Issue 7

August 2018

Registered Charity Number 1165651

Newsletter

ring 20 research & support uk

Bumper Issue!

Special points of interest:

- Highlights from our 1st ever Gala Ball (see centre spread)
- Exciting potential patient led research
- Fun in the sun at our family picnic
- Epihunter has been launched!

Firstly apologies that this issue is slightly late, but there's a good reason—we have so much to report!

Some of what you'll read in this issue you may already be aware of, such as our first ever Gala Ball, but we're so proud of the Sullivan family and their incredible fundraising achievements to-date that we wanted to share some highlights with you and also bring you Chris and Claire's own take on why they've embarked on such an incredible journey. They have inspired some of our other families to join in not only fundraising activities, but also representing us at events—which is key to our mission to grow our community and increase awareness of r(20). Check out our events pages 4&5 to see what our volunteers have been up to...

Our Research Fund Campaign has exceeded expectations; even though we have a way to go, the target is in sight and you are helping to make it increasingly possible.

Our momentum is continuing with the appointment of Kim Alliston our new Fundraising Officer volunteer who has a strong track record in achieving results for small charities such as ours.

We bring you news 'hot off the press' on the potential research opportunities that Don is exploring with the Patient Led Research Hub (PLRH) at Cambridge University examining whether the Ketogenic Diet is a possible treatment option for r(20) - we know that a number of you have tried KDT to varying degrees of success, some almost life changing, but yet there is no reference in medical literature to reflect patient experience. We want to change this. This really is patient led research in the making!

In terms of patient engagement, I've included some updates on my recent patient advocacy activities as part of EpiCARE and how we hope this will improve outcomes for many patients with rare and complex epilepsies, including patients with r(20). You'll also see I've been liaising with Genomics England on how we might improve diagnostic rates for r(20) in the future.

We are pleased to include an interview with Tim Buckinx from Epihunter—Tim is one of our member families and a true innovator!

Lastly thank you to all contributors who bring life to the stories and to Andrea for her help with collation of this issue.

I hope you enjoy reading this bumper Summer issue!

Allison

Co-founder/Secretary

INSIDE THIS ISSUE:

Research Fund Campaign update

Wow! We are now a third of the way to achieving our target to fund a 2-year Biomarker and Natural History Study that could comprise around 100 r(20) patients from around the world—the largest study on r(20) by far—and see the creation of an international database.

This wouldn't be possible without the tremendous efforts of our member families who've raised the majority of this money through their own fundraising events. **Wherever you are in the world, you can contribute to make this research happen!**

https://mydonate.bt.com/events/researchfundcampaign

We are in regular contact with Prof Sameer Zuberi to explore how we can begin this research. We'll keep you posted...

So far we have raised £46,551 towards our £138,106 target! Thank you for your support.

Keep up the great fundraising effort !

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Research

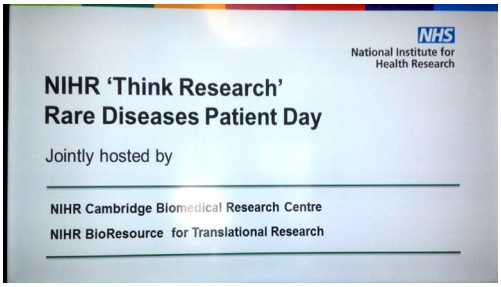
Two great examples of why attendance at events not only increases our learning and networking, but can lead to future collaborations.

National Institute for Health Research (NIHR)

In March, Don attended The NIHR 'Think Research' Rare Diseases Patient Day hosted by Cambridge Biomedical Research Centre and NIHR Bioresource for Translational Research.

Here's what Don had to say about the event:

Great Day of networking and learning #RareDPD - more on why data is central to research, new data consenting laws, the NIHR Bioresource for translational research centre - studying the link between genes, the environment, health and disease @cambridgebrc. Great talk on improving transition from Prof Allan Colver from Newcastle University check out the toolkit involving young people in research www.northumbria.nhs.uk/dahtoolkit



PLRH

We have now submitted a proposal based upon Don's initial research dissertation work in nutritional science in collaboration with the Patient Led Research Hub (PLRH) at Cambridge University Clinical Trials Unit Cambridge University Hospitals NHS Foundation.

Don is also hoping to submit a final abstract on his dissertation work ('The mechanistic plausibility of and clinical implementation of Ketogenic Diet for patients with r(20) syndrome and similar syndromes') at two scientific meetings in 2018/19 so keep an eye out for this in the near future.

Sharing some of these findings and recommendations from Don's work, prepares the way for potential pre-clinical trials in relation to the ketogenic diet (KDT), dietary interventions and patient registry for r(20) patients.

Initially we are looking to PLRH to submit 2 survey's on our behalf — one to health professionals in the UK and the other to r(20) patient families across the world to obtain their views and experience of ketogenic diet.

In addition, we have also been able to engage with a clinical outreach librarian at Royal Papworth Hospital to help with a current review on published literature on r(20) /rare/refractory childhood epilepsy, KDT and its use in the UK.

