Issue 6

December 2017

Registered Charity Number 1165651

Newsletter

ring 20 research & support uk

2017: A phenomenal year!

Special points of interest:

- Hear about what our Volunteers have been up (page 2-3)
- Catchup on the Sullivan family fundraising (pages 4-5)
- Highlights from our recent r(20) families conference (pages 6-11)

This year we've really seen a difference in our group's activities—from hosting a first ever r(20) families conference in the world, to launching a Research Fund Campaign, I think you'll agree 2017 has been a phenomenal year for Ring20 Research and Support UK CIO and all those who benefit from our patient support group!

We got some fantastic feedback from our families and speakers for the conference and everyone that attended said they'd come again and would recommend the event to other families. Yes, we crammed a lot into the weekend, especially the presentations and next time we will aim to get better at our time management, - but we just had so much to share with you all and didn't want to leave anything out! We also got some great ideas about things you'd like to see/hear at future events such as hearing more from families about their experiences and having parent group sessions. Our young people seemed to enjoy the breakout session which went better than expected, especially with the inclusion of he siblings—who's views are important too. We know that lots of new friendships were made that weekend, which is fabulous!

You'll have seen from our regular weekly emails that we've been releasing the video footage every few weeks and will continue to do so over the coming months—there are 28 videos in total, so we didn't want to overwhelm you all at once! Based on your requests we are talking to our video production company and should be able to make some DVD's available, potentially at a small cost, so do let us know soonest if you'd be interested in receiving a DVD so we know how many copies to make.

The conference certainly seemed to be a pivotal point for many in terms of supporting us - see below for the impact it's had on our Research Fund Campaign.

As Christmas approaches, I hope you've finalised preparations —all your cards are sent, presents wrapped and have the turkey out of the freezer? However you choose to spend the festive holidays, have a truly wonderful Christmas with family, friends, neighbours and we look forward to, what promises to be, an exciting 2018!

Yours

Allison

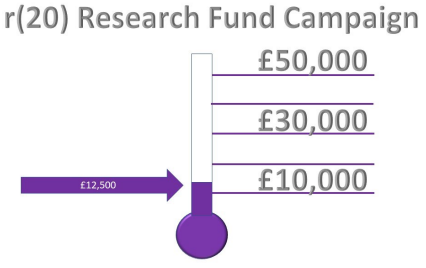
Co-founder/Secretary

INSIDE THIS ISSUE:

Research Fund Campaign update

Our Research Fund Campaign has really taken off; in just a few months we've exceeded our first £10,000—in fact we've surpassed £12,500!

Much of this has been raised by the Sullivan family, who's efforts you'll hear more about inside, but we've also received cheques from Lakeland for over £1,000 and a St. James Place Foundation grant of £2,500. Together with over £500 from the Jeans for Genes 50:50 income scheme, various donations and fundraisers, this is truly a remarkable effort and spurs us on to our first target of £50,000.Gemma continues to submit grant applications on our behalf; in fact we have an assessment for a new grant in Jan for up to £10k. Wish us luck!



LAKELAND

ST. JAMES'S PLACE FOUNDATION

Events

Dale attended the Rare Disease Patient Network Annual Meeting in Cardiff (Oct '17) representing r(20)

On 23rd Oct, Dawn and Allison had the fantastic opportunity to attend the Cambridge Rare Disease Network (CRDN) Summit 2017 at Robinson College, Cambridge.

On arrival we handed over our patient journey posters (see below) and got ourselves signed in to some new tech to follow the conference on our phones/tablets, whilst in the auditorium. The new Glisser technology allowed us to follow all the speakers slides on our devices, and/or to make personal notes, to interactively ask questions during the talks (as well as see questions posted by others and 'like' them) and post slides with comments directly on *twitter*. It was pretty cool technical innovation, if not a bit hectic to keep up with everything!

The event was opened by Alistair Kent OBE, Chair of CRDN.

