Public Engagement and Partnership

Social media

The IMAGINE-ID social networks have gone from strength-to-strength, acting as both a platform for interested and recruited families to engage with the research team, as well as other professionals and organisations. IMAGINE ID was invited to take part in a social media scheme through the organisation *Rare Revolution Magazine* in March 2019. This scheme used the #tuesdaytakeover phrase and allowed the IMAGINE ID team to access the *Rare Revolution* social media channels (Twitter, Facebook and Instagram) to post about the study and raise awareness to their thousands of followers for 24 hours. This was a fantastic opportunity to inform a different, but engaged audience in rare diseases, and generated positive outcomes including an influx of families joining the study.

The IMAGINE-ID Facebook account https://www.facebook.com/imagineid.study/ currently has 815 individuals following our activities and usually receives approximately 1-2 messages a month from an interested family or family that have already been recruited and asking for various updates. Facebook is usually a platform for families to view, after they have joined the project, as many families with intellectual disability and rare disease use Facebook's closed group facility for specific genetic conditions affecting their children.

The IMAGINE-ID Twitter account https://twitter.com/imagineidnews currently has 956 'followers' and has been a particularly excellent tool in promoting the project to genetics and intellectual disability support group and charity followers. As part of our collaboration with many of these groups, we send through an IMAGINE-ID promotional pack which includes suggested social media posts and photos detailing the project (in the limited number of characters available on Twitter) which has proven extremely useful in the recruitment of patients and dissemination of our research, both nationally and internationally.

Collaboration with research groups, patient groups, media and charities

As part of our patient recruitment strategy, we have engaged with many research and patient support groups and charities. We have often exchanged details and agreed to promote organisations on our newly launched website or via social media. We create personal promotional packs that are sent to organisations with suggested text and accompanying photos intended for websites, newsletters, Facebook and Twitter. Here is a list of the current organisations with which we have collaborated, and which have promoted IMAGINE-ID to their members:

100,000 Genomes Project https://www.genomicsengland.co.uk/about-genomics-england/the-100000-genomes-project/
1p36 deletion support group (closed Facebook group)
2q23.1 deletion and duplication disorders (closed group)
2q37 deletion group (closed Facebook group)
2q37 deletion syndrome blog http://2q37-deletion-syndrome.blogspot.co.uk/
22q Awareness Days https://twitter.com/22qAwarenessDay
22q11 Ireland https://www.22q11ireland.org/
7p22 deletion support group http://www.7p22del.com/
Action Medical Research for Children https://www.action.org.uk/about-us/contact-us
Angelman Syndrome Ireland http://www.angelman.ie/
Angelman Syndrome Foundation USA https://angelman.org/about/contact-asf/

Angelman Syndrome Support Education & Research Trust (ASSERT) http://angelmanuk.org/
Bridge the Gap- SYNGAP1 Education and Research Foundation http://www.bridgesyngap.org/
BINGO (Brain and Behaviour in Neurodevelopmental disorders of Genetic Origin) research project http://www.mrc-cbu.cam.ac.uk/bingo/
Cambridge Rare Disease Network http://camraredisease.org/
Catalan Association of 22q11 syndrome http://www.22q.cat/
Centre for the Advancement of Positive Behaviour Support http://www.bild.org.uk/about-bild/contactus/
Cerebra https://www.cerebra.org.uk/
Child Growth Foundation http://www.childgrowthfoundation.org/
Chromosome 18 Registry and Research Society Europe http://www.chromosome18eur.org/
CLIMB http://www.climb.org.uk/
Cornelia de Lange Syndrome Awareness https://www.cdlsawareness.org/
Cri du Chat group http://criduchat.org.uk/
Cure CDLK5 http://www.curecdkl5.org/
Deciphering Developmental Disorders (DDD) study https://www.ddduk.org/
Enable magazine http://enablemagazine.co.uk/
Family voice, Peterborough https://www.familyvoice.org/
FindaCure http://www.findacure.org.uk/
Fragile X Society http://www.fragilex.org.uk/form
Genetic Alliance UK http://www.geneticalliance.org.uk/
Genetic Disorders UK http://www.geneticdisordersuk.org/
GenomeConnect https://www.genomeconnect.org/
Joubert syndrome (https://twitter.com/jsrdf)
KBG Foundation http://www.kbgfoundation.com/contact-us.html
Kids http://www.kids.org.uk/
Kidz to Adultz https://www.kidzexhibitions.co.uk
Kleefstra Syndrome group http://www.kleefstrasyndrome.org/
Learning Disability Carers Community http://www.ldcarerscommunity.org.uk/
Learning Disability Today https://www.learningdisabilitytoday.co.uk/ldt-home
Max Appeal! http://www.maxappeal.org.uk/
MecP2 syndrome France http://www.duplication-mecp2.fr/
Medella Mental & Behavioural Health 22q11.2 http://medellahealth.org/contact/
Parenting special needs https://parentingspecialneeds.org/contact/
Phelan-McDermid Syndrome http://www.pmsf.org.uk/contact-us/
Pitt-Hopkins Syndrome support group https://pitthopkins.org/
Pos'ability magazine https://posabilitymagazine.co.uk/
Potocki-Lupski Syndrome Outreach Foundation http://ptlsfoundation.org/
Prader-Willi Syndrome Association UK http://www.pwsa.co.uk/
PTLS Foundation https://twitter.com/PTLSfoundation
PURA Syndrome group https://www.purasyndrome.org/
Rare Disease Review https://www.rarediseasereview.org/
Rare Diseases South Africa http://www.rarediseases.co.za/
Rare Revolution Magazine http://www.rarerevolutionmagazine.com/
RareConnect http://www.rareconnect.org/en
Rett Syndrome http://www.rettuk.org/
Ring20 Research Support http://ring20researchsupport.co.uk/
Rubinstein-Taybi Syndrome Support group http://rtsuk.org/meet-the-team/
Simons VIP Connect https://simonsvipconnect.org/
Smith Magenis Syndrome Foundation UK http://www.smith-magenis.co.uk/
Special Needs Jungle https://www.specialneedsjungle.com/

