

Newsletter

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Special points of interest:

- Find out what happened at our UK families meeting (full write up inside)
- We are keen to encourage you to join us as members, see sign-up link on page 2
- Can you help?—see our fundraising update for what others have been up to on your behalf...(page 7)

Our 1st 3 months...

Wow...its been only 3 months since we setup our newly formed charity: **Ring20 Research and Support UK** and what a busy start we've had!

From putting r(20) on the map, starting a patient support group forum, to our families meeting (**now on YouTube**), launching a brand new website (with lots of updated medical information on r(20) to be included!) and creating some initial literature—its been a whirlwind, but very exciting too!

Very importantly we have enlisted the help and support of a UK paediatric neurologist, Dr Sophia Varadkar, who we hope will help to represent us and guide us in our efforts to engage other Health Professionals interested in r(20) and to further raise awareness of the condition.

I hope you'll agree that the efforts have all been worth it! And its with much thanks to

the input and hard work of our volunteer trustees, Don, Chris and myself.

Many of you know Don and me, as we've been volunteering for families affected by r(20) for a number of years in a previous capacity, but as Chris is new to you all, we've asked him to write a short piece in this newsletter to introduce himself.

This newsletter is aimed at all our families and supporters, some of whom may not have access to, or may have missed some of our Facebook updates. There's lots of new information here too...

I hope you find this publication interesting and will continue to support us in our mission to help YOU?

Yours

Allison Watson

Co-Founder / Secretary

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Introduction to our Treasurer:

Chris Garn

"Being semi retired now I have been interested in charitable work for the last few years and have previously carried out some volunteering work at Oxfam (retail) and Thames Chase Trust (financial and administrative duties).

As a chartered accountant for some 45 years I have previously held several financial controller positions within international group companies and acted also as finance director/company secretary in medium sized companies.

My hobbies include amateur dramatics, keeping fit, all sport and chasing my young grandson around.

Earlier this year Allison my niece and Don asked me to join the new charity as trustee



mainly to help with financial and secretarial matters of which there are several to deal with in a small charity. r(20) requires further research and those that have the condition and their families, relatives and medical practitioners need continuing information about developments and the positive benefits provided by mutual support groups.

This is an extremely worthwhile cause and I intend to help Allison, Don and members to achieve the charity's aims.

From small acorns large oak trees grow... “
Chris

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r(20) Families UK Meet

Last month saw our first r(20) families meeting, held at Young Epilepsy here in the UK for families affected by the condition.



Our families

Sat 17th May 2014 @ Young Epilepsy, Lingfield, Surrey

The meeting was added on to the end of Young Epilepsy's own Parent's and Families Information Event, where we exhibited as

Ring20 Research and Support UK for the first time.

We met with other parents of children with epilepsy, networked with organisations similar to ourselves, whilst actively participating in the day itself and various workshops on offer.

As there are so few families in the UK (and indeed Europe), we were never expecting a large group for our meet, however we managed 3 out of a possible 5 families attending on the day itself, which is great!

I'm sure more will make it to the next one.

If you haven't already signed up, to become a member, join us here:

<http://eepurl.com/RtyQP>



Introduction by Don Gordon

Don Gordon opened the meeting, introducing the new charity, our name and logo.

Why have we setup a new charity? As you are aware the Ring Chromosome 20 Foundation is being wound up and there is no organisation in the UK or abroad that we are aware of that provides support for families affected by r(20), for which we perceive there to be a continued need. We are setup to operate on a purely voluntary basis run by our three Trustees: Don Gordon, Allison Watson and Chris Garn (Treasurer).

Our purpose or 'mission' is to aim to provide a new support network for families affected by r(20) and signpost families and Health Professionals to the latest information on the condition and to continue to raise awareness. As our name suggests, our 'vision' is to engage in and share future potential research opportunities, which as a patient support group is definitely possible, as researchers need patients to carry out their research and obtain funding for potential studies.

We have just launched our new website:

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

The cost of construction of the website has very fortunately been gifted to us in advance by a kind benefactor who has offered to run a marathon next year to meet the build costs.