We enjoyed presentations from two patient support groups:

- Kay Parkinson CEO of CRDN and Alstrom Europe who inspired the patient journey poster competition, as previously a chance poster presentation by Kay had led to collaboration to develop a drug treatment for Alstrom Syndrome.
- Daniel Lewi CEO of the CATs Foundation and new research into Tay-Sachs and Sandhoff disease being undertaken at Cambridge

The patient journey poster competition

A great innovation of the conference, and real highlight for us was the patient journey poster competition. Prior to the conference we were 1 of 33 different patient groups who took the time to produce posters to summarise their rare condition and their own patient journey. I (Allison) have never produced a poster before and have no background in Graphic Design, but with some guidance from the lovely CRDN team I gave it a go, because I felt the importance was in getting the word out about r(20) syndrome, *how it affects us and what we want*, <u>not</u> the quality of my artwork!

Even more excitingly five poster authors were selected to give a five-minute lightning talk during the conference, telling the audience a little about their condition and their three wishes for the future. Unfortunately, our poster wasn't shortlisted as we were up against some very stiff competition, but it was great to hear their wishes, which were often pertinent to all!

The whole event was a great showcase for rare diseases, and some of the latest innovations. There were talks on:

- Developing orphan drugs (drugs for rare diseases) from Sobi a pharmaceutical company who claim to 'listen to patient communities'
- drug repurposing; we heard from Dr Rick Thompson from Findacure who suggested that 'The future IS drug repurposing', given that the cost of bringing new drugs to market is ever increasing and the number of new drugs being developed in decline. Rick gave a very simple explanation of what drug repurposing is:
- gene editing that can cure some rare diseases (currently being used for specific single genes only at this stage, so not applicable for r(20) syndrome, but an amazing concept just the same!)
- the 'impatient patient revolution' and the power of patient support groups
- the advent of telemedicine and the need for a Rare Disease Nurse Network.
- Mendelian – a new powerful rare disease search engine for health professional

A key question was asked of the audience:

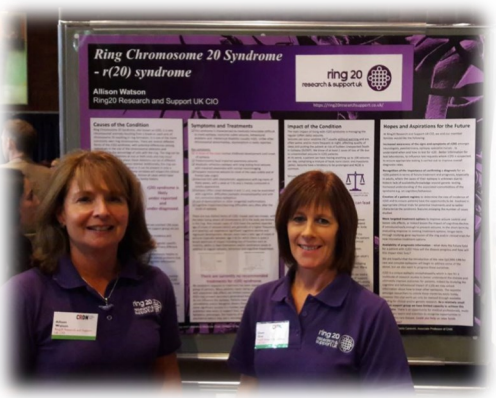
A key question was asked of the audience:

How do we get #RareDisease into the mainstream?
TV campaign, changing the language of disease,
#education choosing right media GP training...

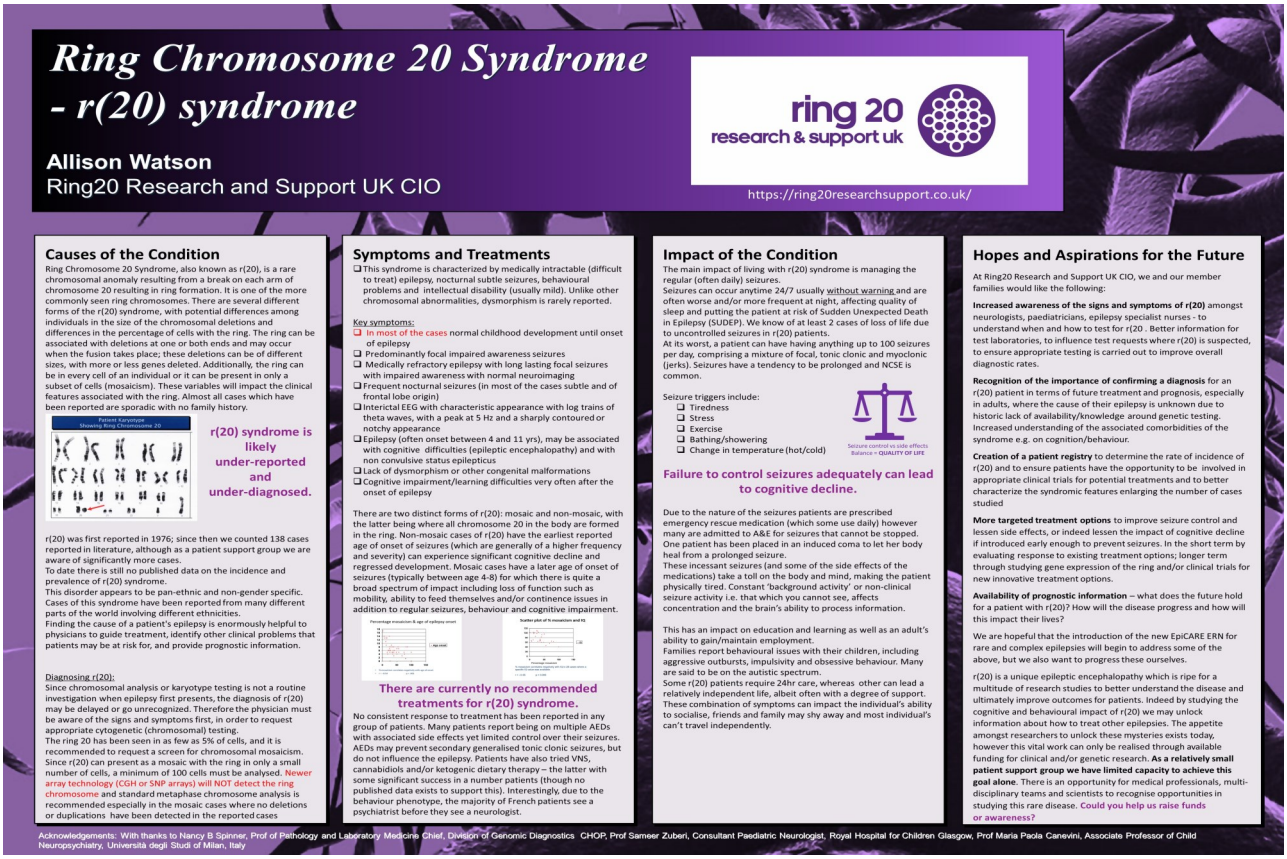
Did you know that Rare Disease is more common than cancer and AIDS combined?