We are very excited to be working with PLRH who have a broad and well resourced facility.

Patient Led Research Hub

Aim

- Patients often know more about disease and lifestyle needs than many medical professionals
- Crucial insight into research priorities

PLRH aims to provide resources, expertise & infrastructure to develop patients' own research ideas into high-quality clinical trials

- Working in partnership, 'lived experience' improves research credibility, reliability & implementation
- No specific research focus: ethos to develop all technically feasible proposals

Research

Sanger Institute—Wellcome Genome Campus - 5th March

Allison was invited by CRDN as a thank you for submitting our patient journey poster at their last summit; here's her report...

Allison had a fantastic day at the Wellcome Genome Campus Sanger Institute, Cambridge learning about the latest in human genome sequencing including Deciphering Developmental Disorders (DDD) project and the 100,000 Genomes Project. It was an insightful presentation and tour seeing the DNA sequencing centre where the magic happens! Unfortunately for those with ring chromosomes like our families, they still can't detect ring chromosomes with Whole Genome Sequencing (WGS) using this fantastic technology YET, but it's on their 'to do' list...our aim is to highlight this as a matter of increased priority

To that end Allison spoke with Dr Richard Scott, Clinical Lead for Rare Disease, 100,000 Genomes Project, Genomics England, discussing ideas for improvng diagnostic rates for ring chromosome disorders across England. Dr Scott recognised that the rate of diagnosis of r(20) shouldn't be in decline, which is potentially what we are witnessing.

A number of initiatives are being proposed by NHS England including standardising and centralising genetic testing (currently it's too widespread), as well as redesigning genetic test systems/processes and request forms/ negative test result further suggested tests.

The update to the Genomic Test Directory is expected to be in place by the end 2018 . Discussions with expert groups will commence early summer '18 in regards how this will be approached and what the requirements are. Dr Scott suggested that we should contact Genomics England to see how we can ensure r(20) syndrome is included in the Directory.

Opportunities arising out of the new initiatives proposed may include the logging of cases of each rare disease patient identified, which could help with epidemiology and/or patient database recording in future.

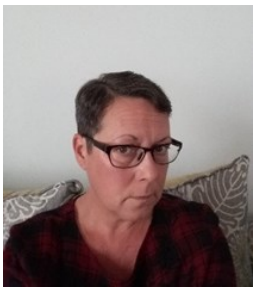
Allison also took the opportunity to ask if NHS England are aware of the need to test for 100 cells for r(20)? Dr Scott was aware himself, however given the current non-standard approach to testing, this may not be followed in all centres.

Paediatricians/neurologists need to know the appropriate techniques to use to diagnose a ring chromosome disorder in epilepsy, in order that they request appropriate testing.

Allison is aware from supporting our families that many cases of r(20) have been diagnosed by chance, including her own son, David — hopefully our voice will bring about change as Allison highlights this in her role as a patient advocate for EpiCARE— getting the message out across Europe.

It is vital that health care professionals are able to recognise the Signs and Symptoms of r(20) to ensure patients don't remain undiagnosed or under reported and get the appropriate care they need.

Our Volunteers



We are very pleased to welcome on-board a new member of the team—Kim, our Fundraising Officer. We've asked Kim to work alongside Gemma, to increase our capacity in submitting grant funding applications.

Initially we've asked Kim to focus on trying to secure us funding to support our everyday running costs i.e. the monies that we need simply to keep the charity afloat and to support activities such as hosting an r(20) stand and attending events to continue to raise awareness. We also want to increase our social media presence, vital in today's world to be heard and we'd like more tools to be able to support member fundraising more—as you're doing an incredible job!

I'll let Kim introduce herself...

My name is Kim Alliston, I am married to Marc and we have one daughter, Sophie, who is all grown up and flown the nest!

I am an experienced and successful Bid Writer, until recently, I worked for a local Essex children's charity, and in those 9 years I raised in excess of £5 million pounds for them.

I am now working as a full time, independent, freelance Bid Writer, (Active Fundraising Solutions) and my aim is to continue to help charities raise vital funds to sustain the services they offer, and look forward to supporting smaller charities and voluntary organisations to "Flourish and Grow"

Some of my past and present clients include: Braintree Mencap, Braintree Youth Project, BENS Homeless Shelter, Essex Dementia Care and Essex Respite & Care Association.

I have worked for many years networking with other small charities and Not for Profit Organisations and I am looking forward to working with Ring20 Research and Support UK and to try and secure some funding to support the vital work they do.

STOP PRESS!....£5,000 grant from Souter

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Events

RARE DISEASE DAY 2018
28 FEBRUARY
Theme: Research

Rare Disease Day—Birmingham

Dawn Gray and Jackie Aley attended this event at Birmingham Children's Hospital. Here's what Dawn had to say about the day...

It was a great day even though it got cut short due to the weather. I met a young man who was studying Learning Difficulties at Stoke. I told him about us. Another man sat by me who suffers with no immune system. He has built up a group over the years like we have done. I still find it hard to believe we are so ultra-rare.

