HEADLINE NEWS

ISSUE 6

Working towards a world where the physical, psychological and social impacts of craniosynostosis and rare craniofacial conditions are no more

Headlines
Craniofacial Support

In this issue

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Disclaimer

From the Chair

This is my first issue of Headline News as chairman, so it would be remiss of me not to acknowledge the wonderful work and achievements of Wendy Edwards, the outgoing chair.

Wendy has really helped put our charity on the map. The Research Committee that she is now leading is making great headway under her expert leadership - she has certainly left very large boots to fill! We are still very fortunate to have her husband, Charles Edwards, expertly looking after our finances in his role as treasurer.

It's been a challenging couple of years hasn't it? Nobody knew or understood the vast impact that Covid 19 would have on our lives and the way we work. This represented an especially testing time for all involved in the charity sector, where face-to-face fundraising became an almost impossible task – but as they say, 'necessity is the mother of invention' and the pandemic made us all sit down and think about how we could adapt to the situation we found ourselves in.

And that's exactly what we all did! We entered the new year escaping the confines of the various restrictions in place with a reinvigorated optimism about the future, armed with new and creative ways to raise money and with Headlines being in as strong a position as it has ever has been.

2021 allowed us to focus our efforts on revamping our website too. It's got a fresh new look. If you haven't seen it yet I encourage you to check it out at www.headlines.org.uk. Many thanks to our trustee Darren Sloane for all his efforts getting this over the line.

The Family Weekend back in September proved to be a fantastic event. This was the first time that my family and I had attended the event. It gave me a chance to talk with other parents about their experiences and it was marvellous to witness first-hand the positive effect that Headlines Craniofacial Support has on people's lives. It was also amazing to see the children having fun and enjoying the various activities available. If you haven't managed to make it to the Family Weekend in the past – you are definitely missing out!

As I come to the end of my first year as chairman, I'd like to say a really big thankyou to everyone involved with Headlines, from the trustees to the countless fundraisers and volunteers out there - your work and continued dedication helps us help those who need it most, and I am extremely proud and humbled to be involved with such a wonderful group of people.

I look forward to meeting as many of you as possible at the next Family Weekend and would like to wish you and your families well for the rest of the year ahead.

Paul Cornell

Director's Cut

Another year into the pandemic and our work has continued apace: this time twelve months ago, we were still pulling together plans for a new Research Fund and its launch in the early summer of 2021 was one of the highlights of our year.

Although it is only a drop in what is a very deep ocean of funding that we need to be able to answer some of the major questions about craniosynostosis and its treatment, it marked an important start – and, importantly, demonstrates the commitment we have made as a charity towards achieving our long terms goals. We hope it will provide our members and potential funders with the opportunity to make a genuine and lasting difference to the future of all those we are here to support.

You can read more about the Fund and its plans on p12. However, I could not let mention of it pass without also paying tribute to the chair of our Research Committee, Dr Wendy Edwards, who has done so much to take Headlines forward over recent years in her former capacity as chair of trustees. We shall miss her input on the board, but we are delighted that we will continue to benefit from her expertise in the new role.

In addition to the progress we have made with the Research Fund, 2021 also saw the launch of a major new piece of work looking at the needs of adults with craniosynostosis. As Bruna Costa explains on p10, the ACCORD project is a direct response to the findings of the research that she and her colleagues at the Centre for Appearance Research completed for us in 2019. Almost three decades after our first families came together to set up Headlines, we hope that this exciting new project will help us develop better support for those who've since reached adulthood and enable us to remain relevant, whatever the life stage of our members.

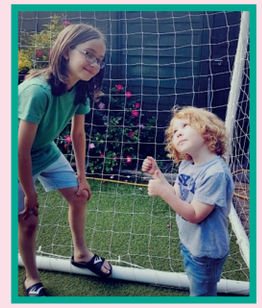
Finally, as you'll see on p14, we were delighted to be able to run our first Family Weekend for over two years in September, when we were joined by friends old and new for a weekend of activities at the Pioneer Centre in Shropshire. It was fantastic to see so many of you in person after the challenges of the pandemic, and we hope to be able to welcome you back for another blast of fun later this year!

Helen Keller famously remarked that on our own, we can do little, but together we can achieve so much, and that has certainly been true of the past twelve months. As staff, Donna and I continue to be humbled by the generosity and kindness of those we have the privilege to support on a day to day basis – our thanks to everyone who make our work such a pleasure.

Karen Wilkenson-Bell

Buddies for life

Last year we were touched to receive an email about George and Christopher, two young members with sagittal craniosynostosis who attended the same primary school in Reading and had been 'buddied' by the school.



Both boys had had surgery at the John Radcliffe Hospital, Oxford, a year apart, albeit at different ages.

Unfortunately, although the pandemic meant that the usual buddy activities in school such as assemblies, reading in the classroom and playing together at break weren't possible, they had still managed to forge a lasting friendship and support each other using Zoom and video messaging to have chats and share books.

Christopher's parents, Gemma and Marcus, said at the time: "it is lovely getting the chance to watch them bond digitally as normally this would be just in school and we would miss out. It took us so long to get Christopher diagnosed and the decision to have the surgery was the hardest thing we've ever done, so seeing him with Geo is just amazing – they have a totally different bond."

Geo's parents, Joanna and Paul added: "The boys instantly developed a great bond. When Geo had surgery as a small baby, it was a scary time for the family, but we knew he was in good hands with the excellent team in Oxford. To see Geo flourishing at school makes us so proud of him."