At the end of the day there was time for a well-deserved glass of wine to reflect on the day with all attendees and a chance to meet up with a great bunch of like-minded people – representatives from other patient support groups; we call ourselves the 'Rare Revolutionaries' as we all subscribe to Rare Revolution magazine and are advocates for change in our own Rare Diseases! We look forward to meeting up again at future events and keeping in touch on social media. These guys teach us so much and the power of information sharing and networking is invaluable.

Roll on the Rare Revolution!



Events (continued)



Our r(20) patient journey poster—displayed for the first time at CRDN

(click on the image to download a copy)

Our Volunteers

We rely on our volunteers, as without them we couldn't run our patient support group and provide the service we do for you.

As well as Dawn and Dale's activities that you see on these pages, Jon Dunn continues to help support our website, Marie Ward has recently agreed to begin to help out in the new year with some of our communications and you'll read on the next pages all about how the Sullivan family!

We recognise how busy our families are, so we are additionally seeking a volunteer Communications Officer. We have also been advertising for a volunteer Fundraising Officer to support Gemma in her work of researching and submitting funding applications for us.

A mention must also go to Ann and Andrea who very kindly gave up their weekend in August to help with the organisation of our families conference—ensuring everyone and everything was in the right place, at the right time (no mean feat) !

Andrea also helped with the preparation and production of a lot of the conference materials. Subsequently Andrea created feedback survey's for our attending health professionals and exhibitors to complete and all of the feedback forms that you kindly completed at the conference were input and collated by Jamie Gray (Callum's brother).

Lastly we have to thank Andrea for helping us produce this newsletter...

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Fundraising

The Sullivan fundraising total is currently an amazing:

£7,250!

Enthused from our r(20) families conference, Chris and Claire Sullivan (one of our member families in Scotland) have been hard at work fundraising for our Research Fund Campaign. They have enlisted the help and support of their friends and colleagues, their workplaces and local businesses to put together a fabulous program of events over the next year.

Here's what Claire has to say...

With the support of family and friends, we're excited to update that Jessica's Sparky Halloween Party was a huge success, and has us well on our way to trashing our 12 month RESEARCH fundraising campaign target of £20,000.

This one event has raised £1,550. Matched funding and a very kind donations has taken the event total to over £2,000.

This is the first of many events to follow over the course of the next 12 months.

We have also since held a bake sale and raffle at SSE, raffled Noel Gallagher concert tickets at the WEBATA Speakers Night, and a Christmas jumper/bake sale at Advance Construction head office.

