

Newsletter

ring 20
research & support uk



New horizons...

Special points of interest:

- Checkout our recent fundraiser's efforts, as well as lots of fund-raising ideas (pages 4-7)
- Find out about the NEW! European Reference Network (ERN) for rare and complex epilepsy (page 10)
- Latest Research news (page 11)

Having been established as a patient support group for 3 years now, 2017 is shaping up to be our biggest year so far!

In August we are hosting our r(20) families weekend conference in Liverpool; the first ever such event held in the world. With families attending from Canada, US, Belgium, Netherlands and Germany as well as the UK, this will truly be an international event. If you still want to purchase tickets please see our feature article on page 9 **Forthcoming Events**; very limited availability remains.

Late last year we recruited two new volunteer Managers: Gemma and Tomi, to help us with our funding and project work respectively. Inside we tell you a little about what they've been busy doing (see pages 4 & 9).

Not content with hosting a world first, we are also continuing promising discussions with eminent researchers regarding the potential to participate in a specific research project into r(20) syndrome – see our feature article on **Research** page 11.

Whilst we progress these potential opportunities, significant funding will be required to move this forward to the next stage. We will need to raise between £50,000-£150,000 in the next 6-12 months—no mean feat for a

small charity such as ours with no track record in research projects - but we must aim high to make a difference!

Gemma's on the case, however any funding applications submitted are likely to be more successful if we can demonstrate our own fundraising efforts as a support group. So, we have put together a 2-page spread in this issue on **Fundraising Ideas** (pages 6&7) to see if you can do your bit to help us too?

Also inside is some first insight into the new **Epilepsy Research Network (ERN)** for rare and complex epilepsy launched in January and how we are getting involved as patient advocates as part of the ePAG group (see page 10). We hope to bring you more as this develops.

All this new and exciting activity, alongside the day to day running of the charity, is quite an ask for those who volunteer their time to make this happen and to whom we are truly grateful. See what they've been up to in our regular articles **Our Volunteers** (pages 2&3) and **Events** (page 8).

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AGM 2017—save the date

Our next AGM is planned to take place at our families conference in Liverpool on Saturday 5th August 2017.

For those attending the conference, this will be an opportunity to attend in person and hear first hand a summary of all the great work undertaken last year and be presented with our end of year accounts.

If you are not able to attend the conference or AGM, then if you have registered your full details with us i.e. you are a member, then look out for the proxy voting form that will be

emailed to you in due course.

If you are registered as a supporter, but would like to become a full member with voting rights, you will need to complete your full registration details with us here: <http://eepurl.com/RtyOP>

We look forward to meeting with you in person at our AGM.

Yours
Allison Watson
Co-Founder / Secretary

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Our Volunteers



We really value the work that our volunteers carry out on our behalf.

Here's what they've been up to...

Dawn—BCH— Rare Disease Day Feb '17

Dawn hosted an r(20) awareness stand at this event in Birmingham for us; here's what she had to say about the day:

A really good day for us at Birmingham Children's Hospital today. Bit of a bad start as everyone went to the marquee, so not many people visited our stand, but then at lunch there were more people, then lots in the afternoon. I only had 3 leaflets left of the folded coloured ones at the end of the day!

I met Jo Williams a nurse at BCH—she has a sister with epilepsy. I told her about the ketogenic diet. She took a card and gave me her email address so she can look at epilepsy. Spoke to a mum of 8 and 13 year old in Cardiff who has partial absence seizures. Her seizures can be set off to certain music. I told her about heat setting our youngsters seizures off or extreme cold.

A lady called Ann said to contact her; she's from www.bild.org.uk the British Institute of Learning Disabilities. A man called Peter Salisbury is who to contact regards their Epilepsy network—he can maybe help us? (Note from Editor—we are following this lead up...)

I also spoke to a young man from Oxford University who is studying rare diseases. He was interested in r(20) so took some information leaflets—he had not heard of R20 before.