Since that first email, we've had regular updates from the families, and the boys have been able to meet in person as restrictions have lifted.

Last year , Geo invited Christopher to help him celebrate his 5th birthday in his garden where they played football and games and enjoyed cake together. They also had a play date and picnic in Christopher's garden where they enjoyed playing football and tennis and learning about growing fruit and vegetables.

Geo is now in Year 1 and doing fantastically at school. Christopher is 12 and has moved on to secondary school where he has a much longer school day with lots of clubs and homework. However, the boys have stayed in touch, sharing chats and gifts at Christmas and sending each other updates via video messaging. They are both looking forward to the warmer weather and the chance to meet up again soon.

As Christopher's mum Gemma explains: "We hope the boys have been able to provide some kind of inspiration or support for others who may be at a different stage in their journeys to our families".

Eva's Story

Talented teenage artist Eva Thomas tells us about her experience of growing up with craniosynostosis and how this has shaped her outlook.

"When I was 9, I underwent surgery to remove a metal wire from my head, but this wasn't my first time on an operating table. That same wire was put in place 8 years prior, following substantial reconstructive surgery to support my altered forehead plate.

Although the wire didn't need to be removed, it would constantly cause me dull head pain due to my cranial bones shifting as I grew older. This is just one way that craniosynostosis has affected my life, as following the surgery, I've still experienced some challenges surrounding my appearance and condition.

Although it might not seem like the most serious condition, the same surgery I underwent to treat it claimed the life of a young girl I was in hospital with at the time. I know I am very lucky to have had a successful surgery and yet, I can't help but compare myself to people who aren't affected by the condition.

I'm 16 years old now, and my facial features are slightly skewed and tilt to the right. My condition may have affected my self-esteem, but it has not affected my abilities nor my opportunities, as I'm about to sit my GCSEs at school. I have been offered further corrective surgery to fix the asymmetry of my face, and I'm planning to go ahead with it, as I think I'll be a lot happier.

I also have the option of therapy sessions to combat my insecurities surrounding my appearance.

My condition definitely impacted my childhood - from biannual trips to a specialist hospital (only to have a team of doctors' prod and grab my head!), to having a 'lazy eye'. Despite it not being a life-threatening condition, I've had to adapt around it.

'I can't help feel that if craniofacial differences were normalised, people wouldn't find them so shocking'

Having metal wires in my head has meant that I could never get involved with practical science lessons involving electricity in school, nor was I ever allowed to attempt anything too physically demanding as a young child, such as cartwheels or handstands, for my mother's fear of damaging my head.

Despite these minor setbacks, I've never actively faced discrimination due to my craniosynostosis, though throughout primary school, and for a while after, I was sometimes bullied for my appearance, even being told my face was "lop-sided".

As part of the active citizenship project in my school, we've been encouraged to campaign for a cause we're passionate about, and I know first-hand about the lack of representation of visible difference in the media. The reconstructive surgery required for craniofacial conditions is often tortuous, and yet such conditions receive little to no media attention. Even though the condition is far from mainstream, it still affects roughly 1 out of every 2200 live births. And yet the only time I ever see people like me is in the waiting room of a craniofacial hospital ward!

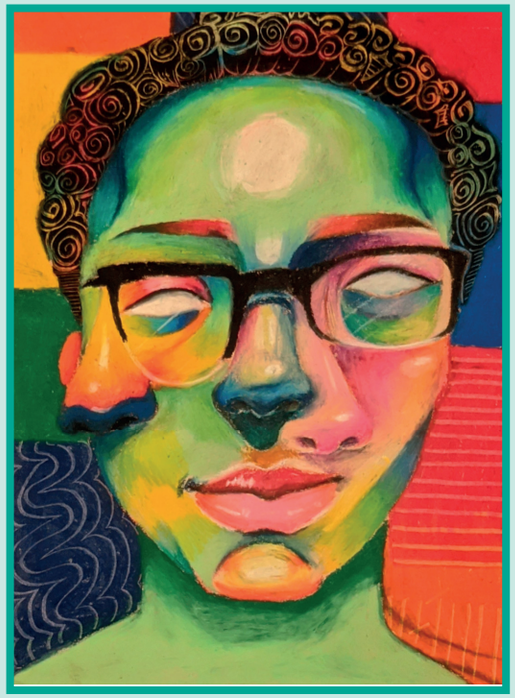
I can't help but feel that if craniofacial differences were normalised, people wouldn't find them so shocking, and thus people with visible differences wouldn't feel so alienated and ostracised.

My goal is to shed some light on this issue, as I feel inclusivity could be promoted more, whether that be through seeing more people with visible differences like mine on the TV and social media or reading about success stories in newspapers and magazines .

By sharing my story, I hope to bring greater awareness to visible difference and shed some light on the stigma associated with what is sometimes referred to as 'deformity'. I also hope that more young people with similar conditions will feel relieved that there are others out there with craniosynostosis, campaigning for their cause.

Although I'm not sure what I want to do in the future, I'm very passionate about art and I intend to take it to at least A-level, even if I can't make a living out of it. The irony is, I solely draw or paint symmetrical faces. I think all faces are beautiful, even if not in a conventionally attractive way, then at least in an artistic one.

