bbsuk

Bardet-Biedl Syndrome UK

Conference 2024

Saturday 8th June

10:00am-3:30pm

Information Pack

Welcome

On behalf of the Bardet-Biedl Syndrome UK team, it is our pleasure to welcome you to the 31st Annual Conference taking place via webinar at 10am GMT, on 8th June 2024. We are thrilled to have you join us for a day of insightful discussions, knowledge sharing, and community building.

Bardet-Biedl Syndrome UK is dedicated to supporting individuals affected by Bardet-Biedl syndrome (BBS), a rare genetic disorder. Our mission is to provide guidance, foster research, and enhance the quality of life for those living with BBS. This annual conference is a cornerstone of our efforts.

This year's webinar promises a comprehensive program with diverse presentations from leading experts in the field. During the day, we will hear from renowned specialists about the latest research developments and clinical advancements in BBS; gain valuable insights from personal experiences; and learn valuable ways to manage BBS from professionals.

Inside this booklet, you will find a programme with presentation timings, technical information about how to join us on the day, and profiles of speakers.

Your feedback is invaluable to us. A link to a survey to share your thoughts on the event can be found at the end of this booklet and will be sent out via email after the conference. We also encourage you to stay connected with BBS UK through our website and social media channels for ongoing updates and resources.

If you would like to show your appreciation with a donation, a link can also be found within this booklet, along with information about how to make a regular donation as a ‘Friend of BBS UK’. As a charity, we rely on contributions to continue our services. This year, our support services, including our annual conference, are needed more than ever. We need your help to continue our work and provide the support that our BBS community deserves.

To our speakers, we offer our heartfelt thanks for helping us to make this conference possible. Their expertise and commitment to the BBS community are instrumental in advancing our collective understanding and support of those affected by Bardet-Biedl syndrome.
We look forward to spending a day with you all, learning, sharing, and, most importantly, connecting!

Warm regards, The BBS UK Team

Attendee information

General

The BBS UK Annual Conference 2024 is being hosted by the BBS UK team via Zoom as a webinar. All those who have registered to attend the event will have received a link via email to join. Information about the day and the form to register can be accessed via the BBS UK website.

Attendees do not have their cameras activated and cannot be seen or heard by anyone however they can choose to contribute using the chat function and Q&A facilities.

Conference programme timings are for guidance only and may be subject to change.

Questions

Questions can be submitted in advance via email: admin@bbsuk.org.uk, text or telephone: 07784 922654. We will also take questions during the conference via Zoom chat and the Q&A facility. If there are any questions that we are unable to answer on the day, we will endeavour to provide a response to you as soon as we can after the conference.

Technical Information

We will use Zoom, the same platform we used to livestream our conference in September 2023.

When you registered, you will have received an email from Zoom with a link for joining the conference on the day. If you have not received this or can't find the email, please contact us by no later than 12pm on Friday 7th June so that staff have time to resend the email to you. It is important that you save this email.

You can either download the Zoom app on your device, or you can access the conference through your web browser (Google Chrome, Safari, Internet Explorer etc.)

If you're new to using Zoom, we recommend the **'Joining a Zoom meeting'** video on YouTube or the **'Joining and participating in Zoom Webinars'** written guide from Zoom.

Please test your internet connection and speakers before the conference. Our staff will be running the conference and might not be able to help on the day.

If you have any technical queries with regards to accessing the conference, please contact Natalie Braunton before 12pm on Friday 7th June through email: admin@bbsuk.org.uk, text or telephone: 07784 922654.

Social media

We welcome you to post about the conference on social media! Please use the hashtag #BBSUKConference2024 on any of the platforms listed below:

- **Facebook:** www.facebook.com/bardetbiedlsyndromeuk
- **Instagram:** bardetbiedlsyndromeuk
- **X:** @BBS__UK
- **LinkedIn:** www.linkedin.com/company/bardet-biedl-syndrome-uk

Feedback form

The BBS UK Team work hard to improve the conference based on your feedback. We will send you the link to the online feedback form after the conference via email or you can follow this link. Thank you for taking the time to complete the feedback form.