I met Oliver Timmis (from Findacure) who knows of our patient support group and also Emily Clarke (Freelance Genetic Counsellor) who had met Allison and Don previously; she was from Genetics Disorders UK and had her support dog with her as she is partially sighted. I chatted with Rachel from Rare Revolution magazine, which was nice to speak with her—she said the next issue will be published in March. I spoke of r(20) to a consultant from Cumbria. He took leaflets and I asked if he could put a poster in the hospital for us. I believe he was a neurologist (didn't get a chance to get his name—sorry!)

A Dr who runs the clinics of rare diseases in Birmingham talked of transition and said it is not easy, especially when they reach 25! She told me of a story of how she got 2 patients together in clinic that would never have met.

So good to share an illness with others.

I took a leaflet from the ladies by me. One lady works or used to work with Prof Helen Cross in research. When passing Larissa Kerecuk she said "Hello Dawn!" How can she remember so many people?

I went to a few of the talks, not all as more people came to the stand during the afternoon. I picked up leaflets etc.

It took me 2 hours to get home. A long day but worth it.

Dawn Gray

Dale—Leadership Symposium

March 2017

For the 2nd year running Dale represented our patient support group at this weekend event in London. The event comprises a networking dinner with representative leaders from 100 rare disease charities from around the UK, to facilitate networking and information sharing, followed by a whole day of talks on the Saturday on an array of topics including some specifically pertinent to our group:

- ◆ How will Brexit impact on UK medical research and patients with genetic disorders?
- ◆ Managing health professionals' engagement with a complex syndrome.
- ◆ Giving a voice to patients affected by genetic disorders and the charities that support them
- ◆ Punching above our weight: How extremely rare disease patient groups can plan for success.
- ◆ Harnessing the impact of the genetic disorders community to increase awareness and raise more funds.

Here's an account of the event from Dale's mum Claire, who accompanied Dale at the event:

Had a great weekend in London for the Global Leadership Symposium conference. I can't tell you how proud I was of Dale as he went and talked to everyone to tell them about his condition and to put the word out there about Ring Chromosome 20. Listening to him speak about r(20) and hearing the passion and dedication in his voice took my breath away and made me very proud. Strangers were coming up to me after listening to Dale's life story and were saying how proud there were of him, even though they didn't know him and how they wanted a Dale in their lives made me speechless and quite emotional on times.

I know I have said it before but Dale you inspire me more 'n more each day and I am truly blessed to have you as my son xxx

Claire Ward



Our Volunteers... (continued)

Tamzin—social media update/ Jon—Website+

Tamzin and Jon Dunn continue to both support our group.

Jon keeps an eye on our website hosting, updates the r(20) map and supports us in all things 'techie', whilst Tamzin is responsible for the regular Twitter and Facebook posts you see.

More recently Tamzin has taken over some of the regular email communications that you receive from us, relieving Allison to perform other core activities for the day-to-day running of the group. Additionally Tamzin has become a fundamental and valued member of the project team assisting with organising of our families conference, primarily as our communications manager. Not only has Tamzin taken on responsibility for emails, but also media communications and advertising of the event. This has included submitting her son Kieran's story to Epilepsy Action for publication in the June edition of Epilepsy Today which has a circulation of thousands of people living with epilepsy in the UK. What better way to highlight awareness of r(20) syndrome and our conference! See here for link to Kieran's story—'[Looking for Answers](#)'

Tamzin is doing all of this despite her family being unable to attend the conference themselves due to a pre-arranged family holiday. We are extremely grateful to Tamzin for the commitment she makes to attending our fortnightly teleconference calls and all of the ac-

tions she takes away to complete.

We hope you enjoy receiving the regular emails and follow us on social media? /if you have any feedback on the content of things that you'd like to see/know more about please drop Tamzin a line at

