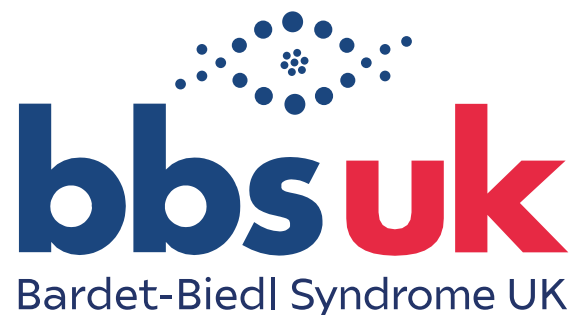


Annual Conference Report 2024



Save the date!

25 - 27 April 2025



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Foreword



On behalf of the BBS UK team, I am delighted to share with you a report from our 31st Annual Conference, held via webinar on 8th June 2024. It was a privilege to have so many of you join us for a day filled with insightful discussions, knowledge sharing, and the strengthening of our community ties. Although we all missed not being able to meet up in person, it did mean that we were able to welcome attendees from the comfort of their own homes, and from all around the world, including America, Germany, France, Spain, Netherlands, Ireland, and Australia. We had 217 registrations for the event, with attendance reaching 130 on the day.

This year's event featured a comprehensive program, showcasing diverse presentations from leading experts in the field. We had the pleasure of hearing from renowned specialists who shared the latest research developments, clinical advancements, and practical advice on managing BBS. A highlight is always the personal perspectives which resonate with so many of us; we are incredibly grateful to those who shared their journey with us in this way. Summaries of all the presentations and personal perspectives can be found within this bumper report; the full recordings can be found on the BBS UK YouTube channel and accessed via our website.

To our speakers, we extend our heartfelt thanks. Your expertise and dedication are invaluable to the BBS community, and your contributions have greatly enhanced our collective understanding and support for those affected by Bardet-Biedl syndrome.

BBS UK is here to support all those living with the syndrome, their families and carers and the professionals involved in their care and relies on the generosity of our supporters to continue providing vital services, including this annual conference. If you would like to contribute, information on how to make a donation or become a 'Friend of BBS UK' can be found on our website. <https://bbsuk.org.uk/donate-to-bbs-uk/#donation-options>

As you will read, the Charity has so much to offer in terms of information and support services so please do stay connected, make sure that you are on the mailing list to receive news, and we really hope to see you at future events.

Laura Dowswell
Chair, BBS UK

SAVE THE DATE:

Planning has already begun for Conference 2025, which will be held across the weekend of 25th - 27th April 2025, at the Hilton Hotel, Northampton. Further details will be available on the BBS UK website in the coming weeks and months; we look forward to seeing you there!

Introduction to Bardet-Biedl Syndrome (BBS)

Dr Shehla Mohammed obtained her degree in Medicine from Pakistan and has subsequently worked in the NHS for 34 years, training in paediatrics, before specialising in Clinical Genetics. She was an ICRF Research Fellow in Cancer Genetics prior to taking up a consultant post at Guys and St Thomas' Hospital, London.



Dr Mohammed has a longstanding interest and experience in rare genetic disorders, and in the care of children and families with life-limiting conditions. She has been involved in setting up and running the national BBS clinic for adults with Professor Phil Beales since its inception in 2010 and oversaw the move of the service to its new home in the Rare Disease Centre (RDC) at St Thomas' Hospital in 2018. Dr Mohammed is also involved in the running of other highly specialised services at the RDC.

Until 2017, Dr Mohammed was Head of Service of the Guy's Regional Genetics Service for 20 years. She represents the genetics speciality on local, regional, and national committees (member of highly specialist committee of NICE) working on national policy developments and research.

Dr Shehla Mohammed provided an introductory overview of Bardet-Biedl syndrome (BBS) including the genetic basis, clinical features, diagnostic criteria, inheritance patterns, and the support available at the multi-disciplinary BBS clinics.

Genetic Basis and Inheritance of BBS

Basic Genetics:

Each cell contains a nucleus with chromosomes that are made from DNA. DNA carries our genetic information and gives instructions for how the body grows and develops. Genes are present in pairs with one set of genes inherited from each parent.

BBS Inheritance:

BBS is an autosomal recessive condition, meaning both parents are usually carriers of the spelling mistakes in both copies of the BBS gene for a child to be affected. Carriers have one normal copy and one with the spelling mistake in the BBS gene (also called a mutation). Carriers are generally healthy and without health implications. Each pregnancy has a 25% chance of producing an affected child if both parents are carriers.

Genetic Variability:

Over 22 BBS-related genes have been identified, with BBS1 and BBS10 being the most common. Genetic mutations can cause varying symptoms among patients, even with the same mutation.

Clinical Features and Diagnosis

BBS is a rare genetic disorder impacting multiple systems. Symptoms can vary widely, which can make diagnosis difficult.

Primary Features:

- **Rod-Cone Dystrophy:** A condition that affects the retina in the eye, leading to progressive sight loss.
- **Polydactyly:** Extra fingers or toes are common in BBS patients.
- **Learning Disabilities:** learning and development may be delayed and/or impaired.
- **Renal Abnormalities:** Kidney issues, including structural and functional problems.

Secondary Features:

- **Speech and Learning Difficulties:** Challenges in communication and processing information.
- **Obesity:** A tendency to gain weight and constant feelings of hunger (hyperphagia).
- **Hormonal Issues:** Problems with hormone regulation affecting various bodily functions.
- **Balance Issues:** Difficulty with coordination and balance.

Diagnostic Criteria:

Diagnosis can be via genetic testing, or via clinical diagnosis, which requires the presence of four primary features or a combination of primary and secondary features.

Genetic Testing and Family Planning

Testing Options:

- **Carrier Testing:** Siblings and family members can undergo genetic testing to determine if they are carriers of the BBS gene. This is particularly useful for family planning and understanding genetic risks.
- **Prenatal Testing:** Two main types of prenatal tests are available:
 - **Chorionic Villus Sampling (CVS):** Conducted around 11 weeks of pregnancy. This test involves taking a small sample of the placenta to analyse for genetic abnormalities.
 - **Amniocentesis:** Usually performed at around 16 weeks, this test involves extracting a small amount of amniotic fluid to test for genetic abnormalities.
- **Pre-implantation Genetic Testing (PGT):** This is a specialist procedure where couples undergo IVF with genetic testing of embryos before implantation. Only embryos without the BBS gene mutation are implanted, reducing the risk of having a child with BBS.
- **Detailed Prenatal Scans:** Specialised ultrasound scans can detect physical anomalies associated with BBS, such as extra digits or kidney abnormalities. However, not all features of BBS may be picked up by a specialist ultrasound scan.

Implications for Families

Informing Decisions: Knowledge of carrier status and potential genetic risks enables families to make informed decisions about family planning.

Non-Invasive Options: Some couples may choose not to pursue invasive testing, opting instead for detailed ultrasound scans to monitor for features of BBS.

Postnatal Testing: For couples who opt out of prenatal testing, postnatal genetic testing can be performed using a sample taken from the baby's umbilical cord after birth.

Support and Counselling

Genetic Counselling: Genetic counselling is offered to help families understand the implications of genetic testing and support them with their decision-making process. Counsellors provide comprehensive information about the risks, benefits, and limitations of different testing options.

Decision Support: Whatever choice couples may wish to make; the BBS clinics are there to support and explain their options and provide support for their decision. This includes explaining complex genetic information in an accessible way and discussing the potential emotional and practical implications of each option.

Personalised Guidance: Couples considering having children are provided with personalised guidance to make choices that align with their values and circumstances. This includes support for those who decide against testing, those who choose non-invasive monitoring, and those who opt for more detailed genetic testing.

Advancements and Clinical Support

Clinical Services:

BBS clinics, established in 2010 with BBS UK's support, have significantly improved patient access to specialised care. Located in London and Birmingham, the clinics provide comprehensive support and coordinated services for BBS patients. For more information visit the BBS UK website, www.bbsuk.org.uk.

Research and Therapies:

Ongoing research and patient observations have led to a better understanding of BBS and the development of new therapies. Continued learning from clinical experiences is crucial for advancing BBS treatment and support.

Dr Mohammed thanked the audience for their attention and participation and expressed appreciation for the collaborative efforts in BBS research and clinical care.

Update on Research and Study of BBS

Professor Philip Beales obtained his degrees in Genetics and Medicine from University College London. He undertook postgraduate training in both general medicine and paediatrics before specialising in Clinical Genetics. With colleagues at Baylor College of Medicine in Houston, he discovered the first gene (BBS6) to cause BBS. Since then, at least 22 genes are now linked to the syndrome.



Professor Beales is based at The Institute of Child Health/Great Ormond Street Hospital where he heads the Cilia Disorders Laboratory. Together with collaborators from Europe and North America, his group have made major advances in our understanding of the causes of the Syndrome. This includes the notion that abnormally functioning cilia (small finger-like appendages on cells) lies at the heart of BBS. The challenges that lie ahead involve understanding how dysfunctioning cilia contribute to various syndrome aspects. These discoveries have brought closer the goal of designing treatments, for example, to prevent further visual deterioration or weight gain.

