Focus

Issue 7
August 2017

Chair's foreword

Dear Members,

I am pleased to introduce the 7th edition of our Focus magazine. A lot has happened since the last Focus magazine was released in February. We had the London Marathon in April, which raised an amazing £220,000 for MACS, you can read more about this on page 9. Then at the beginning of May we had our Family Weekend. This year we had a number of break-out sessions on the Saturday and the feedback we received suggested you all thought that was a positive change so we plan to build on that in 2018. You can read more about the Family Weekend on pages 4 and 5. Next year's Family Weekend will be taking place in Windsor and booking has now opened so please go to **http://macs.org.uk/macs-2018-family-weekend-booking-form/** to book your place. Also in May we talked to you about our 'MACS: The Future' research and encouraged you to tell us your views in a survey. We were blown away by both the quantity and quality of the responses we received. Fay's article on page 12 tells you a bit more about the survey results along with revealing the winner of the week-long caravan holiday! In June, MACS dad Tim Lupton ran his 100th marathon for MACS – you can read all about this incredible achievement on pages 10 and 11. Later in June, we won one of the FSI Small Charity BIG Impact awards thanks to the fantastic responses to our survey – Fay talks about this a bit more on page 20. In July, we revealed our new website along with our brand new video (if you haven't seen them yet, please check them out at www.macs.org.uk). RideLondon took place at the end of July and raised a fantastic £18,000 (and counting) for MACS. Now we are into August, we are running our activity holidays – first the Caldecotte trips and then the sailing trip later in the month.

So, as you can see, it has been a very busy time for MACS but I wouldn't have it any other way. I hope you enjoy this edition of the Focus magazine and I hope to see you at a MACS event soon.

Robbie

Introducing our new Trustees

Caroline Hornby
Caroline lives in Nottinghamshire with her husband Steven and their 3 children Jake 6, twins Joshua and Reece 3 who has Bilateral Anophthalmia and is partially deaf with growth development delays.

Caroline and her family found out about MACS after birth of their twins in Salisbury, Wiltshire where the charity have provided valuable support and guidance and now feels she would like to help out and be more involved within the MACS community.

Caroline sits on the Caravan Sub-committee and volunteers with the administration of the caravans in Cornwall and Dorset. She is currently working towards a CIPD diploma and in her spare time she enjoys sewing and cross country. Caroline is honoured to have become a Trustee and says: "MACS has provided valuable support and guidance to our family and now I feel I would like to help out and be more involved within the MACS community".

Caroline Hornby

Becky Hardisty

Becky Hardisty
Becky is a MACS adult whose family have been MACS members since she was seven. Becky was born with bilateral Microphthalmia and Coloboma. Her elder brother, Matthew, was born with bilateral Anophthalmia.

She currently works as a social worker with adults with a range of disabilities and needs. She lives in West Lancashire with her son Connor and partner Oli.

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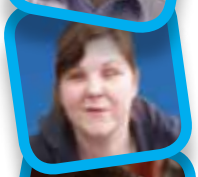

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New member gallery

Say hello some of our newest young members…

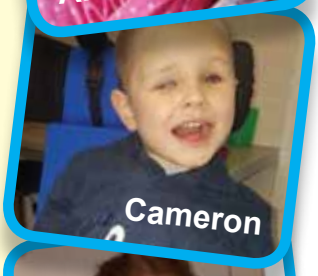
Joshua is 6 months old and has Microphthalmia in his right eye with nystagmus and a cataract. His mum is Anna and they live in County Durham.

Annie will be 1 next month. She has Microphthalmia in her right eye. Annie's mum is Sarah and they are from Northern Ireland.

Cameron is 4 and a half, has Bilateral Microphthalmia with Norrie Disease and lives in Bristol with parents Carla and Adam.

Bailey is 5 years and 10 months. She has Bilateral Coloboma, right eye Microphthalmia, optic nerve Hyperplasia and Charge Syndrome. Bailey's Mum and Dad are Lianne and Gordon and they live in Southern Scotland.

Finn is 3 years old and has Microphthalmia in his right eye. Finn's Mum and Dad are Micheala and Shane. They all live in Northern Ireland.



Contents

‘One of the best weekends ever!’: Family Weekend 2017

As anyone who has been will tell you, the Family Weekend is the highlight of the year in MACS. It’s always fantastic to meet old friends and new, knowing that you are surrounded by people who understand you, your family and the things you are going through.

This year was a mix of old and new. Old favourites like the chocolate fountain, fancy dress (there were some fantastic pirates and princesses!), and of course, MACS the Monkey made an appearance. But this year, the Saturday programme was a bit different.

The AGM was much shorter, and then we held a number of breakout and workshops, including sessions on prosthetics, a session on supporting your visually impaired child, a workshop on the future of MACS. This new format was really successful: instant voting feedback for all these sessions was 100% positive, and one attendee said ‘the subtle change to the weekend’s itinerary worked very well - one of the best weekends ever!’

We tried some new things with our younger guests, too. Young adults aged over 16 attended a confidence building session, Dare2Lead, whilst our older children aged 10-18 spent the day at Rock and River, canoeing, climbing and having a fantastic time. And the younger children got the chance to enjoy the sensory bus (an old favourite) as well as all sorts of exciting animals at the mobile zoo.

On Sunday evening, we were once again blown away by the amazing voices, jokes and funky dance moves of our young superstars in ‘MACS has got talent”. All the young people who got up on stage to perform in front of hundreds of people were incredibly brave, and we were all very proud and inspired by them.

We can’t wait for family weekend 2018 to see our friends again and make new ones. And next year, we’ll be in a new venue! (But for all of you who have a special place in their hearts for Park Hall, don’t worry, we plan to be back in 2019!)

MACS Achievement Awards 2017

Sunday night at the Family Weekend saw the announcement of MACS Achievement Awards. The winners were picked across four categories: 0-6 years old, 7 to 12 years, 13+ years and Sibling awards. We received as many as 26 nominations showcasing some amazing achievements, and picking the winners was not an easy task!

