Issue 8

Apr 2020

Registered Charity Number 1165651 (England and Wales) SC048893 (Scotland)

Newsletter

ring 20
research & support uk

Please note our phone number has changed to:
+44 7385 292797

We are excited to bring you this newsletter, which I hope you'll agree is jam-packed full of good news!

In fact, so good, we had it printed so you can share it with all your family and friends.

INSIDE THIS ISSUE:

To Research and beyond...

At the time of writing we find ourselves living in unprecedented times, facing the challenges of lockdown due to COVID-19 and actively practicing social distancing. Our lives are all being disrupted with children home from school, parents working from home or being laid off, loved ones isolating and/or shielding from this devastating disease and food stores being depleted. Many of us face challenges every day, living with someone with r(20) and coronavirus simply adds to the burdens. Our hearts and thoughts go out to you all. As a patient support group we are here for you, so please pick up the phone, send an email or engage on social media. Remember, you are not alone in this crisis. We hope this newsletter brings you Some-thing more positive to focus on....we've been incredibly busy at Ring20 and we hope that you've been able to follow our progress through our regular enews and social media channels. Research has been a key objective for us in the past 12 months and we have lots to report back on, not least our Research Fund Campaign. In early 2019 we conducted our surveys and workshop into the use of ketogenic dietary therapy in r(20), which has now been published in Epilepsia — our 1st ever medical publication! We're starting to initiate some real interest to work together with other organisations on research and one of the areas we are exploring is the challenge around diagnosis and finding out how 'rare' r(20) really is. We're working to improve diagnostic rates through raising awareness as well as influencing new developments in next generation sequencing. Increasing diagnoses not only ensures we are a louder patient voice, but also allows Ring20 to increase the reach of our support services to even more families in need. And of course, makes us more interesting to researchers. Speaking of support, it was fabulous to see so many families at our comic workshop in November. We're currently working with Rossie and our Youth Group to produce the final comics— free copies will be issued to every one of our member families. Our forthcoming r(20) families conference in Brighton (9-11th Oct), is still in the pipeline although we will consider government/ travel guidance to determine if indeed we should postpone.

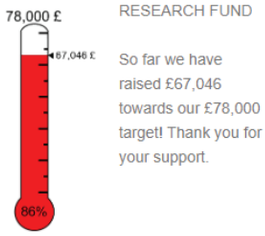
We couldn't bring you all of this without the help of a super team of volunteers – but with Don currently on sabbatical and losing Nik you'll be hearing a lot more from Dave, Audrey and me in the coming months.

Here's to Ring20 in 2020....! *Allison*

Research Fund/Funding update

Just look at our totaliser! We have much cause to celebrate with the launch of our first ever research study funded by you, our members.

We know you have many initial questions about what this research project is all about and so we've tried to answer these inside.



theBigGive CHRISTMAS CHALLENGE 3—10 DECEMBER

JEANS FOR GENES DAY

Kudos to you, we raised an amazing £9,500 in the Big Give Campaign before Christmas. Together with a £5,000 grant from Jeans for Genes, its now possible to bring you our next r(20) Families Conference in 2020.

HUGE THANKS!

Introducing our Medical Advisory Board

I am really pleased to introduce to you the new Medical Advisory Board for Ring20 Research and Support UK CIO. The Board provides advice to the Trustees on research into r(20) syndrome applications and proposals, requests to the Charity for collaboration, identifying opportunities for research and sharing clinical developments (new research, new treatment options and newly published guidelines) relevant to Research and Support UK CIO.

The Board is chaired by myself, Dr Sophia Varadkar, Medical Advisor to the Charity.

I am delighted that a group of experienced and senior clinicians with expertise in the field of neurology and neuroscience across children, young people and adults, have also agreed to join me.

They are:

Dr Dulmini Birkett, Consultant Paediatrician with Expertise in epilepsy and Neurodisability,

Dr Archana Desurkar, Consultant Paediatric Neurologist

Dr Ailsa McLellan, Consultant Paediatric Neurologist ....and;

Dr Arjune Sen, Consultant Neurologist

We very much look forward to working with the Trustees and the charity and to meeting you at engagement events in the near future.

Find out more about our board members here: https://ring20researchsupport.co.uk/about-us/medical-advisory-board/

DID YOU KNOW... ?

we have a MEDICAL ADVISORY BOARD at Ring20

Dr Archana Desurkar

Dr Ailsa McLellan

Dr Arjune Sen

Dr Dulmini Birkett

And Our Volunteers...

Dave Soderquest—Events/Project Manager

Dave joined our team last October, just before our Families Workshop event in Birmingham. He agreed to come along and meet with our families and healthcare professionals to really get thrown in at the deep end and learn first hand about the sorts of things we do as a charity. Dave was immediately put to use helping to coordinate our hugely successful Big Give Christmas Challenge Campaign and is now working on helping to produce our r(20) comic books, spearheading our r(20) Youth Project. Dave's professional skills means he keeps us all organised and now chairs our regular leadership team calls to keep us focused.

A little bit of background about Dave...

