

The BBS UK Annual Conference 14th-15th May 2021

**Gareth
loves
to cook!**



**Clinics Service
and Fundraising**
Updates from
BBS UK

**Access to
Information**
Sarah wins a
legal challenge



BBS UK Recruit New Trustees

We welcome Liam Wilkie and
Dr Shehla Mohammed to the Board

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Newsletter

Spring 2021

Welcome to the BBS UK newsletter, Spring Edition.

Foreword



Welcome to the 2021 Spring Newsletter. It certainly feels like it has been a long winter but the arrival of spring brings with it a sense of optimism and the prospect of a return to some sort of normality after a very challenging few months for us all. Many of you by now will have received your first and possibly your second COVID-19

vaccination, which hopefully has eased anxiety for those who have been shielding.

Since our last edition, BBS UK has been busy as have our members, who have demonstrated amazing resilience and continued to support us despite the unprecedented disruption of the pandemic.

Fundraising efforts have been fantastic as always, and you have all been keeping active at home whether it be cooking, exercising in your living room, knitting, or participating in online social activities. The BBS UK team have had an eventful few months preparing for our first ever online conference, developing our resources and recruiting two new trustees to the Board. The face-to-face clinics service has resumed at St Thomas' Hospital and the distribution of home testing equipment has been launched.

I would personally like to thank everyone for pulling together through the last few testing months. I hope you enjoy reading this newsletter, there are plenty of uplifting articles demonstrating the strength of our members and reminding us of the good days that lie ahead!

Happy reading!

Stefan Crocker

Chair of the BBS UK Board of Trustees



bbsuk

Bardet-Biedl Syndrome UK

CONFERENCE 2021



bbsuk Annual Conference on
ZOOM!
Bardet-Biedl Syndrome UK
14th – 15th May 2021

We are very much looking forward to welcoming you all to our Annual Conference across Friday 14th and Saturday 15th May held on the Zoom platform as a Webinar. Although we are unable to meet in person, we are not going to let that stop us from coming together as a community to catch up with all things BBS!

This year we are holding a 'Professionals Conference' on Friday 14th May aimed at professionals working with those who have BBS, to improve their knowledge and understanding of the Syndrome. Those with BBS, their families and carers are also welcome.

On Saturday 15th May is our Annual Conference, open to everyone to join but specifically will benefit individuals with

BBS and their families and carers. On both days, the conference will be held on Zoom as a Webinar which means you will be able to follow the presentations however you won't be visible to other attendees. You will have the option to ask questions using a 'chat' function (which enables you to type the question) or alternatively you can submit questions in advance via the following email: admin@bbsuk.org.uk.

Across the two days we will hear personal perspectives from our members as well as talks from the following professionals:

- Professor Phil Beales: Update on Research and Study of BBS
- Dr Bob Haws: Centre of Excellence for BBS, USA

- Dr Jane Waite: Person Characteristics and Wellbeing in BBS: Interim Results
- Sarah Flack; Principal Dietitian, GOSH: Supporting weight management in BBS
- Professor Tim Barrett: BBS Clinics Home Testing Equipment
- Dr Elizabeth Forsythe: Update on the Setmalenotide Study
- Dr Emma McGibbon & Anne-Marie Walker: Anxiety & low mood: recovering from social isolation
- Dr Helen May-Simera: Update on Urine Study
- Tonia Hymers & Liz Loughery: Membership Audit and 5 Year Plan

How to Register

To register, go to: www.bbsuk.org.uk

Upon registration, you will be sent a link that will give you access to the events. On the day of the event, you will simply need to click on the link from your chosen device and enjoy the presentations from the comfort of your armchair. We look forward to joining you there!

If you have any questions about the conference or would like to submit a question in advance please get in touch via admin@bbsuk.org.uk

The webinar is free and the first 100 registrants will be eligible to receive a free delegate's pack containing key information resources and merchandise.