In the category of MACS child aged 0-6 years, we had five fantastic nominees and the winner was **Faith Moyle**, aged 6, who had overcome her shyness to sing in front of hundreds of people. Her mum says “From being a shy reserved girl to this in the space of a year is a tremendous achievement for Faith and I’m so proud of her confidence and bravery”.

In the category of MACS child aged 7-12 years, we had nine nominees all of whom had achieved great things this year. The winner, **Amy Sharpe**, had needed to have a lot of operations this year but has managed to stay positive and smiling throughout. Amy’s mum added that besides bravely battling through her operations Amy had also been going through some hard times at school but is determined to work her way through this, and when she puts her mind to something she usually succeeds!

In the category of MACS person aged 13+, we had 2 brilliant nominees. The winner, **Megan Reed**, had decided to take up white-collar boxing and despite her eye sight and lack of depth perception, both key to the sport, she powered through with the training to the best of her ability and did well in her fight in November. She lost the match by one point but that didn’t matter because she proved she could do something when she was told she couldn’t,

Dare2Lead

***Dare2Lead* was a half-day workshop aimed at MACS members over 16 and covered journey to success, overcoming challenges and reaching potential. The session was run by CEO of *Dare2Lead,* John Loughton and delved into the realm of confidence, future life achievement and leadership styles, overcoming negative mindset, being influential, communication techniques, positive mindset and general confidence building with the aim to help transform skills in job seeking, creating change in society and wider life goals. This was a first for MACS and we were extremely excited to offer this experience to our MACS participants.**

MACS young person **Laura Eggins** attended the session. This is what she had to say:

What did you enjoy the most about the session?

Looking back on the *Dare2Lead* workshop the whole session was enjoyable from start to finish. The bit that I enjoyed the most was when we were in groups and we had to draw our ideal person and write inside what our ideal boss would be. This made me think about what I would like in a boss when I go to work.

Laura Eggins

What are the most interesting things you learnt?

One of the most interesting things that I learnt during the session was that **you can succeed in what you put your mind to.** Also, I learnt that if I keep a positive mindset it will help me to transform my skills in to my future job searches.

Do you think it will help you in your future pursuits?

The session has certainly helped me and will help me with my confidence when I take part in future events and pursuits.

What did you enjoy most about the Family Weekend?

At the Family Weekend the thing I enjoyed the most was the Pirate and Princess meal on the Saturday Evening. I enjoyed the evening because it is a chance for me to dress up and I enjoyed seeing what ideas other families come up with. They were all individual and different and some of them made me laugh.

which is inspiring and an amazing achievement in itself. Meg also raised £200 for a local children's hospice in the process!

In the category of MACS sibling, we had 3 nominees all of whom were wonderful help to their siblings and parents. Our winner, **Ruby Gadsby Holt**, aged 11, helps her mum care for her younger sister whenever she is needed. Her mum says that "Ruby puts her sister Darcy before anything else and explains to her friends when they ask why her sister is different. She finds it hard sometimes when we are always at hospital but always tries to be helpful with Darcy. Despite being a young carer, she still manages to keep on top of her homework and see her friends".

Congratulations to all our MACS nominees and winners, you all deserve to feel very proud of yourselves!

Professor Nicky Ragge's eye genetics research update

Many MACS families will know Professor Nicky Ragge, who is a medical advisor to MACS and has been leading a national research programme into understanding the genetic basis to anophthalmia, microphthalmia and coloboma for over 14 years. She currently divides her time between her work in the NHS as a consultant clinical geneticist with specialist interest in eyes, and her research team at Oxford Brookes University. MACS has been supporting her research for many years, and this was paramount in the identification and characterisation of several key genes involved in eye development and MACS conditions, including SOX2, OTX2, STRA6, BMP4, BMP7, FOXE3, VSX2 and OLFM2.

This research would not be possible without the participation of affected families in this research, many of which are MACS members. Professor Ragge has now translated the research into new genetic tests available on the NHS.

A genetic diagnosis has been found for nearly a third of the families participating in our research programme, higher if the condition affects both eyes. The diagnostic rate is increasing with the pioneering work of the team in new gene identification, coupled with rapid advances in genetic testing methods. Receiving a diagnosis is often very helpful for families, who then have an explanation of why the eye anomalies occurred in the first place, and can receive accurate genetic counselling regarding recurrence risk, and access prenatal testing if desired. It also provides the families with more information about the particular conditions and recommendations for any screening that might be necessary. Although treatment with replacing eye tissue or enabling a small eye to grow is not possible right now, understanding the genetic pathways is a crucial step to enable Professor Ragge and others to explore new avenues towards this in the future.

One of the advanced testing methods Professor Ragge's group has developed is the Brookes-Birmingham Eye Gene Panel Test, which looks at changes in over 500 genes involved in eye development, and will be analysed by both clinical scientists in Birmingham and the research team in Oxford. The Eye Gene Panel test is now available through the NHS at Birmingham Women's Hospital to all MACS members nationwide, and can be initiated after a single appointment with Professor Ragge, during which she will also be able to recruit interested families into her research, as well as into the 100,000 Genomes Study, which may give some results in the future.

For all families in the study, the research team will be continuing to re-evaluate results and will test the samples that families have provided until a diagnosis is found. The research team would like to welcome new families to their research programme as with every family they recruit they learn more about different genes and are closer to find treatment options. **If you are interested, please call or email Professor Nicky Ragge's MACS funded family co-ordinator, Dr Dorine Bax (01865 484413, dbax@brookes.ac.uk).** Dorine is always pleased to hear from families that are already part of the study (or think they were recruited many years ago), and will give them an update.

Who's who in Prof. Ragge's team

Professor Nicola Ragge is a Consultant Geneticist, Birmingham Women's Hospital, Birmingham; honorary Consultant, Dept of Ophthalmology, Queen Elizabeth Hospital, Birmingham, and Professor in Medical Genetics, Oxford Brookes University, Oxford.