As a new charity a key aspect for us will be

funding. We will be looking to apply for grants e.g. EU, National Lottery and corporate sponsorship, alongside fundraising activities whereby we'll be looking to you our members and supporters to help us. Securing sufficient funding will allow us to provide the best service we can to you; initially this is required purely to cover our start-up costs. We strongly encourage you to become a member if you haven't already signed-up. Membership is open to all those aged 18 years or over, is FREE and entitles you to vote in our Annual General Meeting (AGM) - so you can have an active role in how you would like the charity to move forward.

Join us here: <http://eepurl.com/RtyQP>

Only 9 families are currently on our subscription list even though we are in contact with some 80+ families worldwide, so in order to stay in touch and to actively to show your support, please take a few minutes to sign-up today - its FREE and no obligation. Membership numbers are frequently asked questions in many grant applications, so the greater our following the greater our chance of success for any bid we make!

We have created a new brochure which introduces our new charity, a PDF copy may be provided on request. We hope to be able to distribute these to encourage new members!

Activities by Allison Watson

Allison then went on to explain some of the initial activities we have been engaged in:

Facebook

We have launched a new Facebook page where we will be posting all our latest news: <https://www.facebook.com/Ring20ResearchandSupportUK>

Patient Support Group

Our patient support group forum is very active, which is really encouraging to see families sharing stories and thoughts and generally helping each other. We have over 60 members in our CLOSED group.

If you haven't already joined, join here: <https://www.facebook.com/groups/798475916833994/>

Website

As Don mentioned, our website is now LIVE with a landing page comprising our contact details. The main pages of the site are currently under construction, which we hope will be beneficial to new and existing contacts affected by r(20). The site will include exciting new updates to medical information being provided by Dr Syed Hosain and Nancy Spinner respectively. We have recently been in contact with Nancy Spinner whose team are continuing their research into ring chromosomes, which is very encouraging!

"We are dedicated to continuing research on Ring Chromosomes and are exploring multiple research questions. To update you on our research, we have received numerous Ring 20 samples from families and from bio repositories. We are currently in the process of isolating and classifying these samples to determine percent mosaicism and general characteristics of the ring within each sample. To date, we have been successful in cloning a mosaic sample from a single patient and isolating the two cell types present – ring containing and non-ring containing cells. These samples were sent for numerous types of genetic analysis to answer different questions pertaining to how a cell reacts to the presence of the ring. By using different technologies (such as RNAseq, WGS, SNP Arrays), we hope to answer multiple questions in a variety of ways. This multi-pronged approach will allow us to more comprehensively understand the ring, which we hope will eventually lead to a better clinical understanding of how to best manage Ring 20 Syndrome."

teristics of the ring within each sample. To date, we have been successful in cloning a mosaic sample from a single patient and isolating the two cell types present – ring containing and non-ring containing cells. These samples were sent for numerous types of genetic analysis to answer different questions pertaining to how a cell reacts to the presence of the ring. By using different technologies (such as RNAseq, WGS, SNP Arrays), we hope to answer multiple questions in a variety of ways. This multi-pronged approach will allow us to more comprehensively understand the ring, which we hope will eventually lead to a better clinical understanding of how to best manage Ring 20 Syndrome."

Research

We are also providing a coordination and translation service (thanks to Allison's daughter Jessica for her Spanish linguistic skills J) to a PhD student in Lima, Peru – Juan Carlos Marin, who with his team are conducting a research study in his country which we hope to hear more about as it progresses.

"I am Mg. Héctor Herrera Reynoso, head of the Human Cytogenetic Department in the Centre of Molecular and Genetic Epidemiology at the Universidad Nacional Mayor de San Marcos. We are interested, (Juan Carlos and the team) starting from next year in studying the relationship that exists between the presence of Ring 20 and epilepsy, by using [Nancy Spinner's] research carried out in this field as a reference."

We are liaising between Juan Carlos Marin and Nancy Spinner, a Nancy is kindly assisting with genetic aspects of the study.



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

r(20) Treatments Survey—patient perceptions

In the 2 weeks leading up to our families meeting we launched a survey asking for your experience/perceptions on medications and treatments for r(20). A fantastic 25% of you responded in that short period – so THANK YOU! – and Allison presented the initial results to the group.