Programme

10:00am	**Welcome:** Laura Dowswell, BBS UK Chair
10:05am	**Introduction to BBS** Dr Shehla Mohammed
10:25am	**Update on Research and Study of BBS** Professor Phil Beales
10:55am	**Respiratory Ciliopathy and BBS** Professor Claire Hogg
11:20am	**Q&A**
11:35am	**Comfort Break** (10 mins)
11:45am	**A Sibling's Perspective** Greg Dowswell **Supporting Siblings** Clare Kassa, CEO, SIBS (Pre-recorded) **Q&A**
12:30pm	**Artificial Intelligence for Accessibility** Hector Minto, Microsoft **Q&A** with Hector Minto & David Prince
1:00pm	**Lunch Break** (30 mins)
1:30pm	**Fundraising Round-Up** Samantha Serido, BBS UK
1:45pm	**Irritable Bowel Syndrome (IBS) and BBS** Kelly Slater, Dietitian
2:05pm	**A Journey of Resilience and Triumph** Bilaal Ali
2:20pm	**Comfort Break** (10 mins)
2:30pm	**Update on Setmelanotide** Dr Elizabeth Forsythe
2:45pm	**BBS Gene Therapy & Clinical Trials** Dr Arlene Drack
3:10pm	**Q&A** Dr Arlene Drack, Dr Elizabeth Forsythe & Panellists
3:25pm	**Closing Remarks** Laura Dowswell, BBS UK Chair
3:30pm	**Close of Conference 2024**

Speakers

Laura Dowswell, Chair of BBS UK

Laura has been married for 32 years to Nick and together they have two adult children, Maria and Greg. They live by the sea at The Witterings on the south coast and enjoy the beach life all year round.

The family became involved with the charity after Maria was diagnosed with BBS at age 14 and they attended their first conference 13 years ago. Laura says they were amazed at the amount of support and information they received and the lifelong friends they made. As a family they were determined to ‘give something back’.

They've supported several fundraising events and ventures for BBS UK with both Laura and Nick being support crew for the Cilia Cycling trip last September. Laura has been a trustee of the Charity for several years and was Treasurer for four years, seeing many changes over this time. She would like to continue being the Chair, supporting BBS UK in its growth and ensuring good governance.

Doctor Shehla Mohammed, Guys and St. Thomas’ NHS Trust

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 Guy’s.

Dr Mohammed has a longstanding interest and experience predominantly in rare genetic disorders and in the care of children and families with life-limiting disorders. She has been involved in setting up and running the National BBS clinic for adults with Prof 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’ in 2018. Dr Mohammed is also involved in the running of other highly specialist services at the RDC.

Until 2017, Dr Mohammed had been 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.

Professor Phil Beales, Great Ormond Street Institute of Child Health Guys and St. Thomas' NHS Trust and Co-founder & CEO at Axovia Therapeutics.

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.

In 1999, he published the largest survey of BBS symptoms with the help of members of BBS UK (then LMBBS). This culminated in the identification of many new features not previously described in the medical literature and enabled Professor Beales to propose new diagnostic criteria, with the expressed purpose of enabling an earlier diagnosis for many more children. 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 to prevent further visual deterioration or weight gain.

Professor Beales has been medical advisor to BBS UK since 1996, was made President of the charity in 2005 and is a founder member of the recently formed BBS UK Scientific Advisory
Board. In 2010, he established, with the support of BBS UK, National Multi-disciplinary Clinics, a comprehensive genetic testing platform for all persons with BBS in England and Scotland.

Professor Claire Hogg, Royal Brompton

Prof 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. Prof Hogg 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.

Her expertise extends to leading clinical trials: as the UK lead for the first clinical trial in PCD as part of an EU programme grant [BESTcilia: FP-7], and as PI for the biomarker project for the first mRNA therapy trial in PCD [ReCode Therapeutics 2024]. She was lead for the BEAT-PCD Training School [COST Action, 2015-2019] and is Chair of the BEAT-PCD consortium “PCD bioresources”” work-package, aiming to develop an international PCD bioresource which can be used for testing novel PCD specific therapies. Prof Hogg has also supervised many higher degrees [MD(Res) and PhD’s], MSc and BSc students to successful completion of projects, writing of dissertations, publications, and presentations.

In her clinical practice, diagnosing and managing children with primary ciliary dyskinesia [PCD], she works closely with the other services looking after patients with complex primary ciliopathies, such as Bardet Biedl Syndrome [BBS]. In this session she will discuss the clinical presentation of respiratory symptoms in patients with BBS, the diagnostic pathway to screen patents with BBS for a respiratory ciliopathy and management strategies learnt from patients with PCD.

Greg Dowswell

Greg is a video producer currently based in Surrey. He is the only sibling of Maria (30) who was diagnosed with BBS at the age of 13. Greg will be talking about his experience of growing up with a sibling who has a disability.

Clare Kassa, Sibs

Clare Kassa joined Sibs as CEO in 2018. 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 both 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 an adult sibling – she has an older brother with a learning disability. Clare is passionate about sibling support and wants to ensure that the important role siblings play in their families is properly recognised.