Nicola Ragge trained in Paediatrics before entering Ophthalmology. After her general Ophthalmology training, she undertook Fellowships in Paediatric Ophthalmology, Neuro-ophthalmology, Adnexal Surgery, Ophthalmic Genetics at centres including: UC, San Francisco, Children's Hospital Los Angeles, Great Ormond St Hospital and Moorfields Eye Hospital, London. She started to specialise in understanding the genetics of congenital eye anomalies, mainly anophthalmia and microphthalmia in 1999 whilst working in the Adnexal Dept at Moorfields Eye Hospital with Professor Richard Collin and set up a joint multidisciplinary clinic. In 2003, she was awarded a Senior Surgical Scientist Fellowship to establish a research programme in developmental eye genetics in Oxford whilst working as Consultant Paediatric Ophthalmologist in Moorfields and Birmingham Children's Hospital. She undertook specialist training in Clinical Genetics to become dual accredited in Genetics and Ophthalmology, and now works as a consultant geneticist, with a special interest in developmental eye genetics and runs a national laboratory research programme on Eye Genetics in Oxford. She has identified new genes and syndromes involving eye anomalies and has established new rapid diagnostic testing for these genes. She has wide-ranging research interests and has published over 80 papers. She has worked as Associate Editor of the Journal of Medical Genetics and is on the Committee for Eye Genetics Group (UK). She is medical advisor to MACS (Microphthalmia, Anophthalmia, and Coloboma Support).

Dr Dorine Bax is the MACS funded research and family co-ordinator in Professsor Ragge's team. She studied

Continued overleaf...

The Oxford research team, from left to right Dr Dorine Bax, Dr Fabiola Ceroni, Professor Nicola Ragge, and Dr Richard Holt (photo by Sheona Bellis).

Who's who in Prof. Ragge's team continued

Biomedical Science in Amsterdam, the Netherlands, and was awarded a PhD from the Erasmus University in Rotterdam, The Netherlands, for her research on the molecular biology of oesophageal cancer in 2005. She then continued her career in cancer research in the UK, working as a postdoctoral researcher at the Institute of Cancer Research in London and the University of Oxford, researching the molecular genetics of childhood brain tumours and lung cancer respectively. She moved to the human genetics field in 2015 to work in Professor Nicky Ragge's team, where she manages the growing research study.

Dr Richard Holt is a Research Fellow in Professor Nicola Ragge's team. After graduating from University College London with a degree in genetics, Richard worked for a year as a research assistant before undertaking a DPhil on the genetics of asthma at the Wellcome Trust Centre for Human Genetics, University of Oxford. After completion of his doctorate in 2007 he subsequently held postdoctoral positions within the university investigating the genetic causes of autism (Wellcome Trust Centre for Human Genetics) and retinal degeneration (Nuffield Department of Clinical Neurosciences). Since 2013, Richard has worked as a postdoctoral research scientist and Research Fellow investigating the genetics of developmental eye disorders within the group of Professor Nicola Ragge, based at Oxford Brookes University. (In addition to his work in genetics, Richard has maintained an active interest in multidisciplinary research, with a particular interest in the ethics of genetic testing. He is a member of the Oxford Forum for Science and Religion, and has obtained a diploma in theology at Regent's Park College, Oxford, and an MTh in Religion, Ethics and Society from the University of Winchester).

Dr Fabiola Ceroni is a Research Fellow who graduated from the University of Bologna in 2010, where she received both a Bachelor and a Masters Degree *cum laude* in Cellular and Molecular Biology. In 2011, she joined a PhD program developed in collaboration between the laboratories of Professor Elena Maestrini, at the University of Bologna, and Dr. Dianne Newbury, at the Wellcome Trust Centre for Human Genetics, University of Oxford. During this time, she investigated the genetic bases of two related neurodevelopmental disorders, autism and Specific Language Impairment. After completing her PhD, she continued her work on these disorders for a further two years, before joining the group of Professor Nicola Ragge in June 2016. Her current research is focused on the identification of genes causing severe congenital eye anomalies. In addition, she has a particular interest in the application of the latest high-throughput sequencing technologies both to identify these genes and to translate the findings into clinical practice.

Virgin London Marathon 2017

On 23rd April 2017 138 runners represented MACS in the Virgin Money London Marathon, tackling the 26.2 mile running challenge and raising over £200,000 for MACS in the process.

On the morning of the race the team gathered for a group photo commemorating the journey they shared in preparation for the event, and as they did so emotions ran high with nervousness and excitement in equal measure. The gun went off and the runners set off on a 26.2 mile challenge that would take them past some of the London's greatest landmarks. The London Marathon is known for its incredible crowd support with many thousands of people cheering along the course. Team MACS were also supported by two dedicated cheering points in the second half of the run, giving them a much-needed boost when they needed it most.

The first MACS runner to cross the finish line was Jarrod Logan, who came all the way from Australia to run for MACS and completed the run in an incredible 3 hours 12 minutes and 40 seconds. At the finish the runners were greeted by MACS Trustees Jenny Lupton and Gillian Bramley who took the chance to tell them more about MACS and thank them for their hard work. Despite the heat, all of our runners successfully completed the course and the final MACS runner finished in just under 8 hours.

The runners enjoyed the experience and commented:

"Brilliant experience. The run was very tough but the experience and support was great. It was inspiring to meet other MACS runners at the start and MACS families at the end."

"I think that MACS is an amazing charity and everyone made me feel so welcome and the support was outstanding! They made my experience the best!"

As a group, Team MACS 2017 has raised an incredible £200,000 so far, and the figure is still rising as their friends and family continue to sponsor them for their efforts.

If you would like to take part in the 2018 London Marathon you can already register your interest on our website: http://macs.org.uk/london-marathon-2018-interest-form/

MACS Dad, Tim Lupton, completes his 100th Marathon

Tim Lupton has been a member of MACS since 2005 and completed his first marathon in April 2010, at the time thinking that it would be his one and only marathon. Seven years later, on 23 June 2017, Tim completed his 100th marathon! We caught up with Tim to see how he is feeling having reached this incredible milestone.

What was the highlight of your road to 100 Marathons?

I think there are 2 best parts of my road to 100 marathons. First is being able to spread the word and awareness of MACS and the MACS conditions, and secondly is to run at the events where there are so many other MACS runners, such as London Marathon, Brighton Marathon, and even the half marathons at Reading and Royal Parks. The feeling of achievement when running these events is multiplied in my eyes as you are also aware that you have raised awareness and much needed funds when collecting the fundraising.