Dave started his professional career with a degree in Chemical and Polymer Engineering from Loughborough University and moved into Information Technology as companies started to introduce computer systems to their factories and offices. This led to a very interesting and enjoyable career of developing, managing and supporting business systems and technology in life sciences research and development in pharmaceuticals, the NHS and charities as a Project Manager, Business Analyst and Operations Manager.

His favourite project was the implementation of a High Performance Compute (HPC) facility to support genome analysis for the Cambridge Research Institute - a joint development between Cambridge University, Addenbrooke's Hospital and Cancer Research UK. The technology was easy – arranging to keep the facility cooled when all the systems started data analysis was more interesting.

Now retired (sort of) he has taken on a role of volunteer Project / Events Manager in Ring 20, though can still be seen, on occasions, cycling though the wilds of Essex and has a keen fascination for music, history and science fiction.

Audrey Everiss—Administration Officer

A new year and a new volunteer, Audrey joined our team in January 2020. Audrey applied for the role of Administration Officer, which in our small organisation means being responsible for 'a little bit of everything!'. Undaunted by the long list of tasks that she might have to turn her hand to, Audrey has embraced the challenge taking on a key role as your contact for all future fundraising events and enquiries. Importantly Audrey has taken onboard our bookkeeping to keep our accounts in order for our Treasurer, Kim, as well as helping us to organise Brighton 2020.

As you can see Audrey and Dave have quite different skillsets, both of whom are valuable assets to our charity, and we couldn't operate without either of them.

A little bit of background about Audrey...

Originally from Edinburgh, Audrey has spent much of her working life in administration jobs, employed mostly in local government and with he Ministry of Defence. After a severe case of "empty-nest syndrome" when her son left home to join the Army, she decided on a career change and went to university. Five years later, with a degree in Biological and Health Sciences and a Post-Graduate Certificate in Education completed, she began teaching.

Working for Edinburgh College, she started out teaching English, Maths and IT as part of the college's Literacy and Numeracy Support Team. She then joined their Mental Health Outreach Team and spent the last twelve years teaching the same subjects in community bases in the city to people with mental health problems. She retired recently and moved to Colchester to be close to her son and his family. Although new to the voluntary sector, Audrey is very keen to offer her skills to Ring20 at what appears to be a very busy and exciting time in its journey.

Meet key members of our Ring20 team...

you'll be hearing a lot more from Dave and Audrey in future

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Natural History & Biomarker Study

As we go to press we acknowledge that there will be an inevitable delay in starting this research due to the outbreak of COVID-19

We hope to bring you news in the not too distant future of when we can get this up and running...

We are really excited to be funding our very first research study into r(20) syndrome, which will be starting very soon, made possible through all of your fantastic fundraising efforts since 2017. But what's the research all about and what do you need to know? Here we try to answer some of your questions...

What is the name of the study?

The study is entitled: "The Ring Chromosome 20 Natural History & Biomarker Study - a model for understanding learning disability in the epilepsies".

Where and by whom will the study be conducted?

The study will be led by Prof Sameer Zuberi, a Consultant Paediatric Neurologist from Glasgow University Hospital, together with his colleagues: Dr Dorris (Consultant Neuropsychologist), Dr Forbes (Consultant Neuroradiologist) and Dr Brunklaus (Consultant Paediatric Neurologist)

Professor Sameer Zuberi

What is a Natural History Study and what is a Biomarkers and why are they important?

In medicine, a natural history study is a study that follows a group of people over time who have a specific medical condition or disease. A natural history study collects health information over time to understand how the medical condition or disease develops and to give insight into how it might be treated. A natural history study is often submitted when applying to the FDA or other regulatory agency as a baseline, to show the disease course for untreated patients.

A biomarker is a biological molecule found in blood, other body fluids, or tissues that is a sign of a normal or abnormal process, or of a condition or disease. A biomarker may be used to see how well the body responds to a treatment for a disease or condition. Biomarkers are important because Biomarkers are characteristics of the body that you can measure. So, your blood pressure is actually a biomarker. Biomarkers are very important to medicine in general. We're all used to going to the doctor and getting all our test results, right, and even imaging — x-ray results or CAT scans — those are biomarkers that tell how the body's doing, and they're measurable. Biomarkers are integral to drug development; they're really critical, because we need to measure the effects of investigational drugs on people during the clinical trials. And the way we do that is to look at their effect on biomarkers. And so it's really important that we have a wide range of biomarkers that can measure everything we want to know about the effect of the investigational drug in people.

What are the main aims of the study?

There are 4 key aims of the study:

- To collect data on all aspects of r(20) including seizure onset, seizure types, response to medication, cognition, behaviour and EEG;
- To measure the cognitive and behavioural profile of people with r(20);
- To identify biomarkers for the cognitive and behavioural impairment which can be used as outcome measures in future trials of therapy;
- To develop and curate an international registry of Ring Chromosome 20 patients to inform future research studies.

What is a patient registry and why do we need one?

A patient registry simply stores an individual's personal information and their medical history, and registries are usually disease specific. This allows researchers to more accurately estimate both the prevalence and incidence of the disease, and to determine their natural history, providing answers to the above questions. Once this information is known, it can be used to give more accurate advice to patients, and to improve care pathways, which can lead to improved care and life expectancy, even in the absence of a cure. Registries can be used prospectively to quickly identify patients eligible for a clinical trial, or retrospectively to analyse the effectiveness of an intervention.