Professor Phil Beales began by welcoming attendees from around the world. He acknowledged participants from various time zones, highlighting the global interest in Bardet-Biedl syndrome.

Publication of Diagnostic Criteria

In June 1999, Professor Beales and his team published the first diagnostic criteria for BBS. Drina Parker and BBS UK (then LMBBS) supported this work. These criteria were needed because genetic tests were not available then. Today, these criteria are still used, but genetic testing is now the main method for diagnosing BBS. The criteria help doctors know when to test for BBS or similar conditions.

Role of Cilia and the BBSome

Cilia are tiny, hair-like structures on the surface of cells, and they play a crucial role in many body functions. In the eyes, cilia are essential for vision because they transport proteins in photoreceptor cells.

The BBSome is a group of proteins important for cilia function. It helps move molecules to and from the cilia. Mutations in any BBS genes can disrupt the BBSome, leading to problems with cilia. This causes many BBS symptoms, such as vision loss and kidney issues.

Clinical Trials in Mice

Professor Beales then went on to describe the process and the outcome of a clinical trial in mice. The main goal of the trial was to test a gene therapy for BBS. The therapy targeted the BBS1 gene, the most common mutation in Western countries.

Subjects: The trials used mice, modified to have BBS symptoms, including early weight gain and vision loss.

Groups: There were ten groups of mice. Researchers compared BBS mice to normal mice (wild-type). They tested three doses of the gene therapy: low, medium, and high. The study lasted six months.

Procedures: The therapy involved injecting the BBS1 gene, via a deactivated virus, into the retina. They measured effectiveness using:

- **Electroretinography (ERG):** This test measures the retina's electrical activity in response to light.
- **Optical Coherence Tomography (OCT):** This imaging test measures the thickness of retinal layers.

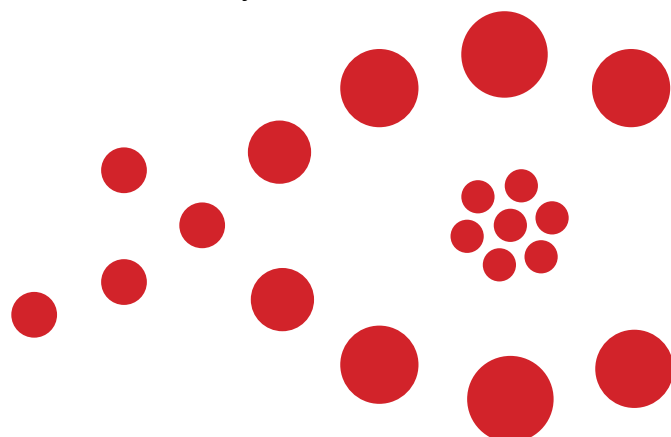
Results

- **Gene Delivery:** The deactivated virus delivered the BBS1 gene to the retina's photoreceptor cells. This was shown by green fluorescent protein and BBS1 gene expression in the cells.
- **Retinal Preservation:** Treated mice had better retinal structure than untreated mice. Medium and high doses worked best in keeping the retinal layer thick.
- **Functionality:** ERG results showed that treated mice had better retinal function. This means the gene therapy maintained both structure and function of the retina.
- **Durability:** The BBS1 gene expression stayed stable in treated mice for six months. This suggests long-lasting benefits.

In conclusion, the studies show that BBS1 gene therapy is safe and effective in preventing vision loss if given early. It is important to remember that this gene therapy cannot restore already lost vision. The results are promising for moving to human trials.

The next step is to organise human trials to test the therapy's effectiveness. Key questions remain about the timing, location, and specific protocols for these trials. These trials will help find the best ways to use the therapy in humans. Professor Beales thanked his team, especially Victor Hernandez, who designed and interpreted the studies. He also thanked Alsa Ventures for their financial support.

The progress in gene therapy for Bardet-Biedl syndrome is a significant breakthrough. With ongoing research and support, there is hope that effective treatments for BBS will, in time, become available. This will improve the quality of life for those affected by this condition.



Respiratory Ciliopathy and BBS



Professor Claire Hogg is Professor of Practice in Paediatric Respiratory Medicine at Imperial College London and Clinical Director in Respiratory Paediatrics at Royal Brompton Hospital. She is clinical lead for the national Primary Ciliary Dyskinesia (PCD) Diagnostic service, in London, and is a leader in clinical research in PCD and novel diagnostics. Many new techniques, including 3-dimensional electron tomography and the validation of immunofluorescence for the diagnosis of PCD, have been developed in her research group and under her supervision. Her group is the first in the world to introduce the use of artificial intelligence to assist the diagnosis of PCD. She has had successive NIHR i4i grant funding to support this work.

In her clinical practice, diagnosing and managing children with PCD, she works closely with other services looking after patients with complex primary ciliopathies, including BBS.

In her presentation, Professor Hogg discussed respiratory ciliopathy and Bardet-Biedl syndrome (BBS), with a focus on how these conditions present in patients, how they are diagnosed, and how they are managed.

Understanding Cilia

Cilia are tiny hair-like structures on cells. They are found in various parts of the body, including the lungs. There are two types of cilia: motile and non-motile.

- **Motile Cilia:** These cilia beat in a coordinated manner to clear mucus and debris from the airways. This helps keep the lungs clean and free from infections. Motile cilia also play a role in other body functions, such as moving eggs through the fallopian tubes in females.
- **Non-Motile Cilia:** These cilia have sensory and signalling functions. They are crucial during the development of an embryo. If they do not work properly, it can lead to organ positioning issues and other developmental problems. For example, non-motile cilia help determine the left-right axis of the body during development.

Primary Ciliary Dyskinesia (PCD)

Primary Ciliary Dyskinesia (PCD) is a condition where motile cilia do not function properly. This leads to various health problems, mainly affecting the respiratory system. Patients with PCD often have chronic respiratory infections, hearing impairments, and fertility issues. They may also have organs in unusual positions because their cilia did not work correctly during development. This can include the heart being on the right side of the body instead of the left.

Diagnosis of Ciliary Disorders

Prof. Hogg described several methods used to diagnose ciliary disorders.

- **Imaging Tests:** Chest X-rays and CT scans are used to look at the structure of the lungs. These tests help identify any abnormalities, such as lung scarring or collapsed areas of the lung.
- **Nasal Brush Biopsies:** This test involves taking a small sample of cells from the nose. The sample is then examined under a microscope to check the cilia's structure and function. Abnormalities in the cilia's appearance or movement can indicate a ciliopathy.

Case Study

Prof. Hogg shared a case study about an 8-year-old boy. He had a mutation in the OFD1 gene, which is linked to Joubert Syndrome, a type of ciliopathy. The boy had severe respiratory symptoms because his cilia did not work properly. The boy had a chronic wet cough, frequent respiratory infections, and difficulty breathing. His chest X-ray showed that one of his lungs was completely collapsed. Further tests showed that his cilia were abnormal, with many being longer than normal and having bulbous tips.

Management Strategies

Managing respiratory ciliopathies involves several strategies to improve the patient's quality of life.

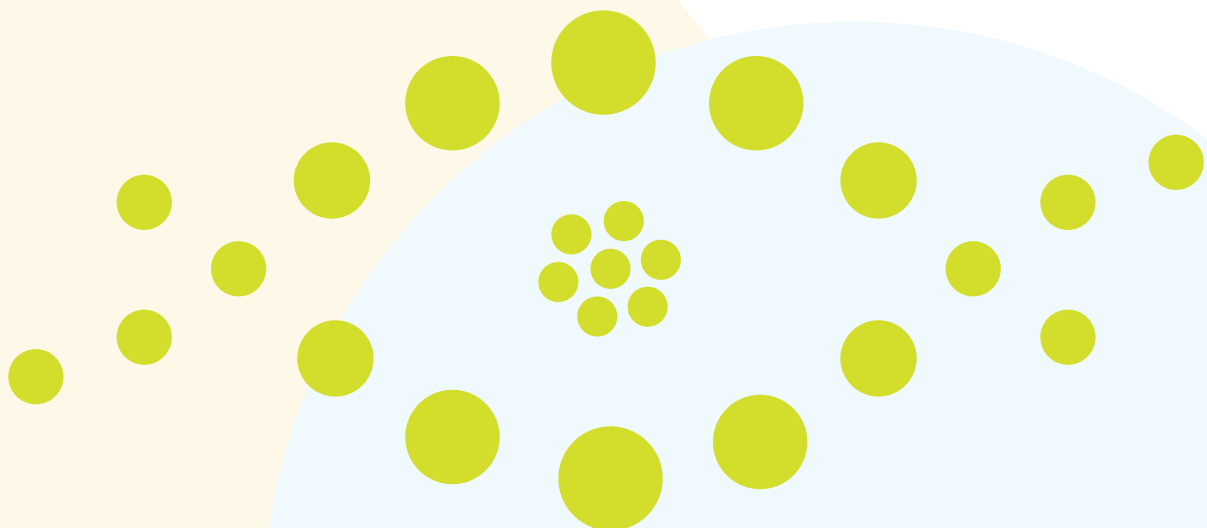
- **Airway Clearance:** It is crucial to keep the airways clear. Techniques like physiotherapy and using a nebuliser with saline help to break down and remove mucus. Physiotherapy can include exercises and techniques to help patients cough up mucus more effectively.
- **Infection Control:** Regular monitoring for infections is important. When infections occur, they are treated with antibiotics. Sometimes, stronger antibiotics given through a vein (intravenous) are needed. Prof. Hogg stressed the importance of early treatment to prevent lung damage.
- **ENT (Ear, Nose, Throat) Symptoms Management:** Many patients have problems like a runny nose, sinus infections, and ear issues. These need to be managed carefully. For example, regular ear check-ups are important to prevent hearing loss due to chronic ear infections.
- **Multidisciplinary Approach:** Care involves a team of specialists working together. This ensures that all aspects of the patient's condition are addressed. This team can include respiratory specialists, ENT doctors, physiotherapists, and genetic counsellors.