Do you have any favourite moments?

That is a hard question with so many different moments to remember over the years, BUT I think I would have to say my favourite moments boil down mainly to family, so reaching the finish lines of the marathons and seeing the family members there, and now having Ben by my side while I guide him around so many of these runs.

What about a favourite event?

There is, in my eyes, 2 categories for this:

My favourite event for a specific reason is Virgin Money London Marathon, this is because in 2010 it was my first and ONLY marathon I'd ever do (little did I know), the atmosphere, support, encouragement and it being my first was enough to lock it in as a very special and favourite event for me.

But I have since then discovered the trails, and these throw a whole new light on running as it works your muscles differently, they are more focused on getting the miles done rather than time (time is not my thing). For this reason I think my other favourite events (that are still going) would be Giants Head Marathon, held by White Star Running in the Dorset countryside or Shin Dig in the Shire in Shropshire held by How Hard Can It Be (and to be honest - VERY!) both

are very demanding courses with lots of inclines and declines hard and uneven under foot, but both are massively rewarding when you finish because you know you have done something absolutely amazing!! I am still trying to convince Kaja to join me on one of these!

What made you start running marathons?

In 2009 I had not run for anything, not even a bus, I was not the right side of 20 stone, and went to London Marathon to support the MACS runners and welcome them back into Horseguards Parade thanking them. It was while we were there that I saw, that what I thought was the marathon, was miles and miles away from the truth, as I had always thought that to run any distance but especially the longer distances you had to look a certain way (Sebastian Coe or Steve Cram) and unless you were like that you couldn't run. But I came away thinking if some of the people I'd seen completing that day for MACS and other charities could do it, Why couldn't I?... So on the train home I made the whole family laugh as I announced that I was thinking of doing the Marathon the following year! After lots of

training and 10k's and half marathons, I was at London and completed it! In the evening, when out at dinner, Jenny asked what it was like going past certain landmarks which I had no idea that I had passed!! But said NEVER again was I doing anything like that… BUT in May the ballot opened… Oh I might as well see… In October I found I didn't get in but I had started training, so I applied for a Charity place again.

At which point did you realise you would reach 100 marathons?

To carry on from the last question I did London every year since 2010 but in 2013 at the London Expo I became aware of these other marathons advertising, and one in particular caught my eye - Bournemouth. Bournemouth is just along the coast from me, and their marathon was a new one just starting in 2013 and it was in October so London in April keep training (or should I say try to) six months later Bournemouth, it sounded in my mind like a plan so I entered and got in. To cut a long story short, running down from Bournemouth AFC to Boscombe (a suburb of Bournemouth), I met these 3 lovely chatty ladies all running together at a slightly slower pace than I was doing but I wanted a quick breather so slowed down and chatted to them as well and found out that the Blue and Yellow club tops they were all wearing, were not a local club but a UK club, and that they all belonged to the UK 100 Marathon Club. I believe I am right in thinking that they had run in over 500 marathon between them. They also told me that there were a lot of other marathons held around the UK in smaller fields than the like of London and Bournemouth in fact groups as small as 50 or so runners, so when MACS decided to celebrate 20 years of MACS in 2014 I decided I'd run 20 events of half marathon or more and ended up running 11 Marathons and 9 half's, that is when I became hooked on taking part, so the following year I did 33 and last year 37 but I have enjoyed every one to a certain degree. If I ever completely stop enjoying them I will just stop.

What are your plans after the 100th marathon is done?

Until I reached about 90 I thought I'd just carry on doing marathons and guiding Ben as he wants. But when in circles of people things rub off on you, sometimes they are good things, sometimes bad things and sometimes they are MAD things! Well one of the MAD things going around in the circles of runners I mix with is the fact that a number of them are achieving, and some are getting quite close to doing, 52 marathons in 52 weeks, so this has perched itself on my shoulder and it is sitting there.

So as Giants Head Marathon is number 100 of the present it will also be number 1 of the 52 for the future!!!

Do you have any words of wisdom to someone who secretly hopes to complete a marathon but thinks it's not for them??

If anyone is considering doing a marathon - DO IT!

To the person doing the marathon, it is not about the day you do the actual 26.2 miles it is about the journey. You will struggle with time, with commitment, with a massive number of things BUT no one said running a marathon was easy, if it was, everyone would be running them. One of the facts that staggers me when I ever see the numbers at VMLM is that ' *"Less than one-tenth of 1 percent of the world's population will dare to complete the distance of a marathon."* That means **99.9% of the world's population will not do what many people who run a marathon, no matter their speed, have done - and that is to run a distance of 26.2 miles'.**

You will never change a marathon but a marathon will change you!

Do you not get bored running?

Yes I do!! But I always find that my mind can be distracted easily by thinking of other things, I normally think about the difference in awareness I can bring the charity while out there doing these runs.

Tim at the finish of his 99th marathon with son Ben who completed his 9th marathon and wife Jenny who completed 5K! 3rd June 2017

MACS is looking to the future...

The MACS team are currently undertaking a project to review the services MACS provides, and understand the impact that we have on our members. Over 180 people participated in the future of MACS survey. We were so happy to hear how MACS has helped so many families. It's also given us some good ideas for where MACS needs to go next, including more support for adults and giving more support to help with self-esteem and resilience.

We also held workshops at the family weekend, where we discussed ideas for the future of MACS, and we also asked children at the family weekend what they liked and what they'd like to be different. The next stage will be interviews with some MACS parents and adults, to get a detailed insight into a range of experiences and views about MACS and how it can help. If you are interested in participating in a detailed interview to share your views and help shape the future of MACS please contact **fay@macs.org.uk**.

We look forward to sharing the final report on the impact and future of MACS with our members when it's finished towards the end of 2017.