The Star Centre is due to open in Birmingham this year. Its an amazing place, not like a hospital, where families can get together with rare disease. Nine consulting rooms, adult changing rooms, one appointment under 1 roof. Up to age 18 so not possible for my son Callum.

So many interesting talks: Sarah Lipett is an author and musician; she told us of her rare disease Moya Moya and has a book coming out soon. Her art work of cartoon characters was brilliant and illustrations were shown through her talk; it made an understanding of what it is like being the patient and seeing it from a child's view. She did a lot of drawing when ill in hospital.— such talent. I have emailed her as she wanted feedback on her talk. I also told her about our group.

Dr Lucy McKay is involved in rare diseases with medical students. She knew about our families conference. Hurray we are getting recognised! I have emailed her and sent her one of our DVD's.

Miriam Al-Attar spoke of one of her patients with a rare disease called TRAPPED. Very sad when they could not get funding for medications but they have now through her help.

A phrase from one talk quoted ***"Nothing about us without us".*** I thought it was good because if we didn't have r20, or talk about it, nothing would be known about it.

Jackie was so touched by this day. She said it makes you want to say something.

Just met some medical students with an interest in R20. Looking to offer their services in some capacity. One of the students previously worked with Dr Zuberi for her work experience so will be making contact with him.

RCPCH conference #RCPCH18

Vanda Clunas, Veronique Ford and Chris Sullivan hosted a stand for us at this 3-day event. Here's what Veronique had to say...

I found the RCPCH Conference very informative and interesting as this was my first time attending. There were many professional people to chat and engage with and all seemed very interested in Ring 20. We managed to get details of a few people who were interested in getting more information and hopefully get involved with Ring 20 in the future. This was very encouraging and made the day worthwhile and feel Ring Chromosome 20 is getting noticed more and more.

English Parliament—Westminster

Allison attended the Rare Disease Day event at the Houses of Parliament on 28th February. The event was hosted by Rt Hon Dame Cheryl Gillan MP, and attended by a variety of patients, parliamentarians and other stakeholders. During the reception there were speeches given by: Parliamentary Under Secretary of State for Health and Social Care, Steve Brine MP, Dr Jayne Spink, and Rt Hon Dame Cheryl Gillan. The event featured the launch of the 'Understanding Children and Young People's Experiences' report by Rare Disease UK, the first report of this kind, featuring the voice of children and young people.

Scottish Parliament—Holyrood

Claire Sullivan attended the Rare Disease Day event at Holyrood on the 20th February. She says: Having had the privilege to represent Ring 20 Research & Support UK at the Scottish Rare disease reception recently, there was one key message that I've taken away from the event that I'd like to share with you all.

On reflection of the event, it's astounding just how far Don and Allison have truly put R20 on the map, whilst continuing to support member families along the way!

Approximately 50% of rare diseases do not have a disease specific foundation supporting or researching their rare disease. Whilst there's so much more for all of us to play our part in, it really is inspiring to see more and more member families offering their time and support, combined we will have a much bigger impact.

The presentation from the event that I'd like to share with you is Rebecca's story, one which accurately describes the challenges of living with a rare disease and establishing a diagnosis.

We must all continue to work together to #raiseawareness and by doing this, we will make a difference !

'I believe you'

https://www.raredisease.org.uk/news-events/ews/all-too-often-the-person-is-lost-behind-the-diagnosis/

Just met Dr Hannah Mshelbwola from South Staffordshire who was interested in Ring20 . She thinks she has a patient with the condition and looking for more info. Getting Ring20 known in the medical world

Events

UK Genetic Disorders Leadership Symposium

A well rounded and comprehensive programme with some great networking opportunities at this event attended by our Chair.
Most notable was Susan Passmore CEO of Prader-Willi Syndrome who was actively engaged in launching a new initiative which we amongst many others have now added our support to. Her campaign highlights the poor provision and deployment of resources for educational health and care plans for young people in the UK. Commonly referred to as Support for special educational needs and disability (SEND) the 2001 code still applies for those who have a SEN statement under part 4 of the Education Act 1996, rather than an education, health and care (EHC) plan

An education, health and care (EHC) plan is for children and young people aged up to 25 who need more support than is available through special educational needs support. EHC plans identify educational, health and social needs and set out the additional support to meet those needs.

Susan has been representing to the Education Select Committee, on behalf of a growing number of organisations of which we are one. The campaign is focussed on the need to secure the right support at the right time and in the right place within all educational settings for children requiring additional support.

When she spoke at the Symposium, the goal was to bring the dreadful state of SEND, and in particular, the poor implementation of the Children and Families Act 2014 to the notice of decision makers and to call for a review.