tamzin@ring20researchsupport.co.uk

To keep up-to-date with what we're up to checkout our website -

<http://ring20researchsupport.co.uk/>

And if you're not already on our R20 Map-

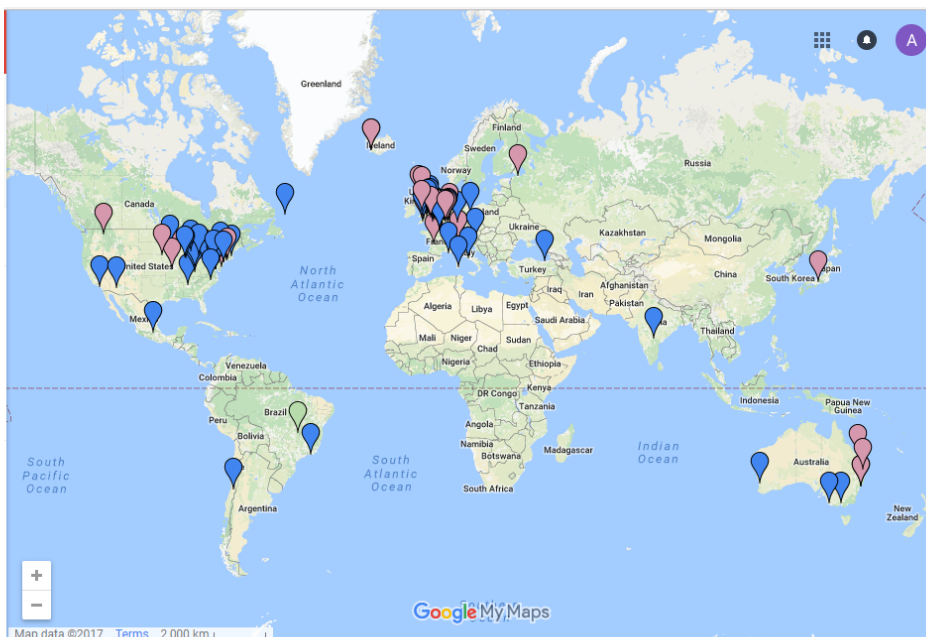
<https://www.google.com/maps/d/view?mid=1qp4LBEeah3UaDOV4j-bEU9DkFGY>

We will be very happy to add you — so please contact us [here](#).



<https://www.facebook.com/Ring20ResearchandSupportUK>

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



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Fundraising

Welcome to our new Fundraising Manager

In the 10 months since our last newsletter we have recruited 2 new volunteer managers to help with the administration and organisation of our group's activities, each with specific roles to play as part of our expanding management team. Here we introduce:

Gemma Underwood
Volunteer Fundraising Manager



Gemma Underwood (MInstF) has over 13 years' experience working in the voluntary sector, for small, medium and large charities, providing a wide range of services to vulnerable people. Specialising in trust, statutory and corporate fundraising Gemma has also undertaken Head of Fundraising roles with responsibility for managing multiple fundraising income streams. As a

consultant Gemma now develops fundraising strategies and submits high quality funding applications for charities across the sector.

Having been diagnosed with an unrelated, genetic condition in 2009, Gemma is keenly aware of the importance of research into rare conditions and having established a fundraising consultancy in 2015 was keen to support a dynamic local charity in championing this cause.

Ring 20 Research Support UK CIO provided an ideal opportunity and Gemma is now our Volunteer Fundraising Manager with responsibility for developing fundraising strategy, submitting funding applications and managing fundraising activities. As a member of the Institute of Fundraising Gemma is committed to adhering to the codes of fundraising practice in all fundraising activities.

Gemma volunteers 1 day per week. She has created a pipeline of suitable grant funders and corporates to apply to, for funding our future projects and activities and she is busy submitting numerous grant funding applications every week for us, as well as providing much needed guidance to the Trustees in this area. Gemma also has lots of other fundraising ideas so watch out for her feature article in the next newsletter.

Grants and Sponsorship

Corporate Sponsorship

Matched funding has recently secured us a:

- £111 donation from Ledger Bennett in support of a cake sale organised by Jon Dunn for Purple Day. Jon nominated us to receive the matched funding, as the direct proceeds of the cake sale were donated to Epilepsy Action.



Bringing together those affected by genetic disorders

Jeans for Genes Grant application

We are really pleased to confirm that we have been awarded our full grant of £5,000 from Jeans for Genes towards our r(20) families conference this summer.

Together with our £6,000+ grant from the BIG Lottery Fund and a further £1,000 from our generous sponsors LivaNova, plus £150 donation from IFDS, we have been able to organise an event far surpassing our original expectations and budget of 6 families!

See our feature article on the forthcoming event on page 9.