Who can participate?

We are aiming to recruit 100 individuals with r(20) into the study, across all ages (children and adults), and from a range of countries. This will be an international study, the largest of its kind ever conducted into r(20).

Natural History & Biomarker Study

When will the research start and how long will it last?

We're just waiting on the final contract signatures, but we aim to be up and running in the next few months. The study will run for a period of 2 years.

Do we have all the necessary funding?

Nearly...we still have approx. £9,000 to find in the next 2 years. Can you help us raise these important last funds? The majority of funding has come from our UK families, and yet the study will involve and benefit everyone with r(20) across the globe. We will happily accept donations and/or funds raised from outside the UK; every penny counts, even small donations can make a big difference.

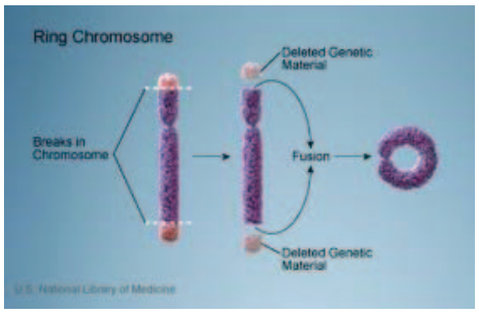
Royal Hospital for Children, Glasgow

What benefits might this study bring?

Patients with r(20) will be followed over a period of time and have their history of living with r(20) formally documented. For the first time we will see how well r(20) responds to different treatments or not, how r(20) changes over time so patient families can have more idea of what to expect as their child grows into adulthood. If we can identify biomarkers they can be used for potential clinical trials to measure treatment response rates and maybe help find a recommended treatment(s) for r(20). The data from our patient registry can be made available (under appropriate consent) to future researchers and we hope that our findings will incite new interest for future research, to seek improved treatments and patient outcomes. We aim to better understand the impact of seizures on behaviour and learning – key problems associated with r(20) – and how they can be best managed. Not only will this benefit those with r(20) but also potentially provide insight to help other complex epilepsies.

How do I sign-up?

As soon as we get started on the study, we will send out details for how you can register to participate. Watch out for this on our social media channels and in our regular enews or targeted email.



References

https://en.wikipedia.org/wiki/Natural_history_study, 2019

https://www.fda.gov/drugs/cder-biomarker-qualification-program/what-are-biomarkers-and-why-are-they-important-transcript (2017, 03 03).

Perdeaux, L. (2014, 04 04). https://www.bhdsyndrome.org/forum/bhd-research-blog/why-are-patient-registries-important/

Report by **Allison Watson**

At Ring20 we believe this study will provide the basic building blocks necessary for future research that have been absent for so long.

A better future with r(20) starts here...

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Research—KDT use in r(20) syndrome

Ring20… leading the way for our patient community

Patient-led research

I had long been intrigued and fascinated by the capacity for the ketogenic diet and its ability to control seizures. Hearing stories from family members and other patient support groups like Matthew's Friends and watching that famous film "First Do No Harm"' starring the American actress Meryl Streep, I was no less curious about how it may relate to ours and similar epilepsy syndromes. After 5 long years of study and graduation in nutritional science and over 15 years of involvement in r(20), I therefore began by writing a research proposal for review by my institution's medical ethics committee at the Centre For Nutrition and Lifestyle Management (CNELM).

Fast forward to March 2018 and a "Think Research" Rare Diseases Patient Day run by the National Institute for Health Research (NIHR) in London. Here I attended a presentation by The Patient Led Research Hub part of University of Cambridge Department of Medicine Clinical Trials Unit.

I was taken by the potential opportunity of us working with them to further develop our research profile and approached them on behalf of our charity which would allow the original part of my research dissertation to be fulfilled. After supplying them with a copy of my research work and methodology behind the dissertation they agreed to undertake a proposal and seek funding to facilitate a workshop and survey. A small grant was secured and after circulating two surveys amongst medical professionals and r(20) families the workshop brought together representatives from these two groups including Dr Archana Desurkar, Dr Thomas Hiemstra, Dr Sophia Varadkar, Emma Williams MBE and four r(20) families in London May 2019.

After reviewing and analysing the results from the surveys and workshop this collaboration and input from medical professionals as well as r(20) families has now been written up.

To summarise my findings:

The Ketogenic Diet (KDT) should be considered as an early intervention treatment option for individuals with r(20) who invariably have drug refractory epilepsy, along adherence to NICE guidelines. Furthermore the current age of r(20) patients (50% 18 years or older) reflects a gap in provision of KDT suggesting immediate need for increased provision of adult KDT services.

In addition, the findings and write up have been further developed to provide a scientific review paper which is awaiting final acceptance in the publication 'Epilepsia'. This publication has been recommended by our medical advisor Dr Sophia Varadkar as being the most widely read relevant publication for neurologists and clinicians. The abstract and review articles are entitled:

"Assessing the Role of Ketogenic Diet in Ring20 Epilepsy: a Patient Led Approach"

You can read the full manuscript here:

https://onlinelibrary.wiley.com/doi/full/10.1002/epi4.12387

I am also deeply grateful to the unstinting and remarkably collaborative support we received from Laura Mader and Dr Thomas Hiemstra of The Patient Led Research Hub part of University of Cambridge throughout the whole project.