If you haven't already seen this a copy can be provided on request to: ring20uk@outlook.com

Some of the key findings include the range and number of treatments (primarily AED's) that respondents are currently on and/or have tried. The average number of AED's being ~

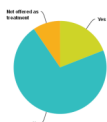
2.5, with many patients on 4 or 5 AED's currently. The number of AED's tried is also quite extensive for many patients, perhaps indicating the trial and error scenario of treating this condition. Another interesting, but perhaps not surprising result is the lack of commonality of AED's in use by our patients, however seizures are known to be uncontrolled in all but one respondent, so how effective is treatment and do treatment benefits outweigh side-effects? We heard at the YE day that where any more

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Vagus Nerve Stimulation (VNS)

- Q4: Do you have a VNS implant?



Ketogenic Diet - perceptions

- Q8: Have you found the ketogenic diet successful in controlling seizures/quality of life?

Of the 8 responses to this question:

- 12.5% - n/a
- 25% unsuccessful
- 12.5% no change
- 12.5% some success
- 12.5% good success
- 25% extremely successful

r(20) Treatments Survey (continued)

than 2 AED's are used, any further are likely to be minimally effective and potentially toxic. (The answer to this may lie in a question for another day...)

The survey is still open, so if anyone who hasn't yet participated still wishes to do so you can find the link to the survey here:

<https://www.surveymonkey.com/s/93Z2SG8>

The greater the total response, the more accurate representation the results will be and therefore the most useful to potentially pass on to interested medical professionals...

Anti-Epileptic Drugs (AED's)

73% of respondents are on multiple AED's currently (average 2.5 AED's per respondent).

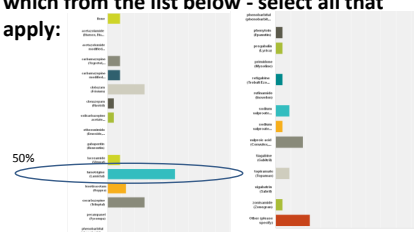
2 respondents are not on any AED's.

Only 1 respondent is seizure free.

Respondents have tried between 0-21 other AED's (prior to their current regime) – average no. of previous AED's tried = 5.6

Anti-Epileptic Drugs (AED's)

- Q1: If you are currently on AED's, please state which from the list below – select all that apply:



Watch the video of our recent Families Event in the UK on YouTube (in 3 parts):

<https://www.youtube.com/channel/UC23QAR4IbvBkJsb4ORt4uQ>

Findacure Workshop

Findacure are a charity supporting patient groups for rare diseases like ourselves and aim to provide the tools and information necessary for such small groups to be successful individually, by providing free workshops on key topic areas of relevance, which individually groups may not have the time, money or resources (or indeed contacts) to find for themselves. Their presence is therefore valuable to us... The day prior to our meet Allison attended a 2nd Findacure workshop in London entitled 'European Commission Funding for small patient groups'. Whilst not immediately relevant to us as a start up new group, it is useful to develop a strategy for our vision and is also a great networking event with similar like-minded rare disease groups. What we took

away was some tactical steps that we can start to take towards our 'vision', such as creating opportunities to meet and/or communicate with other interested parties e.g., medics /Health Professionals and get them on-side. There may be chances to collaborate with other small patient organisations, to share knowledge /costs. It is evident that we need to build our presence in the UK/EU, growing support for our group by identifying new r(20) patients, targeting GP's, hospitals, nurses to refer patients to us and raise awareness of r(20) to increase the diagnostic rate.



Epilepsia—Our story...



In this vein, we have just submitted an article that has been accepted for publication in Epilepsia about living with r(20) and our charity. Epilepsia is a publication distributed to all neurologists around the world and articles are available online.

We are grateful to Carol and Rachel Taylor for agreeing to share their story with everyone.

We will let you know when it goes to print...very soon.

NEW! We welcome Dr Sophia Varadkar to support us!

We are very lucky to have enlisted the support of Dr Sophia Varadkar, a paediatric neurologist from Great Ormond Street Hospital and UCL, to work with us as our medical contact here in the UK, helping us to engage with appropriate Health Professionals to further our presence.

Dr Sophia Varadkar MRCPI,
MSc, PhD
Consultant Paediatric Neurologist and Honorary Senior Lecturer Great Ormond St Hospital for Children NHS Foundation Trust & UCL Institute of Child Health



Questions to our panel

At this point we handed over to our guest panel, Dr Sophie (SV) and Clare Harrison (CH) - Epilepsy Nurse Specialist from Young Epilepsy - to take questions from our families. We've attempted to summarise some of the discussion points below:

(DG) Potassium channel drugs e.g. Retigabine (Trobalt) EU / Ezogabine (Potiga) – US

SV explained that whilst Retigabine when introduced was intended to be a new 'wonder drug' for epilepsy as it worked in a new way (something that hasn't been seen in new AED's), however due to adverse side effects of blue pigmentation to the skin and eyes and potential damage to eyes, it has been withdrawn from the market for further investigation to ensure its safety. It is potentially still available on request.