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

If so, do consider approaching them in respect of any fundraising activity you undertake.

Fundraising... (continued)

Raffles

The Gray family have held a couple of raffles for us; last summer Dawn's box of chocolates raised £53 whilst Callum's Easter Egg raised £67 for us recently.



Arts and crafts

Last summer we had a stall at the Billericay Summerfest and had a tombola and sold Sue's handmade cards raising £40.20 on the day.

Subsequent card sales have raised an additional £28.

Are you inspired to fundraise for r(20)?

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

Collection Tin

The Gray's have also had a collection tin out at a local store for which the proceeds will be counted at our forthcoming families conference.

Donations

We are always extremely grateful to receive any donations no matter how large or small, which make an invaluable contribution to our vital work, ensuring we can continue to provide you with an ongoing service, as well as pursue bigger projects, such as research opportunities.

All donations from UK tax payers are also eligible for gift aid, which means we can claim a further £1 from HMRC for every £4 donated.

giftaid it

Here's some examples of generous families who've chosen to support us through donations since the last newsletter...

Family donations

A member of the Fenner family recently had a birthday but instead of presents they requested a donation to r(20). £200 was raised which was fantastic.

A member of the Dunn family kindly made a £50 donation at Christmas.

As did the Glewes family with a donation of £100.

A member of the Gray family donated £20.

Other donations

A donation from a kind member of the Brewers Club made a £5 donation.

Regular donations

One family makes a small regular monthly donation of £5 towards our work. Again, this valuable contribution is eligible for *gift aid*, so we can claim a further £1 from HMRC every month.

Could you make a small regular donation towards our work? If so, please contact us for a standing order form and gift aid declaration (UK only).



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Fundraising Ideas

Feature article

There are lots of different ways to fundraise for charity, you don't have to be super sporty, have lots of time on your hands, or specialist skills.

Here's just some ideas...

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](http://www.easyfundraising.org.uk/causes/ring20researchsupport)

Fundraising the easy way

easyfundraising
.org.uk

Join *easyfundraising* and raise funds while you spend!

It's FREE to join, simply signup and choose your fundraising partner Ring20 Research and Support UK CIO and download the fundraising reminder.

Then every time you purchase something online, simply activate your donation and a small percentage of the purchase price of your item(s) will be automatically donated to our bank account—at no cost to you!

Get all your family and friends to join too, because the more members we have the more donations we receive.

In the last year we have received over £100 through *easyfundraising*.

eBay.co.uk



Have a clear out and sell your unwanted items on eBay.co.uk and donate a percentage of your sales to Research and Support UK CIO. You decide what percentage you donate from 10-100%.

Buyers are more attracted to items sold that benefit charities.

We also automatically receive gift aid on any items sold too!

Car Boot/Bring and Buy/Garage Sales

A fun way to sell off a few unwanted items, so why not do so for the charity you support? Often the entrance fee can be waived if your proceeds are going to a nominated charity. We can provide you with an official letter of support...

Donate

We accept donations by post, through BT MyDonate, or eBay for charity or PayPal.



If you live in the UK you can also choose to make a regular e.g. monthly donation to us.

All donations are eligible for gift aid if you are a UK tax payer. Please ask us for a gift aid form if you haven't previously completed one.

Host a tea party

..with family, friends or neighbours. Invite a few people round, make some cakes and serve tea/coffee or juice for the younger ones in return for a small donation towards Ring20 Research and Support UK.



or a cake or bake sale...

...or if you're into baking and/or have some friends who can help, go bigger and better and sell cakes at school, or work for donations.

You can use posters from Rare Disease Day, Jeans for Genes Day or just Ring20 all of which can help advertise your event.



Get crafty!

Are you artistic, or good at making things? Could you make items to sell for charity, or even host a stall at a local event?

Plant Sale

Are you green fingered and like gardening...could you grow some plants from seed and sell them to family, friends or colleagues or at school for a small donation?

Sell your unwanted items and raise funds for r(20) on eBay here:
[http://
www.ebay.co.uk/egw/
eBay-for-charity/charity
-profile/?NP_ID=71688](http://www.ebay.co.uk/egw/eBay-for-charity/charity-profile/?NP_ID=71688)

Fundraising Ideas...(continued)

Feature article

Hold a raffle

Buy something small to raffle to friends like Dawn and Callum Gray have done previously, or approach local traders with a fundraising request letter to ask them to donate prizes for your raffle event.