Future events scheduled include:

- Santa Sleigh - Dec 17
- Jessica's birthday - school event - Jan 18
- Pub crawl - Jan 2018
- Race night - Feb 2018
- Work raffle - Mar 2018
- Stirling Marathon - Chris +others- Apr 18
- Gung Ho family event - May 18
- Ben Nevis Climb - Claire +15 people and still more people joining event - May 18

On top of all that, wee Jessica completed the Aviemore (5k) Santa Dash!

Here she is on her run—well done Jessica!

JESSICA'S RING20 JOURNEY

...so far!

Save the date...21st April 2018

The highlight of the Sullivan family fundraising will be a **Ring20 Gala Ball** on Saturday 21st April 2018.

Due to be held at the lovely Radisson Blu hotel in Glasgow, a venue hosting up to 500 people, the event will be kindly sponsored by Advance Construction.

Prof Sameer Zuberi, from Glasgow Children's hospital will be the guest speaker on the evening.

This could be an opportunity for r(20) parents to meet up again?

If you'd like to attend for what promises to be a major r(20) event for 2018, then tickets will become available early in the New Year.

Georgina's handmade jewellery

One of our patient members, Georgina Clunas is hugely talented and makes handmade jewellery, which she sells at craft fairs, in Scotland.

Georgina has very kindly donated 10% of her profits to Ring20 Research and Support UK CIO, which has been matched by her parents. We've also received a generous donation from another Clunas family member.

Do check out Georgina's Facebook page (search for thisgirlcrafts) and/or her website: http://www.thisgirlcrafts.co.uk/

THE SULLIVAN'S ASK TO OTHER MEMBER FAMILIES

Are there any fundraising activities that you could arrange locally to support the Ring 20 RESEARCH fund too?

We can honestly say that we were worried we wouldn't have the time to dedicate to this but now that we're up and running, we finally feel that as parents to a child with Ring 20, we're doing our bit to RAISE AWARENESS and FUNDS..... that will go straight into the new Ring 20 RESEARCH fund.

WE CAN MAKE A DIFFERENCE IF WE ALL WORK TOGETHER. Please get in touch with Allison, Chris or myself if you're keen to find out more. And just think, if these events could be replicated by the families who attended the conference alone, we could be running the same events in numerous parts of Britain, Europe, America, Canada and even Australia with all families looking for the same outcome, a cure, targeted medication or even just a better understanding of Ring 20 Chromosome Syndrome.

You'll be amazed at just how many people and established fundraising groups that have come forward to offer help and donate monies from their proceeds raised at other fund raising events just by raising AWARENESS alone of Jessica's condition.

An example of this is the many coffee mornings and fundraising events that are happening now in our local area to raise funds for the Ring 20 cause.

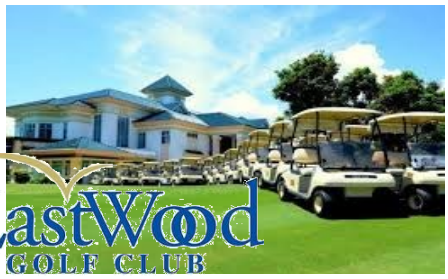
Let's do this!

Could you fundraise to help towards for our Research Fund Campaign?

Or nominate us as a charity for the year to a company you know?

Nominated Charity of the Year

We are very pleased that one of our member families put our group forward at their local golf club AGM, who agreed to support Ring20 Research and Support UK CIO (alongside 2 other charities) as their nominated charities of the year 2018. a third of proceeds raised throughout the year will be donated to us. Thank you in advance to Eastwood Golf Club in Glasgow.



Another of our member families in Wales also successfully nominated us and we are pleased to announce that Tai Tarian Homes have selected us as their chosen charity for 2018. In fact 2 of their staff have already signed up to run the Manchester Marathon in 2018!

Tai Tarian

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R(20) Families Conference 2017

First Ever r(20) Family Conference in the world!

What an amazing weekend!

Twenty r(20) families attended the event, together with 9 health professionals (comprising paediatric neurologists, geneticists, clinical psychologist, ESN and Young Epilepsy transitions worker) representing the UK, Belgium, The Netherlands, Germany, Italy, USA, Canada and even Australia.

A truly international event - we're still taking it all in...

We had an official photographic team onsite to record the event and videos of the speaker presentations, now on our website and YouTube channel. Look out for your email notifications as to when each video is released.