Hector Minto, Microsoft

Hector Minto is the European Director for Accessibility & Evangelist at Microsoft. Hector has worked at the cutting edge of accessibility and assistive technology (AT) for 25 years. Hector's role at Microsoft as Lead Technology Evangelist sees him engaging with major global employers, Microsoft's commercial partner network and a wider set of community stakeholders to showcase inclusive design, product accessibility, inclusive hiring practices, the role of disability employee resource groups and accessibility innovation. A critical part of Hector's role is to learn from Microsoft customers how we can continue to adapt to the changing needs of a diverse population using technology, and to provide this feedback directly to the Microsoft product teams, thus driving greater inclusion.

Samantha Serido, BBS UK

Samantha joined BBS UK in November 2023 as our Fundraising and Communications Officer. She has devoted the lion's share of her professional life to date, to non-profit and health systems management, fundraising, and content development.

Before moving to Manchester, she earned a Bachelor of Arts in Writing in the United States, where she was raised. Samantha also holds a Master of Arts in Social Anthropology from the University of Manchester, with a specialisation in Wellbeing Anthropology. In her spare time, Samantha enjoys hiking, yoga, and spending quality time with friends and family.

Kelly Slater, Dietician, QEH BBS Clinic

Since qualifying as a Registered Dietitian in 2006, Kelly has worked in many specialist areas, including Gastroenterology, Diabetes, Renal, weight management and nutritional support. Having worked in a variety of settings including primary, secondary and community. Kelly has worked at the Bardet-Biedl Syndrome clinic at University Hospitals Birmingham since 2016. Other current Dietetic roles include Primary care and the Dietitian Ambassador for Coventry and Warwickshire Training Hub.

Bilaal Ali

Bilaal Ali is a young man with BBS who is registered blind. He's currently in the second 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.

Dr Elizabeth Forsythe, MRC Clinical Research Fellow (Clinical Genetics), University College London, Great Ormond Street Institute of Child Health

Dr Elizabeth Forsythe has degrees in molecular medical sciences and medicine from Queen Mary University of London. She trained in adult medicine and paediatrics before specialising in clinical genetics at Northwick Park Hospital and Great Ormond Street Hospital, where she is currently a registrar. Dr Forsythe has over ten 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 Children’s 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 Arlene Drack, University of Iowa

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 treatment trials.

Her clinical practice is now focused on paediatric genetic eye disorders, especially inherited retinal degenerations; Arlene will be presenting an overview of her research utilising Bardet-Biedl syndrome mouse models (BBS10) to study disease mechanisms and develop treatments.

Become a member of BBS UK

Please take a moment to ensure you have joined our BBS UK membership.

How will I know if I am already a member?
To become a member of BBS UK, you will have either completed an application form or consented to becoming a member when asked by one of our team. If you have been sent annual general meeting (AGM) voting information in the past, you are already a member. If you are not sure, contact us and we will check for you: admin@bbsuk.org.uk

Benefits of Membership:
BBS UK's members are at the heart of our work. Become a member and you will join a growing group of people who play a part in everything we do. For BBS UK, having dedicated members enables us to meet our aims and objectives more fully. Engaged and involved members ensures transparency, credibility, and expands our impact through increased volunteering and community engagement. The collective strength of our membership not only benefits individual members, but the BBS community as a whole.

Members also have:

- The opportunity to participate in decision-making and shape the direction of the Charity
- The chance to vote to elect BBS UK trustees and to become a trustee
- Access to BBS UK projects including the IT Equipment Fund and Chris Humphreys Memorial Fund

In addition to becoming a member, you can support BBS UK's work in all sorts of ways. There is no pressure to do anything, but if you are keen to take action, we will support you every step of the way.

Options include:

- Taking part in surveys, focus groups and government consultations
- Sharing personal experiences
- Fundraising to support BBS UK's work

Membership is entirely **free** to ensure that everyone who needs our support can access it. To become a member, visit our website www.bbsuk.org.uk and click 'Become a Member' at the top of the home page.

Donate: Friends of BBS UK

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:

- Visit www.bbsuk.org.uk/friends-of-bbs-uk
- Complete the 'Become a Friend of BBS UK' online form
- 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!

Our Publications

Available from BBS UK in multiple formats:

- Introducing Bardet-Biedl Syndrome
  (also available in Turkish, Urdu and Easy Read)
- Medical Information Booklet
- Who We Are and How We Can Help!
- Booklet for Schools and Colleges
- My Life, My Future – A Booklet On Transition
- SEND and EHCP Factsheet

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Bardet-Biedl Syndrome UK
Booklet for Schools and Colleges
supporting children and young people with Bardet-Biedl syndrome in a learning environment

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Bardet-Biedl Syndrome UK
Introducing
Bardet-Biedl Syndrome

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Bardet-Biedl Syndrome UK
My Life, My Future!
A young person's guide to living their life with Bardet-Biedl Syndrome

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Bardet-Biedl Syndrome UK
Medical Information
Booklet

Copies can be downloaded via:
www.bbsuk.org.uk

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