BBS Clinics Update



In my last report, I expressed our gratitude for the NHS and those who work within it, including our BBS clinics teams. We were coming out of a second

lockdown and heading into the Christmas season. Although we were aware that a second wave was possible, many of us perhaps were not expecting it to be as severe as it was; our heartfelt thanks go to all those who provided care and support during such difficult circumstances. Our clinics service was only just starting to resume face-to-face appointments and once again we were fortunate to have the telemedicine service to ensure those who needed an appointment were still able to be seen and in a safe manner.

Recognising the limitations of a telemedicine service in terms of monitoring, as previously reported, NHS England have generously provided funding for equipment for home testing, including a blood pressure monitor and bathroom scales for all, and a stadiometer (a device for measuring height) for the paediatric service. Finger prick blood testing kits will also be used. Unfortunately, the pandemic has hindered progress with the distribution of equipment however we are hopeful that we will be up and running very soon.

A significant development since our last newsletter has been the roll out of the Covid-19 vaccination programme and with it the end of the second shielding period, which may feel exciting for some and prompt anxiety in others. Everyone should continue following the government guidelines for staying safe and move

forwards in a way that feels comfortable for them. Dr Emma McGibbon and Anne-Marie Walker, Clinical Psychologists from the London and Birmingham BBS clinics service, will be discussing anxiety and coping strategies at the BBS Annual Conference on the 15th of May.

Our BBS clinics service at St Thomas' Hospital, London has now resumed face-to-face appointments and the other centres will follow over the coming weeks and months. The pandemic has had a significant impact on appointment follow-up times and we are all looking at how we can provide the best care and support, utilising a mix of face-to-face and telemedicine appointments.

If you have any urgent health concerns, these should always be taken straight to your GP. Your appointment may take place over the phone, but if you feel you need a face-to-face appointment you should be able to request one. You should also consider whether you have any appointments outstanding, for example, do you usually have an eye appointment or renal appointment six monthly or annually? If you think your appointment is overdue, contact the relevant service. Your GP surgery may be able to give you the telephone number you need.

BBS clinic appointments are currently running with 20-24 month follow-ups and patients will be contacted when their appointment is due; we try to give at least a month's notice. If, in the meantime, you have a question for the BBS clinics team, or feel you need to be seen in clinic, contact your Patient Liaison Officer, Angela or Amy who will be happy to help you.

Tonia Hymers

Service Manager: BBS UK Clinics Ltd

I Love to Cook

by Gareth Mainwaring

Hi, my name is Gareth Mainwaring and I love to cook. I like cooking because it's fun and you get to eat your creation after you've finished. If it wasn't for my love of Lego, cooking would probably be my favourite thing.



I have always loved to cook but since lockdown I have been cooking even more. My design and technology teacher sets us a cooking task every fortnight too, so I have been really busy in the kitchen over the last year.

Even when I am not cooking for my design and technology lessons, I still find an excuse to cook. I love to bake homemade croissants for breakfast. It's quite a long process but it is worth it in the end. It's my birthday before long, so I'm planning on baking a baby Yoda themed angel cake. I seriously can't wait!

As part of my BBS, I have a sight impairment. I have Rod Cone Dystrophy and I am totally night-blind. I used to struggle quite a bit with measuring ingredients, so I needed my Mum or Dad's help, but then a very kind family fundraised and donated money to the local County Council to buy some equipment for visually impaired people. This equipment meant I was able to borrow some talking scales and a liquid level indicator, so now I am a lot more independent.