If you have a large workplace, colleagues are often quite happy to buy a few raffle tickets in support of a good cause!

You'll be surprised how generous small, local business can be, from donating gift vouchers, cinema tickets, bottles of wine, chocolates, complimentary passes to health clubs, handbags, flowers, perfume, mobile phones and more...

iPad mini raffle

If you've got a large audience of potential buyers of raffle tickets, why not try purchasing a popular electrical/electronic item (I've tried an iPad mini, in the past).



Sell enough tickets to cover at least the cost of the item e.g. 100 tickets for £5, then one lucky person gets a superb prize and we profit too!

Pack bags at a local supermarket

Get a few family, friends or work colleagues together and spend a few hours packing shopping bags at the tills in return for donations.

It's easy to approach your local supermarkets to ask for a slot to fundraise for charity in this way and we can help provide you with a letter of support.

If there are only a few of you, then a smaller supermarket may make more sense such as Marks and Spencer, as opposed to a Tesco Extra with 20 till points!



It can be quite tiring so make sure you take regular breaks and dressing up or wearing something that promotes our charity e.g. one of our r(20) t-shirts, will help people to understand why you are fundraising.

It's also a great way to raise public awareness of r(20) as some people will stop to ask you about the charity and the condition.

Get sporty!

Run, walk, cycle, swim, skydive...

You don't have to be a sporty individual yourself, but maybe a friend, family relative or neighbour is? Do they know about Ring20 Research and Support UK and that we are always looking for people to help raise funds for us?

Maybe you have some work colleagues who like running marathons, or half marathons, or even 5 or 10k's, sponsored cycle events or other — think about approaching them to ask if they would run for Ring20?

We can provide FREE running vests in return for their generous support.



Other sponsored events

Often popular with school age children/youngsters...and a great way for young people to get involved with charitable giving...

There are lots of ideas that you can use for sponsorship, here's just a few:

- ◆ Sponsored silence
- ◆ Sponsored walk, run, climb, skip
- ◆ Sponsored knit
- ◆ Sponsored sit-in e.g. overnight
- ◆ Sponsored chocolate fest
- ◆ Sponsored goal shoot

The more creative or original the idea, the more likely people are to sponsor you.

Also it's important for your potential sponsors to understand why you are fundraising, so make some home made leaflets/flyers about our charity and r(20) syndrome. You could make these up yourself, or we could help you with logos or templates—simply ask...

Collection Tins

Ask to put out one of our collection tins at a local store or club near you?

Host an event

If you've got more time and energy then host a whole event, such as the Ward family and their friends have done in the past...maybe taking over a local venue such as a pub or club, for a fundraising evening—maybe host a fundraising ball, race night or karaoke and raffle?

Or speak to your local restaurant to see if they'll help you organise a charity dinner or buffet for a set price, where you agree upfront that the charity gets a percentage of the proceeds from the night?

Be inspired!

and have fun supporting r(20)...

Don't forget we have balloons, collecting buckets and tins to help you with your events and to promote the cause you are supporting.



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Events



Midlands Rare Disease Showcase—Oct '16

Dawn and Callum Gray hosted an r(20) stand at this event which celebrated the diverse progress being made within the rare disease community by bringing together patient groups, pharmaceutical and biotech companies, clinicians and researchers. The event was an opportunity to showcase the UK's first Children's Rare Disease Centre being developed within Birmingham Children's Hospital.

Annual Welsh Rare Disease Patient Network meeting—Dec '16

Dale represented r(20) at this event in Cardiff which comprised talks from experts discussing the latest advances in genomics and genomic medicine and from inspirational speakers in the field of rare diseases as well as a great opportunity for networking with other rare disease patient groups, researchers and health professionals.

Cambridge International Rare Disease Summit Oct 2016 (CRDN)

Don and Allison attended this event last year.

The program for the day centred around:

- What can Cambridge contribute to global efforts in rare disease research?
- Who's leading the way in collaborative, innovative research, treatment and care for those with rare diseases?