Families that attended the event, commented that they had found a new extended family!

This event couldn't have happened without the generosity and support of our funders. So we would like to thank the Big Lottery Fund and Jeans for Genes and also our additional sponsors LivaNova for making this event possible.

Also we would like to thank our Exhibitors for providing our families with lots of useful information and take-aways: Nutricia, Epilepsy Action, Young Epilepsy, Matthew's Friends, RareConnect, MPS Tracker.

And we must also mention the numerous local businesses and organisations who donated items for our delegate packs and raffle prizes Nutricia, Atkins, Allied Media, Cineworld, Nandos, Trudi Jones Beauty, Waterstones, Tesco, TK Maxx, Game, Wildwood, Bloomin' Jacks and The Little
Florist.

Weekend where the magic happened...

In the evening we arranged for a close-up magician to entertain our families, while they dined and chatted. Everyone seemed to enjoy this as it appealed to young and old alike. We were all amazed and couldn't work out his tricks?

Here's what Richard (magician) had to say about the evening:

I have performed at many events over the last forty odd years and I can say that two events stick out from all the rest ,the first one was at the Hilton hotel for the orphaned children of Chernobyl who sadly had lost everything that we sometimes take for granted - family and homes. The children were polite well mannered and truly a credit to the parents they sadly lost.

The second one was at the Ring 20 event held at Jury's inn Liverpool city centre on Saturday night the 5th August.

There were families from all around the world Australia ,America, the Netherlands to name but a few . And it was humbling to see once again children with health problems so well mannered polite and respectful. I cannot speak more highly of them and their families are wonderful people.

I also want to wish all the people connected with Ring 20 every success in whatever they do.

Richard t Smith

Ring20 Young Ambassador Award

Feature article

This is an analysis of how the R20 Liverpool Conference for me (Dale Ward). At this conference I was a speaker talking about how I live and cope with R20 (Ring Chromosome 20) as I personally have R20. I decided to surprise my mother that I was speaking so I set up a presentation which I sent to Allison.

So after a long trip from Cardiff to Liverpool in my Stepdad's van we arrived at the hotel where we unpacked, rested and cleaned ourselves up ready for the pre-drinks. At the pre-drinks we met up with other families of R20 being such a rare condition the families were originated from all around the world, we met a family from Scotland, Australia and America which for me was a very good experience.
The pre-drinks was a chance to get to know each of the families and how themselves cope with R20 and it how it affects them. Many of the comments I received were very similar to how I was when I was younger.

I am one of the oldest known to have Ring Chromosome 20 Epilepsy at the age of 24, however that said that some were very similar others were also very different. The pre-drinks was very enjoyable but I could not drink too much as I was speaking the next day, none the less a very f
un and enjoyable night with great company.

The next day was the conference which began early in the morning and had some brilliant and encouraging speeches from the other speakers. I was then called and welcomed kindly to the front to present my speech I went up feeling confident and knowing I was going to do a good job. The only nervy part in my head was that it was my first time being a speaker.
The presentation was put up for me and given control of the slides to me ready for the talk, I had put the presentation slides by age making it easier to explain. I started by talking about how I was before I had epilepsy as I was diagnosed when I was eight, how my life was turned upside down
By becoming very isolated and having very many seizures, this is where it started to get a little difficult talking about my past experiences. I knew and hoped that in the end that this would inspire and encourage young people who are struggling not just with R20 but people struggling with similar conditions. I came off the stage with a big round of applause which surprised me but also made me very proud.

People congratulated me, I found out my mother had been recording me and that my mother as well as others had been brought to tears, I was immensely overwhelmed with pride.
I sat back in my seat and listened to the rest of the speakers which again was very inspiring.

After they had finished I was surprised to find out they had made a Young Ambassador award for me which they again called me to the front and presented to me. This caught me in shock and I didn't really know what to say I shook Don's hand, thanked him and had a photo with him. I sat back down still in shock and admired my award; I couldn't believe what had happened.
I am still very thankful to my good friends at R20 for the award. We then finished the conference and went on a tour of the stadium which I enjoyed a lot as it is one of the stadiums I had yet to tour, the Dixie Dean tributes impressed me the most about the stadium. We then went back to the hotel and got ready for the tea time gala dinner.