SWAN UK http://www.undiagnosed.org.uk/
SYNGAP1 Education and Research Foundation http://www.bridgesyngap.org/#!research-/ch2r
The Challenging Behaviour Foundation http://www.challengingbehaviour.org.uk/cbf-resources/information-sheets/mental-health-problems.html
The Fragile X Society http://www.fragilex.org.uk/
The Judith Trust http://www.judithtrust.org.uk/about-us/
The 22q Crew
Tuberous Sclerosis organisation http://www.tuberous-sclerosis.org/
Unique http://www.rarechromo.org/html/home.asp
VCFS (Velo-Cardio-Facial Syndrome/22q11.2 deletion) http://www.vcfsfa.org.au/
Williams Syndrome Foundation http://www.williams-syndrome.org.uk/
Wolf-Hirschhorn Syndrome group http://whs4pminus.co.uk/

In addition to these collaborations, we have also been featured in a number of magazine publications including:

Rare Revolution magazine included a feature article about the project, written by Professor Lucy Raymond as part of their Winter 2017 "Rare Minds" issue. This followed on from a shorter promotional piece in the Summer 2017 edition of the magazine.

Enable magazine, the UK's leading disability and lifestyle magazine, has published pieces about IMAGINE ID to raise awareness of the project and encourage families to join in 2017 and 2018.

12

IMAGINE ID

stands for Intellectual Disability and Mental Health: Assessing the Genomic Impact on Neurodevelopment.

It is a research study led by University of Cambridge, University College London and Cardiff University that aims to answer a question that parents often ask when they learn that their child has a genetic condition "So what does this mean for my child?"

Its researchers are collecting information about a large group of children with intellectual disability and doing research to find out how genetic changes affect children and young people's behaviour. The study hopes to help with the care of families and children now, and in the future.

This is one family's experience of taking part in the study:

When my son Ethan was diagnosed with a chromosomal abnormality, I immediately turned to the internet to research his condition and find out what it meant for his future, and for us as a family. I even bought a book on the condition. Whilst there was a lot of information available – an awful lot to take in in fact – disappointingly I found that it was overwhelmingly negative.

A long list of learning difficulties and things that he would never be able to achieve, but this didn't fit with the delightful boy sat in front of me.

He may never be able to write more than a few words but he can tell instantly if someone is feeling sad and somehow always manages to lighten a situation by saying something very caring and kind (unless it's his sister and then he just laughs and points – proof that he is just as normal as any other little brother!). He can be so selfless and thoughtful but all I was hearing from both my own research and from the mainstream school he was attending at that time was a long list of problems socialising and difficulties concentrating. When we completed the questionnaires for the IMAGINE ID study, it gave us a chance to describe the positive aspects of Ethan's personality, as well as facing up to his limitations and difficulties.

Coming from a scientific research background myself, and still working for a university today, I can understand the importance of scientific study and I was keen that we do what we can to support any research that could help other families in the future. It took quite a lot of time to fill in the Development & Wellbeing Assessment and the Strengths & Difficulties Questionnaire, but we were able to sit down when we had half an hour or so and complete them in small chunks, a little each night.

When we received our IMAGINE ID results, it was the first report that we had on our son (and we'd had lots by then!) that was a true description of him: his strengths as well as his weaknesses. It was perfect timing actually, as we were going through the difficult process of updating his EHC plan with the objective to move him from mainstream schooling to specialist provision.