How can we put patients and their families at the centre of all discussions?



A thought provoking day of speakers at Robinson college, Cambridge [#crdn2016](#). The introduction from new chair of CRDN Alistair Kent OBE was followed by a panel of 'Powerful Voices' where patient organisations are shown to be integral to making change in rare disease: quote from Ben Howlett MP "patient organisations make massive amounts of change".



We heard from research and pharma and had an opportunity to uncover that whole genome sequencing (WGS) in its current state (as used in the 100,000 genomes project) may not necessarily detect the presence of a ring chromosome and asked what can be done if anything to change that? (we have discussions ongoing with Dr Richard Scott – geneticist at GOSH and Clinical Lead for 100,000 genomes project)

In the afternoon, we learnt about the wonderful facility that exists in Norway run by Frambu – a multi-disciplinary centre for rare disease, centered around the patient and their families, followed by Larissa Kerecuk (amongst others) highlighting the new rare disease centre for children here in the UK, due to open mid-'17.



Throughout the day patient organisations (including ourselves) were actively encouraged to be involved in the discussions. Speakers made reference on a number of occasions to 'Dr Google' – which is now a power tool in helping making a diagnosis in rare disease cases – highlighting the power of the internet and why our rare disease [r(20) must have a presence.

We also got to understand just how important it is for us to establish a patient registry for r(20) and begin to chart the natural history of the syndrome for researchers, clinicians and pharma biotech companies looking to provide better treatments in the future

None of us really knows what the impact of Brexit is likely to be for cross border health initiatives but there is definitely a willingness amongst existing organisations toward collaboration and the establishment of European Reference Networks (ERN's) that work collectively on treatments and protocols to maximise and standardise diagnosis protocols and improve care for rare diseases.

See how we are getting involved in the ERN for rare and complex epilepsy on page 10.

Could you
volunteer to
represent
r(20) and/or
host a stand
at a future
event for us?

If so, please
do get in
touch
(expenses
are usually
covered)

Forthcoming Events



Our families conference is fast approaching!

With only 2 months to go to the big weekend, final planning is well underway. This is truly shaping up to be an international event with families from the UK, US, Canada, Belgium, Germany and the Netherlands attending so far.

The speaker line up includes a number of eminent names many of you will recognise from r(20) research papers. These include Pr Nancy Spinner, Pr Sameer Zuberi and Pr Maria Canevini.

As part of the programme we hope to feature some r(20) Young Ambassadors which will be delivered by Janine Palm from Young Epilepsy and Emma Ninnis (Epilepsy Specialist Nurse) from GOSH in conjunction with some of our young members.

In addition, we are very fortunate to have secured additional exhibitor support with LivaNova and Nutricia and are anticipating more educational contributors over the coming months.

There will also be a great time for everyone to get together at the gala dinner on the Saturday evening with some interesting entertainment planned alongside some great social activity on Sunday morning— you can probably guess what's involved since we will be in Liverpool that a certain musical interlude is afoot.

Without giving too much away this event promises to be very special indeed and we will be keeping you all up-to-date with regular messages.

The event is being filmed so if you cannot attend you will be able to see all the highlights

from the weekend after the event and reach an even wider audience. We really do look forward to seeing as many of you as possible over the weekend **4-6th August 2017**

If you still want to come along you can purchase tickets [here](https://www.eventbrite.co.uk/e/ring20-family-conference-tickets-31372338509):
<https://www.eventbrite.co.uk/e/ring20-family-conference-tickets-31372338509>

Tomi Akintunde
Volunteer Project/Event Manager



Tomi joined us 6 months ago to add value to the team, volunteering 1 day per week of her time.

She has volunteered in the past with Oxfam and BHF as she is passionate about giving back to the society. She has been busy managing the upcoming family conference event leading fortnightly teleconference calls between the project team and taking on various actions herself such as researching venues and securing bookings.

Regrettably for us, Tomi has just returned to paid employment and with a young family to care for her commitment to us is too much; we have therefore had to withdraw her voluntary role. However Tomi has expressed a desire to work with us in the future, personal capacity allowing.