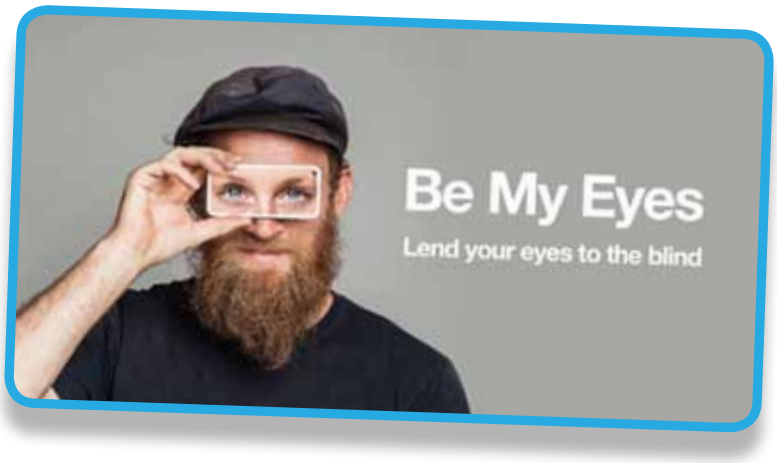
Early findings from our research project: views from parents of MACS kids

- **87% say that their child faces barriers in relationships and friendships as a result of their condition, and 86% say MACS helps their child meet others like them**
- **70% say their child's confidence is affected by their condition, and 62% say MACS has helped their child be more confident**
- **55% say they have experienced mental health problems as a result of their child's condition**
- **89% said MACS made them feel less alone in difficult times**
- **83% said MACS makes them more positive about their child's future**
- **91% said MACS helps them to cope with their child's condition**
- **65% wanted more help to deal with their child's school or local authority**

"Finding MACS when [our child] was born was an absolute ray of light moment, to know that other parents were out there sharing their experiences and that we weren't alone - which we absolutely felt at that time - was completely uplifting."

Congratulations to Claire Wolstenholme who won the week's holiday in a caravan!

Tech Talk



Welcome to the second edition of Tech Talk, the section where we talk about accessibility and accessible technology!

This time we want to look at 2 different apps that are available to download for android and iOS devices called *'TapTapSee'* and *'Be My Eyes'.*

TapTapSee is an app that you can download for android and iOS devices that uses your phone's camera and voiceover/speech technology to identify objects in front of you. You hold your phone in front of the object you want to identify, as if to take a photograph of it, and tap the screen. It will then accurately describe what the item you are looking at is. It is incredibly accurate and can give detail about specific items. For example, it can identify food packaging, money and cutlery and many other things. The great thing about this app is that it is very specific and accurate!

The next app we want to tell you about is an app called 'Be My Eyes'. Unfortunately, this app is only available on iOS devices at the moment but I believe they are working on an android version as well. This app is one that can be used by those who are blind or visually impaired, and sighted too! When you download the app you can register as a visually impaired user, or a sighted volunteer. If you sign up as a visually impaired user you can open the app to be connected with a sighted volunteer when you need help with something. If you sign up as a sighted volunteer you can make yourself available to help blind or visually impaired people. The app connects you live with a volunteer who you can ask to identify things for you, and uses your phone's rear camera so the volunteer can see what you are pointing your phone at so they can help you with what you need.

Both apps are great and very helpful. I personally use the 'Be My Eyes' app more often, but that is just personal choice.

If you have any ideas or would like to see any other apps or accessible technology in future editions of Focus magazine then please feel free to get in touch by emailing me at jonathan@macs.org.uk

Focus on MAC research with Dr Mariya Moosajee

New Medical Advisor for MACS

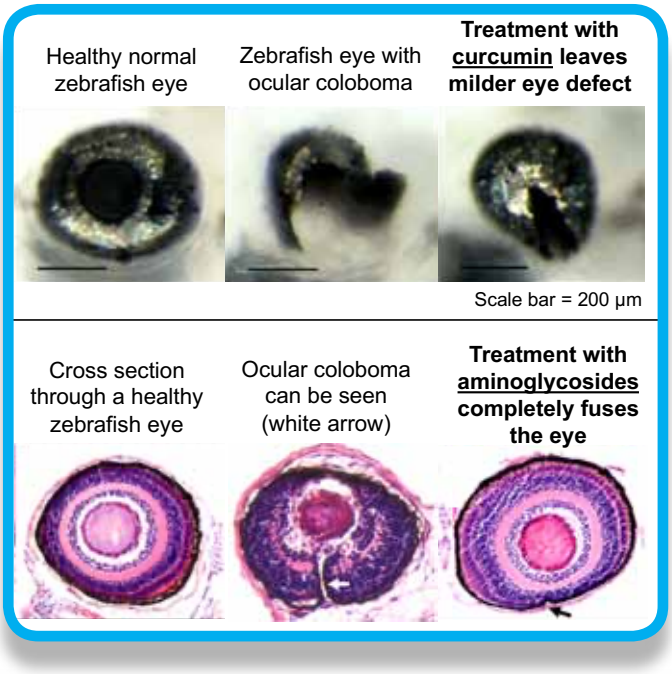
Dr Mariya Moosajee is our new Medical Advisor to MACS. She is a Consultant Ophthalmologist at Moorfields Eye Hospital and Great Ormond Street Hospital for Children specialising in Genetic Eye Disease, and Senior Clinical Lecturer at UCL Institute of Ophthalmology, London. She graduated with First Class Honours in Biochemistry and Molecular Genetics in 2000, Medicine (MBBS) in 2003, and was awarded her PhD in Molecular Ophthalmology in 2009 all from Imperial College London. She completed her National Institute of Health Research (NIHR) Academic Clinical Lectureship in 2016 and became the first Ophthalmologist to be awarded the prestigious Wellcome Trust Clinical Research Development Fellowship. Her current clinical focus is developing a Genomic Medicine service for children and adults affected with genetic eye disease with a particular focus on MAC conditions.