Review of the Talking Scales and Liquid Level Indicator

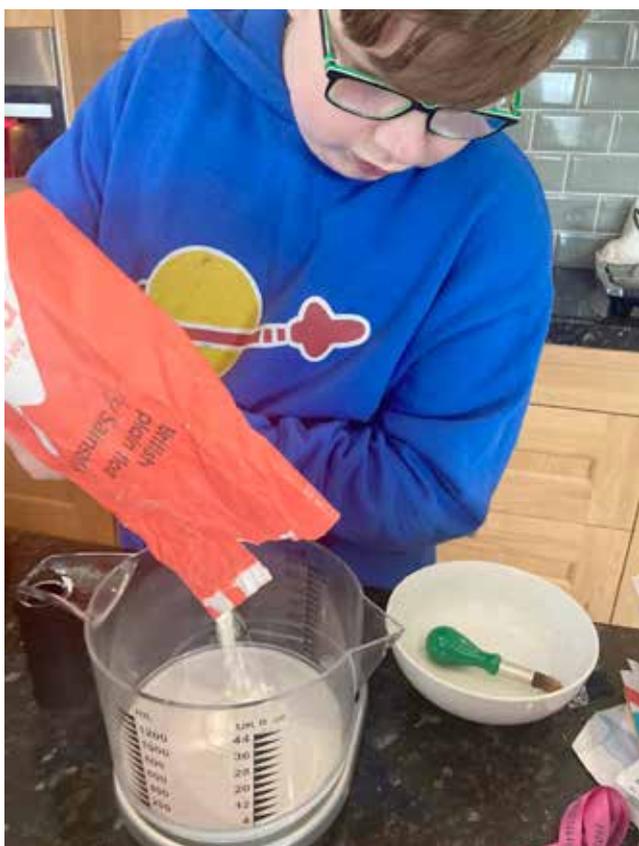
I actually borrowed two different types of talking scales. One measures weight in different units and the other is a jug scale specifically for liquids. On the liquid measuring scales you can choose milk, water or oil, which is what most recipes ask for in terms of liquid.

Both scales are very good and it means that I don't have to struggle to try and read the scales anymore. I also don't have to have anybody to help me. I prefer the main scales as I find the voice on the jug scales difficult to understand. If I was going to buy some scales (which I intend doing) I recommend testing them first to make sure the voice is easy to understand.

I also really like the liquid level indicator - it is a small device that you put on the side of your cup, mug or jug. It has little probes that go down into the cup, mug or jug and it has two levels of alerting you. It tells you by beeping and vibrating when the cup is nearly full and then again when it is full. This is handy if you want to make tea, for example, and you want to leave some space for the milk.

I highly recommend this little device as I can now pour myself drinks and make myself hot chocolate without worrying about spilling anything.

I love these devices; they make cooking so much fun and I am so much more confident doing things on my own now.



My Future

This year I will be choosing which GCSE's to take at school. I am hoping that I will be able to take food technology as a GCSE. I have someone at school to help me, her name is Mrs Daniel and she is wonderful. She will be there to help me when I need her but with my new scales, I will be able to do a lot more for myself, so she can chill out and watch me cook!

I'm not sure if I will be cooking as a career in future. It's definitely a possibility, although I think cooking will probably be my hobby... unless I can get a job in the kitchen in Legoland!

Fundraising Update

Thank you to you all for your continued fundraising efforts, despite the challenging circumstances of the pandemic. Unfortunately, many of the great events and fundraising ideas have been curtailed because of the Coronavirus restrictions which have meant donations so far this year have been slower, however we are grateful to see that this has not dampened your enthusiasm!

As we now cautiously move towards a more opportune period where 'virtual' events will be able to become 'reality' we would like to encourage you to make your plans for sponsored walks, runs, 'back to the office' celebrations, meeting with lifestyle groups again and look at them all as opportunities to have fun, socialise and raise funds for your favourite charity, BBS UK. We currently have two places left for the Virtual London Marathon, in October – why don't you face the once in a lifetime challenge and contact us now?

Christmas Fundraising

Christmas came and went, brightened up by our Christmas Cards, one of the many fantastic ideas from our members. There are 1,000 of them out there and not only did they look good and cheered-up the recipients, but they also helped raise £800 for the Charity! Well done all of those who bought some, we are very grateful. We will be repeating the competition and production of cards this year.