We wish Tomi all the best in her new job!

r(20) family conference

A world first!

Have you got your tickets?





Don / Allison
are elected
ePAG
representatives
for EpiCARE.

This is a 5 year
project and a
very exciting
opportunity for
all those living
with rare and
complex
Epilepsy—
including r(20)
syndrome.

NEW! ERN for Rare and Complex Epilepsies

Feature article

Earlier this year 24 European Reference Networks (ERNs) were approved, including EpiCARE specifically for rare and complex epilepsies.

EpiCARE Network

The EpiCARE network has been developed to deliver highly-specialized diagnostics and care to improve interventions and outcomes in individuals with rare and complex epilepsies.

The EpiCARE group, coordinated by Prof Helen Cross from Great Ormond Street Hospital for Children (GOSH), has been recognised as a ERN on rare and complex epilepsies. The network comprises 28 Health Care Providers, from 14 EU countries.

Dissemination of activities will be core to the network.

NETWORK GOALS

- Full access and utilisation of pre-surgical evaluation and epilepsy surgery;
- An increase in diagnosis of rare causes of the epilepsies;
- Enhancement of identification of patients with treatable rare causes of the epilepsies
- Increase access to specialized care for rare causes
- To foster research on innovative causal treatments in rare and complex epilepsies.

ATTAINING NETWORK GOALS

With a strategy of collaborative working, sharing of expertise and access to advanced diagnostics EpiCARE anticipates an increased number of individuals with refractory epilepsy to have an underlying diagnosis as a cause for their epilepsy.

By collecting information about where such patients exist, the ERN will be able to collate information on clinical presentation and evolution across the life span, constitute cohorts of these rare conditions and advance the development of clinical trials, utilising innovative trial design for small select numbers of patients.

EpiCARE will also continue to increase awareness and accessibility of epilepsy surgery for carefully selected individuals through a continuation of the core network.

For more information on EpiCARE click [here](#)

European Patient Advocacy Groups (ePAG) - The role of ePAGs

Specifically, ePAGs and ePAG representatives:

- Contribute to the ERN Board to provide the perspective of patients on all relevant aspects of the ERN strategy, policy & organisational processes
- Promote and encourage a patient-centric approach in both delivery of clinical care, service improvement and strategic development & decision-making

- Advocate for care that is patient-centred and respectful of patients' rights and choice
- Provide the patient perspective on the application of personal data rules, compliance of information consent & management of complaints
- Ensure that processes to address all ethical issues and concerns for patients are in place, balancing patient and clinical needs appropriately
- Advise on transparency in quality of care, safety standards, clinical outcomes & treatment options
- Advise on overall planning, assessment and evaluation of ERN activities and initiatives
- Monitor the performance of the ERN by reviewing quality indicators such as clinical outcomes of diagnosis and treatment
- Develop an ePAG feedback and evaluation framework across all ERNs to provide patient experience feedback of ERN and healthcare providers' activities
- Monitor and evaluate the adoption of patient feedback by the ERN based on patient experience surveys prioritising the objectives, work plan and service improvement in the network on an annual basis
- Contribute to the development and dissemination of patient information, policy, good practice, care pathways and guidelines
- Contribute to research e.g. defining research areas important to patients and their families and disseminating research-related information
- Identify expert centres to join the ERN as a full member or affiliated partner
- Provide an evidence-based patient perspective on the needs of people living with a rare disease and ensure all rare diseases are considered and included in ERN discussions and activities
- Produce annual ERN evaluation reports

Collectively, ePAGs represent the perspective and interests of European rare disease patient organisations associated with ERNs.



Research

Feature article

(R(20) Research Proposal Prof Sameer Zuberi

We have been continuing to follow up on our lead to make a research project specifically targeted at r(20) a reality.

We received a research proposal from Prof Sameer Zuberi, Consultant Paediatric Neurologist at the Royal Hospital for Children, Glasgow which the Trustees have been busy reviewing.

Don took the opportunity to visit Prof Zuberi and some of his research team in Glasgow in April this year, to discuss the proposal in more detail and to view the excellent facilities.