Dr Moosajee leads a research group at UCL Institute of Ophthalmology focused on advancing our understanding of the genetic basis of normal human eye development and the MAC conditions, using relevant disease models, human induced pluripotent stem cell derived retinal cells and medical bioinformatics. Her research has brought her international and national acclaim winning over 25 prizes including twice winner of the Foulds Trophy awarded by the Royal College of Ophthalmologists, Best Young Scientist Prize by the European Paediatric Ophthalmological Society, and the Members-in-Training Prize from the Association of Research for Vision and Ophthalmology (ARVO). Through dissecting the molecular and cellular pathways of disease, Dr Moosajee and her team have identified therapeutic targets and developed treatment strategies for clinical translation to patients.

Over the past 10 years Dr Mariya Moosajee has made advances in understanding how genes involved in eye development cause MAC and by dissecting the disease mechanisms has developed therapies that have actually shown closure of the ocular coloboma (cleft defect of the globe) and restoration of the normal growth of the eye in animal models. She has generated zebrafish with the same genetic mutations as patients, as the zebrafish share 70% of their genes with humans and have a similar eye structure with an elaborate colour vision system more akin to humans than rodents and mice. Zebrafish allow for the assessment of how MAC conditions arise in very early eye development and can easily undergo treatment to see if it can be modified or prevented.

Curries for colobomas

An example of her pioneering research began by the discovery of a gene called FADD, known to be involved with cell death signalling, as a



cause of ocular coloboma in patients. By looking at the role of FADD in eye development, she ascertained that there were higher levels of cells dying at the site of the ocular coloboma. She then tested a drug called curcumin (active ingredient in the Indian curry spice turmeric), giving doses to zebrafish with ocular coloboma and microphthalmia and found that the severity of the coloboma was much reduced compared to the untreated fish. Dr Moosajee has won over 25 international and national prizes over her career, but she was awarded the prestigious Fould's Trophy from the Royal College of Ophthalmologists UK when she presented this research entitled "Curries for Coloboma" to the scientific and clinical community at their annual national conference.

Antibiotics that can fuse the eyeball

She went on to test the application of a class of antibiotics called aminoglycosides (gentamicin and paromomycin). These drugs not only fight infections but also have the potential to override a particular type of genetic mutation called a nonsense mutation, which introduces an abnormal stop signal in our genes and prevents normal protein being formed. These mutations can cause up to 70% of genetic disease, and was the cause in many MAC patients. The drugs were given to the coloboma zebrafish, and the eye completely fused as a significant amount of normal protein was produced. The drawback of this ground-breaking treatment was that the long-term use of these antibiotics was toxic to humans, causing hearing loss and kidney problems.

Overriding genetic mutations with new safer drugs

However, over the last few years a new class of small molecule drugs have been identified and these are now starting to show very promising results for the treatment of genetic eye disease. One particular drug called Translarna (also known as ataluren or PTC124), which has passed through phase 1-3 clinical trials and been given approval by NICE in the UK to treat children with Duchenne muscular dystrophy. It has a safe profile with minimal mild side effects. This drug is currently in phase 2 clinical trials for another eye development disorder called aniridia, where the iris (coloured part of the eye) and fovea (area of the retina where visual acuity is highest) fails to develop and the patient, who has poor vision, goes on to acquire further problems with scarring of the cornea and raised eye pressure. This trial is underway in the US and Canada and is based on laboratory studies where they showed the eye defect could be reversed in aniridia mice treated after they were born. The results from this trial will be very valuable for future applications to MAC. Dr Mariya Moosajee's team are undertaking the laboratory studies to provide evidence for clinical translation.

Four main research areas:

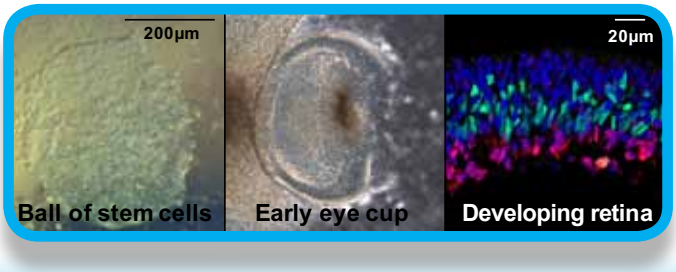
1. To determine the genetic causes of MAC, as <10% of patients receive a genetic diagnosis, and document the natural history of MAC-related conditions to see if specialist care is needed to minimise associated health problems. Families can enroll into the Department of Health sponsored 100,000 Genomes Project which offers the most state-of-the-art next generation sequencing technology called whole genome sequencing for patients. This test screens all 3 billion letters of your genetic code including all 20,000 genes, with the goal of identifying the genetic cause of 50,000 patients with rare diseases such as MAC.

2. To identify chemical changes/marks (called methylation) on our DNA that influence genes being switched on or off at crucial points during eye development to highlight any potential causative environmental factors.

3. Growing human 3D models of the developing eye from stem cells derived from patient's skin to investigate changes in genes that cause MAC and allow us to develop and test new therapies.

4. Developing therapies for MAC using disease models and translating to patients as fast as possible.

Continued overleaf...



The benefits for MAC families will be accurate diagnosis and genetic counselling, and improved care pathways for patients and families. Once we understand the genetic causes, a real focus on treatments can be made.

Dr Mariya Moosajee's research team, (Dr Rose Richardson, Dr Nick Owen, Dr Andreas Mitsios, Dr Adam Ali, Ms Hajrah Sarkar, Ms Dhani Tracey-White and Mr Khilan Khambhaita) are focused on making a difference for MAC patients.

How to get involved with research

As part of their research, they are setting up a patient focus group so that they can receive your feedback about the work they are doing. They want to know what you think is a research priority and help guide their direction. Please do contact them if you wish to find out more information or join the focus group. If you have any questions about the work outlined above, please do not hesitate to contact Dr Mariya Moosajee.

To be seen in our MAC clinic for clinical advice, joining the research study or to participate in the 100,000 Genomes Project, please send Dr Mariya Moosajee an email (Mariya.Moosajee@moorfields.nhs.uk) so she can advise you on how to get seen in her Genetics clinic at Moorfields. She sees both adults and children.

Contact us

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http://www.moorfields-private.co.uk/consultants/mariya-moosajee

For more information on the 100,000 Genome Project, please contact Samantha Lawrence, Research Coordinator

Email: samantha.lawrence@moorfields.nhs.uk

Tel: 0207 566 2260

Barry Stickings receives an MBE

Former Chairman, Barry Stickings, has received an MBE for his services to children with visual impairments.