As of late, I've been working on drawing more 'distorted' faces and trying to battle my need for symmetry. By breaking away from my need to draw 'perfect' faces, it helps me accept myself a bit more, and makes me believe that a person doesn't need to be well-proportioned to be seen as normal, let alone beautiful."



Profile: Greg James

Greg James is a familiar face to many of our families in his role as Consultant Neurosurgeon and Associate Professor at the Craniofacial Unit at Great Ormond Street Hospital (GOSH). Last year he and GOSH colleague Juling Ong also spoke at our first-ever online conference.

We invited Greg to tell us more about what makes him tick.

Can you tell us a little about yourself and why you went into neurosurgery?

I grew up in Bristol and was the first in my family to go to university. I wanted to be a doctor for as long as I can remember - I was (and remain) fascinated by how the body works, in particular how this 2kg lump of fat and water (i.e. the brain) generates our consciousness, emotions, language and behaviour.

I studied at Guy's and St Thomas' and also undertook a 3 year PhD in neuroscience during my medical degree (see the fascination referenced earlier). I had a feeling I might like neurosurgery, but you don't get exposed to it much in medical school. But I clearly remember my first day as a neurosurgery SHO (junior doctor) - it felt like I had 'come home' - I totally fitted into the specialty and never looked back. I am lucky to be married to a beautiful wife who is a psychologist and I have 2 teenage kids.

How long have you been at GOSH?

I've been a consultant at GOSH since 2014, before that I was a registrar and fellow there. A bit like my first day in neurosurgery, I fell in love with the place as soon as I walked through the door, and realised I wanted to treat children not adults - I am inspired by children's positivity, resilience and bravery. I also love the fact in GOSH we work as teams (like our incredible craniofacial team) and the quality and dedication of the nurses, doctors and allied health professionals I work with is so impressive.

What do you most/least enjoy about your work?

Like most surgeons, I most enjoy operating - especially seeing the results when children do well (which, thankfully, is the vast majority of the time). Again, like most surgeons, I'm not so keen on admin - I get hundreds of emails a day and it's difficult to keep up.

What do you enjoy doing outside of work? (if you ever get time off!)

I do work long hours - and when I do get some time, I like to spend it with my family. I do have some interests though: I collect vinyl records and am a (now occasional) DJ, I ride my road bike and I'm a keen supporter of Bristol Rovers football club. I also love videogames - which I now play with my kids, which is great fun!

What has been your proudest achievement?

Craniofacial-wise, definitely introducing the endoscopic surgery (ESCH) to the UK. I remember when I started meeting a family who had paid many thousands of pounds to fly out to the States to get this operation - I thought that was unfair and looked into the data which seemed to show ESCH was effective and safe - and popular with families.

I was awarded a grant from the Worshipful Company of Barber Surgeons to travel to Boston where I learnt the surgical technique at Harvard from Mark Proctor, and was able to introduce the service at GOSH. We've now done over 70 cases with encouraging results and happy children/families, and we've managed to get the helmet therapy fully-funded by the NHS. It has taken a LOT of hard work to get to this point and I'm so proud of our entire team, especially our amazing nurses Andrea White and Kathy Truscott.

What changes would you like to see in the treatment for those with craniosynostosis?

Whenever I go to a craniofacial conference the question people ask is 'what operation to do' for, say, sagittal synostosis. I would like to re-frame that as 'whether' to do an operation at all - I don't think we always understand whether or not our surgery will benefit our children, especially in important domains like intellectual development and speech. My hope for the future is we can offer rapid individualised assessments including genetics and 3D scans to show parents what the future is likely to hold - both appearance-wise and developmentally - so they can make an informed choice about 1) whether an operation is necessary and 2) which operation is best for their particular child.

Can you tell us something about yourself that might surprise readers?

Well, I don't DJ that often any more, so I make up for it by being the operating theatre playlist master. My 'Yacht Rock' playlist is legendary and the theatre staff get upset if I DON'T put it on!

ACCORD: our new project for adults

Bruna Costa, Research Fellow at the Centre for Appearance Research (CAR) at the University of the West of England, Bristol, writes about an exciting new project focused on adults with craniosynostosis.

One of the findings of the research we undertook for Headlines in 2019 was that while children with craniosynostosis and their parents were generally well-supported by the NHS and by Headlines, there was little – if any – dedicated support for them as they reached adulthood.

Worryingly, the study highlighted the fact that adults affected by craniosynostosis reported higher levels of anxiety and appearance concerns than the general population and many had concerns about important issues such as starting a family and gaining employment.

In an attempt to address some of these gaps, Headlines successfully applied for further support from the original funders of the research - the VTCT Foundation – to work with CAR on a major new piece of work looking specifically at the needs of adults.

The overall aim of the 'ACCORD' project (so-called because the term means 'to give someone power and recognition') is to improve the information and psychological support available to adults living with craniosynostosis by developing resources for adults. The work will also include evaluation of the resources to ensure acceptability among adults with craniosynostosis and NHS craniofacial teams.

Work on the new project began in autumn last year, when Headlines put out a call for adults with craniosynostosis who were interested in joining a group to actively contribute to the design and development of the resource, and provide detailed feedback on the finished content.

We were delighted by the level of interest amongst Headlines members, and some 15 people joined us for an online workshop in November to find out more. At that workshop, we also heard from Fine Rolling Media, an award-winning video production agency with whom we plan to work on producing video content.