The IMAGINE ID report was colour-coded and easy to interpret and I was very pleased that the information I had provided in the 'free text' boxes when I completed the questionnaires had been captured and recorded to form part of the report. It meant that I could stop repeating myself by giving anyone new to Ethan a copy of the report, and I felt supported in my own observations of him. Looking back at the report now, 18 months or so after it was produced, I am able to see the progress that Ethan has made and reminded of some of the difficulties we were struggling with at the time that are no longer so much of an issue, which gave me a bit of a boost.

The results also made me appreciate the stress that Ethan has been under sometimes, simply to try and fit in with 'normal' children and everyday life. I knew that he would make noises and strum his lips when we were in crowded situations but I'd only really looked at it from my own point of view; that people were staring or he was being noisy. Seeing the words 'Your child's score for overall stress is VERY HIGH' made me feel that it didn't matter if he was making a noise that gave the person ahead of me at the checkout a minute or so of irritation (and a crick in their neck for staring!) if it helped my little boy cope with the bright lights, background noise and crowds that were making him so stressed that he had no choice but to try his best to drown them out and find his own little piece of calm.

We were able to give a copy of the report to his new teachers and this has helped them to see the bigger picture that is our child and understand what his relatively rare syndrome actually means for him. I hope other parents and carers reading this will be encouraged to register for the study and help the researchers behind IMAGINE ID to obtain their target level of 5,000 participants by 2019.

Hayley Monk
Parent carer to Ethan with Williams Syndrome

Ethan and his sister looking at the book he received for taking part in the IMAGINE ID study

Our involvement with *FIND* magazine – a Special Educational Needs and Disability (SEND) newsletter covering the Lancashire region – arose after a mother of a participant contacted us because she wanted to publish an article describing her experience of taking part in the study with her son. This was published in Spring 2018 and subsequently picked up by other local news coverage including *Cambridge Network.*

Pos'ability Magazine, an innovative disability lifestyle magazine, will feature an online editorial on IMAGINE ID, as well as an advert for the project and a promotional piece about our children's book *Avery*. They have published various pieces across 2017 and 2018.

Autism Eye magazine a publication for parents and professionals with in-depth information and advice on autism published various pieces about the study and our aims/objectives of finding out more on autism in 2018.

We have also made a number of appearances on social media and the news to raise awareness of our study and findings, alongside the help from some of our families involved in the study!

In February 2019, for rare disease day, we shared a short video on Facebook about our research that assesses development through play-based activities, and included experiences of a family with a child with 16p11.2 duplication *(https://animoto.com/play/baR6a8NMV7Z6QiNbdlcrfg).* Professor van den Bree was also interviewed in February 2019 about 16p11.2 research for The Conversation *(https://www.spectrumnews.org/news/deletion-duplication-chromosome-16-segment-may-confer-autism-risk/).* Advance Wales included a feature article on Cardiff CNV research, based on an interview with Prof van den Bree.

In November 2018, Professor van den Bree interviewed about Cardiff studies of 22q11.2DS for BBC breakfast television by Sian Lloyd. *https://www.bbc.co.uk/programmes/m00017qj https://www.bbc.co.uk/news/uk-wales-46292480 https://twitter.com/BBCBreakfast*. Also see *http://www.cardiff.ac.uk/news/view/1371274-awareness-of-22q2018 (Feb).* Additionally Professor van den Bree was interviewed regarding her research on 22q11.2DS by BBC Wales Radio *https://www.bbc.co.uk/programmes/m00017qq.*

For Rare Disease Day 2018, the National Institute for Health Research (NIHR) interviewed Professor van den Bree: *https://www.nihr.ac.uk/news-and-events/support-our-campaigns/rare-disease-day-2018/echo-study.htm*

The Conversation and the Independent included a feature article on CNV research, based on interview with Cardiff PhD student Hayley Moulding in May 2017.

In August 2017 a film was created by our Cardiff team on motor problems and possible physical interventions to improve outcomes for children born with 22q11.2DS. *(https://www.youtube.com/watch?v=7k-Njq95yp0)*

In November 2016, the Cardiff team began a Cardiff University Mental Health Blog, discussing a wide range of things, including the experience of mental health research, mental health and well-being: *http://blogs.cardiff.ac.uk/mental-health/2016/11/09/an-undergraduate-students-experience-of-mental-health-research/*

In addition to sending regular newsletters to the 22 UK Regional Genetic Centres (RGCs) with updates on recruitment, family feedback and findings, we also continue to send newsletters at the end of each year to families. In December 2018, this newsletter was sent to over 2,800 families and included details of our upcoming Family Day (discussed below) as well as a final request to families to share with any further groups or families that may be suitable as recruitment nears closing in mid-2019. If they felt IMAGINE-ID had been a worthwhile study to be a part of and if they knew of other families who would benefit from the study, we asked if they could recommend us, invite a friend or share amongst any patient groups or charities they may be involved with. This was met with plenty of enthusiasm from families, offering to pass on information and promote us in various avenues:

"Your work is so valuable. We will share this with local groups"

We also sent out a mailshot to all families in September 2017 and continue to ask for feedback on the project, as we wanted to share this with the RGCs to show how families are benefiting from the IMAGINE-ID study. We asked if the study had been worthwhile, if they found the personalised report useful and what their overall experience of working with the IMAGINE-ID team had been. We received lots of feedback, with many families commenting on how beneficial they found the report and the extent to which they had shared it:

"Really easy to complete the questionnaire. The report is concise and helpful especially if you don't know how to phrase and express your child's difficulties. I loved receiving the little story book and have read it with my children. Thank you"

"The study completed by IMAGINE-ID team has been extremely beneficial to both our son and our family. It has been extremely useful passing on this information/report to doctors, teachers and specialists. …using this as additional support for Education Care Plans and PIP assessments… has ensured he receives the correct funding on his EHCP and gives his teachers/doctors an in-depth and updated guide to his conditions"

"We found the feedback to be extremely useful. It was wide ranging and gave us a clearer idea of our child's strengths and weaknesses, as well as identifying new areas of need which we hadn't previously considered"

One family also sent in a video, summarising how they had found the experience of taking part in the study, which we were able to share (with permission) with the RGC coordinators and local PI's who attended at our annual RGC Workshop in 2017.

We will continue to keep regular contact with the recruited families to keep them informed about the latest findings in the project and provide opportunities to maintain their engagement.

The F2F component of the IMAGINE-ID study has had wonderful feedback from parents:

'We had the visit today from 3 lovely ladies who completed the assessment needed.'

'I would just like to pass on feedback about how the visit went!'

"Myself and my partner were both very pleased in which the boys were accommodated during the session. Both of the boys have continued to talk about their experience with happiness and tell everyone they have seen today how fun it was. Myself and my partner both felt very comfortable at answering the questions needed and was constantly reassured throughout"

"Thank you for making it such an enjoyable experience for us and the boys"

"The research team were fantastic with my 6 year old Son. They were friendly, helpful, good listeners and knowledgeable. Thank you! Xxx "

IMAGINE-ID Website

Another medium we use to engage with our families is our website (http://imagine-id.org/). We continue to post regular news items on the News/Events page of our website, updating on public engagement events we may be attending, progress of the project or special features which may be of interest to families.

We also continue to receive a large influx of enquiries (on average 18 a week) from interested families coming through the website's "contact us" form, which are sent directly to our secure nhs.net email address. This is a perfect tool for self-referral patients to get in touch with us and for us to be able to send out initial study documentation to the families. Indeed, the majority of our self-referral families are recruited in this way.

Training School

In December 2018 in Skopje, Macedonia, Prof van den Bree, Dr Chawner and other Cardiff team members together with colleagues in Leuven (Prof Swillen), Maastricht (Prof van Amelsvoort), Warsaw (Dr Nowokowska) and Skopje (Dr Markovska-Simoska) developed and organised a Training School for Clinicians and Researchers, to improve understanding and care for CNV carriers.

PPI Events

A focus group was held in London in November 2018 to bring together IMAGINE-ID researchers and families. The focus group aimed to facilitate the opportunity for the IMAGINE-ID team to understand a parent's experience of IMAGINE-ID, as well as to get an idea of their thoughts and ideas for the future of our research. We asked parents to explain why they had wanted to get involved with the study, how they found this experience, their feedback on the personalised report they received, and whether they would find it useful to be involved in IMAGINE-ID in the future. We received lots of feedback, with many families commenting on why they were interested in our research, how useful the report had been and how important it is to follow-up with their children over time.

'I think for us it's about education, about learning, it's about also him understanding his condition.'

'What you tend to get is a diagnosis […] that's great, but what does it actually mean?'

'And I think one of the things that first appealed to me with the project, was having a holistic report about [child's name], because we see so many different professionals that all deal with one little bit of him or one aspect of him, but we've never really had a holistic view from a professional source, so that was really appealing.'

'We are passionate parents who see the benefit, not just to themselves but also to other families who may be on a similar journey to them, to be involved in research like IMAGINE-ID.'

Parents understood the need for research like IMAGINE-ID and for its continuing research, so that we can better our understanding of how their child's mental and physical health changes over time.

'*We're interested in sharing information and the more we find out the more we can help other people, and other people can help us.'*

'There aren't that many people that we know, apart from us today, that have "been there before us", but there'll be many coming after us, so anything we can share, discuss and give assistance, I think why not it's for everyone's benefit'

In addition to the focus group, we encouraged families to give feedback on the study if they were not available to join the focus group. We received lots of feedback again, with lots of encouragement about the importance of IMAGINE-2 and their keenness to be involved.