Sleep Apnoea in Ciliary Disorders

Prof. Hogg also discussed sleep apnoea, a condition where breathing stops and starts during sleep. This is common in patients with ciliary disorders. Obstructive sleep apnoea occurs when the throat muscles intermittently relax and block the airway during sleep. In patients with ciliopathies, obstructive sleep apnoea can be due to nasal congestion and obesity, both of which are common in these patients. Sleep studies are used to diagnose sleep apnoea. These studies monitor breathing, oxygen levels, and sleep patterns. Treatment can include the use of a CPAP machine, which helps keep the airway open during sleep. Weight management and addressing nasal congestion can also help reduce symptoms.

Prof. Hogg concluded by stressing the importance of early diagnosis and specialised care for patients with respiratory ciliopathies. She highlighted ongoing research and potential future treatments, including gene therapy. Gene therapy aims to correct the underlying genetic defects that cause ciliopathies, offering hope for more effective treatments in the future.

Prof. Hogg thanked her team for their work in understanding and managing respiratory ciliopathies. She also acknowledged the patients and families who participate in research and share their experiences to help advance medical knowledge.



Q&A – Professor Phil Beales, Doctor Shehla Mohammed, and Professor Claire Hogg

Does the type of BBS gene affect how severe the symptoms are?

Dr Mohammed: There is a lot of variation among the 22 BBS genes. For example, BBS1 is generally milder, but this isn't always true. Some genes, like BBS10, might cause more severe problems or be associated with specific issues like kidney problems. However, it is hard to predict exactly how severe the symptoms will be, based on the gene type alone, as there is a lot of individual variability.

Can BBS affect pregnancy or the ability to have children?

Dr Mohammed: Yes, BBS can sometimes affect fertility. It is more commonly recognised in men but can also affect women. Women with BBS often have related conditions like polycystic ovary syndrome, which can impact hormone levels and the ability to conceive. However, many individuals with BBS have successfully had children. If there are difficulties, further tests and treatments can be undertaken, often involving endocrinologists who specialise in hormones and fertility.

Will approving gene therapy for one BBS gene make it quicker to gain approval for other BBS genes?

Prof Beales: No, each gene therapy is considered a separate drug, so each one needs its own approval. However, regulators like the FDA (Food & Drug Administration) are exploring ways to speed up the process. If the method used to develop one gene therapy is proven successful, it might help to fast-track the approval process for other gene therapies in the future.

When should gene therapy be given to be effective?

Prof Beales: Gene therapy needs to be given early when there are still some working cells in the eye, specifically the photoreceptors (rods or cones). The therapy can stop the disease from getting worse but cannot revive cells that are already dead. So, it is important to start the treatment while there are still enough healthy cells to preserve.

Are there treatments for advanced eye disease that work no matter what the gene is?

Prof Beales: Yes, there are exciting developments in treatments for advanced retinal degeneration that do not depend on the specific gene involved. For example, the Foundation Fighting Blindness is working on these treatments, and more information will be shared at the upcoming BBS conference in the US. This offers hope for those who feel left out by gene-specific therapies.

A Sibling's Perspective

Greg Dowswell is a 28-year-old video-producer from Surrey. He shared his personal experiences growing up as the younger brother of Maria, who was diagnosed with Bardet-Biedl syndrome (BBS) at age 13. Greg gave an insight into their sibling relationship and how Maria's diagnosis affected their family. He used photos throughout his presentation, describing them to the audience to give a visual representation of their shared memories.



Early Memories:

Greg recounted his early memories with Maria, highlighting their playful childhood before her diagnosis. He mentioned instances of clumsiness during family holidays that hinted at Maria's condition. Greg showed a photo of them at ages four and seven, leaning over the back of a chair and pulling cheeky faces.

Diagnosis and Impact on Family:

Maria's medical journey began with numerous tests in London, causing concern for the family. Despite the focus on Maria's health, Greg's parents ensured he continued to engage in his activities and hobbies, such as Sunday football and swimming lessons. A significant moment was when Maria had a suspected brain tumour, which led to an urgent MRI on Greg's 10th birthday, a day overshadowed by anxiety for Maria's well-being.

BBS diagnosis:

Following Maria's diagnosis, both siblings received counselling, which Greg found beneficial in understanding and coping with the situation. These sessions helped him process the emotional impact of Maria's condition and develop a better understanding of genetics and biology related to BBS. Greg showed a picture of them sitting on a pier shortly after Maria's diagnosis, looking windswept but content.

Adapting to a New Norm:

As the family adjusted to Maria's condition, white canes became a common sight at home. Greg's focus was on his education, while Maria and their parents battled with the local council over her right to choose her own college. They were ultimately successful, and Maria chose the college she wanted. Greg admired his parents for balancing their attention between Maria's needs and his own interests.

Independence and Support:

When Maria moved to a college with supported accommodation, Greg continued his education and developed a habit of offering physical support to Maria during family gatherings. This period marked a shift towards more independent lives for both siblings.

Living Apart and Reconnecting:

Greg described the distance between him and Maria during their college years and early careers. Despite the physical distance, their bond remained strong through occasional visits and phone calls. Maria's move closer to the family in 2019 and the subsequent pandemic lockdown brought the family back together, challenging and strengthening their relationship.

Current Relationship:

Greg expressed admiration for Maria's independence and resilience. He shared his pride in her accomplishments, including maintaining a full-time job while living with BBS. Greg reflected on his own journey, highlighting how growing up with a sibling with a rare condition made him more independent and appreciative of his family. He included a selfie from the Californian desert.

Greg concluded his presentation by emphasising the positive impact of his experiences and the invaluable lessons learned. He acknowledged the excellent job his parents did in managing the challenges and joys of raising both children. Greg's heartfelt narrative, supported by personal photos, offered a powerful insight into the unique dynamics of growing up in a family affected by a rare disease.

Supporting Siblings

Clare Kassa joined Sibs as CEO in 2018. Clare is an adult sibling – she has an older brother with a learning disability. She has a long connection with the charity and was part of the working group to set up Sibs back in 2001. Clare has worked in the voluntary sector supporting disabled children and adults, and their families, for over 25 years. She has delivered workshops on sibling issues, been involved in research, and undertaken direct support work with adult siblings. Clare is passionate about sibling support and wants to ensure that the important role siblings play in their families is properly recognised. Clare shared her personal journey and introduced the work of the charity.



Sibs is the only charity in the UK focused solely on supporting the siblings of disabled individuals. It provides various resources and support through:

- **Young Sibs service:** For siblings aged 7-17, offering information, one-to-one support, and advice.
- **Website:** Offering resources for parents and professionals, and support for adult siblings.
- **Workshops:** For parents on topics such as supporting siblings, sibling rivalry, and planning for the future.
- **Sibling group leader training:** For those interested in running sibling support groups in the UK.

Clare then described some of the common issues faced by siblings:

- **Lack of Attention:** Siblings often receive less attention from parents who are focused on the needs of the disabled child. This is frequently raised by both parents and siblings who contact the charity.
- **Increased Responsibilities and Worries:** Siblings may have more worries and responsibilities than their peers. They often worry about their family's current and future situations.
- **Social and Academic Impact:** Siblings might struggle with sleep, friendships, and schoolwork, especially if they are young carers. About half of all young carers in the UK support a brother or sister.
- **Emotional and Behavioural Challenges:** Siblings might exhibit overachievement, passivity, or occasionally act out, often mimicking their disabled siblings' behaviours for attention. There is also a tendency towards perfectionism among siblings.

Clare highlighted the positive experiences of sibling relationships, including love, care, and loyalty. Many siblings gain valuable skills and often pursue careers in health, education, and social care due to their experiences.