Part of the initiative included a government petition calling for a review, when the Education Select Committee announced an Inquiry into SEND. The initiative is called 'Gideons Charter and a survey about SEND provision addressing the questions that the Inquiry is looking at, are being included in order to quote facts. You will probably not be surprised by the results:

86% of children have an EHC Plan
52% do not believe that the plan specifies all the additional support that their child needs
63% are not receiving all the support specified in the plan
37% found the transition from a statement to an EHC Plan very difficult and 24% found it fairly difficult
35% have not completed the transition although this is well beyond the timescale
69% believe their son or daughter has been refused support through lack of funding
89% have received no support for their 19 – 25 year old to work towards independent living
86% have received no support for their 19 – 25 year old to transition into adult services
82% have received no support for their 19 – 25 year old to access a college
75% of 19 – 25 year olds were not in college
94% have received no support for their 19 – 25 year old to access an apprenticeship
100% of 19 – 25 year olds do not have an apprenticeship
83% of 19 – 25 year olds have received no support at all to access work
100% of 19 – 25 year olds do not have either full or part time paid work

This represents a small but significant opportunity to highlight the needs of many families with rare diseases and individuals such as those affected by r(20). It also demonstrates the power of collaboration within groups such as Genetic Disorders UK, Global Genes and we hope to keep you updated on future progress.
Don also had the opportunity to find about more about future fundraising opportunities through The BIG Give and the UK's biggest online matching fundraising campaign, the Christmas Challenge – keep your eyes open for more exciting news on this in the Autumn.

Listen to Susan's presentation here: http://www.geneticdisordersuk.org/static/media/up/GDLS2018_SusanPassmore_Gideon%E2%80%99sCharter.mp4

Does your child have an EHC Plan and does it work well for them?

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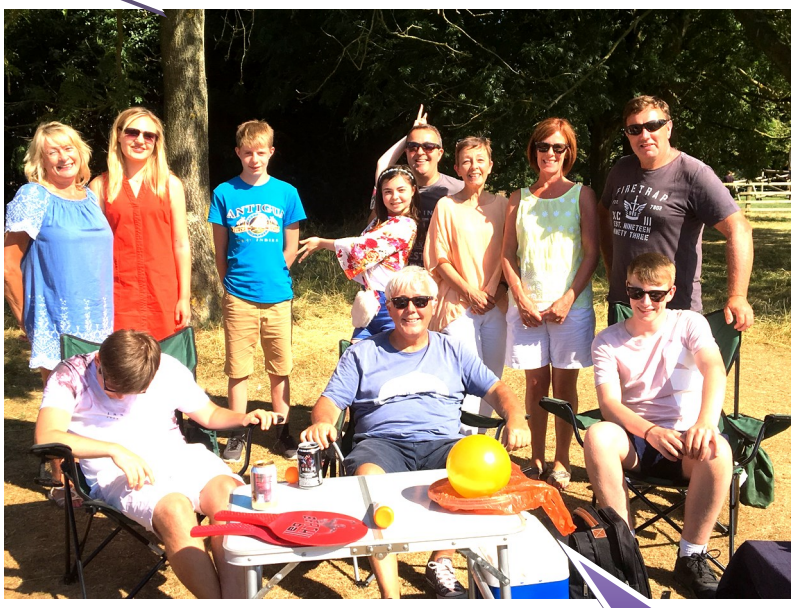
r(20) Family Picnic

Allison hopefully the first of many more times to come! Memories made...



Jane McHugh
So so lovely to meet these people at long last. We had a **lovely time** .thank you Dawn Gray for arranging the picnic today x

Jackie Aley

Fabulous day 😀 xx

Dawn Gray Great to see you all today. Thank you so much for coming. Xx

Jackie Aley So lovely to see you all, amazing get together and hopefully see you all soon xxx

Next informal families meet: 13th Sept 2018— Brussels

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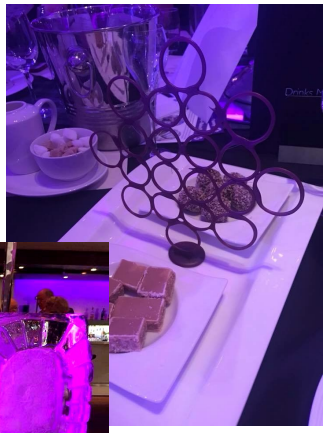
R(20) Gala Ball - TRULY INSPIRATIONAL!

Feature article

Jessica's Ring20 Gala ball, where many Ring20 families were able to join us at the Radisson Blu Hotel in Glasgow and was a hugely successful night. Attended by Prof Sameer Zuberi, our guest speaker for the evening, the event was both hugely emotional and impactful.

We're hoping this has inspired some other Ring20 families to consider taking the baton for a future high profile event..... watch this space!



The Sullivan fundraising total £33,734 Thank You!

Sullivan Fundraising

SSE raffle

Civic Centre coffee morning and raffle £590

Airdrie, Coatbridge & District

West End Bar Airdrie Tartan

Having attended the Ring20 conference last summer, we were inspired to find the time and to set about an ambitious 12 month fundraising journey, Our initial target was to raise £20,000. We're delighted to announce that we have exceeded our initial fundraising target and so far have raised an astounding £33,734. This has only been possible through the generosity of everyone who has been involved along the way. Every donation makes a difference and we can't thank everyone enough.