The gala dinner was very fun as we had some delicious food and a great laugh. We got to know each other much better, I met another Welsh lad who had a son with R20 we have since kept in contact and plan to meet up soon as we got on really well at the conference.
There was a raffle and a magician which made an all round good day.

The next day there was a tour of the Beatles Museum which I could not attend as I needed to travel home but by what I have heard was a very enjoyable day. Knowing that I could not attend the tour with the group my Mother, Stepdad and myself went on a mini tour and had a quite one the day we arrived.
Overall it was a fantastic weekend and when the next pops up sign me up!

Many thanks goes out to Allison Watson, Don Gordon and the rest of the team who have put in their time and effort to make this happen.
Dale Ward

Here's what Dale Ward had to say about the event...

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Welcome Reception

On the Friday evening, after having travelled from far and wide, our r(20) families met for the 1st time, for informal drinks and nibbles. It didn't matter that no-one knew each other previously, everyone came together as one, forging new friendships. They now describe themselves as a 'new extended family' and have vowed to keep in touch!

For families to continue being a support for one another. Families will forever and always be the light in the darkness and will always help each other see the rainbow through the storm. Well done!!








All excellent speakers, extremely interesting. I have very much enjoyed listening to everyone.

Gala Dinner

After a long and intensive day of listening to presentations, everyone was ready to relax and let their hair down. It also provided time to chat about the weekend's events, sharing thoughts and experiences of living with r(20) with others who know and understand what you're going through. Its really comforting when you say 'My son/daughter does that too!"

We kicked off with welcome drinks and all enjoyed a 3-course meal, whilst being entertained by our magician. For a bit of fun and fundraising, we hosted a game of 'Heads and Tails', where everyone joined in and of course, we had a raffle with some amazing prizes, including weekend spa breaks, a pair of ICE watches, £50 of shopping vouchers, hamper and more...








Fab event and learnt so much and met amazing people. Not always easy to talk about this condition & the impacts but felt very comfortable

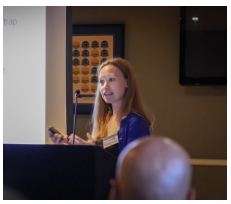
Conference Highlights—Everton FC

Amazing location. Loved it!

A few memories of a hugely informative day! A massive thank you to all our speakers who gave up their weekend to be with us and enlighten our families, and each other on the latest information on r(20) syndrome.

It was important to us to share video coverage of the day's events, so more families and health professionals can benefit from this new knowledge on r(20) —not just those that were able to attend in person.

Very well structured and organised!



Feedback from our speakers:

What for you, were the highlights of the conference?

- ◊ State of the art on clinical and genetics of ring 20 syndrome. Patients and families' inputs
- ◊ Meeting with overseas experts and hearing from colleagues. It was also great to network with families.
- ◊ Meeting all the families' Children/teenagers/siblings talking about current issues they have and the impact of Ring 20

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Would you recommend this to another r20 Family or come again? 100% of respondents said YES!

Young People's Breakout Session

Ring 20 Weekend away: Break out session

The aim of the break out session was to provide engaging activities for each young person present, including their siblings. All the activities were planned to ensure everyone was able to participate regardless of age, ability or circumstances.

The break out session was planned, so it was a free space to talk away from parents. The focus was for each young person to develop strategies of how to deal with the current pressures they face, whatever challenges they each have. To begin with everyone discussed what their dreams were for the future. The aim was to support each young person in believing in themselves. They were then put into three groups:

Living with Ring 20 can have a detrimental impact on a young person's self-esteem and wellbeing. This can result in low aspirations, poor engagement at school/college and issues achieving independence into adulthood. It is important that parents regularly talk to their young person and help set goals for them to gradually become as independent as they can be, so they can reach their full potential.
It was also very evident from the session, that being a sibling of a young person with Ring 20 also faces psychological struggles and challenges. Young people will naturally want to help care for their sibling. However, it should not ever be expected that the sibling takes on the role as care giver. It was also apparent that the siblings are also given individual time on their own with their parents, in order for their needs to be met.