Sadly, plans to host an in-person event in London in December were thwarted by omicron, but the 'core' group continued to meet online to work with us on the development of content as we moved into the New Year. By the time this magazine goes to press, we hope to have a framework for both written materials and potential video content, to enable us to work on the production of the videos across the summer.

It's been an exciting start – and the enthusiasm of Headlines members so far means that we are more convinced than ever of the importance of this work. We're looking forward to reporting on the final outcomes next year, so watch this space!

Zoe's Story

One of the members contributing to the ACCORD project is this issue's cover star, Zoe Hilton-Webb, whose family have been involved in Headlines since Zoe was tiny. Zoe will be familiar to families who've been at our last two Family Weekends as one of the team of childcare specialists running the creche.

Zoe was born with Apert syndrome, a genetic (syndromic) form of craniosynostosis, which is estimated to affect around 1 in 65,000 newborns.

As a result of her condition, Zoe has hearing and visual impairments and problems with fine motor skills (the co-ordination of the small muscles in her hands and fingers).

Throughout her life she has had to deal with people's stares, comments and negative attitudes, but her condition has never stopped her from doing what she enjoys.

She achieved good GCSEs and got into college, eventually going on to complete a part-time degree course with The Open University.

"As a child I found being at school incredibly challenging." Zoe says, " I was bullied a lot because I looked different from everyone else. I had to move schools at the beginning of Year 4 and then again in the middle of Year 7, because I was really struggling with the situation and the school were not very helpful. I received counselling to help me deal with the bullying and with my anxiety.

My anxiety also resulted in me finding it difficult to talk to people and build relationships. However, once I left school and went to college, things improved and I became more confident.

As a person with a visible difference, many people I have met have tried to persuade me to have further plastic surgery. However, I am happy with the way I look and have never had a personal problem with my appearance. I am pleased that my closest family members and friends have not made me feel that I need to change the way I look."

As an adult, I faced challenges in particular getting into employment. It took me six years in total to finally find permanent employment. I am really enjoying the fact that I am working with children, which is something I love. But it has not been easy.

I am not surprised that the Headlines study showed that adults who have craniofacial conditions may have real difficulties in finding employment. Employers sometimes cannot see past the person's appearance and therefore make assumptions that they will not be good enough to do the job."

Research Fund

2021 saw the launch of something quite special for Headlines – our first-ever Research Fund. Here our former chair of trustees and now chair of the new Research Committee, Dr Wendy Edwards, tells us how this important milestone was reached and what the future might hold.

Research was woven into Headlines' core objects when it was first registered as a charity back in 1996: in addition to providing support and raising awareness, our constitution states that Headlines exists 'to preserve and protect the health and promote the welfare of people with craniosynostosis and rare craniofacial conditions by facilitating research that seeks to advance understanding, ensure the provision of quality care, and identify the best treatments for craniosynostosis and rare craniofacial conditions'.

When I became a trustee in 2014, research was something we supported reactively, rather than in active way, largely because as a tiny charity with very limited resources, we did not consider ourselves 'big' or financially secure enough to fund research ourselves.

As an experienced research manager, this frustrated me a little: although the conditions that Headlines support are considered 'rare' and therefore unlikely to command the attention of vast swathes of the research community, how could we ever start to address some of the basic questions about the conditions if we didn't actively encourage more research?

An opportunity to change the status quo emerged in 2016, when Headlines was invited by the National Institute for Health Research Cleft & Craniofacial Clinical Studies Group (NIHR CSG) to take part in an exercise to produce a list of patient-focused priority research questions. This was led by Headlines member Ingrid Lawrence with support from myself and several other members.

Through a process of consultation (which some readers will no doubt remember!), we eventually came up with a list of 'Top Ten' research questions that were then validated as unanswered by a range of expert health professionals and researchers in the field of craniofacial research. This list became our guide point, and two of the questions subsequently formed the basis for a small-scale study undertaken on our behalf by a team from the Centre for Appearance Research (CAR) at UWE Bristol, which examined the long-term psychological impacts of craniosynostosis in 2019.

That study - which not only unearthed unmet need, but also led to the production of new support materials - illustrated the value of directly steering research and helped persuade our board that we should look to create mechanisms for funding our own.

Thankfully, by that time we had also arrived at a point where the charity had built up sufficient reserves to ensure my fellow trustees were able to agree to earmark funding to do so – and finally, as I came to the end of my tenure as chair, we were able to realise my long-held dream of creating a Headlines Research Fund.

I am delighted that we have been able to persuade several eminent independent experts to join our new Research Committee – their insight will supplement the expertise brought by trustees Dr Mehran Moazen and Dr Caroline Hilton and our 'expert by experience', Charlotte Ashby, as we move forward with our plans. I am also pleased that in the months since announcing the Fund, we have launched a 5-year strategy and made our very first award, details of which you can read about below.

Of course, there remains much to be done – not least in encouraging more top quality researchers to consider applying – but I am confident that the excellent start we've made on this important journey will continue!"

Sarah Kilcoyne, Principal Specialist Speech and Language Therapist at Oxford Craniofacial Unit was the recipient of our first-ever grant, for a study entitled 'Language and communication in children with non-syndromic sagittal synostosis: A sibling control study'.

How do we know if sagittal synostosis impacts on language development?

It's an important question and one this new study should help answer. Sarah and her team at Oxford will be inviting parents to join if they have a child with sagittal synostosis, and another child of the same sex without sagittal synostosis.