'Going forward with it, the more we can learn […] the better informed as to why one child has one condition and another child is different or why it's so varied. That's why I think the study [IMAGINE-ID is] doing is so important.'

'I think [IMAGINE-2 is] relevant because the situation changes, diagnoses change'

'If the study does continue I would very interested. Five years on will be very interesting to see where the 5/6 year olds are, as they will then be 10/11.'

'I have plenty of time to be able to answer questions in any follow up studies, and would be happy to help.'/ 'I am happy to spare time completing further questionnaires.' / 'I would be able to spend a few hours answering questions in the follow up.'

Contributions to Events and Wider Collaborations

There have been several events in the past year that IMAGINE-ID teams have either directly hosted, or to which we have contributed as part of a wider collaboration.

2019 (March) Prof Lucy Raymond presented at the Cambridge Autism Genetics Symposium

2018 (Feb) Prof van den Bree presented IMAGINE-ID findings for the Centre for Genomic Medicine, Manchester.

2018 (Feb): Prof van den Bree invited to present for the Centre for Genomic Medicine, Manchester.

2018 (Feb) Prof van den Bree invited to present at NMHRI Centre Neurodevelopmental Disorder Research Day at to celebrate the opening of the new Child Development Centre, Cardiff.

2017 (Nov) Prof van den Bree presents CNV research findings to the Belgium Ambassador in the UK, Ambassador Rudolf Huygelen, Cardiff.

2017 (Nov) Dr Samuel Chawner presents at the Unique rare chromosome organisation North-West family day.

2017 (May) Dr Samuel Chawner and other IMAGINE-ID Cardiff team members took part in the Cardiff Rare Disease Showcase Lightning Presentations.

2017 (May) Speaking of Science. PhD student Hayley Moulding in the Cardiff group was interviewed about genetics, aimed at general public and students.

2017 (May) Welsh Government - Mental Health Awareness Week. PhD student Hayley Moulding in the Cardiff group gave a presentation to staff members, Policy makers, service users.

2017 (May) S4C - "Matt Johnson - Depression and Me". PhD student Hayley Moulding and IMAGINE-ID team members in the Cardiff group gave a presentation to the general public.

2017 (Feb) Welsh Government - Mind Matters. PhD student Hayley Moulding in the Cardiff group gave a presentation to staff members, policy makers, service users about genomic disorders.

2016 (Dec) PhD student Hayley Moulding in the Cardiff group gave a presentation about genetics to teachers and European partners for the Erasmus+ - European Commission Office, Wales

2016 (Dec) Science Café. PhD student Hayley Moulding in the Cardiff group gave a presentation the the general public about genetics.

2016 (Nov) Scouts Neuroscience Night contribution by Cardiff PhD student Adam Cunningham.

2016 (Nov) Dr Samuel Chawner and PhD student Hayley Moulding from Cardiff contribute to the ESRC Social Science Festival Panel Debate.

2016 (Oct) van den Bree, M.B.M. Findings from Our Studies of 16p11.2 Deletion and Duplication. Presentation at 16p11.2 family day, Cardiff.

2016 (Oct) Erasmus+ - Ysgol Bro Eirwg. PhD student Hayley Moulding in the Cardiff group gives presentation on genetics to teachers and primary school teachers.

2016 (Oct) Genetic Alliance meeting for families affected by 16p11.2 CNV. Presentations and volunteering by the Cardiff IMAGINE-ID team.

2016 (Sept) Dr Samuel Chawner is Brain Bee volunteer for project with 6th forms students.

2016 (July) Professors Hall and van den Bree and Dr Chawner presented for clinicians of the All Wales Genetics clinic.

2016 (July) Dreams Workshop
PhD student Hayley Moulding in the Cardiff group gave a presentation about genetics to students of Grangetown Primary School.

2016 (Jun) van den Bree, M.B.M. New advances in research on copy number variants relevant to psychiatry. Royal College of Psychiatrists International Conference, London.

2016 (June) Dr Samuel Chawner presented at the Unique rare chromosome organisation family day in Scotland.

2016 (June) Life Sciences Quiz. PhD student Sinead Morrison in the Cardiff group gave a presentation about genomic disorders to secondary school children.

2016 (Feb) van den Bree, M.B.M. Intellectual disability and psychiatric disorder in 22q11.2 Deletion Syndrome (22q11.2DS). Meeting for Intellectual Disability clinicians by the National Centre for Mental Health (NCMH), Cardiff.

Regional Genetics Centre Knowledge Exchange Workshops
October 2016 and September 2017
Workshops organised and led by the IMAGINE ID team to discuss genetics research, how research is coordinated within the UK NHS Regional Genetics services and provide an opportunity to exchange tips and network with other professionals working in genetics.