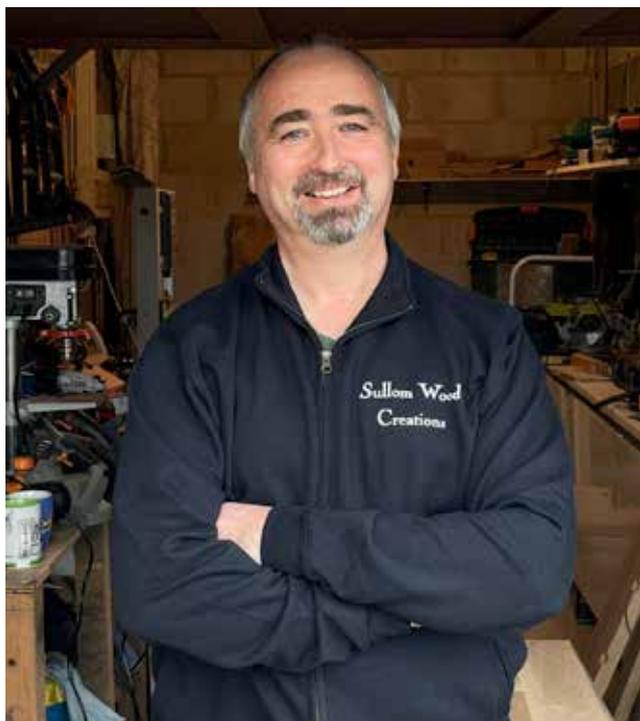
Christmas decorations by Emma Eesha

You may also want to have a think about other Christmas, festival, or community fundraising activities. Member Emma Eesha, made wooden angels with the BBS UK logo for the Christmas tree in her local church and raised £20.00 despite services moving online. Thank you Emma.

Best Bloomin' Sunflower Competition 2021

Our new Sunflower initiative reflects the optimism we are showing as we face the challenges of 2021 and as usual the response has been magnificent. Information about the initiative and how to take part can be found elsewhere in this newsletter and on our website, www.bbsuk.org.uk/fundraise. Flowers will be blooming everywhere!

Corporate donations



A long-time supporter of BBS UK, Nick Dowswell (Sullom Wood Creations) recently completed a commission and his delighted customer gave a donation to the charity of their choice, which was of course, BBS UK. Nick Dowswell set up his business in November last year and named it Sullom Wood Creations, after the area in the Shetland Islands where he grew up. Working with his father, Nick developed a love of wood and making things by hand which is reflected in his beautiful and bespoke product range made from reclaimed timbers.

We rarely receive corporate donations yet many businesses have a social enterprise programme, especially the bigger organisations. Do you know of any companies who could support us? Perhaps you work for a company with a 'matched giving' scheme? Christine Saxon, Fundraising Officer would be happy to speak with them!

We have highlighted several stories of individuals who have thought of different ways to support BBS UK, including decorating Christmas trees in a church, corporate donations from a new company and knitting and selling soft toys. There are many more of you who have been raising funds and awareness in fun and unique ways and we would love to tell everyone; please let us know what you have been doing and we shall pop it on the website or in the next newsletter to inspire others.

And finally.....

Last year, through good governance, caution and your support, BBS UK not only managed to survive the risks of much reduced finances and keep our services going, but were also able to recruit to a new advice worker post. After five years continued growth, income is however greatly reduced and we need your support more than ever. We rely on fundraised income and regular giving is crucial for meeting core costs. If you cannot do an event why not consider becoming a Friend of BBS UK instead? The services of BBS UK are needed now more than ever before, so please support us however you can. Thank you.

UK Rare Diseases Framework: how BBS UK are showing their support

On the 9th January 2021, the Department of Health and Social Care published the UK Rare Diseases Framework, which replaces the previous UK Strategy for Rare Diseases published in 2013.

The Framework represents a commitment from the governments of England, Scotland, Wales and Northern Ireland to deliver a shared aim to improve the lives of people living with rare conditions, so the diagnostic odyssey no longer exists, patients and clinicians know far more about rare conditions, and care is of the highest standard possible.

But what does this policy mean for those with Bardet-Biedl syndrome?