There are many outside influences that can impact on language development in children, but by studying children from the same family, the only difference between them will be the presence of sagittal synostosis. The team at Oxford hope this will help them provide parents with better information about what to expect for their child's language development if they have a child with sagittal synostosis.

We wish Sarah every success with her study, which will run for 18 months, and we look forward to reporting on the outcome in future issues of Headline News.

Friendship and fun: our Family Weekend returns!

After a hiatus of more than two years, we were delighted to welcome members back to the Pioneer Centre in Shropshire for our Family Weekend over the weekend of 17/19 September 2021.

Concerns about the weather proved unfounded, as the sun arrived and surprised us all by staying for the whole weekend. Spurred on by the sunshine, our families enjoyed a whole range of outdoor activities, from high ropes and climbing to raft-building and archery.

Friday evening frolics were led by our ever-entertaining volunteer MC and quizmaster John Connett, and we were treated to an impromptu singalong around the campfire on Saturday night, thanks to long-standing members Michael and Margaret Hebden, whose hilarious rendition of 'Ging Gang Goolie' transported many of us back to the damp canvas of our youth.

On Saturday we were also joined by some very special visitors in the form of Ewoks and Imperial Starfighter Pilots from Star Wars. We are not entirely sure who enjoyed this bit the most, but as the photos suggest, it seems it wasn't just our younger members!

As ever, we had a wonderful team of play specialists for the creche – ably supported this year by Zoe Hilton-Webb, whose story we feature on p11.

In a new departure, we were also able to offer sessions specifically for members of our Young Person's Network this year, a number of whom were with us for the Weekend.

All in all, we had a wonderful time – and as well as thanking those who joined us, we'd like to extend an extra special thanks to the Coleman Family, whose amazing fundraising efforts enabled us to offer free children's places to everyone for the very first time.

Plans are underway for our next Family Weekend in September, so if you would like further information, please contact Donna via info@headlines.org.uk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EDELRID

Futureheads: vitual surgery to advance the treatment of craniosynostosis

Back in October 2021, a group of members from our Young Person's Network took part in a special workshop organised by one of our trustees, Dr Mehran Moazen, Associate Professor in Biomedical Engineering at UCL in London, which looked at skulls and the causes of craniosynostosis.

Here, Connor Cross, a PhD student from Dr Moazen's team who helped deliver the workshop, tells us more about their ground-breaking work.

"The skull's open midline joint that extends across the top is responsible for its upwards and sideways growth. If this joint (known as the sagittal suture) fuses too early, the skull becomes long and narrow. In rare cases, problems with brain development can arise, as the level of pressure inside the skull may build up. This early fusion is known as sagittal craniosynostosis and is the most commonly-occurring form of craniosynostosis.

Different Specialist Craniofacial Units have developed different approaches to reconstruct the skull. Each approach helps to guide the skull to grow in height, shorten the length, expand across the width, and release any pressure across the brain.

Of course, surgeons can't practice these approaches on their patients before they operate. Recently, however, computer models of 3D skulls have been shown to be a promising tool for enabling Specialist Units and biomedical engineers to perform "virtual surgery", mimicking the surgery across a 3D skull, before treating patients.

Our team here at UCL's Department of Mechanical Engineering, led by Dr Moazen, has pioneered a novel computational platform that enables craniofacial surgeons to optimise treament of sagittal craniosynostosis by helping estimate the shape of the skull and the pressure inside the skull as the child grows up after surgery. The models we have created are very powerful tools and can be used to compare different "virtual surgeries" and their possible outcomes.

Funded by the Rosetree Trust, I have been working in collaboration with several Craniofacial Units worldwide, including Oxford, Paris, Gothenburg and Olsztyn (Poland) to focus on 9 different surgical approaches for the treatment of sagittal craniosynostosis. The models can estimate what the skull shapes and internal skull pressure levels may look like across each of these approaches six years after the surgery was performed.

Using CT images of a 4-month-old patient skull before surgery, we created a virtual model which was then checked against follow-up CT images of the same patient, to confirm the accuracy of the approach.

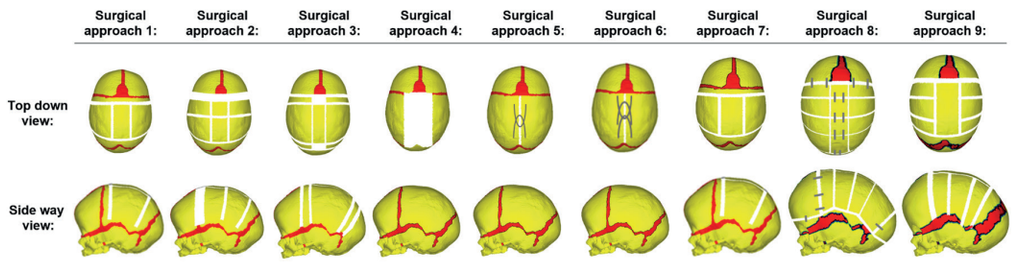
The nine 'virtual' techniques included the use of springs to help widen the skull (known as spring-assisted cranioplasty), and correcting the skull shape during surgery with the aid of special fixtures, which disappear naturally after surgery (known as total calvarial remodelling).

The team were able to accurately predict skull growth up to the age of six years, and the rate of skull healing, across the different surgical scenarios. The model was also able to analyse the pressure build-up inside the skull, allowing the team to estimate if one approach may result in higher pressure than another.