(TD) medical marijuana

SV explained that the use of medical marijuana is being explored as a potential epilepsy treatment in the UK and other countries and indeed Professor Helen Cross is involved in a trial into its use (dosage) and efficacy. Medical Marijuana is available for MS sufferers so it is known to have therapeutic effect for other conditions.

(AW) "Brain Power" – essential oil

CH warned of the care that should be taken with the use of certain essential/aromatherapy oils that may exacerbate seizures. However neither CH or SV were specifically aware of "Brain Power"

(AW) Survey findings – polytherapy and range of treatments being advised for r(20) patients – is it because patients are being treated as individuals and/or potentially as a result of lack of joined up thinking between medics because the condition is so rare?

SV answered that the list of drugs tried is not surprising as it represents the main 1st line epilepsy treatments and the thinking in terms of polytherapy vs. monotherapy is 'less is more'. In respect of the range of treatments our respondents reported, though unable to say specifically, it may be due to the fact that the condition is so rare and medics see so few patients that they have limited information available to them.

We asked how we might change this and endeavour to create more joined up thinking amongst Health Professionals. A number of possible suggestions were put forward by both SV/CH:

- Adult Neurologists - whilst it is perceived paediatric neurologists are aware of r(20), adult neurologists and/or epileptologists may be less informed (as previously not so targeted). AW can attempt to assess this as David transitions to adult services over the coming months. Collectively we could target through the International League Against Epilepsy (ILAE)
- Paediatricians - it was suggested that maybe we should target paediatricians, to raise their awareness of the signs/symptoms list, such that they more readily refer and/or arrange appropriate diagnostic testing for potential patients. Maybe through the Royal College of Physicians & Child Health (RCPCH).
- Unique/ContactaFamily – we should connect with other like-minded support organisations, who can flag newly diagnosed patients to our group.
- Geneticists – raising awareness of correct diagnostic testing.(AW will speak to SV offline to identify relevant representative organisation)
- Community Paediatricians – another group of health professionals who can be contacted via the British Association of Community Child Health (BACCH)
- ESNA – CH suggested we could also ensure we target epilepsy nurse specialists through their group (ESNA). Some future questions for surveys/research suggested were:
What drugs worked well? And (as importantly)
What drugs made you worse?
It would be useful to obtain a of rate of incidence (epidemiology) of r(20) in the UK. This may be possible to arrange a survey via the College of Paediatricians (of which he BPNA is an offshoot), who can ask for cases on specific conditions. The British Paediatric Surveillance Unit (BPSU) is a further smaller offshoot and it may be possible to engage a junior doctor to pose some research questions on our behalf.(We might like to ask for Dr Hosain and Nancy Spinners contributions for suitable questions)



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Feedback...

And finally, a comment from one of our families attending:

"The day was very helpful; so many families with children so ill with epilepsy; meeting GOSH and was very good to talk to consultant."

We hope that you have found this interesting and/or useful? We've tried to relay as much information that was shared on the day with you, but it is never as good as actually being there!

You can see that much has happened in the 3 months since we established ourselves and

there is lots more to do! If you feel willing or able (in any way) your offers of help/support would be gratefully appreciated – we cannot do this alone!

If you would like to be a part of a future r20 support group meeting, please do let us know and any ideas on what you would like us to cover and where you think we should hold this?

Your feedback is important to us, so do please send us your thoughts and suggestions. Our patient support group exists **for you** and can only be successful **because you make it so**.

Would you be willing to support us by making a donation?

Or

by printing and distributing some of our brochures at your local hospital?

Contact us if you can help...

Brochures for families and flyers for Health Professionals

We have produced a new brochure for new families who wish to familiarize themselves with our new organization.

The leaflet introduces us and has links to our website, Facebook page, support group forum, map etc..so families and/or professionals affected by or have some connection with r(20) may contact us and join our activities.

