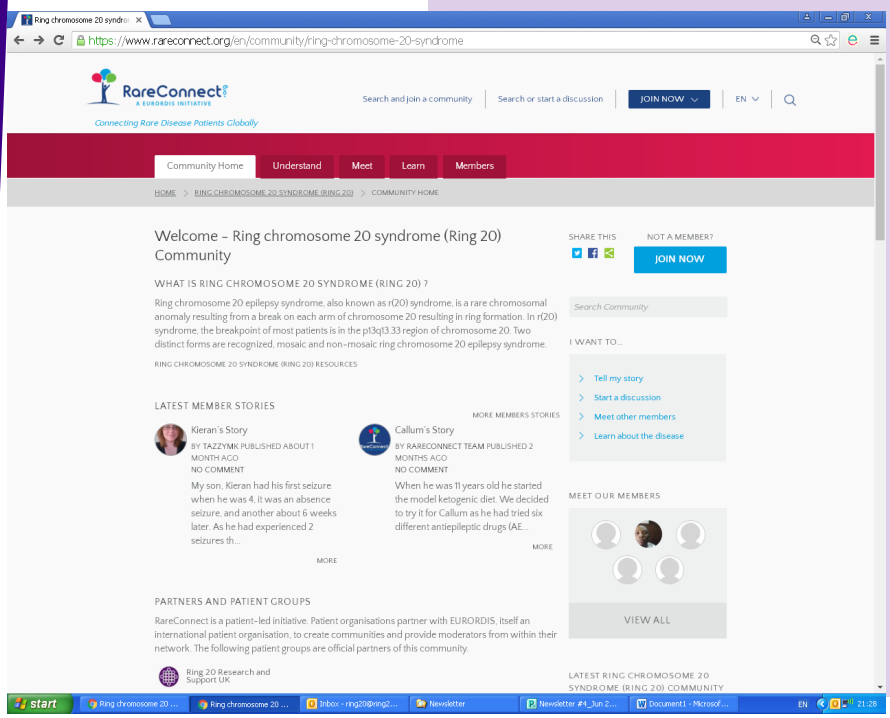
This exciting new venture has attracted the interest of neurologists from around Europe, who have previously engaged in, or continue research into r(20) syndrome including Dr Biraben and Dr Semah from France, Dr Gil-Nagel from Spain and Prof Canevini from Italy—all of whom have joined the community and are awaiting your input.

Future opportunities include:

◊ The ability to host webinars—our aim will be to identify suitable topics of interest to you, our members and invite guest speakers to host a session with real-time Q&A. If you miss a session it can be recorded, so historic sessions can be accessed at your convenience.

◊ Creation of a library of all published medical articles on r(20), in full abstract form, available on request—so you can access all there is to know about the condition.

What are you waiting for—JOIN NOW!



Research

Prof Helen Cross – r(20) research study opportunity with Prof Sameer Zuberi

A timely encounter with Prof Helen Cross OBE who you all know as the one of this country's leading authority on epilepsies in the UK and possibly abroad.

Allison and Don had a meeting with Prof Cross earlier this year and we are engaging behind the scenes to bring about a research project in collaboration with Sameer Zuberi from Scotland who has reportedly over 10 r(20) clinical cases to publish. Projects like this are usually many years in the making so being able to talk directly with Prof Cross at this meeting allowed the impetus and interest to be maintained for this important work.

Significant funding may be required if this project takes off—but that won't stop us pursuing this vital work.

We have also been in discussion about potential participation in a European Reference (ERN) for complex epilepsy.

BPSU audit

As a result of attending the last 2 conferences, Don has now engaged with Richard Lynn from the British Paediatric Surveillance Unit (BPSU) to find out how we might get an audit undertaken of all diagnosed r(20) cases in the UK. Audit requests can be submitted to BPSU for consideration and can cost ~£12,000, taking 2 yrs to conduct. The audit comprises a number of questions that are sent out to centres around the UK to record known cases of r(20). We are very lucky in that Dr Archana Desurkar has expressed an interest in helping us with this study and we are hoping that we will be able to make a submission request to BPSU shortly.