Whist we've now carried out numerous events, a special mention must go to our hugely successful Race Night and Gala Ball. Our Race Night in February brought in a total of £4,100, completely blowing our expectations.

This was then followed by Jessica's Ring20 Gala Ball in April 2018 where a staggering £16,100 was raised. The ball was also an amazing platform to raise awareness of the Ring20 condition and to hopefully encourage others to further fundraise to progress research.

All of our events have helped to significantly raise awareness of Ring20 within our local community. Throughout our fundraising journey, we've pledged to our wider community to support with ongoing fundraising.

Reflecting on our year of fundraising, we left the Ring20 annual conference at Goodison Park in the summer of 2017 as a family knowing we have the medical professionals, we have the strategy,

and we have the leadership to make a difference, all we need now is the funds to make it happen,' It's been so reassuring to see, that as a community of families, we are beginning to make this a reality...but we need ALL member families to help on this journey, no matter how big or small you are able to contribute, on an ongoing basis.

In addition to our fundraising events, as a family, we have also found the time to support and provide a presence at the Holyrood Rare Disease Day and then most recently RCPCH day in Glasgow, where awareness and wider networks were further developed. This combined, can only help put Ring20 Chromosome Syndrome firmly within the medical arena.

Forthcoming Events::

- Ben Nevis Climb - 1st Sept '18
- Loch Lomond Swim—25th Sept '18

Future fundraising events now lined up and will bring a close to our current fundraising campaign:

- Craig Burns, a close family friend was inspired by our story at the Gala Ball and has signed up to swim the Great Scottish 10k Swim on the shores of Loch Lomond on Saturday, 25th August.
- A number of volunteers are climbing Ben Nevis on Saturday, 1st September, this will be a growling challenge for a group of us amateurs.
- And finally, just by raising awareness, another well established local fundraising forum has recognised our cause and has confirmed that they will donate a contribution from their Speakers Night due to take place in November 2018.

We are already getting our thinking caps on for future events and considering the kilt walk next Summer.

Claire and Chris Sullivan

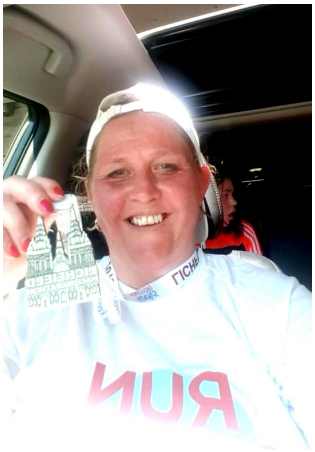
Other Fundraising

Sangster family—Open House

The family held an open house on Monday 26th March selling cakes for epilepsy awareness day. They did really well raising £375 and donated the money that they raised to the Ring 20 Research and Support UK CIO.

Could you fundraise to help towards our Research Fund Campaign?

Marie Ward—Lichfield Half Marathon



We are super proud of mum Marie Ward raising over £1,143 running her 1st ever event - the Lichfield half marathon no less!

Marie says:
That was the hardest thing I have ever done, walked more than I would have liked but still a respectable 3hrs. Thank you for all your support, donations and words of encouragement - don't know about smashed it but certainly finished it xxx

Gray family—Easter Egg Raffle/Sue's Cards

Callum seems to want to do this every year now!! We sold the raffle tickets then the deal I had with Callum was he folded all the tickets and did the draw. This kept him occupied!!

He really enjoys doing this. I can say it is an easy way to raise money. Buy an Easter egg and some raffle tickets and sell them to friends, family, neighbours.

We raised £229—all amounts help.

Allison gave me some hand made cards made by Sue I have them in a basket at work so I can sell them for r(20). In April there was £27 made from the cards. I am still selling them and there are more funds to pay in. **Thank you to Sue for sending us these lovely cards.**

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Charity of the Year—Tai Tarian

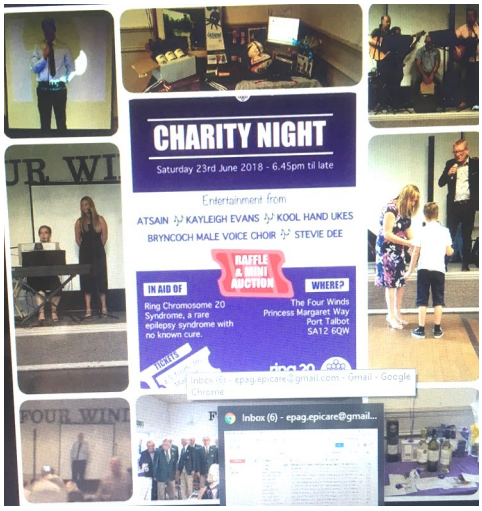
Mark & Jo – Ring20 Research & Support Fundraising 2018

After attending the first ever Ring 20 conference/weekend in Liverpool August 2017 and having the opportunity to meet other Ring20 families, hearing from all the medical professionals, gave comfort to us that we were not alone. We felt hopeful and felt compelled to speak out about the devastating disease. Hoping that research could be undertaken for a better future.