Robbie Crow, MACS Chairman, had the following to say "We are thrilled that Barry has been recognised for his contributions to MACS as Chair, a position which he held from 2005 until 2015. During his time as Chair Barry made phenomenal contributions to MACS, including starting up a grants service and co-ordinating three charity balls. Barry was part of the team that made MACS into the welcoming, family orientated charity it is today and this is testament to the love and effort he put into the organisation. In my opinion Barry is very deserving of his MBE and the charity, along with all our member families, congratulate him for this fantastic honour! Barry's mission was always to 'help even one family who felt like he and his wife Kelly did when Toby, their son, was born'; I think we can safely say, 'mission: accomplished!'. Well done!"

Video gaming documentary starring MACS adult Toby Ott goes viral!

MACS member Toby Ott has recently appeared in a documentary entitled Gaming Through New Eyes. The 14-minute film shows how Toby can play and enjoy computer games despite having bilateral anophthalmia. A recent review said *"Absolutely incredible documentary, keep up the great work. He's inspiring to everyone and it's so heart-warming to see him enjoying his favourite hobbies."*

Toby says:

For those in MACS that do not know me I am Toby and I was born with bilateral anophthalmia.

I have been a fan of video games since I was 5 years of age. I started off by playing a game in an Arcade on holiday. I spent all my pocket money in doing so playing against Scorpion. I have also fought countless foes on Playstation 1, 2, 3 and now 4. Over the years I have always wondered why these games never have any audio description track to help blind players like myself. TV, films and the theatre have audio description so why not games?

An assumption no doubt that the blind cannot play video games!

In my quest to learn more about games I searched through YouTube and I discovered a gamer who commentates the story as he plays. The game he was commentating was from my favourite series of Final Fantasy, which I have been playing for years. Yes, I can play this game! His descriptions opened up a new world for me in understanding this game in more depth. In fact, many parts I was not even aware of. So I emailed him and a friendship began. His name is Berk Isel and he is a film maker. He contacted me and said he is a director and makes documentaries and adverts. Berk said he would like to make a documentary about me with a view to presenting it to the video games industry. His vision is the same as mine: for them to consider adding an audio descriptive element to games.

So Berk and I have made a documentary which he has presented at several Film Festivals.

The film is called Gaming Through New Eyes and has now won several awards. As a result I was contacted by the creator of a game called Doom, another favourite of mine. He has said that he is passing the film onto the many contacts he has in the gaming world. He agreed that the gaming industry has been lacking in the area of audio description on all levels for younger and older players alike.

I know video games do not always have the best press. However, many games have been proved to help those with various disabilities, such as autism. This is a very positive move forward.

I am extremely proud of my input into this documentary, which has so far has been viewed over 90,000 times on You Tube and over 3 million times overall. Yes, 3 million on other sites around the world. I hope it will continue to influence change. In fact, it has already prompted one company to start installing audio description onto a new game. I hope in the future all video games will have an audio descriptive track to help the visually impaired enjoy games just like their sighted peers. For me, I still beat Dad's friends and have high ranking scores on lots of different games.

For now I am continuing to help game makers on a voluntary basis to test new games and I hope by doing so will encourage more visually impaired to take up gaming and one day see all games audio described.

To view the video search Google for "Gaming through new eyes" or visit Berk's YouTube channel dansg08.

Family Weekend 2018 - MACS is coming to Windsor

Bookings are now open for the 2018 MACS Family Weekend being held at the De Vere Beaumont Estate, Windsor. With the MACS Family Weekend coming to the South of England for the first time in over 17 years, we can't wait to bring our unique event to more smiles than ever! Situated near to Lego Land and Thorpe Park, the De Vere Beaumont Estate will offer a fantastic experience for all of our families.

The event will take place from Friday 4th May to Monday 7th May at De Vere Beaumont Estate, Burfield Road, Old Windsor, SL4 2JJ.

Book now to secure your place! Discounted early bird bookings, are available until 1st December 2017. The early bird booking rate is £40 per person per night (for the first 2 adults and any adult siblings of the MACS young person) this is for lunch, dinner, bed & breakfast. After the 1st December the rate will increase to £50 as per above.
http://macs.org.uk/macs-2018-family-weekend-booking-form/

Children and Young People Services Strategy at Moorfields Eye Hospital - survey for parents and carers

The team at Moorfields Eye Hospital are developing a strategy for children and young people's services across all of their sites. However, before they can do that they would like to seek views and experiences of both the young patients and parents. The team recognise that parents and carers are as important part of this strategy as the young patients themselves, and would therefore like to hear your views to better understand what is most important to you as a parent or carer of a young patient at Moorfields.

This is your chance to have your say and help Moorfields improve their young patient services. Do use this opportunity and share your views by completing the survey here:

https://www.surveymonkey.co.uk/r/CYPparentsurvey2

Please volunteer for us!

Whether you are looking for a regular volunteering role or something to do as a one-off, whether you want to get out and meet people or help from home, whether you have a specialist skill to offer or want to add to your skills-base, if you want to help we would love you to join our team of wonderful volunteers.

At the moment we have volunteers doing administration, data analysis, event planning, IT configuration, community fundraising and much more. There is no end to the ways you can help us support our 1500 members around the country. So if you are passionate about making a difference to the lives of children born without eyes or underdeveloped eyes and their families then please email **volunteering@macs.org.uk** and join our team.

Fundraise in your community

Are you interested in raising funds and awareness for MACS in your local community? If so, why not join our Community Fundraising Team?

There are so many ways to raise funds and awareness in your local area. In the past we've had supporters bag packing, pub quizzing, cake baking, bucket collecting, concert singing, hamper raffling... There really is no end to the ways you can help raise funds and help children born without eyes or with underdeveloped eyes across the UK.

Our team have lots of ideas already but we'd love to hear yours too. So whether you'd like to plan an event, help out at a bucket collection or do something completely different please get in touch to join our team and we will help you every step of the way.

Help fundraise for MACS with no cost to you!