Report by **Donald Gordon**

Research—NCARDRS—self-registration

The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) records those people with congenital abnormalities and rare diseases across the whole of England.
This registration service:

- provides a resource for clinicians to support high quality clinical practice
- supports and empowers patients and their carers, by providing information relevant to their disease or disorder
- provides epidemiology and monitoring of the frequency, nature, cause and outcomes of these disorders
- supports all research into congenital anomalies, rare diseases and precision medicine including basic science, cause, prevention, diagnostics, treatment and management
- informs the planning and commissioning of public health and health

We've been talking to the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) about piloting a new self-registration service with them for r(20) patients/patient families. We do hope that many of you will participate to help inform with a view to ultimately improving health services for those living with r(20) in England. If this is successful, then we may reach out to the public health services running similar programs in the devolved nations and perhaps into Europe and beyond?

Why are we doing this?
We found out that our national rare disease registration service holds no record of anyone with r(20) syndrome and we were shocked, but not surprised. A relatively new service in England (and similar services operate in Scotland, Wales and Northern Ireland) GPs and doctors are asked to report patients diagnosed with a rare disease before the age of 5. Herein lies the problem, many r(20) patients aren't diagnosed until after their 5th birthday. We want to get r(20) patients on the map, so that our national health service recognises that they exist and need care.

What's in it for you?
Collecting information helps NCARDRS/Public Health England better understand rare diseases like ours to help make sure that people living with these conditions receive the best possible individual care. The national register is also used for research and planning.

Who is eligible?
The service is available to all our families who are registered and/or treated in England – as they are part of Public Health England.

How can I contact NCARDRS?
To start the process you will need to send an initial e-mail to the NCARDRS team e-mail address (phe.ncardrsrd@nhs.net) requesting to register in relation to this pilot. You will receive a reply with instructions on what to do next. Safe transfer of information is extremely important to the NCARDRS team. General e-mail is not a secure method of data transfer – messages sent in this way will not be encrypted and may be at risk of interception. Because you will be sending your personal information to NCARDRS they want to give you an option to transfer your information securely, and to do this they use an encrypted e-mail system called Egress switch. In future NCARDRS are hoping to be able to offer an online registration system for patients to register with them.

What information will you be asked to provide?
If the patient is reporting themselves NCARDRS will ask for the following information:

- Name
- Date of birth
- Sex
- Address
- Rare disease
- Treating Clinician
- Specialist centre / hospital
- Confirmation that they agree to NCARDRS holding their data
- Confirmation that they are the patient Confirmation that they understand NCARDRS will contact their clinician for further information.

If the patient is the person's child, NCARDRS will ask for the following additional information.

- The name of the person who is registering the child.
- Confirmation that they are the patient's parent
- Confirmation that they understand NCARDRS will contact their child's clinician for further information.

Want more information before you decide?
The NCARDRS patient leaflet is available at the following website link:
https://www.gov.uk/government/publications/national-congenital-anomaly-and-rare-disease-registration-service-introductory-leaflet

Registration is open to all r(20) patients living in England

(we hope to extend this to the other devolved nations of the UK)

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r(20) Breakfast Symposium—Global Keto

A unique opportunity to reach a large number of healthcare professionals from around the world, to educate and update them on r(20) syndrome diagnosis and treatment

All families and health care professionals are aware of the importance of reaching a timely, correct diagnosis. We all know that a patient has much to lose when there is a misdiagnosis or late diagnosis.

In the case of Ring Chromosome 20 Syndrome, the standard panel tests for genetic epilepsy, comprising the latest techniques, cannot yet diagnose a case of ring chromosome in the mosaic form – the most common form of the disease. At the same time, the use of long established microscopy based testing (specifically Karyotyping) is declining as new gene based testing is introduced and, consequently, with the advance of genetic testing, the rate of diagnosis of r(20) is likely in decline.

Due to lack of research there are no clinical practice guidelines for doctors to follow in the case of possible presentation of r (20). Given the current lack of information the charity is proposing to Host a Breakfast Symposium at the forthcoming 7th Global Symposium for Medical Ketogenic Dietary Therapies in Brighton on 6th - 10th October 2020 to address these issues.

This Breakfast Briefing will be held on the morning of Thursday, 8th October. Three speakers have agreed to attend, and make presentations at the Symposium on behalf of Ring20 Research and Support UK CIO:

Dr Sameer Zuberi - Consultant Paediatric Neurologist at the Royal Hospital for Children and Honorary Professor in the University of Glasgow.

Dr Nancy Spinner - Chief of the Division of Genomic Diagnostics at Children's Hospital of Philadelphia.

Dr Manny Bagary - Consultant Neuropsychiatrist, Barberry National Centre for Mental Health, Birmingham.

We intend to fund translation of key material into a number of languages to enable families, globally, to benefit from this work.

We will also fund an Information Stand at the main exhibition in the Symposium and host supplementary sessions comprising families and health care professionals on the weekend from Friday through to Sunday. This will enable us to ensure:

Effective engagement with leading Health Care Professionals at a key event.