So far 2018 has been a positive year for us Cumberlin-Edwards family. An amazing opportunity arose to raise awareness of Ring20 when colleagues and friends supported our cause and chose Ring 20 Research and Support UK as the Annual Charity 2018 at our work place Tai Tarian.

With our committed volunteers, the start of our fund raising journey at Tai Tarian began Tuesday 13th February 2018 – Pancake Day, selling a mixture of bakers pancakes with lots of different toppings including fresh fruit, squirty cream, syrup, Nutella etc. 150 pancakes went on sale at 10am sold out by 10.40am. Alongside a raffle, we raised £272.

As Ring 20 is ultra-rare, and under diagnosed, it was important for us to highlight Rare Disease Day so on Wednesday 28th February this year we decided on a well-loved cake sale. Luckily we have absolutely fantastic cake bakers at Tai Tarian who gave absolutely amazing donations.



On Sunday 8th April two of our colleagues Simon Jones and Paul Davies at Tai Tarian felt so inspired by James's story they decided to run the Manchester Marathon. We were immensely proud that they did this for us and as Simon's first marathon he was an inspiration to us.

At Easter weekend over 40 people joined us on a delicious Charity curry night at the Panshee Restaurant in Skewen on Maundy Thursday. It was £10 for a starter and a main course. Half of the total cost of the bill was returned to the Charity. A thoroughly enjoyable night.

Since January we have had a driving force team of family members including Steve, Gill, Les, Lana, Daniel, Katie, Emma & Jaqueline - planning and organising a charity music night. On Saturday 23rd June at The Four Winds, Port Talbot we had an evening of fantastic non-stop live music. This included the Bryncoch Male Voice Choir, duo female singers Kalex, a group Atsain and singer Stevie Dee. Stevie Dee had also made a CD of his songs which he had been selling and made over £200. We had a raffle and an auction of brilliant donated gifts. To highlight the horrors of Ring 20 and the purpose of our fundraising we showed a video of James' story, made by my brother. It was an emotional but enjoyable evening at the same time. We had a lot of support and positive feedback which was encouraging. We raised an amazing £4,031.60.

Back at Tai Tarian on Friday 29th June the Annual Summer BBQ - organised by Caroline Davies and her team of helpers - took place on a scorching Summers day. With fantastic variety of food from burgers, sausages, onions, cheese and salad. There were even fruit kebabs and lolly's. Very well organised and a fantastic turn out. Caroline also organises and maintains the work's tuck shop and donates the money to charity on a monthly basis as well.

With all this hot weather in July and to celebrate the tennis at Wimbledon, on 12th July our committed team of volunteers - Carol Maunder, Hannah Phillips & Carly Lewis who have done a fantastic job with all their organising the events - decided strawberries and ice cream, along with scones with jam and cream and cake sale was a must.

We are extremely grateful to be supported by James' family and friends who are in the Port Talbot Fire Cadets and after hearing about our fund raising decided to donate £200.00 of their subs money, which is so generous of them.

We are looking forward to arranging some more fundraising ideas for the coming months at Tai Tarian. For our next fundraiser my manager, Mark Windos will be completing a Bog Snorkelling challenge on 26th August and will be doing this in aid of Ring 20 Research & Support UK. We look forward to supporting him.

We would like to say a huge thanks to our fantastic family and friends and colleagues for all their support.

We are looking forward to presenting the cheque at the end of the year to Ring 20 Research and Support UK.

Charity of the Year—Eastwood Golf Club

Press Cuttings

My name is Veronique and I am the parent of Cara Ford who has Ring Chromosome 20 Syndrome.

Cara and I decided we wanted to arrange a fundraiser for Ring 20 and raise awareness of the condition. We agreed to organise a golf charity day at Eastwood Golf Club in Glasgow, where I am a member. The golf club generously donated the course and club house on Sunday 24th June 2018.

The days format consisted of a Texas scramble and we managed to secure 22 teams of four players. We started the day by selling raffle tickets and having an auction, which people gave generously. At The halfway house we had a BBQ which the burgers were donated by Ians Breakfast Club and he cooked them all with support from my friend Denise Murray, there was also home-baking donated by various members.

On returning to the club house I delivered a speech explaining what Ring 20 was and what Ring 20 Research & Support UK was trying to achieve. I also gave a little information on what we had been through ourselves as a family and gave out some leaflets.

The response was fantastic people felt they were well informed and listened with great interest.

This event would not have been possible without the generous donations of all my family, friends and of course Christine Mcconnell who was so supportive from start to finish.

In the end we managed to raise £3,210.00 which was incredible, I am so appreciative of all the support and generosity of everyone and hope this helps towards achieving our goals at Ring 20 Research & Support UK.

NEWS

Health

Teeing off to help research into illness

The family and friends of a young Newton Mearns woman who suffers from a rare medical condition have rallied round to support a fund-raiser this weekend.

Cara Ford, 23, was diagnosed at the age of six with Ring Chromosome 20, a chromosome disorder which causes seizures that often misdiagnosed.