Teen siblings face unique challenges, including constraints on social time, school expectations, and pressures from parents. Clare shared insights from teenage siblings about their experiences and the need for support from schools and families.

Adult siblings also experience varied challenges, such as balancing care responsibilities with personal life, dealing with childhood experiences, and often feeling misunderstood. Sibs provides support around self-care and well-being for adult siblings.

Resources that Sibs offer:

- **Online Support:** Sibs offers e-books, peer support groups, and guides on topics like mental capacity and future planning. These resources are designed to support both young and adult siblings.
- **Educational Materials:** Sibs provides resources for schools to support young siblings, including films, presentations, and worksheets, which are freely downloadable from their website.

Ways to help a sibling feel more connected:

- **Self-Identification:** Encouraging siblings to identify and connect with the broader sibling community. Many siblings are unaware that there is a larger community that shares their experiences.
- **Celebrity Siblings:** Highlighting public figures who are also siblings, such as Jo Wiley, Alicia Dixon, Lewis Hamilton, Rag'n'Bone Man, and Jack Grealish, to foster a sense of community and understanding.

Clare concluded by inviting attendees to explore Sibs' resources and contact the Charity for more personalised support. She encouraged signing up for the newsletter and following Sibs on social media for updates and information on events and research. Clare emphasised the importance of recognising and supporting the unique needs of siblings, who often play a crucial but underappreciated role in families with disabled members.

Overall, Clare's presentation underscored the significant role of siblings in families with disabled members and the comprehensive support Sibs provides to address their unique challenges and needs.

Sibling Q&A

The responses have been condensed for clarity.

Greg, at what age did your parents explain fully what BBS was? How did they go about it?

Greg: I think it must have been when Maria was initially diagnosed. I was only told what Mum and Dad knew. As I got older, it was gradually explained to me in more depth.

Clare: From about age seven, siblings start to ask questions. Having good, uncomplicated information is really helpful for siblings. At Sibs, we have got loads of resources such as book lists and reading material because it's really nice for siblings to be able to read about other children who share some of those similar experiences.

Greg, having been a sibling to someone with BBS, is there anything that you found to be particularly helpful that your family did?

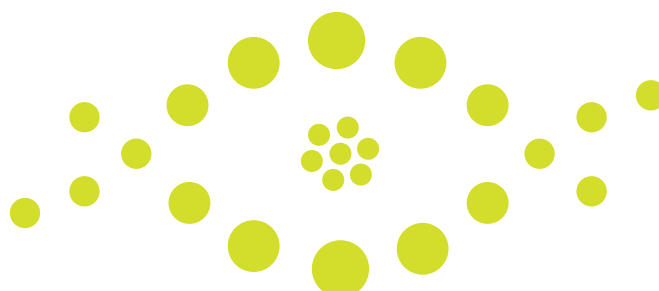
Greg: Yes, there were my own hobbies which were always fulfilled; being part of the sports teams or playing drums. That made me feel valued as a youngster. Mum and Dad gave me amazing support to do my own things as well as juggling everything that was going on with Maria.

Greg, did you feel protective of Maria growing up? Did having BBS in your family make you more aware of disabilities in general?

Greg: Yes, there certainly was an element of protection over Maria, though that was more apparent as I got older, because I was still the younger sibling. I became more aware of other people with disabilities. When I met other people who mentioned having a sibling with another disability, it became a good opening point to talk and empathise with each other.

Claire, could you tell us more about the events you organise for adult siblings and how to access them?

Claire: We advertise everything through our monthly newsletter. We have a new set of sessions that are going to run this autumn for adult siblings around a range of topics, including things like advocacy around behavioural challenges, on safeguarding issues, and on mental capacity in the law. Every couple of years we run an adult sibling event. The last one was in October last year, but a brilliant opportunity to bring together adult siblings face to face. We also have support groups and a Facebook page with over 500 members for those who prefer online interaction.



Artificial Intelligence for Accessibility

Hector Minto is the European Director for Accessibility at Microsoft. With over 25 years of experience, he works to make technology accessible to everyone and advocates for adding accessible solutions into mainstream technology. He also advises the UK government and many organisations on how to include accessibility in their digital plans. His career is dedicated to using technology to help people with disabilities.



Hector Minto talked about how artificial intelligence (AI) can help people with disabilities. He focused on the importance of making digital experiences accessible, as technology is a big part of our lives. His colleague David Prince, who spoke about AI and accessibility at our last conference, supported the session by answering questions in the chat function of Zoom.

Windows Accessibility Features:

Hector started by talking about the built-in accessibility features in Windows. He showed how to open these settings by pressing the Windows Key + U. Features like Dark Mode and High Contrast Themes help users with vision problems.

Seeing AI:

Seeing AI is a smartphone app that uses the camera to read text aloud, recognise handwriting, identify colours, and even navigate buildings. It can also recognise and describe people's faces. Seeing AI is free and works on both iOS and Android.

Be My Eyes:

The Be My Eyes app connects users with real life volunteers or AI to provide assistance. For example, AI can describe screens, pictures, and documents. Be My Eyes is free and can be downloaded on Windows and mobile devices.

Copilot:

Copilot is a tool that works with Microsoft Edge and other platforms. Copilot summarises and answers questions about web content. It can describe images and interpret charts, making digital content easier to understand. This tool helps users with disabilities interact with the web more easily and can be very helpful for students.

OneNote:

OneNote is a digital notebook that comes free with Microsoft 365. Hector demonstrated how to use OneNote to convert photos of text into accessible formats. He gave the example of using OneNote to convert text from a photo of an invoice, into text that your computer can read out for you.

Q&A Section:

One question we have received is about the cost of accessibility tools. What is Microsoft doing to help bring down these costs?

All the features I have shown today, like the accessibility tools in Windows, are built-in and available for free for those who have Windows. We are also working with assistive technology companies to reduce their costs. By supporting these companies with our technology, we help them keep their products affordable. Additionally, we fund global programs to make sure assistive technology reaches under-served areas. This helps reduce costs in regions where importing these technologies can be very expensive.

What support and training is available for these tools. What resources does Microsoft offer?

We have a lot of support and training options. First, we have the Disability Answer Desk (DAD), which is a free service offering support for accessibility issues. We also have the Enterprise DAD for organisations. Sometimes features aren't enabled, simply because people don't know how to do it; the DAD helps with that. Microsoft Learn and LinkedIn Learning offer free training courses on assistive technologies; these courses are accessible and cover a wide range of topics.

How can someone use AI tools if they don't have a touchscreen?

If you don't have a touchscreen, you can use keyboard shortcuts. For example, you can navigate using the tab and arrow keys. This method requires some training, but it's very effective. For additional help, charities like AbilityNet offer training and support to both individuals and companies. Additionally, resources such as Microsoft Learn and LinkedIn Learning offer free courses on various assistive technologies.

BBS UK: The Royal National Institute of Blind People (RNIB) also have resources to help.

How can I get AI to read an invoice that is an image?

If you have an image of an invoice, there are a few ways to get AI to read it. You can use the Seeing AI app, which allows you to point your camera at the invoice and have it read aloud. Alternatively, you can use OneNote. Take a photo of the invoice, upload it to OneNote, and use the Immersive Reader to have it read to you. Both methods are effective and use AI to make the text accessible.

Hector wrapped up the presentation by urging everyone to try these AI tools. He emphasised how AI can make technology more accessible and invited the audience to explore tools like Copilot.

Hector and his colleague, David, will be hosting an online workshop for a more detailed and interactive overview of these technologies. If anyone is interested, please email us at admin@bbsuk.org.uk to register.

List of applications and resources:

AbilityNet

AbilityNet is a UK charity that helps everyone, regardless of ability or age, to use digital technology. Their free online resources and a network of over 450 community-based volunteers help individuals with any disability, of any age, to use all kinds of digital technology.
<https://abilitynet.org.uk/>

Be My Eyes

Be My Eyes connects blind and low-vision users who want sighted assistance with volunteers and companies anywhere in the world, through live video and artificial intelligence.
<https://www.bemyeyes.com/>

Copilot

Microsoft Copilot is an AI-powered productivity tool developed by Microsoft. It serves as your everyday AI companion, helping you find information, create content, and enhance your work experiences.
<https://copilot.microsoft.com/onboarding>

Microsoft Disability Answer Desk – for users i.e. you/children, carers

The Microsoft Disability Answer Desk is where customers with disabilities get support with Microsoft Office, Windows, and Xbox as well as 3rd party products like JAWS that run on Microsoft software i.e. your Windows PC. Visit online or call 0800 026 0584.
<https://www.microsoft.com/en-gb/accessibility/disability-answer-desk>

Enterprise Disability Answer Desk

The enterprise Disability Answer Desk is a support resource for organisations that have questions about the accessibility of Microsoft products and product conformance with accessibility standards. The support team can help resolve issues relating to assistive technology and functionality of products for users with disabilities, as well as find conformance documentation.
<https://support.microsoft.com/en-us/accessibility/enterprise-answer-desk>