Did you know that whenever you buy anything online - from your weekly shop to your annual holiday - you could be collecting free donations for MACS? Or that you can donate to MACS every time you do a websearch?

MACS have recently partnered with two companies who will donate money to MACS every time you use their sites to shop online- **Easyfundraising** and **Savoo.**

Every time you use **Savoo,** either by searching online, using their voucher codes or shopping online, they will donate money to us without it costing you a penny.

Easyfundraising have over 3,000 shops and sites on board ready to make a donation, including Amazon, John Lewis, Aviva, thetrainline and Sainsbury's – if you shop through easyfundraising, these companies will donate to MACS and it won't cost you a thing!

You can find out more about both of these companies on our website: **www.macs.org.uk/want-to-help/give-while-you-spend/**

Small charity… BIG impact! MACS wins national award!

We are absolutely thrilled to announce that MACS were crowned winners in the first ever national Small Charity, BIG impact awards, as part of Small Charity week, fighting off competition from over 300 other charities. Robbie Crow and Fay Skevington accepted the award at a ceremony in London on 25th July.

The FSI Small Charity Big Impact Awards are a way to highlight the work of small charities (those with a turnover under £1.5 million) and the impact they make on a local, national and international level.

Our members know the difference that MACS has made to their lives, so it's fantastic to the impact MACS has being recognised nationally. As well as training, advice and a new video about MACS (see opposite), our prize includes a book containing information about MACS and other award winners being delivered to Number 10 Downing Street.

The judges, a range of funders and well respected charity leaders, said:

"MACS won their category as they clearly showed how they measure their impact, use it to improve their services and share it to build trust. We would like to congratulate MACS on their award and their fantastic work."

Robbie Crow, MACS' Chairman, said

"We're honoured that the transformational impact we have on children born without eyes or underdeveloped eyes has been recognised by this award. Above all, the award is testament to the time, energy and love poured into the charity by our family of volunteers, trustees and members."

We're really proud of MACS, and hope you are too - MACS would be nothing without its members and volunteers!

MACS kids star in new video

As part of our prize, MACS were given the opportunity to have a film professionally made about the charity. Five MACS families brought their little super-stars for a day of filming in London, and created an amazing film which highlights the work of MACS.

You can see the film on our homepage: www.macs.org.uk – please share it with all your friends to spread the word on Facebook!

eBay shoppers donate to MACS

Also as part of Small Charity Week, MACS won a competition to have our fundraising message featured at eBay checkout. We were really pleased that our 90 character appeal helped us to raise £994 from generous eBay shoppers.

How our grants are helping MACS kids

Harry is four years old and was born with microphthalmia in his left eye, cataract, PHPV and later developed glaucoma. His right eye is unaffected. We have been patching his right eye to try and develop the little vision he has in his left eye. It's been a real daily struggle and on some days we have only achieved 10 minutes!

We heard about the MACS grants and applied for an iPad for Harry to use whilst patching. It's made a massive difference; it holds Harry's attention and he is able to follow and play simple games on it when patched. We have since managed to achieve two hours of patching which we never thought would be possible and at his last check up his vision had improved so we are over the moon!!

Thank you MACS – you have made a huge difference to our everyday lives. The iPad has made patching become less of a stressful experience for Harry and we hope it will help his vision to continue to improve further.

Thank you again MACS!

Louise

This is a long overdue massive thank you for MACS' contribution to the purchase of the amazing *Prodigi Connect 12 magnifier* for our daughter **Jessica**. She is getting on really well with it and having much greater access to print and school work. We are very grateful for the assistance to buy it. She really likes using it too so that's a great bonus since she's quite stubborn about using equipment she doesn't like!

Here is a photo of Jessica planning her school residential trip a few weeks ago. I hope it gives a good example of how much the Prodigi has changed her access. Very best wishes

Flo (mum)

New book about MACS conditions – What makes you, you?

After the initial shock, the tears, the hundreds of questions and then finally the processing, I was left with one question; how do I help with the journey? Our daughter Aurora was born with Microphthalmia affecting her right eye. As a family we received so much help from MACS, especially in the first few very raw months. I immediately realised I wanted of course to help my daughter in any way that I could, and secondly I wanted to give something back to MACS in return for their support. Being a teacher I found myself very early on thinking about what school might be like for Aurora, and what it could be like socially, as children ask those inevitable questions, stare and wonder why Aurora was different. I scoured the internet for books on the subject and found that there were very few, so set about writing and illustrating one myself, initially just for Aurora. With the help of both Jenny and Teresa I was put in touch with two wonderful families, The Crofts and The Kavanaghs, whose children went on to become the two other main characters within the book and ensure each of our MACS conditions were represented equally.

Writing the book was easy as I knew the message I wanted to promote, but I had to teach myself very quickly using an online course how to use a professional illustrating programme. I was able to scan my initial sketches and then transform them into cartoons of their real-life counterparts. The book took in total around 6 months to create, and wasn't without its frustrations, but I am extremely happy with the end result. The message itself promotes the fact that our three gorgeous MACS children, George Croft, Charlie Kavanagh and Aurora, do have something that makes them different but that this difference should be celebrated like the differences between all children and all people should be.

MACS kindly agreed to make the book available to parents and children and it has proven to be very popular. Already around 30 preschools have this book and are using it to help introduce MACS conditions to children in their EYFS classes! All credit has to go to the wonderful children that feature within the book, and indeed to all our MACS children! Seeing all the MACS posts and photographs make me so happy, and seeing what our children can aspire to and do is incredible and inspiring when we look towards Aurora's future! I hope the book is some small help to anyone who needs it. Thank you to MACS for taking it on and for everyone who has already helped get the message out there to schools. Watch this space as I am about to start the next one; aimed at a more physical, social look at what our MACS older children/teens experience!

For a free copy of "What Makes You, You?" for MACS members, please contact jenny@macs.org.uk


MACS
What Makes You, You?

Microphthalmia, Anophthalmia & Coloboma Support
Registered address: MACS, Suite 472, Kemp House, 152 City Road, London, EC1V 2NX
Registered charity number: 1161897

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