FREE Patient Registry

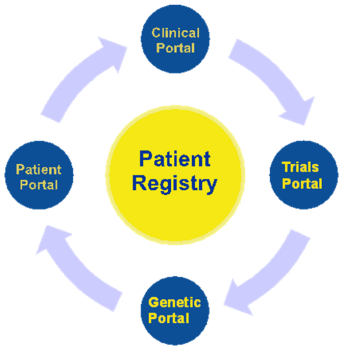
There is currently no patient registry for r(20) syndrome and we believe that this is an essential tool for all involved in patient care. At ECRD Don was referred to OpenApp an online system that is free to rare disease communities tor the 1st 100 cases. Read on to found a little more about OpenApp:

Why you need a Patient Registry

Rare diseases are highly heterogeneous. International interoperable registries are particularly important for rare diseases. They bring together a small patient population which can be used to initially complete a natural history of the disease and then engage with academia and pharma companies. Patient Registries, ultimately, improve the quality of life of patients. Registries allow clinicians to discover the best care path for a disorder and then measure patient progress along that care path. Patient registries constitute key instruments for the development of natural history studies, the improvement of patient care, healthcare planning and quality of life outcome - *"If it can be measured, it can be managed."*

From Registry to Rare Disease Management System

Our vision is that a patient registry ought to be optimised for a disease and not limited by geographical area. It should be a system not just for research but for patients and clinicians. We believe that a registry should evolve into a patient-centric, longitudinal record which is an ideal solution for:



- ☐ Patients
- ☐ Clinicians
- ☐ Clinical trial studies
- ☐ Genetic research

Using this eHealth patient centric approach, registries can integrate with multiple different systems including hospital based labs, radiography departments and IT systems. Doctor's notes, multi disciplinary team reviews and automated assessment triggers can be integrated into the registry giving a longitudinal view of each patient's disease history along with real-time comparisons to health factor norms. With OpenApp Registry, every patient registry can become a virtual centre of expertise, highly optimised around the target disease. OpenApp Registry is the ideal ICT infrastructure to develop a European Reference Network (ERN).

Potential collaboration with r(14) group

Don an Alison had a Skype meeting with Marco Crimi Scientific Coordinator for the r(14) support group in Italy to discuss potential for future collaboration on ring chromosome projects. Watch this space...

Epihunter

Some of our families are helping out with research providing input and feedback into the concept of a new device for children that will detect absence seizures whilst at school.

Awards

Our own Allison Watson was nominated earlier this year for a Young Epilepsy Champions Award, for being inspirational to those around you in the epilepsy community.


Champions Awards 2016

Young Epilepsy Champions

Allison Watson

Congratulations! You have been nominated for a Young Epilepsy Champions Award 2016 for being inspirational to those around you. Everyone at Young Epilepsy commends you for this incredible achievement.

Carol Long
Chief Executive

Better futures for young lives with epilepsy

Now recruiting…

We are currently advertising for a professional fundraising volunteer to help us with researching, completing and submitting significant grant funding applications—up to £50,000 per annum.

If you've just been reading the section on research you'll appreciate that we have some serious funds to raise if we are to realise our ideas and make research happen.

We continue to appreciate all your fundraising efforts and these are equally as important to continue to fund the running costs that allow us to support you and provide an ongoing service: through phone/email support, website updates and provision of up-to-date information etc.

So please keep doing what you are doing—as we wouldn't exist without your help!

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.

Ring20 Research and Support UK CIO

26 Headley Chase
Warley, Brentwood
Essex CM14 5BN
England

Phone: 00 44 1708 403620
E-mail: ring20@ring20researchsupport.co.uk

Find us on the web:
http://www.ring20researchsupport.co.uk/

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

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