Microsoft Learn

Microsoft Learn allows you to learn how to use Microsoft products and services through task-based, interactive learning. They have hundreds of free courses, in 23 different languages.
<https://support.microsoft.com/en-us/accessibility/enterprise-answer-desk>
Look here for accessibility courses: Browse all courses, learning paths, and modules -

<https://learn.microsoft.com/en-gb/> (MS Learn)
<https://learn.microsoft.com/en-us/training/browse/?terms=accessibility> (All accessibility courses)
<https://learn.microsoft.com/en-us/training/modules/introduction-to-accessibility/> (Intro to Accessibility)
<https://learn.microsoft.com/en-us/training/paths/accessibility-fundamental/> (Accessibility Fundamentals)

OneNote

Microsoft OneNote is a note-taking software that is part of the Microsoft 365 suite. To access OneNote, you must have Microsoft 365 on your device.
<https://support.microsoft.com/en-gb/office/accessibility-tools-for-onenote-37ea8231-860c-4a9e-b04b-f14ee4e1c3bd#:~:text=Learn%20about%20accessibility%20tools%20for%20OneNote%20and%20how%20to%20make>

Seeing AI

Seeing AI is a free app for IOS & Android that narrates the world around you. Designed with and for the blind and low vision community, Seeing AI assists with daily tasks from reading, to describing photos, to identifying products, and more. The app continues to evolve with input from the community and AI research advances.
<https://www.seeingai.com/>

RNIB

RNIB is a charity that offers support to blind and partially sighted people across the UK.
<https://www.rnib.org.uk/living-with-sight-loss/>

Education:

Microsoft accessibility tools for Vision: <https://support.microsoft.com/en-us/topic/accessibility-tools-for-vision-b3c57606-e0af-46d2-97b4-fa6b5fba4fa1#:~:text=This%20page%20lists%20sight-related%20accessibility%20tools%20to%20help>

Watch or listen to videos from Microsoft on Accessibility go to:

<https://www.youtube.com/@MSFTEnable>
Hector talking on Accessibility:
<https://www.youtube.com/playlist?list=PLtSVUgxl06Kp6YmGI9pIgE3V9bFeAFU1t>

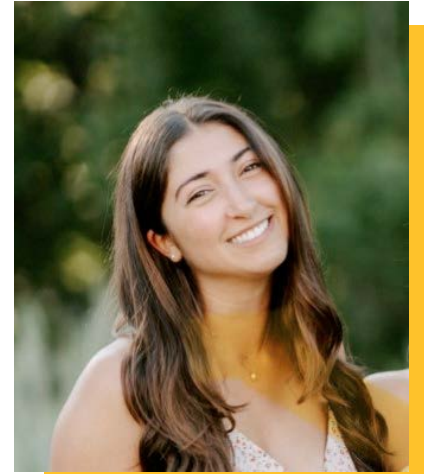
People to follow on LinkedIn:

(10) Jenny Lay-Flurrie | <https://www.linkedin.com/in/jennylf/>
(10) Michael Vermeersch | <https://www.linkedin.com/in/michael-vermeersch/>
(10) Hector Minto | <https://www.linkedin.com/in/hectorminto/>

Fundraising Roundup

Samantha was BBS UK's Fundraising and Communications Officer from November 2023 to September 2024.

As a relatively new member to the team at the time, Samantha was in awe of the remarkable work achieved by the Charity and the wealth of information shared at the conference. Samantha started by highlighting BBS UK's achievements over the past year and then spoke about a new fundraising campaign called "Rare Not Alone" and other ways the community can raise funds for the Charity.



Advice Service

Over the past year, BBS UK provided support to over 100 people through its Advice Service, with our registered social workers supporting families and individuals with complex issues across areas including education, housing, and future planning to name just a few.

Newsletter and Publications

The Charity delivered its newsletter to over 840 recipients, containing personal stories, medical updates, and opportunities for community connection. BBS UK also produces a range of publications, including a Medical Information Booklet, Booklet for Schools and Colleges and a Transition Handbook, ensuring accessibility and utility through their accreditation with the Patient Information Forum.

Clinic Support Service

BBS UK's clinic support service assisted over 450 families, with patient liaison officers providing essential support, before, during and after their clinic appointment to enable them to make the most of their appointment.

Global Conference Engagement

The Charity connected with over 100 conference attendees from several different countries, emphasising the importance of such events in driving change and improving lives. These meetings provide a platform for discussing the future of living with BBS and available support, fostering a sense of community among those with the rare condition.

Support Through Donations and Campaigns

Samantha highlighted the vital role of supporters in enabling the charity's work, noting that BBS UK relies entirely on donations. She recounted the success of the "Carry the Gene" campaign, which raised over £80,000, showcasing the community's potential when united. She encouraged further support through spreading awareness and engaging local businesses for donations or fundraising events.

Rare Not Alone

Samantha introduced the "Rare Not Alone" campaign, encouraging personalised fundraising efforts that connect with supporters' interests. This campaign is designed to highlight the

individuality of the BBS community while fostering a sense of unity and support. Participants can choose any activity that they enjoy, and which aligns with their personal interests or hobbies. Whether it's cooking, dancing, or gaming, the key is to create events that bring people together and raise awareness and funds for BBS UK.

The “Rare Not Alone” campaign is versatile and inclusive, allowing participants to engage in a way that is most meaningful to them. Here are some suggestions for how supporters can get involved:

- ✓ **Social Gatherings:** Host a meet-up such as a coffee morning or lunch where guests can donate to attend.
- ✓ **Sports and Physical Activities:** Organise a charity run, walk, or cycling event. Smaller-scale events like yoga classes, dance classes, or hiking trips can also be excellent ways to engage people.
- ✓ **Creative Arts:** Hold an art exhibition, photography contest, or craft fair with proceeds going to BBS UK. Local artists and creatives can contribute works, adding a unique local flavour to the fundraising.
- ✓ **Gaming and Virtual Events:** Set up a gaming marathon, or a virtual quiz night. These events can reach a broad audience and are perfect for supporters who prefer digital interaction.
- ✓ **Educational Workshops:** Offer workshops or classes in areas of expertise, such as cooking, painting, or technology. Participants pay a fee, which is donated to the cause.
- ✓ **Community Projects:** Collaborate with local businesses like pubs or cafes to donate a portion of their proceeds from special events or sales. This not only raises funds but also increases community awareness.

Challenge Events and Regular Donations

Sam asked that supporters consider BBS UK during their next challenge event sign-ups, such as marathons or hikes, to raise awareness and funds. She also emphasised the impact of becoming a Friend of BBS UK by giving regular donations, no matter how small, which provide crucial, reliable funding for the charity's ongoing efforts.

One-Off Donations and Gift Aid

For immediate support, Samantha encouraged one-off donations and stressed the importance of taxpayers ticking the ‘Gift Aid’ box, which allows the charity to claim an extra 25p for every £1 donated, significantly boosting contributions over time. To donate, please visit our website <https://bbsuk.org.uk/donate-to-bbs-uk/#donation-options>

Samantha concluded by thanking all supporters, whose collective contributions enable the charity to support families affected by BBS. We look forward to sharing your fundraising efforts in the next newsletter.

Irritable Bowel Syndrome (IBS) and BBS

Kelly Slater is a highly experienced dietitian working with BBS patients at the MDT clinic at Queen Elizabeth Hospital, Birmingham. Kelly's presentation aimed to provide practical dietary and lifestyle advice for managing IBS symptoms effectively.

IBS is characterised by a range of symptoms, including abdominal pain, bloating, and changes in bowel habits persisting for at least six months. IBS is the most common disorder of the digestive system, affecting between 5% and 20% of the general population.

IBS is a complex condition without a single definitive test for diagnosis, which can make it difficult to determine the cause. A diagnosis of IBS should not be made without medical investigations to exclude other conditions. This ensures that the symptoms are accurately attributed to IBS and not to another underlying issue.



Dietary and Lifestyle Recommendations:

- ✓ **Eat regular meals:** Eat three regular meals a day and avoid late-night eating to allow the body to rest and repair overnight.
- ✓ **Drink enough:** Drink six to eight glasses of fluid daily, focusing on water and herbal teas, while avoiding sugary drinks.
- ✓ **Limit alcohol:** Limit alcohol intake to no more than two units per day and have at least a couple of alcohol-free days each week.
- ✓ **Reduce caffeine:** Reduce caffeine intake to no more than two mugs per day, gradually decreasing if currently consuming more, to avoid withdrawal effects.
- ✓ **Avoid fizzy drinks:** Reduce the intake of fizzy drinks as they can make gas and bloating worse.
- ✓ **Eat healthy food:** Avoid high-fat foods such as fried foods, crisps, chocolate, cakes, fast foods, and pies, as they can negatively affect bowel habits.
- ✓ **Fruit and vegetables:** aim to reach 5 portions of fruit and vegetables a day, but limit fruit to no more than 3 portions throughout the day.
- ✓ **Chewing habits:** Avoid chewing gum and ensure food is chewed thoroughly to aid digestion.