Throughout the study, the team have collaborated with distinguished surgeons from around the globe, from Sweden to France and the UK. These surgeons have shown significant interest in the study, seeing it as a valuable tool to help them select the best treatment for sagittal craniosynostosis based on the model's predictions.

Although the results are very promising, more work needs to be carried out to test the validity of these models. Just as importantly, while the focus of this study only considers patients with sagittal craniosynostosis, there will be opportunities for the team to investigate other forms of craniosynostosis in the future, using these kinds of studies as their baseline.

It's exciting work and we look forward to sharing more of our results as they unfold."



Fundraising Fun

Little Maxwell (Maxy) Coleman was born with Apert syndrome. Auntie Katie Tang decided to organise a 4x4x48 Challenge to raise funds for Headlines, raising over £6,500 to help fund free child places at last year's Family Weekend. This year dad David Coleman is picking up the baton and has already raised more £3,000 – go David!!

Fundraiser Michael Jordan ran the London Marathon for Headlines, raising almost £1,300 for us – well done Michael!!

Lucy Holland raised over £500 for Headlines by swimming half a marathon last September.

Lucy was diagnosed with craniosynostosis in 2000 and underwent surgery at Alder Hey Children's Hospital.

Lucy graduated with a degree in Psychology from the University of Manchester, and now works in the field of children and families research.

"I am forever grateful to Headlines as well as the staff at Alder Hey for what they have done for me" she says. And thank YOU Lucy – we are grateful for your amazing fundraising efforts!

After several postponements, our team of fearless fundraisers finally made it to the top of the UK's tallest sculpture in September as they completed the long-awaited Headlines Challenge Abseil in London's Olympic Park to mark CranioAwarenessMonth 2021.

Leading the team was our former chair, Wendy Edwards, who made the 80 metre descent with son Ben, who was born with sagittal craniosynostosis. Joining them were intrepid members Holly Crookes, Mitch Camp, Laura Melles, Louise Hebden and Rebecca Meddick. Between them, the team raised over £4,000 to – a massive thanks and well done to all our dare devils!

11 year old Grace Turner-Cox's baby cousin Henry was diagnosed with craniosynostosis in 2020 — so Grace decided to run the equivalent of a marathon a week for Headlines, raising over £2,500 to date.

Grace was the very first person to receive one of our all-new running shirts, available to anyone pledging to raise more than £300 for us. Here she is trying out her new shirt - we think you're definitely Amazing Grace!!

Every year, pupils at Coteford Junior School in Pinner run a café to raise money and learn about the different skills involved in planning and running a cafe.

This year, one of the children asked if they could nominate Headlines as their chosen charity as their cousin had craniosynostosis. "Everyone thought it was an amazing cause," says teacher Katerina Taylor, "and we wanted to raise awareness. We had to do lots of preparation - making menus, choosing and tasting smoothies, deciding on cakes and entertainment . On the morning of our cafe, we had the two other Year 5 classes come to visit for an hour. In the afternoon, we ran a delivery service for the school and the children were designated to a specific class and staff members so everyone was covered."

In the event, the school raised almost £400 for Headlines. Thank you Year 5 – we think you are all fantastic!!

In her role as Captain of the Ladies Section of Chipping Sodbury Golf Club, Pauline Fletcher helped raise over £500 for Headlines.

"We were unable to do many of the usual events due to the Covid restrictions" she explains, "but fortunately we had one fundraising day when we played 9 holes and all had afternoon tea."

Pauline is pictured presenting the donation to our Vice Chair, Barry Fletcher – who just happens to be Pauline's son! Our thanks to Pauline and all the members of Chipping Sodbury Golf Club – we are very grateful indeed for your support.

WILL YOU FUNDRAISE FOR HEADLINES?

As a very small charity, we rely entirely on donations to fund our work, so every penny raised makes a real difference. Whether you're having a party, holding a bake sale or running a marathon, there hundreds of ways you can support us. And don't forget to tell us about your fundraising efforts – we'd love to hear from you!

Leo's Journey

Back in October 2020, at our first-ever online Conference, we heard from surgeons Greg James and Juling Ong about the use of helmet-assisted endoscopy at Great Ormond Street Hospital.

At the conference were Claire and Mark, who had contacted our helpline a few weeks earlier about their baby son, Leo. Here we learn about the family's journey and what has happened since.

Claire and her husband Mark were worried. Their beautiful baby boy was 6 weeks old and they could not escape a nagging feeling that something about his headshape didn't quite seem right.

When she'd mentioned her concerns to the hospital paediatrician when Leo was born, Claire was assured there was nothing to worry about and that baby Leo's head was fine. Over the following weeks, she was reassured by various midwives and her GP. "Like most new parents,* Claire says, "I accepted what the doctors and health professionals told me and went along with it. But that intuitive feeling didn't go away, so late one night, I started googling. Straight away,I saw pictures of other babies with exactly the same triangular head shape as Leo's. It was just so obvious. Like most people, I'd never heard of craniosynostosis, but in that instant, I knew that was what he had".

Claire and Mark set about arming themselves with as much information as they could about the impacts of the condition and treatment options.

Claire re-visited her GP, who initially referred Leo to Birmingham Children's Hospital, where the diagnosis was confirmed. Leo had metopic synostosis: the suture which runs from his front fontanelle (soft spot) through the forehead to the top of the nose was fused, causing the irregular triangular shape that had so worried them.