Cambridge Science Festival
March 2017, Cambridge
Professor Raymond presented an evening talk entitled 'Parents as partners in research', in which she discussed the innovative method in which we are collecting data from parents and utilising their expertise in their children's behaviours by means of online assessments. The evening also included a reading of the children's book AVERY.
In addition, we had an exhibition stand at the "hands-on" weekend event; an interactive session wherein we showcased the latest assessments available in the project and engaged with patients and families.

Living with a Rare Disease – British Paediatric Surveillance Unit
March 2017, London
IMAGINE-ID exhibited at an informal networking event, focusing on rare childhood diseases and disorders, that brought together patients and carers, healthcare professionals, researchers and policy-makers.

BRAINFest,
June 2017, Cambridge
IMAGINE-ID team exhibited at the interactive showcase event of the Cambridge Neuroscience Festival of Brain Science, which was designed to bring together neuroscientists from across Cambridge University. This also included a VIP invite-only evening reception, where we engaged with high-profile and influential attendees.

Public voices shaping health research
June 2017, Ely
IMAGINE-ID exhibited in the pop-up village at this public involvement forum, focusing on the importance of patients, carers and the public in the future of health research.

Charity/Support group Family Days *e.g. UNIQUE, Ring20, PTEN*
June, August & November 2017, February 2018
Warrington, Liverpool, London
We have attended various patient group and charity family days this year including those hosted by the rare chromosome charity UNIQUE, as well as Ring20 and PTEN groups. These events provide a wonderful opportunity for us to meet with families and to provide information about our study to relevant families.

Findacure Rare Disease Showcase
May, August and October 2017
Cardiff, Cambridge & Newcastle
Members of the team presented a lightning talk at the Cardiff and Cambridge events to highlight the study to attendees from local pharma, patient support groups and research groups. At the Newcastle event IMAGINE-ID had an exhibition stand to provide information to local researchers and clinical staff.

Cambridge Rare Disease Network Summit
October 2017, Cambridge
Exhibited at the annual CRDN conference meeting in Cambridge, joining other members of the rare disease community, including patients, charity/support group representatives and researchers.

Surrey NHS Developmental Paediatrician Study Day
November 2017, Woking
Following communication with some community paediatrician's eager to recruit to IMAGINE ID, we were invited to attend the Surrey NHS Developmental Paediatrician Study Day. Here we presented the project and engaged in valuable discussion with the local community paediatricians.

Kidz to Adultz North
November 2017, Manchester
A new venture for the team this year was attending one of the Kidz to Adultz Exhibitions, hosted by Disabled Living. These events are totally dedicated to children and young adults up to 25 years with disabilities and additional needs. We had an exhibition stand, providing information and spoke with many parents and carers, as well as professionals who support and work with them.

NHS Clinical Genetics Study Day
February, 2018, Cambridge
Exhibited IMAGINE-ID study material at the Cambridge University Hospital Clinical Genetics Study day, attended by healthcare professionals and researchers.

Cardiff Rare Disease Showcase
May 2017, Cardiff
Dr Samuel Chawner presented a 5 minute 'lightning talk' at the event hosted by Find A Cure charity to other researchers, industry and patient groups in Cardiff Bay.

Pint of Science
May 2017, Cardiff
The Cardiff University team was involved in the organisation of the first ever Pint of Science festival in Cardiff which brought researchers to a local institution within the city to talk and engage with the public about science and the current research currently being undertaken in the University.

NHS All Wales Medical Genetic Service Multidisciplinary Team Meetings – The Cardiff University team have organised collaborative multidisciplinary clinical meetings with local medical genetics services.

Library of Imagined Genes
September 2017, Cardiff
The Cardiff University team was involved in the second instalment of this interactive performance and installation helping the public understand the relationship between genetics, behaviour and emotions. The team also helped with the first instalment back in 2016 which took place in Green Man Festival.

22q Ireland European Alliance Meeting
October 2017, Cardiff
Several members of the Cardiff University team presented at a European meeting for patients and families affected by 22q11.2 Deletion Syndrome.

The World Congress of Psychiatric Genetics
October 2017, Orlando
Samuel Chawner showcased the work of IMAGINE-ID at an international conference to showcase the IMAGINE-ID study. He was awarded early career presentation finalist and a Guarantors of Brain travel grant.

Cardiff Rare Diseases Patient Networking Day
October 2017, Cardiff
The Cardiff University team had an exhibition stand open to the public at the event. The event held talks from expert speakers discussing the latest advances in the field of rare diseases and also had patient's talking about their experiences and perspectives on living with these conditions.

MRC's Millennium Medal Science Showcase
Professor Jeremy Hall attended the event and showcased the IMAGINE-ID study as a "lifelong mental health" exhibitor. This entailed raising awareness of the study to parliamentarians and policymakers.