Kelly suggested keeping a food and symptom diary for a couple of weeks to help find out what might be causing the IBS. She also recommended trying probiotics for four weeks to see if they alleviate symptoms. Different brands contain different types of bacteria, so it is important to stay with one brand for the four weeks.

For those with persistent symptoms, Kelly mentioned the low FODMAP diet, which should only be followed under professional supervision due to its restrictive nature. Additionally, she highlighted the potential benefits of hypnotherapy or cognitive behavioural therapy (CBT) for stress-related symptoms.

IBS within the BBS community:

Within the BBS clinics service, clinicians report a high number of patients with BBS who have IBS symptoms. However, there is little to no research supporting this. Kelly emphasised the need for more studies and increased awareness. She advised patients to consult their GPs and seek referrals to dietitians, while also noting the challenges dietitians might face due to their limited knowledge of BBS. Kelly wants to spread awareness about BBS among dietitians to improve patient care and to ensure proper diagnosis and management.

A Journey of Resilience and Triumph

Bilaal Ali is a 21-year-old from Sheffield with BBS who is registered blind. Bilaal is currently in the placement year of an accounting and finance degree at Sheffield Business School, is an avid sports enthusiast and plays blind football and tennis.

Bilaal was recently featured in a Financial Times article entitled 'Dismantling barriers to disabled business students' and has also shared his experiences on the radio. He believes that at the core of his achievements is his unwavering belief in the power of hard work and perseverance.



Bilaal spoke about the challenges and triumphs he has experienced from childhood to his current life as a university student.

Early Life and Education

Bilaal was diagnosed with BBS at birth which brought with it significant challenges. He and his family received support from hospitals and doctors in Sheffield. He attended mainstream primary and secondary schools, where he enjoyed subjects including maths, PE, music, and food and catering. During secondary school, when he had more sight than he does now, Bilaal used various strategies to cope with his vision impairment, such as coloured pens, whiteboards, and tactile diagrams.

Challenges and Adaptations

During his teenage years, Bilaal struggled with accepting his sight loss and initially resisted using a cane. A significant turning point came in 2017 when he broke his ankle, leading to a long recovery period. This injury, coupled with being registered blind, motivated him to start using his cane and embrace his condition. He found solace in sports, particularly blind football, which boosted his confidence and physical health.

Academic Journey

Bilaal's GCSE results were not what he had hoped, and this really affected him mentally. Despite this, Bilaal persevered and excelled in his Business Level 2 and 3 courses at college. He is currently studying Accounting and Finance at Sheffield Hallam University, where he has completed his second year and is preparing for a placement. Support available has included note-takers, sighted guides, technology training, and cane training.

Achievements and Opportunities

Bilaal's hard work and determination earned him the Inspirational Students Award in his first year at university. He has engaged in various opportunities, such as a work experience with the Financial Times through the Thomas Pocklington Trust and a job placement with Nicholson and Co. Accountancy as an assistant accountant. He has also appeared on BBC Sheffield and contributed to a university project book titled 'Rising Above.'

Sports and Hobbies

Outside of academics, Bilaal is an avid sports enthusiast. He plays blind football and tennis, goes to the gym, swims, and boxes. He enjoys attending football matches, playing the djembe drum, and collecting DVDs and Funko Pop figurines.

Bilaal concluded his presentation by emphasising that nothing is impossible and encouraged the audience to work hard and never give up. His story is a testament to the power of perseverance and resilience in overcoming life's challenges.

Q&A Section

You mentioned that you were reluctant to use your cane initially but then later on you did start to use it. Do you have any tips or advice for someone who is starting to use a cane?

Don't worry about what everyone else will think. When I was using it, I was thinking, what are people going to think of me? Are they going to act differently towards me? Then, when I started using it properly after my ankle accident, people became more aware and helped me more. I realised there's no point in not using it.

What do you think has helped to improve your mental health?

The best thing for me is my sports - like football, tennis, gym, and swimming. Also, my faith and religion. Praying and reading the Quran helps me a lot.

Do you have any other sports that you would like to try?

There's a blind sport called Showdown that I have tried once or twice. There's a table and you hit a ball across the table, and you have to get it into a hole. Then there's another blind sport: goalball. One of my friends is going to be setting up a club in Sheffield, hopefully, so I can do that as well. Then there's blind cricket as well. There's a lot of sports available out there for me to do.

What type of support did student support services give you at college and university?

At college, the support services were better. At university, they still help with course materials and other needs, but there are often delays and issues with accessibility. It's not perfect, but I manage. There could be better awareness. Sometimes, I get graphs with poor descriptions or images that I cannot see.

How did accessing work for the first time make you feel?

I was nervous and scared at first, but I found ways to work around my vision impairment with tools like JAWS and ZoomText. It showed me that there are always ways to do something.

Update on Setmelanotide

Dr Forsythe has degrees in molecular medical sciences and medicine from Queen Mary University of London as well as a PhD focused on Bardet-Biedl syndrome from University College London. Dr Forsythe trained in adult medicine and paediatrics before specialising in clinical genetics and becoming a consultant across Guys & St Thomas' Hospital and Great Ormond Street Hospital with a focus on Bardet-Biedl syndrome. Dr Forsythe has 15 years research and clinical experience in Bardet-Biedl syndrome and has been involved in the BBS Clinics in London since their inception in 2010. Her PhD in Bardet-Biedl syndrome was supported by the Medical Research Council and the Great Ormond Street Hospital charity.



In addition to her clinical work, Dr Forsythe is a passionate advocate for patient engagement and sits on the board of the Ciliopathy Alliance UK as well as BBS UK Scientific Advisory Board. Dr Forsythe was an investigator on the trials for Setmelanotide for people living with Bardet-Biedl syndrome in the UK and has a specialist interest in developing therapies for rare diseases.

In her presentation, Dr Forsythe talked about Setmelanotide for BBS. She explained who can use it, its side effects, and how to use this new treatment.

Weight and Hunger in BBS

Dr Forsythe started by talking about how the body controls hunger. She explained that signals from different body parts go to the brain to control appetite and energy use. In people with BBS, the signals that tell them to stop eating do not work well. This causes increased hunger and trouble controlling food intake.

How Setmelanotide Works

Setmelanotide works by fixing the faulty hunger signal pathways in patients with BBS. It acts on specific receptors to signal the brain to reduce appetite, helping to manage hunger and weight.

Clinical Trials and Approval

Dr Forsythe talked about the clinical trials for Setmelanotide. Two people from the UK and others worldwide took part. These trials checked if the drug works and is safe. On 22nd May 2024, Setmelanotide was approved by the National Institute for Health and Care Excellence (NICE) for treating obesity and hyperphagia (excessive hunger) in people with BBS.

Who can get Setmelanotide?

Age: Patients must be 6 to 17 years old when starting treatment. Those who start in their teens can continue as adults, but at present, Setmelanotide is not approved for prescribing in adult BBS patients.

Medical Need: The treatment is for those who cannot maintain a healthy weight despite following a healthy diet and doing regular exercise.

Safety: The patient's medical history and blood tests are checked to ensure it is safe to give Setmelanotide.

Side Effects

Dr Forsythe grouped the side effects into three main types:

1. **Injection Side Effects:** Pain, redness, and irritation at the injection site.
2. **Common Drug Side Effects:** Nausea and stomach pain, which usually get better over time.
3. **Skin Colour Changes:** Changes (darkening) in skin colour because the drug affects receptors that control skin colour.

Setmelanotide Clinics

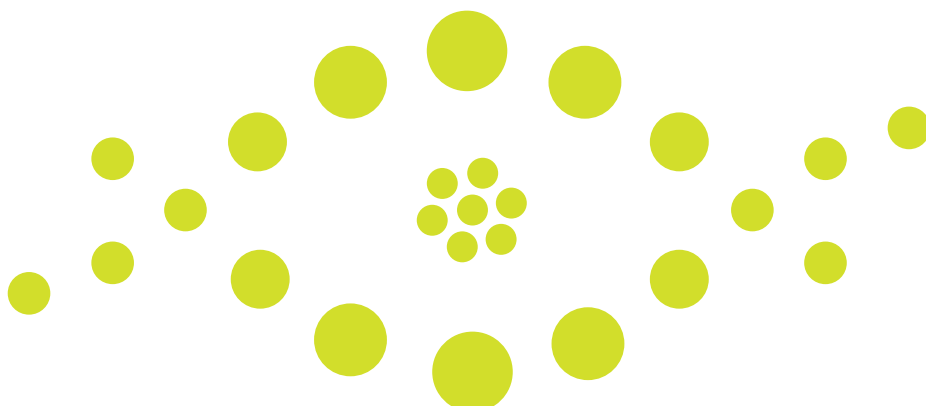
Dr Forsythe announced special clinics for Setmelanotide starting in autumn 2024 at Great Ormond Street Hospital and Birmingham Children's Hospital. The process will be:

- **First Appointment:** Discuss Setmelanotide, its effects, and eligibility. Patients will have thorough assessments and blood tests.
- **Second Appointment:** Learn how to give the injections and get the first dose at the clinic.
- **Home Care and Follow-up:** The drug will be delivered to the home, with ongoing support and follow-ups by local paediatricians.