Publication of translations of key information from the Symposium, both on site and subsequently in documents, videos and on the ring 20 website, to provide enhanced interaction for the patients, supporters and healthcare professionals globally.

It is understood that there will be approximately 500 delegates at the Symposium and this provides an excellent opportunity to reach a significant number of these key professionals.

The 7th Global Symposium for Medical Ketogenic Dietary Therapies has been organised by Matthews Friends (a UK charity, established in 2004, providing Ketogenic support for drug resistant epilepsy) and widely publicised to health care professionals related to epilepsy and neurology ensuring a wide and appropriate attendance. Ring20 will also ensure additional publicity through its contacts with global Rare Disease networks, epilepsy groups, EpiCARE ERN and IBE and its established social media (Facebook, Twitter and LinkedIn) feeds.

Arranging the Breakfast Symposium, in agreement with by Matthews Friends, enables Ring20 to fully leverage this opportunity to reach a spectrum of interested healthcare professionals from across the globe without leaving the UK ourselves.

r(20) Families Conference—Brighton

Families Conference 2020
Fri 9th Oct – Sun 11th Oct 2020
Hilton Brighton Metropole Hotel
Kings Road, BN1 2FU
Brighton, UK

We are delighted to share some more information with you about this year's r(20) Families Conference in Brighton.

Cost is £35 per person (same for both adults and children). Included are lunch on Sat and evening meals on Friday and Saturday.

All families attending will need to register through the Global Keto 2020 website: https://globalketo.com/product/parent-carer-information-day/

As we go to press we are still planning this event, however we will consider government guidance and travel restrictions and if necessary postpone to 2021

Here are some of the highlights:

Friday 9th October – arrival 6pm	**evening**—Buffet Dinner with Guest Speaker, Professor Sameer Zuberi
Saturday 10th October 9-5:30 pm	**morning** – an opportunity to attend the Global Keto Families Day sessions to learn more about Ketogenic Dietary Therapy: https://globalketo.com/family-day-program/ or...have some free time in Brighton **afternoon** – r20 Families Conference/AGM with guest speakers, Dr Nancy Spinner and Dr Manny Bagary **evening** – Gala Dinner (from 7:30pm) with guest speakers - to be announced
Sunday 11th October – 9-1:00pm	**morning** – r20 Families Conference (continued). Guest speaker Ilumina. More speakers/workshops to be announced...

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Families/Comic Workshop Nov 2019

The Jasper Carrot Suite in Birmingham City Football club proved an excellent venue for the Ring20 Families Workshop 2019 on Saturday, 23rd November, with a great turnout of over 50

Events of the day included :

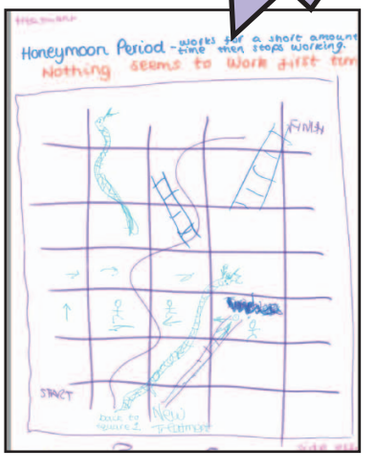
- A session to design the content for an accessible patient information booklet to help patients, carers and health care professionals through diagnosis, support and treatment;
- A question and answer session with medical experts
- the Annual General Meeting
- the launch of group's participation on the Big Give to run from 3rd to 10th December (which proved extremely successful in reaching its funding targets).

Rossie Stone, founder of Dekko Comics, led workshop sessions where all attendees could present not only their feelings and concerns regarding Ring20 but also develop innovative ideas, which could be grown by Rossie into easily accessible and enjoyable comic book form to be published and circulated to patients, health care professional and the wider audience of carers, friends and supporters.

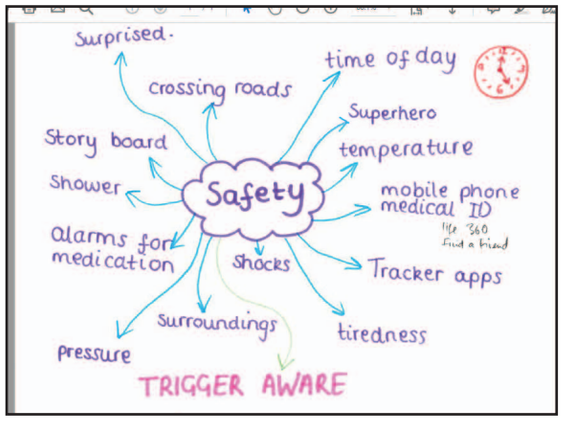
Ideas included thoughts on the characters to be portrayed, key areas to be addressed and, potentially, interactive games, including a maze and snakes and ladders, to guide those involved through the support process.

Our r(20) Youth Project is actively meeting regularly to develop ideas from the workshop into the comics to be printed

FREE copy of our comics will be sent to every r(20) member family



DEKKO COMICS



Families/Comic Workshop Nov 2019

There was lots of very good interaction in the workshops, led by Rossie, who has taken away the results to be developed and fed back to Ring20 in the first quarter of 2020.