Genetic Disorders UK Leadership Symposium
March 2018, London

IMAGINE-ID exhibited at the Genetics Disorders UK Symposium, which gathers leaders of UK-based genetic disorder charities and patient support groups.

Cambridge Science Festival 2018
March 2018, Cambridge
IMAGINE ID exhibited at the Cambridge Biomedical Campus, where the focus is more on life-changing research conducted in the labs, hospitals and pharmaceutical companies on the site and how this, along with developing new treatments and medicines, is benefiting society. Again, we displayed the latest assessments available in the project and engage with patients and families.

Kidz to Adultz Arena Events 2018
Coventry, Farnborough, Bristol, Manchester (March, May, July and November 2018)
Exhibition stands at Kidz to Adultz events to raise public awareness and encourage recruitment into the project for families with affected children and young adults.

RareFEST exhibition
December 2018, Cambridge
Exhibition stand at international event to promote rare disease awareness and research to the public, families, patients, managers and CEOs of international patient support groups and charities.

***Seattle Club* Conference**
December 2018, Shrewsbury
Poster presentation and exhibition stand at annual conference for international researchers to present research in intellectual and developmental disabilities.

Angelman Syndrome Support Education & Research Trust UK Family Conference
August 2018, Coventry
Exhibition stand at national family conference for individuals with Angelman syndrome to promote awareness and research to eligible families.

Leicester Local Offer Live
May 2018, Leicester
Exhibition stand at regional event to promote rare disease awareness and research to the public, families, patients, managers and CEOs of patient support groups and charities.
Genetics Research Matters: Training and Development Event UK Clinical Research Network
June 2018, London
The study coordinator from Cambridge gave a training session to all UK Genetics-based trainees, research nurses and coordinators through the UK CRN informing them about IMAGINE ID, the recruitment procedures and the latest findings, as well as feedback from the families of what it is like to take part in the study.

NIHR Clinical Research Network East Midlands and West Midlands Inaugural Genetics Speciality Day, January 2019, Lichfield
Presentation to research practitioners and early career researchers to publicise successes and challenges of the study and share good practices about involving the UK CRN in study processes.

Book produced for distribution, free of charge, to all participants in IMAGINE-ID

Avery

Author and Illustrator: Marta Altes
In collaboration with Prof Lucy Raymond
Published: March 2017

The idea of writing *Avery,* a children's story about having a rare disease, emerged whilst applying for research ethics approval for IMAGINE-ID. To recruit children with a rare disease into research, parents are asked to consent on behalf of their children. Standard consent forms are legalistic and formal, and adapting these for children usually involves simplifying the language of the consent. This standard approach does not fundamentally address the process of children's understanding and does not adapt consent to children's own methodology of learning and discussing their concerns. Prof Raymond worked in collaboration with Marta Altes, a children's book illustrator, to produce a children's story where the issues of taking part in research are presented and discussed in an approachable way. The intention was to produce high quality literature as a tool or vehicle for more profound discussions between parents and children about their rare disease. Although the IMAGINE-ID project is the source of inspiration for the book, the material is deliberately generic to be used by anyone considering recruiting children into rare disease research in general.

By June 2017, copies of *Avery* had been sent out to all retrospective participants, after which we were able to focus on sending out to participants at time of them joining the study. Copies are sent

to all under 16's and Consultees. Over 1,900 copies have been sent directly to participating families so far.

Due to overwhelming interest in *Avery*, not only from others in the rare disease and genetic disorders communities, but from fellow academics, hospital departments, patient support groups and the general public, we felt there was a much wider reach of *Avery*, as the story is not specific to any particular disorder.

We received numerous requests to make *Avery* available to purchase and from July 2017, it became available to buy online via Cambridge University e-sales store.
https://onlinesales.admin.cam.ac.uk/product-catalogue/products/schools-faculties-departments-and-institutions/cimr/avery

Avery is available to purchase within the UK (£8) and internationally (£10) – both prices include postage. To date, over 420 copies of *Avery* have been sold online so far for a total sum of nearly £3200. Costs for the postage, packaging as well as the book itself need to come out of this figure but any money arising from these sales will be used within the study – possibly to fund a further print run or a family day for the participants.

Recipients have ranged from pharmaceutical giant Roche, Cambridge University Alumni, schools and libraries, family members and genetic disorders groups. Our feedback form shows that these people have heard about *Avery* through social media – promoted by ourselves, patient support groups and the rare disease community - as well as from our website, various news articles and word of mouth.

Copies of *Avery* have also been sent to numerous groups and organisations in order to promote the study and aid recruitment. UNIQUE has received just under 200 copies, over 200 copies were sent to Child Development Centres across the country, every RGC were sent 5+ copies, and a copy has been sent to many rare disease charities/support groups, whose families are eligible to join the study. Avery has also been sent to news/media groups who have gone on to write articles and promote the book and raise awareness of the research.