Other Treatments

For those who cannot use Setmelanotide or want other treatments, Dr Forsythe suggested persevering with diet and exercise. There are other drugs available, including Wegovy and Mounjaro. Last year, Dr Foggensteiner talked about BBS patients using Wegovy with good results in weight management.

Dr Forsythe ended her talk with hope for more weight management treatments for people with BBS. She said that new drugs will likely give more options and better results for managing weight and hunger in the BBS community. BBS patients are encouraged to discuss the latest treatment options for obesity and hyperphagia with their clinic team at their next BBS appointment.



BBS Gene Therapy & Clinical Trials

Dr Arlene Drack is a clinician-scientist specialising in paediatric ophthalmology, with a subspecialty in genetic eye diseases. For 15 years Arlene did primarily clinical research and medical/surgical patient care, was Chair of Paediatric Ophthalmology at The Children's Hospital, Denver, and directed Paediatric Ophthalmology Postdoctoral Fellowship programs.

In 2008 Arlene was recruited back to the University of Iowa to start a paediatric genetic eye disease service and to be a co-investigator in the pivotal Phase 3 gene therapy trial for voretigene neparvovec, now Luxturna®, for RPE65-associated Leber Congenital Amaurosis.



This trial, which resulted in the first FDA approved ocular gene therapy, is proof of concept that “untreatable” causes of childhood blindness are treatable and forms the basis for her work in studying mechanisms and developing treatments in animal models and participating in human clinical trials.

Her clinical practice is now focused on paediatric genetic eye disorders, especially inherited retinal degenerations. Dr Arlene Drack presented her work on gene therapy for Bardet-Biedl syndrome (BBS), focusing on the BBS10 gene. She was excited to join the conference and explained that her research aims to find treatments for BBS and related eye conditions.

Leber Congenital Amaurosis (LCA) and Gene Therapy

Dr Drack started by talking about Leber Congenital Amaurosis (LCA), a condition that affects the eyes from birth. It causes poor vision and involuntary eye movements. She explained how LCA can be diagnosed early using a test called an electroretinogram (ERG). Dr Drack shared a success story of a girl with RPE65-Associated-LCA who, after receiving gene therapy called Luxturna, was able to walk through an obstacle course without help. This treatment was approved in 2017 and was a big step forward for gene therapy.

BBS and Retinal Degeneration

Dr Drack then discussed Bardet-Biedl syndrome (BBS), which also causes vision problems but starts later in life. She showed how ERG tests can diagnose BBS before significant vision loss occurs. This helps in planning treatments early.

Gene Therapy in BBS10 Mouse Models

Dr Drack worked with Val Sheffield, MD, PhD to develop mouse models to study BBS10. These mice showed similar eye problems to humans with BBS10. Dr Drack's team then injected gene therapy into the mice's eyes, which led to significant improvements in their vision. The team used an OCT scan to see the layers of the retina and found that treated mice had better retinal structure as well as function measured by ERG, compared to untreated mice. They also used a swim test to measure the mice's vision, showing that treated mice could find a platform in water much faster than untreated ones.

Challenges and Future Directions

The COVID-19 pandemic posed challenges, but the team received special permissions and funding to continue their work. Dr Drack emphasised the importance of understanding the best age for treating BBS patients with gene therapy. They are currently studying nine BBS10 patients in order to understand the natural course of the retinal disease so endpoints can be chosen for human clinical trials. Dr Drack explained that ongoing research aims to compare the retina thickness between humans and mice, helping to refine treatment approaches. This study will provide insights into how BBS progresses and how gene therapy can change its course.

Moving Towards Human Trials

Dr Drack outlined the steps needed to move from mouse models to human trials. The team is developing new gene therapy vectors and conducting safety studies to ensure the treatments are safe for humans. Once these studies are complete, they plan to start small pilot studies to assess safety in humans. After successful pilot studies, they aim to conduct larger clinical trials. The goal is to have a treatment similar to Luxturna that can be widely available to BBS patients. The team is working with regulatory bodies and collaborators to make this happen.

Dr Drack concluded by thanking the patients and families who have supported their research. She acknowledged the daily challenges faced by those with BBS and reiterated her commitment to finding effective treatments. She thanked the audience for their attention and support, highlighting the collaborative effort needed to advance this important research.

Q&A - Dr Elizabeth Forsythe, Dr Arlene Drack, Dr Shehla Mohammed and Prof Phil Beales

How is Setmelanotide administered in children?

Dr Forsythe: It is a subcutaneous injection, a daily injection under the skin. These drugs are still in development, moving from daily injections to possibly weekly injections and maybe even tablets in the future.

Is there an advantage of Setmelanotide compared to Wegovy?

Dr Forsythe: It's a good question. We don't know because we have never done a trial comparing the two directly. Setmelanotide works on the pathway that isn't functioning well in BBS. Wegovy works on a different pathway, and we have limited data suggesting it also works for people with BBS. To know the difference, we would need a big study comparing the two. The aim is to have options for everyone because different people will respond differently to different drugs.

How can people get prescriptions for Wegovy?

Dr Forsythe: It depends on where you live. We recommend you talk to your BBS

clinician, ideally the endocrinologist. There is a shortage, but it will improve over time as production meets demand.

As the retinal treatment appears to work with BBS10, will you now be trying the treatment with other BBS types?

Dr Drack: As with most things in science, we can only work on what we can get funding for, and so I have funding for BBS10. Prof Beales has funding for BBS1. We hope that if we show promise with these types of BBS, it will spur funding to work on the other types. These BBS genes all make proteins that work together, so it makes sense that if we can treat some of them, we should be able to treat all of them.

Prof Beales: I absolutely agree. It is also a bandwidth issue; it is a full-time job to get these first gene therapies out. We have to do them sequentially. The amount of work required before even considering going into the clinic is considerable.

Is there a difference between the UK and US gene therapy studies?

Dr Drack: It might be a difference in the type of BBS. BBS10 might be able to turn some of the cells back on, and BBS1 cannot. It is subtle and different between each gene.

Prof Beales: Yes, we have not seen that type of improvement, so it might be a difference in the way the product or vector was developed or a gene difference. It is probably the gene difference.

What are the timescales for gene therapies for BBS1 and BBS10?

Prof Beales: We hope it's not 10 years, but we don't know. There is a lot to get through before starting clinical trials, including safety confirmation and regulatory approval.

Dr Drack: I echo what Phil said. We work daily to move it forward, but it's like moving a boulder up a hill.

Are there any promising prosthetics for RP and other retinal dystrophies?

Dr Drack: There is a lot of work being done. Various visual prostheses can make the nerve fibre layer in the retina sensitive to light. There are different kinds of retinal implants, but nothing is routinely available yet.

Are there any gene therapies for other symptoms of BBS, such as urology?

Prof Beales: Not yet. There are efforts being made to address kidney problems, but challenges remain in getting the therapy to stay in the right cells for long enough.

Dr Drack: Agreed. Treating the whole body is tough. Even with systemic injections, targeting specific organs like the eye is challenging. Better vectors might help in the future.

Are there any countries where there are no people with BBS?

Prof Beales: I'm sure there are, but BBS is found worldwide. Some islands might not have any cases, but in places like the Barrow Islands, it is very common.

Dr Mohammed: Many patients go undiagnosed for years. Some only have the eye phenotype of BBS and nothing else, making diagnosis difficult.

Conference feedback

Your feedback is vital to the success of our conferences and is something we come back to year on year in our planning and development. Thank you to everyone who completed an evaluation form. Here's a summary of your feedback:

As a result of attending our online conference:

- **97%** feel better informed about ongoing research into BBS
- **82%** feel better informed about the management of BBS
- **76%** report having greater awareness of the support and information provided by BBS UK
- **64%** feel more confident managing the challenges of BBS
- **76%** feel more connected to the BBS community
- **97%** would attend or recommend attendance at a BBS UK event in future

Comments received:

"...attending online means more people are able to join and watch from all over the world and ask questions... it was a great setup as you're able to ask questions in real time."

"I was really doubtful that I would enjoy this conference as much, because I like the community of our in-person conferences... however, I have to say that the quality of the information presented was fantastic. What I also liked was that my daughter with BBS was able to sit in on some of the presentations which normally she would not have been able to, as she would have been out on the activity day. Thank you!"

"Well done everyone that helped to organise or took part in presenting, what a fabulous job! Really interesting, informative, and professional. I especially look forward to hearing the personal perspectives, which are always inspiring and so heart-warming. Thank you."

Fundraising: we need your help!