Subsequent to the Workshop, a contest was launched to design our "ring 20" character for the campaign. And the winner , chosen by the members of our Medical Advisory Board, is

Otsi by Georgina Clunas

Congratulations to you, Georgina — we love Otsi! And thanks to everyone else who took part and gave our judges a really tough job!

We are looking forward to the comic being published in the summer—keep an eye on our social media platforms for further updates.

The "Ask the Experts'–Q&A session for all" was led by Dr Sophia Varadkar, the specialty lead for the Epilepsy Unit and the Children's Epilepsy Surgery Service (CESS) at Great Ormond Street Hospital who is a Medical Advisor and Trustee for Ring20.

The session covered a range of subjects, including the very topical issues and opportunities recently raised in the media regarding the NICE recommendations on cannabidiol / CBD (Epidyolex), especially with regard to those people with epilepsy.

The main meeting finished with a presentation of Ring20's involvement in the Big Give where the group is specifically aiming to raise money which will enable ring 20 to take an active and effective part in a Scientific Symposium in Brighton (the 7th Global Symposium for Medical ketogenic Dietary Therapies) in October 2020.

This will enable Ring20 to **further expand** awareness through engagement as part of a global epilepsy event for medical treatments, utilising the potential of a large international audience of researchers, neurologists, epilepsy nurses, dieticians and others.

The day was rounded off by a guided tour of the Birmingham City football stadium itself and a dinner in the Jasper Carrot Suite.

Report by **Dave Soderquest**

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Hear how some of our r(20) representatives have been attending events on your behalf

Rare Disease Day

On the 25th February me and my bud attended a reception at the Senedd for Rare Disease Day 2020. The evening started with a networking session for the first 15 minutes. During that time, we met a scientist who believed in a connection between the immune system and rare diseases. We talked a little more, had some refreshments, then took our seats to listen to the first speaker, Angela Burns, who discussed screening of Rare Film Finalists.

After this we were given an update on the Cross Party Group for Rare, Genetic and Undiagnosed Conditions. Next, the Chair of Rare Disease UK, Dr Jayne Spink, spoke of their launch to provide every person with a rare disease an alert card. Sioned Lewis, who has been affected by a rare pituitary condition, talked of her experiences of how the system helped her and what can be improved.

After Sioned, the Chair of Rare Disease Implementation Group, Dr Graham Shortland, gave a brief update on bloodspot scanning and how it is progressing. They believe they could potentially spot if someone is more likely to have a rare disease by testing all newborns. After Dr Shortland had his time in the spotlight, Angela Burns brought the session to a close by making the listeners aware of past and future events.

During the session, my attention was brought to the staggering statistics concerning rare diseases, such as: 1 in 17 people will suffer from a rare disease and that there are over 6000 known rare diseases. Other subjects and projects were mentioned only briefly due to time constraints, but this conference seemed to me to be a great networking opportunity.

Report by **Dale Ward**

Festival of Genomics

The Festival of Genomics started in 2016 and, now in its fifth year, the Business Design Centre in London hosted over 100 Speakers, 50 Exhibitors and 1,800 attendees on 29th / 30th January for its latest incarnation.

With over 90% of attendees, from charities, the NHS and other healthcare professions and researcher groups, attending free it was an excellent opportunity to keep up to date with, and meet people at the leading edge of, genomics and its potential to improving diagnosis and care for sufferers of a wide range of major and rare diseases. Professor Dame Sue Hill, Chief Scientific Officer for England, responsible for the healthcare science workforce in the NHS and associated bodies, lead off with an excellent presentation on the moves in the NHS for the use of genomics in the personalisation of medicine. Covering a wide range of topics from new and innovative techniques, through the need for effective Patient, Public and Professional engagement to the massive infrastructure required to manage and analyse the vast amount of data produced and requiring storage, the talk was much appreciated by a standing room only audience.

The Festival was actually set up by Rich Lumb, Founder/CEO at Front Line Genomics, following the death of his father from mesothelioma, a rare, aggressive form of cancer. A number of other speakers and exhibitors this year were keen to present their own views on progress and future objectives in this rapidly moving field. This wide range of other speakers covered topics ranging across basic research; consulting in paediatrics with children with rare diseases; the changes required within medicine to match and manage these new challenges and visons as to how personalisation of medicine can be realised effectively. The realisation that, while genomics offers a wide range of opportunities, existing testing, such as karyotyping, has not yet been superseded and will need to remain was raised on several occasions.

For me, it was an excellent opportunity to catch up with friends and former colleagues, and make new contacts with groups, not only around the UK but around the world. For the first time, I met a Chinese company working in this area. The most important message that came out of the Festival was that this world continues to advance at a massive rate, seriously enhancing our tools and techniques in research and medical support for Rare Diseases.

The Festival of Genomics & Biodata

Report by **Dave Soderquest**

Improving diagnosis for r(20) syndrome

We know that there are only 150 cases of r(20) syndrome from across the world documented in medical literature and yet as a patient support group we support over 100 families on our mailing list and that's typically most families who can speak English because that is the language we at Ring20 communicate in currently (something we are exploring to change). r(20) is perceived to be very much under diagnosed and under-reported meaning there are many people who have an undiagnosed (or even misdiagnosed) cause for their epilepsy who may actually have r(20) but have never been properly tested for the disease. For example many of the signs and symptoms of r(20) are very similar to those reported in Lennox Gastaut Syndrome, which is simply the name for a collection of specific symptoms including a similar spike slow wave EEG pattern and prolonged periods of non-convulsive status epilepticus (NCSE).