Feedback on *Avery*

The response to *Avery* has been overwhelmingly positive.
A link to SurveyMonkey is sent out with the book and below is a summary of the feedback we have received so far from 42 respondents:
Has the book helped you feel more included in the wider rare disease community? Yes = 93%
Has this book helped you/your family to talk more about rare diseases? Yes = 78%
Rate the book (out of 10) AVERAGE = 9.1

Written feedback comments have also been sent in to us:

"It's the most wonderful story ever. I can't describe how much it describes our family – my little girl is excited to show her teachers."

"What a lovely book! …It brought tears to my eyes. A lesson learned, we are not all the same and how boring it would be if we were!"

"The words just sum it up for our family and such lovely illustrations. Thank you, we will enjoy sharing it with others."

"It is a beautiful book, I adore the images. The message is so clear and powerful… I am certain my daughter will love the book and it will help her to know and understand that as a family we are part of an aviary, not flying solo."

"It is a lovely way to express a difficult and poignant issue. We will cherish it."

"Your book is a very good way of telling children that life can also be different."

and perhaps the most touching comment is from a brother…

"Mum, have you read this book? You've got to read it! It's just like me and Aiden. The little bird finds things difficult like Aiden does, but the brother bird is always with him, helping him, just like I help Aiden."

Avery has been featured in…

There has been much interest in *Avery* following its publication which has prompted requests for interviews and features in the media and other publications.

Professor Lucy Raymond has been interviewed on BBC Radio Cambridgeshire and That's TV Cambridge. A review was also features on the ITV news website (http://www.itv.com/news/anglia/update/2017-08-31/academic-writes-childrens-book-to-help-kids-with-genetic-illness/).

We have been approached to write promotional pieces on *Avery*, and have been featured in both the Rare Revolution Magazine Summer 2017 issue (http://edition.pagesuite-professional.co.uk/html5/reader/production/default.aspx?pubname=&edid=ed3b359c-0955-4560-8c76-60e7a9e5b6cd) and Pos'ability magazine (Feb/March 2018 issue).
The University of Cambridge has taken a particular interest in *Avery*, featuring reviews in Cambridge University Alumni e-newsletter and the Horizon (research highlights) magazine (https://issuu.com/uni_cambridge/docs/issue_34_research_horizons/4), as well as on the University's Medium page (https://medium.com/@cambridge_uni/avery-the-little-bird-helping-children-understand-genetic-illness-af8a12ef1adf)

Avery has also featured in a number of rare disease patient support e-newsletters, such as the Cambridge Rare Disease Network newsletter (May 2017).

In addition to this, *Avery* has been reviewed on a variety of scientific blogs, including Sciblogs (https://sciblogs.co.nz/tag/avery/), Enseqclopedia (http://enseqlopedia.com/2017/10/explaining-rare-disease-bedtime-storytime/) and Bionews (http://www.bionews.org.uk/page_859698.asp).

Family Day 2019

As a thank you to our families for taking part in the project and to provide a rare opportunity for families to meet each other and share their experiences we are organising a special Family Fun day to take place on Sunday 7th July 2019. This day has been organised in partnership with charity, UNIQUE, and will include arts and craft workshops led by Avery and children's books illustrator, Marta Altes. There will also be some science-led activities, a reading corner, puzzles, soft play, a silent disco and specific sensory needs play equipment. It will also be an opportunity for parents to speak to the researchers, find out more about the project and disseminate the findings to a wider

group of families. Places have been reserved for over 200 people to attend the day, with many more on the waiting list.

Policy implications

Prof Lucy Raymond is an invited member of the recently established Rare Disease Working Transition Group chaired by Prof Sue Hill, Chief Scientific Officer for NHSE. This group is advising on the policy, needs and provision of genetic tests for the NHSE from 2018 onwards. With respect to the provisioning of genetic testing for intellectual disability IMAGINE-ID has proved an invaluable source of data to inform planning. *The age of diagnosis of a genomic CNV abnormality has systematically decreased from 10yrs to 2.5 yrs. over the past 10 years* whilst the age of diagnosis of single gene defects other than Fragile X has remained at 7-9 yrs.

Based on the IMAGINE-ID data and that of Genomics England, provisioning of whole genome sequence analysis for moderate to severe intellectual disability is recommended.

Work in the Rare Disease Working Transition Group has continued through 2017-18. We aim to establish service specifications including novel methods of triage, to identify who should be offered whole genome sequence analysis, or microarray assessment only. We envisage data from the on-line medical questionnaire we are introducing shortly to support phenotyping on-line will inform this process, and that the on-line triage procedure will become routinely used by community paediatricians.