We're calling on you, our amazing BBS UK community, to help us raise funds to keep our services going. By donating money or organising a fundraising event, you can contribute towards our:

- **Advice Service** – a support and advocacy service that helps people with BBS to access the local support they need across health, education, and social care, including negotiating support packages, helping with housing issues and benefits applications, and with future planning.
- **Annual family conference weekend** - brings members of the community together with interested professionals and experts to provide updates on the latest research and the opportunity to participate in tailored workshops.
- **Newsletters and conference report** - provides up-to-date research, information, guidance, personal perspectives, and details of our projects and fundraising activities.
- **Publications** - including our Medical Booklet, Introducing BBS and Booklet for Schools and Colleges leaflets which are available in various formats for accessibility.
- **Attendance at external events** – educating professionals and members of the public about BBS and BBS UK at events such as Sight Village and Retina UK.



Got what it takes to be a top fundraiser? Join our Countdown to Conference competition!

During the countdown to our annual conference in April 2025, we're challenging our fundraisers to a competition to raise the most money for BBS UK.

Every fundraiser who pledges to raise at least £100 will receive a BBS UK T-Shirt and fundraising pack, and the three top fundraisers will receive a limited-edition BBS UK hoody!

Organise a fundraising event

We're asking for fundraisers to choose one of the following ways to raise money:

1. Host a meetup

It can be as simple as inviting family and friends around for a cup of tea, cakes and a chat, or more involved, for example by organising a party.

The Sloane family host multiple Afternoon Teas each year. They make their own food and drinks and charge a ticket price for attendees. You could do this at your home, or get a local business involved and ask for free use of their space. You can even ask your local supermarket if they offer any schemes for discounted baking goods when raising money for charities.

2. Get outdoors and walk, cycle, run, or ride for BBS UK!

Many of you will remember the huge success of the We Carry the Gene campaign, where the community were asked to get out walking, running, riding and cycling while being sponsored to reach a set distance.



You can do this too by organising a sponsored walk or other activity, getting family and friends and even your dogs involved, and spreading awareness of BBS along the way.

Tanja, her daughters and friends walked 100km in June along with their dogs and horses, raising money and talking to people about BBS.

Get your organisation involved

If you're employed or have a close relationship with a particular business, you can ask for their support in fundraising.

Team building fundraisers!

Your company can engage in exciting team-building fundraisers for BBS UK. Imagine challenge events like donating 1p for every step walked during an office day or training together for a group bike ride. Get creative with events such as 'Fancy Dress Friday', where employees contribute to BBS UK to join in the fun!

Checking out your company's fundraising resources!

Discover the potential within your company by checking if there is a company foundation or a match giving programme. These resources can significantly amplify your impact!

Sponsoring our events or projects!

Make a lasting impact by sponsoring projects like our annual family conference or Clinics Support Service. Your company's support will ensure we can continue to be there for every family affected by Bardet-Biedl syndrome.

Choosing us as your charity of the year!

We would be thrilled and honoured to be selected as your company's 'Charity of the Year'. Together, we can raise awareness and provide crucial support for those impacted by Bardet-Biedl syndrome.

Donate monthly

By making a regular monthly donation or raising funds, you will be helping BBS UK to plan ahead and make long-term developments to our much needed and highly regarded support, advocacy and information services. A regular gift allows us to plan strategically as we know day in, day out that we will have a steady income that we can rely on.

To become a Friend and regular giver:

1. Visit our website at <http://www.bbsuk.org.uk/friends-of-bbs-uk>
2. Complete the 'Become a Friend of BBS UK' online form
3. Setup a monthly standing order with your bank. Make sure to use your name as a reference so we know who to thank!

Thank you to our Friends for their generous support!

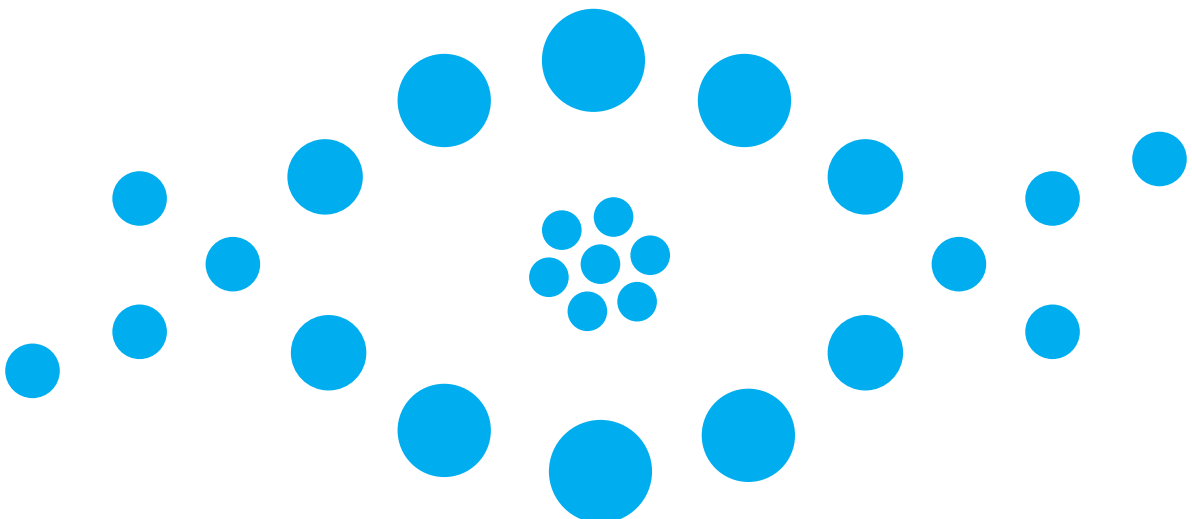
Spread the word on social media

Most people haven't heard of Bardet-Biedl syndrome, and so by sharing our social media content, you can help to inform people about BBS, and the amazing work we do here at BBS UK.

Let people know they can donate to us directly through our website: <https://bbsuk.org.uk/donate-to-bbs-uk/>

We're here to help with your fundraising efforts! Team BBS UK can help you set up your JustGiving page and will provide support along the way.

Contact us at fundraising@bbsuk.org.uk



Community Audit: Your Voice Matters!

Every three years, our charity conducts a Community Audit. Through this survey, you help us to learn about your experiences with Bardet-Biedl syndrome (BBS) so we can better support you.

By completing the Community Audit, you will:

- ✓ Help guide our future projects and services
- ✓ Show us how our work is making a difference in your life
- ✓ Provide valuable information about living with BBS

There are two versions of the survey:

1. For **parent carers**, available [here](#) and via this QR code:
2. For **people with BBS (adults or children)**, Available [here](#) and via this QR code:



Your feedback will make a big difference! The information you share will be used to shape the future of our charity and make sure we are meeting your needs.

Thank you for your time and support. If you need any help with the form, please contact admin@bbsuk.org.uk.

Thank You

Our heartfelt thanks go to the incredible speakers who generously gave their time to share their insights, expertise and research. We are especially grateful to those who shared their inspiring personal perspectives, which resonated so well with us all.

Our thanks also to our wonderful BBS UK team of staff and trustees for their commitment and efforts throughout the year that enable us to put on such high-quality events.

Most importantly, we thank all of you, who continue to support our work, and give so much back to the BBS community. We hope that you have all learned something that will help or inspire you in your day to day lives as someone living with the syndrome or supporting someone with BBS, personally or professionally; it is uplifting to know that there is such a large community eager to learn, share experiences, support each other and work together to make life better for those with Bardet-Biedl syndrome.

Our annual conference and all the other services we provide really are invaluable and rely on your donations and fundraising – if you would like information on how to donate or support the work that we do, visit the BBS UK website.

SAVE THE DATE!!

BBS UK Conference 2025 will be held, in-person, across the weekend of
25th-27th April 2025 at the Hilton Hotel, Northampton.

We will once again be live-streaming the event on Saturday 26th April, so if you cannot attend in-person, you can still join us by watching and listening through the livestream. Further announcements to come!

Contact Details

General Information

admin@bbsuk.org.uk

General Contact/Enquiry:

Natalie Braunton

07784 922654

natalie.braunton@bbsuk.org.uk

Mon and Tues: 9:00am – 5:00pm

Thurs: 9:30am – 3:30pm

Fri: 9:30am – 2:30pm

Patient Liaison Officer:

Birmingham BBS Clinic Service

Amy Clapp

07591 206787

amy.clapp@bbsuk.org.uk

Mon - Thurs: 9:00am - 3:30pm

Patient Liaison Officer:

Birmingham BBS Clinic Service

Laura Davis

07512 198484

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Fundraising

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Operations Manager

Tonia Hymers

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Mon – Thurs: 8:30am – 4:30pm

Advice Service

Rebecca Perfect

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Mon - Thurs: 10:00am - 2:45pm

Patient Liaison Officer:

London BBS Clinic Service

Angela Scudder

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Mon - Fri: 9:00am - 3:00pm

BBS UK Social Worker / Patient Liaison Officer

Shirin Memi

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Tues - Thurs: 9:30am - 2:30pm

www.bbsuk.org.uk