But how rare is r(20) and why are doctors not diagnosing more cases – there are lots of advances in genetic testing right?

Right and wrong – its correct that there have been incredible advances in genomics and next generation sequencing techniques to identify many existing and new genetic mutations and therefore more patients with rare diseases are now receiving accurate diagnoses for the first time; incorrect because these fantastic new techniques cannot yet detect a ring chromosome with no change in sequencing, as seen in many patients with r(20) syndrome in the mosaic form. Doctors still have to request a karyotype test (blood test) for chromosomal mosaicism and submit 50-100 cells – more than usual - to confirm a diagnosis of r(20) in the mosaic form, however this testing technique is being phased out in the western world of medicine in favour of the newer techniques. Hence we suspect the diagnostic rates of r(20) are likely in decline, bucking the trend against most other rare diseases.

What are we at Ring20 doing about this?

We've been speaking about this problem at events and to interested parties to see if we can find someone to help us solve the conundrum. We've had some very positive interest from industry and are in discussion with several parties, including Illumina who are responsible for developing Whole Genome Sequencing (WGS) technology in the UK and abroad; WGS is the latest technique in genomic diagnostics. We're at the very early stages but hope to work together on a new research project to find a method to detect what changes in the genome make the ends of the chromosome 20 stick together to form a ring structure. To investigate the problem Illumina will need access to blood samples from people with r(20) to work on, so we are considering linking consent for this into our Natural History Study and also collect samples through the NIHR Bioresource Unit where we aim to setup a new r(20) patient cohort for future research opportunities – we'll tell you more as this unfolds...

Won't it take years to change diagnostic techniques – what can we do now?

It is likely that it will take some time to find a new way to detect r(20) more easily. In the interim, the best approach is to raise awareness of the diagnostic challenge by speaking at events to doctors, geneticists, researchers, industry, pharma and others. Only by speaking about the problem and how we can overcome today – by sharing the signs and symptoms of r(20) and highlighting the need to request karyotyping – can we expect to improve diagnostic rates. Much of the work Allison is doing through the EpiCARE ERN involves just this and there are numerous initiatives unfolding this year to improve education, training and awareness amongst healthcare professionals (read page14 on the latest EpiCARE initiatives for r(20))

Why is it important to have a diagnosis anyway, we don't have any recommended treatment or cure for r(20)?

If you didn't have a diagnosis of r(20) you wouldn't have access to our patient group where we signpost you to the latest/most accurate information on r(20) and provide mutual support by connecting you with other r(20) families through our Facebook group, RareConnect forum, individual introductions or even face-to-face at one of our events. Having a specific diagnosis will be an enabler to future research participation and if we can identify a greater number of affected individuals, we will become a louder voice, much more interesting to researchers and grant funders. We also recognise that having a diagnosis or r(20) an also be a barrier to treatment, for example in accessing new medications that come to market e.g. the recent advent of medicinal cannabis being a case in point, specifically in the UK where it is only currently licensed for patients with a diagnosis of Dravet Syndrome or Lennox Gastaut Syndrome.

NIHR BioResource
Rare Diseases

The rate of r(20) diagnosis is likely in decline.

WE ARE WORKING TO CHANGE THIS SITUATION!

ring 20
research & support uk

At Ring20 we are driving change to influence the future of all with r(20) . This is patient-led research in the making ...

EpiCARE ERN update

Raising the profile of r(20) across Europe

The EpiCARE year ahead for r(20) syndrome

As you know, I am co-lead for the Patient Advocacy Group (EPAG) for the EpiCARE European Reference Network (ERN) for rare and complex epilepsies. ERN's have been in existence for nearly 3 years now, but what difference has this made for those with r(20) and what work is taking place that will benefit you now and in the future. Well, following the recent AGM in Lyon we can report that there's actually quite a lot going on, where do I start…?

Monthly virtual patient consultations are scheduled for experts across Europe to discuss non-surgical cases referred to them where a diagnosis or treatment is urgently needed for a severe or complex case. EpiCARE has a healthcare pathway along which patients can be referred by their own doctor, firstly within their own national healthcare systems and then ultimately up to the ERN if a more specialist expert opinion is required.

If you are struggling with severely worsening symptoms which your local team cannot resolve, try approaching your healthcare team to ask them to refer your case upward to your national EpiCARE centre – a list of EpiCARE centres across Europe can be found here: https://epi-care.eu/members-of-epicare/. If you do not get any help, contact Allison who may be able to put you in contact with a specialist at your national EpiCARE centre.

We know that patients are being considered for r(20) diagnosis through these virtual consultations which is progress indeed!