We have copies available in PDF format for circulation if you'd like one, or feel able to distribute any on our behalf to help raise awareness then please let us know..

Also in progress are flyers targeted at Health Professionals summarizing updated signs &

symptoms of the condition and information regarding diagnosis, appropriate genetic testing and treatment

We hope to be able to produce these to distribute at various vents/conferences and to mail to targeted Health Professionals.



Subscriptions and Donations

We are now able to accept donations in the UK, on which we will be able to claim Gift Aid, should you be eligible. We would also welcome regular subscriptions (via Standing Order or Direct Debit).

Form available on request.

Should you wish to support our work and help other families like yourselves who are keen to

have the latest information on r(20) and/or are interested in promoting research into the condition, then any contribution you are able to make (no matter no large or small) is gratefully received!

We hope to be able to accept donations from outside the UK very soon, via a PayPal donate facility that is being setup on our new website.

giftaid it

Grants and Sponsorship

Our first grant application has been submitted to Ford Britain Trust for a small grant of £250 to assist with our start-up costs. Our application is currently under consideration, and we are keeping our fingers crossed that we will be successful, however we know that they are always oversubscribed.

Going forward we will need continued funding for essential annual running costs of the charity, including website hosting/updates, telephone costs, printing of literature, insurance, fundraising equipment, conference attendance fees—this is without looking forward to our mission of promoting research opportunities. As you know we are run on a purely voluntary

basis, so all funds raised are invested back into the charity to support our aims.

We have been offered assistance to make an application for funding from the Big Lottery, so this is where we'll likely apply next...

Do you work for a company that offers Corporate Sponsorship to small charities? If so, would they consider supporting us and/or award you matched funding on any monies that you raise when you host your own fundraiser e.g. sponsored event or cake sale.

Don't be shy, apply NOW!

**Organise a fundraiser
for us**

**—no matter how large
or small your idea...
every penny helps!**

Fundraising Update

As well as applications for grants and sponsorship we will be relying on members and supporters to help us raise funds.

This is an essential source of income to us and is very highly valued, not only by your Trustees, but also by all our other members—as we are all helping ourselves.

Indeed if we can demonstrate a willingness amongst our own members to support ourselves, this will make us more successful in future funding applications!

Those of you who've been following us on Facebook will know about some of the initial fundraising activities that have already borne fruit in our first 3 months, these include:

- £45 personal donation.
- £75 from an Easter Egg raffle organised by the Gray family.
- £225 proceeds from a cake sale at Ithaca College—London.
- And £576 (so far...) from the Gray family's recent coffee morning event.

Here are some photos—I think you'll agree the cakes look very tempting...mmm? And what a fantastic effort—we are extremely grateful!



To aid future fundraisers we've purchased a small number of logo'd collection buckets and boxes and also some balloons. These are available to borrow, should you wish...

We've also been lucky to have been nominated charity for the Year, by the Brewers club in Romford, Essex (a retirement association who meet monthly and collect for a chosen charity at each meeting)

Future planned events include:

An Indian Curry Night that I'm organising at the Rajmoni, Brentwood, Essex on Wed 20th Aug—do come along and support us if you are within travelling distance. More details on our Facebook page, or contact me directly.

Mark Saggs (our website developer) running a marathon next year on our behalf.



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Merchandising...coming soon

We hope to be able to offer you the chance to purchase T-shirts and Polo shirts sporting our new logo in the very near future.

We are currently organising costs/printing and will be providing an order form for you to purchase items real soon...

A small percentage of the production costs will go towards our fundraising pot—so by purchasing you will not only be able to show all your family and friends that you are supporting us, but also you will be doing so knowing you have helped raise funds for our cause to continue to support families like yourselves in the future!

If you are thinking of organising a fundraising event—what better way to demonstrate who your collecting for and look professional at the same time!



The Gray family sporting our new polo shirts at their recent fundraiser!

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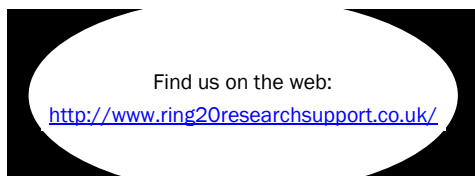
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We hope you have enjoyed reading our very first newsletter.

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 here:

<http://eepurl.com/RtyOP>



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.

*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.