The condition is so rare that there are only 138 known cases worldwide and just three families affected in Scotland. But her mother Veronique says that she suspects there are more cases which are currently going undiagnosed and being treated with drugs which may be ineffective.

She said: "We just trying to raise awareness of the condition, as I believe there are more people out there although there are only 138 cases world-wide. Cara was diagnosed at six but before that it was thought that it was just epilepsy she had although there were other suggestions and she can't now get a job as she has seizures daily."

Veronique has also organised a golf tournament, with barbecue and raffle, at Eastwood Golf Club this Sunday, with funds going directly to Ring20 Research and Support UK to help finance genetic research. Further information about the condition and how to donate can be found at ringaoresearchsupport.co.uk

WELL DONE

Could you nominate us as a 'Charity of the Year' to a company you know?

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EpiCARE ERN update

If you want to know more about EpiCARE, check out their website here:

https://epi-care.eu/

As announced at our Families Conference last year, Allison is now also volunteering as a European Patient Advocate Representative (EPAG rep) for **EpiCARE**—the new **European Reference Network (ERN) for rare and complex epilepsies.**

This new role has kept Allison rather busy in the last 6 months, here's why:

The role as an EPAG representative stemmed from a recommendation from Prof Helen Cross to become involved in helping a wider number of families affected by rare and complex epilepsies.

The newly formed EPAG team in 2017, comprised approximately 12 representatives from across Europe led by Isabella Brambilla from Dravet Italia ONLUS and Emma Williams MBE from Matthew's Friends. Meetings via Skype or WebEx call takes place every 2 months to discuss progress and actions for the group.

Allison attended the EpiCARE AGM in London, in March with representatives from all of the 28 Epicare participating centres across Europe, together with various health care professionals who are leading and/or participating in one or more of the 16 Work Packages (WP). It was an incredibly insightful meeting hearing from each WP lead present their progress over the initial 12 months of EpiCARE and their objectives for the coming year. It was clear that as an EPAG we weren't really very involved initially — so we have since worked hard to change that. A lead and deputy EPAG rep were appointed to each WP, connecting them with their respective WP lead, in order to bring the patient and family voice to the table for future meetings and discussions. Allison is lead (or deputy lead) EPAG rep for the following WP's:

Laboratory diagnostics, Neuropathology, Dissemination, eDatabase and Research.

Emma Williams had to step away from her role as deputy coordinator for our EPAG earlier this year, as she was already extremely busy in her role as the WP lead for dietary therapies. As a consequence yours truly was asked to step up.

As deputy EPAG lead, Allison now helps organise and facilitates the bi-monthly meetings for EpiCARE EPAG. Her role also includes contributing at the monthly EPAG coordinators meetings, as well as attending EURORDIS events such as in Vienna (May) as a EURORDIS volunteer. At this event EPAG reps from each of the 24 different ERN's came together to discuss common goals and priorities across all ERN's. The next face-to-face event is in Brussels, when the EPAG coordinators will work as a group to further ratify findings from Vienna and formulate ways forward to put these into practice.

Allison has a seat on the Steering Committee for EpiCARE which meets monthly to discuss progress and Is invited to influence decisions by representing the patient's perspective.

What does this role do to help patients with r (20) ?

Certainly our presence within EpiCARE and the EPAG group, is getting r(20) noticed. Whilst we represent the views of all rare and complex epilepsy patients, our unique representation of some of the ultra-rare epilepsies, like ours or those even less common, presents a voice for those with no patient registries/databases, no clinical trials, no guidelines or recommended treatments and even in many cases no patient support group. This is where we believe EpiCARE has a role to play, to bring about equity of knowledge and treatment for all patients.

So what has EpiCARE delivered so far?

One of the really exciting developments has been the launch of the CPMS (Clinical Patient Management System). Clinicians from across Europe meet fortnightly at 'virtual' meetings where they discuss complex patient cases with a group of experts . The system allows them to share information on the patient in an anonymised fashion, such as test results, EEG, MRI scans etc, so they can discuss best practice in treating those patients. The system introduces cross-border care, where location has no boundaries—if the experts are not 'in country', then that is no longer a barrier to optimum care.

And what is next for EpiCARE?

Work gathers apace in the Targeted Medical Therapies Work Package and working parties are underway looking into delivering new or improved guidelines for the treatment of rare and complex epilepsy patients. There are also proposed updates on each rare epilepsy as listed on Orphanet—the trusted site for information on all rare diseases.

Allison is currently working very closely with the Prof Lieven Lagae on the eDatabase Work Package, offering her services to help project manage and deliver requirements for a European-wide patient registry that will inform future research, clinical trials and/or epidemiological studies.

What about EPAG?

EPAG are working on presenting Patient Priorities to Prof Cross and the EpiCARE Steering Committee, assimilated from a recent survey (if you recall we asked you all to contribute?). Our aim is to use these priorities to set objectives for EpiCARE, against which they can be measured going forward. Matt Bolz-Johnson from EURORDIS is helping our EPAG formulate these objectives, the idea being that patients and patient families should have a say in what makes EpiCARE a success. After all, EpiCARE has been created ultimately for the benefit of patients, not the health care professionals!