Look out for the following initiatives this coming year:

- launch of **r(20) information leaflets** on the EpiCARE website (1 for healthcare professionals), 1 for families. These summary leaflets are designed to provide an overview of the syndrome for newly diagnosed patients and the healthcare professionals who manage their care and signpost to trusted locations for further information and support.
- **A webinar on r(20) diagnosis and treatment** – which will be co-presented by Prof Zuberi and Allison, on Thurs 25th June 4-5pm (UK time). Sign-up here: http://bit.ly/r20webinar its FREE!
- **r(20) Orphanet updates** – the 'Go To' website for rare diseases - for r(20) syndrome (last updated 2006 so much overdue)
- **r(20) case studies** will be used for **specialist training** for epileptologists (Level 2)

In addition, the EPAG group have been working together developing:

- **patient journey's** to be accessed from the EpiCARE website which includes one for r(20) which we've recently asked our members to contribute to validate.
- Recommendations for EpiCARE to address the '**common unmet needs**' of patients with rare and complex epilepsies, including those with r(20). We are calling for a more holistic approach to care, involving multi-disciplinary teams, prescribing guidelines to avoid over-prescription, more awareness of the family support needs, mental health issues and to address the gaps in transition and adult services. A presentation was made in Lyon, which was favourably received by the doctors who've asked us to create a webinar on the subject…watch out for this. We've also submitted an abstract and poster at the forthcoming European Congress for Rare Diseases (ECRD) meeting in Stockholm in May, for which Allison is lead author (this meeting is now being held online due to COVID-19).
- I continue to explore prospective research opportunities with Prof Helen Cross and also possible new leads with adult neurologists in London, as well as developing a research strategy for Ring20.

I think you'll agree the future for r(20) syndrome is looking more positive?

Fabulous Fundraisers!

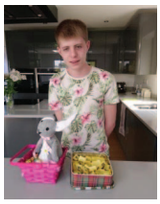
As you all know, our research and support services would not be possible without the amazing fundraising efforts of our members, their families, friends and work colleagues! *Here are some recent and planned events:*

3 Peaks Challenge—Carl Darby

On 16th March staff at IMI Truflo Marine held a Bake Sale and Raffle to raise funds in support of their forthcoming challenge. Amazingly they raised a staggering £1,136 at with 50% coming from matched funding from IMI Truflo Marine—THANK YOU!!!!!! On May 7th, 8th and 9th a team of eight climbers, including Carl and James, backed by a support team of eight, will climb Ben Nevis (7th), Scafell Pike (8th) and Snowdon (9th). This is not only an amazing physical challenge, but also a massive logistical feat!

Easter Egg Raffle—Dawn/Callum Gray

Dawn and Callum Gray held their (now annual) Easter Egg raffle and raised £100!



Raffle and Tombola—Nicola Middleton

On 30th March a raffle and tombola planned by Nicola Middleton at North Tees and Hartlepool NHS Foundation Trust were postponed due to coronavirus, however the raffle was able to take place 'from home'. Amazingly £636 has been raised with the tombola is to be rescheduled post-lockdown.

Pub Fundraiser—Dale Ward

Dale Ward was due to host a Pub Fundraiser on 24th May however this event has had to be postponed due to COVID-19.

Ring20 Skydive

Unfortunately our Ring20 Skydive has been put on hold due to COVID-19.

HUGE thanks to everyone concerned! We love sharing photos of your events.

If you would like to get involved in fundraising for Ring20, please get in touch with Audrey to find out how we can support you with Ring20 branded items to help your event go with a bang! Contact:
audrey@ring20researchsupport.co.uk

26th April 2020

Here's how you can contribute to keep our charity fundraising alive during lockdown.

the 2.6 challenge
Save the UK's Charities

#TwoPointSixChallenge

https://twopointsixchallenge.justgiving.com/get-

Facebook Fundraisers!

We'd like to take this opportunity to thank all the individuals and families who have setup Facebook fundraisers over the last year for Ring20.

Celle's Birthday Fundraiser £90.78
Dale's birthday fundraiser £180.00
Donna Reed's Birthday Fundraiser £35.21
Georgina's fundraiser £20
Heather's Birthday Fundraiser £24.84
Jackie's birthday fundraiser £285.00
Ring 20 Skydive £765.17
Daniel's 2019 Challenges £1193.05
Katie's birthday fundraiser £169.00
Melissa's Birthday Fundraiser £349.40
Tsuyoshi Nagatoã £74.96

£3,343.22

ring 20
research & support uk

Everyone can help with fundraising…even during lockdown!

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.

fit4change

Recycling FOR GOOD CAUSES

How about recycling?
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 on eBay and donate 10-100% to Ring20.

ebay FOR CHARITY

https://www.charity.ebay.co.uk/charity/Ring20-Research-and-Support-UK-CIO/71688

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

THE ESSEX LOTTERY

Ring20 Research and Support UK CIO

26 Headley Chase
Warley
Brentwood
Essex
CM14 5BN

Phone: 00 (44) 7385 292797
E-mail: ring20@ring20researchsupport.co.uk

Regular Donations

Regular donations really can help make a difference to what we can achieve as an organisation and deliver vital services to you. Complete our form here: https://www.ring20researchsupport.co.uk/wp-content/uploads/2016/03/StandingOrderForm.pdf Even a small amount can help!

We hope you have enjoyed reading our latest newsletter.

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

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

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

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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