"We were advised that Leo's condition would require him to undergo a procedure called cranial vault remodelling", Claire continues, "and that this would normally be carried out when he was about 18 months old. However, this seemed a very long time to wait and even though the specialist team we'd been referred to assured us he would be fine, we were concerned about the possibility of him developing pressure inside the skull and about the impact of the condition on his development.

In the course of our research, we'd read about less invasive surgical techniques that needed to be undertaken before 6 months of age, while the baby's skull bones are still very soft, and wanted to find out more".

Claire contacted Headlines. "They explained that endoscopic surgery was at that time only offered at Great Ormond Street," she says, "but by sheer coincidence, Greg James and Juling Ong, two of the specialists who carried out the treatment at GOSH, happened to be speaking at the Headlines conference the following month". In the meantime, Claire requested another referral by her GP to GOSH as she and Mark were convinced that endoscopic surgery was going to be the best option for Leo. A few weeks later, Claire and Mark logged into the conference and listened to the surgeons as they gave their talk. "Greg and Juling set out all the pros and cons, carefully explaining that this treatment wasn't suitable for everyone, and that the helmets that were required to accompany the surgery were not available on the NHS, so had to be paid for privately. It was so helpful".

After discussing the options through after the conference, Claire and Mark felt that they had all of the information they needed to make an informed choice and asked GOSH to put them on the waiting list for endoscopic surgery. Leo had his operation when he was 5 months old and was only the ninth child with metopic craniosynostosis to have the surgery.

Leo was fitted for his helmet 2 weeks later. The treatment was initially very difficult, as the helmet is deliberately more 'aggressive'", applying pressure to the forehead and around the sides of the head in order to force growth in the right area.

"Wearing the helmet aggravated Leo's surgery wound", explains Claire, "and resulted in a horrendous sweat rash."

As growth over the first 6 months is so rapid, the family also had to visit the helmet clinic in London at least every 2 weeks to have the helmet adjusted as Leo's head grew.

Leo has now been wearing his second helmet for almost a year. This has slightly more ventilation and although Leo does not like wearing it, it is hoped he will be finished with treatment when he reaches his second birthday in July 2022.

Leo still has to visit the helmet clinic every few weeks for adjustments, but the growth is slowing down.

Looking back, the first two years of Leo's life have been more difficult than the family could ever have imagined: Claire describes receiving the diagnosis and having to put Leo through the procedures as 'heart breaking'. However, "although Leo's head may never look completely 'normal'" she says, "the transformation has been phenomenal and we are convinced that having the endoscopic surgery was the best thing for him and his development.".

The family are waiting to see what the future brings, but as Claire reports with a smile, Leo is currently enjoying being a toddler "and causing all sorts of mischief!".

Since Leo began his treatment, Alder Hey Children's Hospital has also started offering endoscopic surgery. The NHS has now agreed to fund helmet therapy for those who require the treatment.

Unsung heroes: genomic testing for craniosynostosis

In the NHS, genomic testing for rare conditions is offered through a network of Genomic Laboratory Hubs (GLHs): Clinical Scientists Helen Lord and Dr Tracey Lester from Oxford tell us more about this crucial but often overlooked service.

Between them, Genomic Laboratory Hubs provide testing for all of the disorders described in the National Genomic Test Directory, including tests for craniosynostosis.

As craniosynostosis is very rare, genomic testing is offered by two GLHs aligned to centres with national and internationally-recognised expertise in research into these conditions, and co-located close to two of the four national craniofacial specialist clinics in England.

The genomic service for craniosynostosis is also recognised as a Highly Specialised Service (HSS), meaning that all cases across England and the devolved nations are funded centrally, allowing equitable access to patients in the diagnostic service regardless of their location within the UK.

The two specialist testing laboratories are located within the Central and South GLH (in Oxford) and in the North Thames GLH (in Great Ormond Street Hospital). This article focuses on the testing within the Oxford Genetics Laboratories.

Although craniofacial testing at the North Thames Genetics Laboratories began in 1999, this service was introduced in Oxford in September 2002. At the time, only a handful of genes had been reported in association with craniosynostosis, so testing consisted of looking for common variants in the FGFR1,

FGFR2, FGFR3 and TWIST1 genes, which could provide patients with a molecular diagnosis of Pfeiffer, Crouzon, Apert, Saethre-Chotzen, and other syndromes that are caused by variants in these genes.

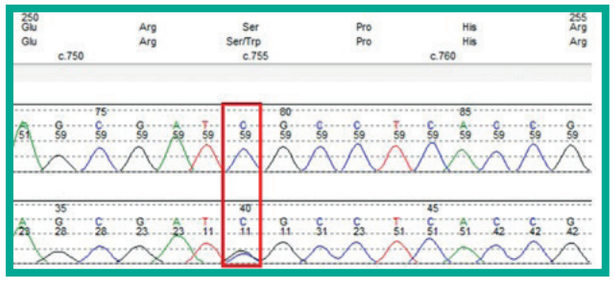


Figure 1: Above is a sequencing trace from a patient with Apert syndrome. The top trace shows a normal control for comparison, and the bottom trace is the patient. The red box shows that in the patient two different peaks are present, which represents a variant at this position. In this case the patient has been shown to have the c.755C>G p.(Ser252Trp) pathogenic variant which is seen in 2/3 of cases with Apert syndrome.