Emma Ninnis (Clinical Nurse Specialist GOSH)

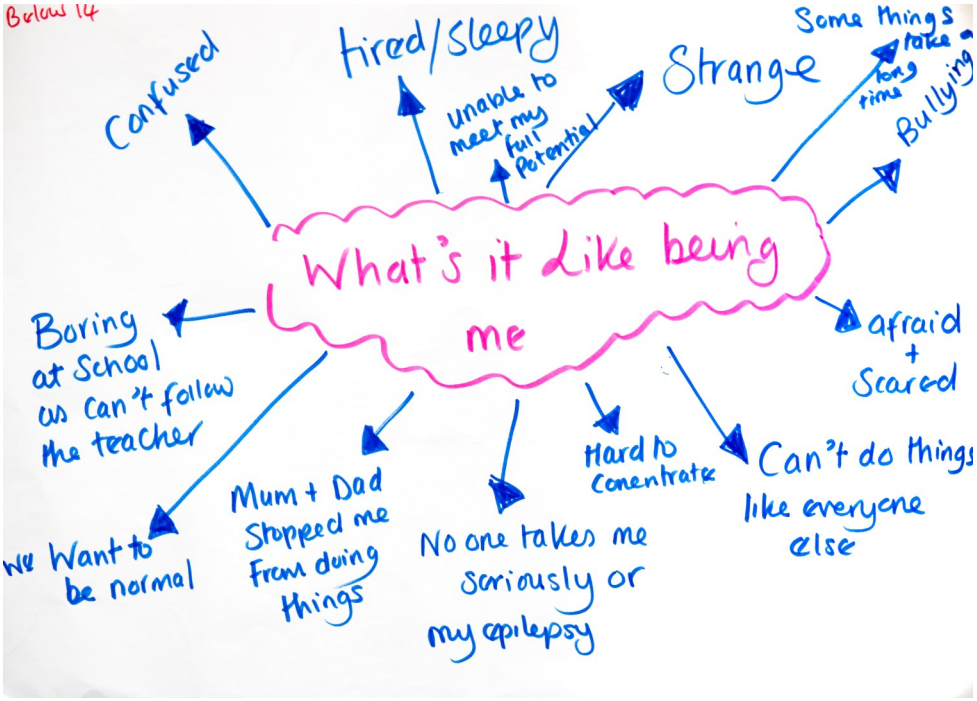
David and Georgina presenting 'What we want!'

The aim was to support each young person in believing in themselves. The participants were then put into three groups:

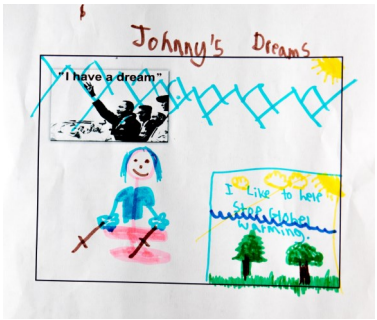
Under 14s: what's it like being me: This group was very confident in their answers and aware of the challenges they each faced. They also each discussed their fears and the barriers of living with r(20)

Siblings: brothers' and sisters' opinions matter: Initially this group was quite reluctant to discuss the topic because they did not want their parents knowing their worries. We reinforced the idea that it is important to talk about these issues and that the session was a safe space. After this was explained, the young people all felt able to contribute to the discussion

Over 14s: what we want: This group all wanted independence. They were very much aware of potential barriers to achieving future independence. They also wanted to discuss how to make things better for other young people



Our young peoples work in action...



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Beatles Museum and Goodbye!

On the Sunday morning, some of us a little weary from a very busy couple of days, we strolled down to the Beatles Museum on the historic Albert Dock.

A truly interactive experience suitable for all the family.

The morning gave us time to reflect, but before we knew it, it was time to say goodbye, as we all went in our separate directions home.

We hope you all enjoyed the whole weekend as much as we did and certainly from your feedback we hear you'd like to do this again!

Essex Lottery

We now participate in the Essex Lottery, with 50% of funds raised from our supporters going to Ring20 Research and Support UK CIO.

Its only £1 per week and you don't have to live in Essex to play (although you do have to be in the UK). You have a chance of winning up to £25,000! For more information or to signup to support our cause: https://www.essexlottery.co.uk/support/ring20-research-and-support-uk-cio

Data Protection Update

Many of you will be aware that there are new EU regulations for Data Protection coming into force in May 2018. As a result we need to know you're still happy for us to keep in touch.

We will be sending out an email shortly requesting you to update your contact details and provide consent for us to contact you.

Please do respond before its too late, as failure to do so, may result in us losing contact.

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.

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

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