EPAG coordinators meeting Vienna

EpiCARE AGM—London 2018

Epihunter: how a bright idea turned into a solution for absence seizure detection!

Interview with Tim Buckinx, founder of Epihunter and dad of Daan.

Can you share something about your family and yourself?

We are a family of four: my wife Cindy, our oldest son Jeroen, Daan who's now 13 years old and myself. My wife is a teacher and prior to founding Epihunter I helped drive forward a global digital initiative at Bose. Daan has always been very curious and adventurous: always climbing in trees and chatting all the time. In March 2011 when Daan was 6 years old, he suddenly started having epileptic (generalized motor) seizures. They occurred every 10 minutes, day and night. A few months later he was diagnosed with Ring20. After many changing cocktails of medication, he now 'only' suffers from absence seizures every day.

How does Daan feel about the absence seizures?

About three years ago, during a father-son bedtime conversation, a "people are always angry with me" theme came up. At first I couldn't quite understand why he felt that way until one school morning I was helping Daan getting ready for school. That day I reacted stressed after asking him three times to put on his clothes and he responded: "Why are you angry at me?"

Then I understood: What I had repeated three times, Daan had only heard once. I realized that this situation happened everywhere, even at school and this lead to big frustrations. A few evenings later, Daan gave me the solution: "Papa, you're in digital. Can you make a light that turns on when my brain switches off?"

Indeed a bright idea!

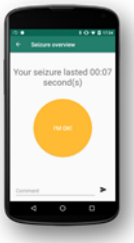
Yes, that straightforward question made my private and professional life merge into one; I simply had to do something with it. I read about EEG technologies, spoke with medical experts, other parents, teachers, and children with epilepsy. I learned that we all have the same need: objective information about seizure frequency. And I discovered that consumer EEG headsets existed that measure brain activity and are being used for meditation, focus and concentration exercises. A hackathon at the pharma company UCB provided the confirmation I needed to take Daan's idea to the next level. In May 2017 the company was founded and in August 2018 we made our solution available across Europe.

How does it work?

The child wears a thin and comfortable EEG headset. No fluids needed, turn it on, put it on, wait for two beeps and you're ready to go. The Epihunter smartphone app analyses the EEG information it receives from the headset and turns on the smartphone light during an absence seizure or impaired awareness seizure.

Epihunter's nice and thin EEG headset

In the classroom, the teacher knows which information or instructions the child misses and can repeat these later. The child feels better understood and experiences fewer misunderstandings. Parents are now reassured that seizures do not go unnoticed anymore at school. When using Epihunter at home, parents and the child gain more insights on seizure frequency and occurence. And finally parents can visit our secure web portal that provides them with a clear overview of seizure activity.



Screenshot from the Epihunter smartphone app for Android.

Is Epihunter already available?

Yes! Epihunter is now available across Europe. Our subscription plans include an EEG headset, continuous improvements and additional functionalities. Visit our website www.epihunter.com

Link to Daan's story:

https://ring20researchsupport.co.uk/personal-stories/daans-story/

At Epihunter we strongly believe in close collaboration with families living with absence epilepsy. We want to deeply thank all the families in Belgium and the Netherlands who tested Epihunter for many and many hours! You can be very proud: Together we have the potential of making a truly great step forward in research on absence epilepsy.

1 in 3 people with epilepsy

Despite medication, 1 in 3 people with epilepsy still live with unpredictable and uncontrollable seizures. Thanks to Daan's great idea, Epihunter is now available to make epilepsy matter less, starting at moments that matter most.

Our initial focus is to help children, yet Epihunter can be used by people of all ages with absence seizures or impaired awareness seizures. Our company's next milestone is medical certification so Epihunter can also be used to support the neurologist and epileptologist in the diagnosis and optimisation of medical treatment.

Everyone can help with fundraising...

You don't have to host an event or even leave home to raise funds for us...

Do you shop online? Then are you signed up to any of the following:

easy fundraising .org.uk feel good shopping

amazonsmile You shop. Amazon gives.

Are you more of an outdoors person? Then have you got:

fit4Change - this app on your mobile phone raises 5p for every mile you walk, run or cycle. You don't have to be super fit—just use when walking the dog, going to the shops or school run.

How about recycling?

Recycling FOR GOOD CAUSES

Could you give out any leaflets for us e.g. at your local school, workplace, friends/neighbours, or even in clinics or GP surgeries? We can earn money from old watches, unwanted gold, silver or costume jewellery including damaged items and foreign or UK bank notes

Request leaflets from ring20@ring20researchsupport.co.uk

These guys also accept used stamps.

Have unwanted items? Sell them and donate 10-100% to us:

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Or play the **Essex Lottery**—£1 for a chance to win every week! We receive 50% of ticket sales when you support us: https://www.essexlottery.co.uk/support/ring20-research-and-support-uk-cio

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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.

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

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