In brief, the research may look to delineate in detail the natural history of Ring Chromosome 20 Epilepsy Syndrome and identify biomarkers for the cognitive and behavioural impairment which can be used as outcome measures in future trials of therapy.

Also, to develop and curate an international registry of Ring Chromosome 20 patients to inform future research studies.

Part of the visit included a tour of the facilities which includes a 7 Tesla Magnetic Resonance Brain Imaging (MRI) unit. The 7T scanner based in the Imaging Centre of Excellence (est. 2017) at the Queen Elizabeth University Hospital and Royal Hospital for Children campus in Glasgow is one of the few 7T scanners worldwide in a clinical hospital setting.

There is enough wire wrapped around its huge magnets to stretch from London to Brussels.

When it is operating, those magnets - but not the patient - will be cooled to minus 269 Celsius.

That is the temperature of deep space. So this particular corner of Glasgow will become one of the coldest places in the universe.

The unit of strength of a magnetic field is a Tesla (T), named after the Serbian-American pioneer Nikola Tesla.

This is the most powerful imaging technology available and is only now becoming available in select centres for research in human disease. This means unparalleled detail when looking at brain structure in people with Ring 20 syndrome.

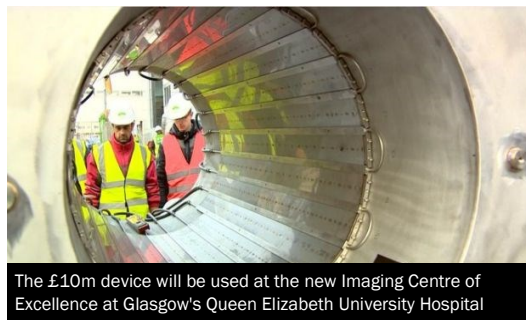
- Are different brain areas affected in terms of volume and the connections they make?
- Do these changes relate to the profiles of individual patients and do

they change with age in the cohort?

- Are they better preserved in people with less severe epilepsy?

This is an exciting development and represents a significant milestone in our charity's aims and mission as well as the wider rare disease community.

Although this project requires significant financial resource we are hopeful that with the help of our fundraising manager and other partners this research will become a reality within the next few years.



The £10m device will be used at the new Imaging Centre of Excellence at Glasgow's Queen Elizabeth University Hospital



The 7 Tesla MRI scanner gives a much more detailed picture of the human body



Don's Health Culture Research Project

Our Chair and Co-Founder is currently engaged in his final year of a nutritional Science Degree at The Centre For Nutrition and Lifestyle Management (CNELM)

As part of this he is hoping to complete a dissertation thesis project later this year looking at the research question of 'common pathophysiological mechanisms *surrounding dietary interventions for seizure management in r(20) epilepsy syndrome* ? A Literature Review.

Don is hoping this may be a precursor for attracting future research funding to examine this area in collaboration with medical professionals and groups such as Matthew's Friends. Look out for more news in your inbox, or for more information contact at don.gordon@ac.cnelm.co.uk.

ring 20
research & support uk



r(20) Activists Campaign

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

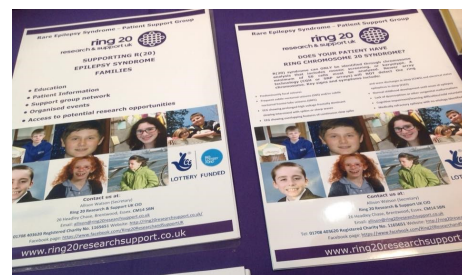
Posters have gone up in clinics around the UK:

- 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, wherever you are in the World?

Please ask us for more details.

Can you help...?

To continue our work and build on what we have already achieved requires more and more demands on our scant resources.

We are always looking for additional help, from the day-to-day running of the group, to larger projects, events, fundraising and more.

You don't necessarily have to have specialist skills; a willingness to give some of your time and energy is all that is often required. Training and support is available, plus out-of-pocket expenses are usually covered.

If you think you can help us or know someone who can, then do please get in touch:

allison@ring20researchsupport.co.uk

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 sign up 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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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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Any medical information included herein is merely to signpost readers to information that is freely available. Ring20 Research and Support UK do not provide medical advice and patients seeking such, should always consult with their own medical team.