A great deal has changed since 2002 due to extensive research and the application of new technologies, allowing more causative genes to be discovered and incorporated into the diagnostic service. Since 2019, following the introduction of the National Genomic Test Directory, testing methodologies have been adjusted so that both laboratories now offer the same testing to all patients.

The new streamlined testing protocol consists of an initial test with a small in-house gene panel including the core genes tested since 2002, alongside three other genes: EFNB1, TCF12 and ERF. These seven genes together make up the more common causes of craniosynostosis, and this panel can make a diagnosis in around 25% of eligible patients tested. Cases with suspected Apert syndrome are tested separately with a highly specific targeted test.

If the initial testing is negative, for more complex cases where there is multi-suture involvement or other presenting features, a larger gene panel (currently comprised of 55 genes) is analysed to try and reach a diagnosis. This panel is reviewed annually to ensure any newly- discovered genes associated with craniosynostosis are incorporated, ensuring more patients with rare craniosynostosis syndromes are diagnosed. Although only small numbers have been tested via this panel, so far a diagnosis has been made in 20% cases tested in the 3 months up to March 2022.

It was also recognised that specific testing for SMAD6 was necessary, due to the large number of cases of syndromic and non-syndromic craniosynostosis involving the midline sutures. Analysis of this gene alone has been shown to provide a diagnosis of SMAD6-related craniosynostosis in 4.3% of cases with metopic/sagittal synostosis (data January 2020-December 2021).

This is now a recognised test within the test directory and is available for all eligible patients.

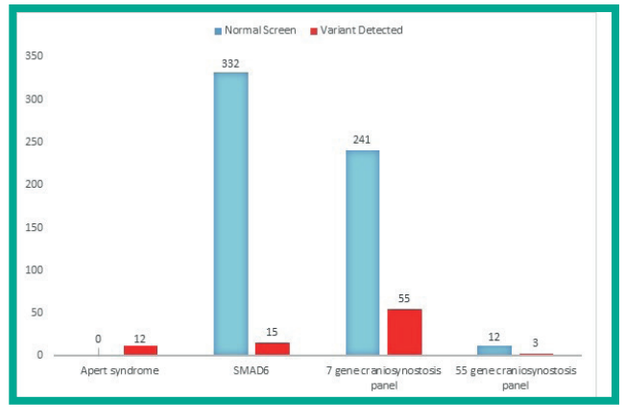


Figure 2: The chart above shows the number of patients with a normal result or where a variant was reported, for each of the respective panels tested in the Oxford laboratory between January 2020-December 2021.

The introduction of more comprehensive testing in the NHS will therefore allow for more diagnoses to be made in children with complex craniosynostosis syndromes in a shorter time frame than ever before.

You can find out more by visiting the Oxford Laboratory website www.ouh.nhs.uk/geneticslab, or the Great Ormond Street Laboratory website www.labs.gosh.nhs.uk/laboratory-services/genetics/molecular-genetics-service

Have you visited our online shop?

Headlines products are available on our website, with safe, easy ordering and special deals! Every single purchase you make helps us in our work to support those affected by rare craniofacial conditions.

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We have an active panel of members who assist us by reading and assessing our information materials – if you'd like to join, drop an email to info@headlines.org.uk and we will add you to our list.

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We are always on the lookout for suitably qualified members to join our board of trustees – we're particularly keen to hear from people with a finance/accounting or legal background, but would also love to hear from anyone who feels their skills could help us achieve our future ambitions.

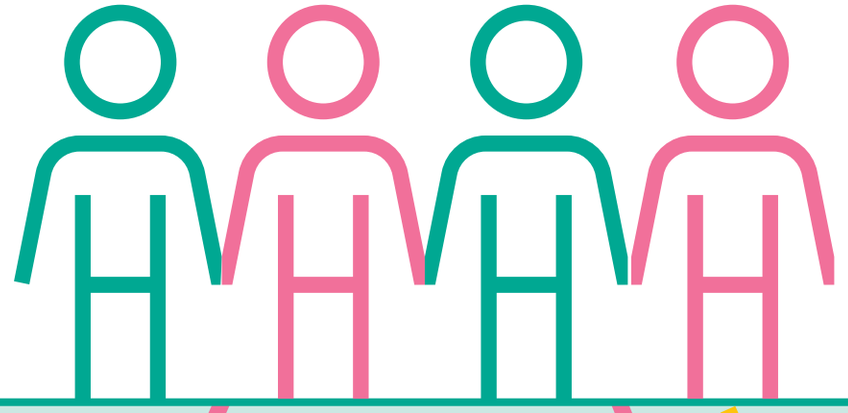
If you're interested, you can download information on what's involved in becoming a trustee on the Charity Commission website www.gov.uk/government/publications/the-essential-trustee-what-you-need-to-know-cc3

Fundraise for us

Headlines relies entirely on donations to fund its work – we receive no government funding. Whether you're having a party, running a marathon or holding a bake sale, every bit you raise can make a real difference. For free, downloadable posters, visit www.headlines.org.uk/fundraising

Take part in research

Taking part in a research study can be a very interesting and rewarding experience – for further information on current studies, check out www.headlines.org.uk/get-involved-in-research



Headlines is the only national charity supporting those affected by craniosynostosis and rare craniofacial conditions. As well as providing information for parents, carers and families, we also offer a confidential helpline and regular newsletters, conference and information days and an annual Family Weekend.

Join us!

Membership is free.
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