On 21st January 2021, we attended the Westminster Health Forum Policy Conference and listened to talks from Genetic Alliance UK and other patient organisations. We wanted to understand a bit more about this new framework and how BBS can help to deliver against the key aims and criteria that have been set out to improve diagnosis and the access to and co-ordination of patient care.

The framework was informed by the 'National Conversation on Rare Diseases' survey, which was launched by the

Minister for Rare Diseases in October 2019. Many of the BBS community took part in this survey and your responses will have enabled the government to better understand the greatest challenges faced by those living with, caring for, or developing treatments for rare diseases.

To address these challenges the Framework sets out four high-level priorities for rare diseases in the UK over the next five years.

Priority 1 - Helping patients get a final diagnosis faster

People with a rare disease like BBS often present with unusual patterns of common symptoms that can be hard for a GP to recognise; there can often be trips to multiple specialists before a final diagnosis is reached. This can take years and can have a big impact on patients, their families and the NHS. The Framework hopes to improve access to specialist care to speed up diagnosis and avoid potential misdiagnosis.

Priority 2 - Increasing awareness of rare diseases among healthcare professionals

BBS is a rare condition with only approximately 560 people in the UK currently diagnosed. BBS are therefore dedicated to raising awareness of the Syndrome to support quicker diagnosis and better patient care. We have developed a comprehensive range of information resources and over the last

12 months have completely rebranded and re-structured our website. Some of the booklets we have produced which are available to support the training and education of healthcare professionals include:

- Medical Information Booklet
- Introducing Bardet-Biedl Syndrome
- Booklet for Schools and Colleges

Priority 3 - Better coordination of care

BBS UK are committed to achieving the best possible care for patients and are passionate about improving the lives of all those affected. We acknowledge that living with BBS and managing appointments and health services can be a complex and time-consuming job. Many of you are unable to work or learn because of this impact. We endeavour to do all we can to minimise this burden and have this year recruited an Advice Worker to provide advice and information across the areas of education, health and social care.

As a team, we also offer support in respect of the fluctuating COVID-19 pandemic, ensuring members have access to accurate and up to date information in an accessible format and providing support with accessing local services.

The BBS UK clinics support service is also constantly striving to improve the patient experience and works closely with patients and the NHS in this respect.

Genetic Alliance UK have done excellent work in this area through their Coordinated Care Of Rare Diseases (CONCORD) study which can be accessed via their website:

www.geneticalliance.org.uk/our-work/healthcare-and-delivery/coordinated-care-of-rare-diseases-concord/.

Priority 4 - Improving access to specialist care, treatments and drugs

The 'National Conversation on Rare Diseases' survey has identified a number of issues in relation to access to specialist care, treatment and drugs, which have also been included in the Framework as underlying themes. These are:

- Continue to champion the patient voice
- Pioneer research, so that the potential of cutting-edge science can be harnessed and translated into frontline clinical care outcomes.
- Use digital tools, data and technology to improve efficiency, patient experience and research
- Maximise collaboration with the rare disease community in the UK and across the world to drive better outcomes for patients
- Ensure alignment with wider policy so that rare disease issues are recognised across government

Those affected by BBS in the UK are fortunate to have access to a specialised clinics service of which BBS UK has a unique role as an NHS funded third-sector partner. At each clinic, patients are seen by a broad range of clinicians including an ophthalmologist, nephrologist, dietician, clinical psychologist, geneticist, speech and language therapist and endocrinologist; the aim being to provide patients with specialised and expert attention with a focus on diagnosis, early intervention and good health management.

BBS UK's role in the clinics service includes providing information and support to patients and their families

and carers before, during and after their clinic appointment. Our focus is on ensuring individuals are adequately informed about the Syndrome and feel well supported. BBS UK representatives attend each clinic, offering emotional and practical support and ensure that patients and their families and carers are made aware of relevant benefits and social support services. Alongside this we are constantly striving for improvement and understand that listening to patients and taking onboard feedback is a key part of this process. BBS UK regularly conducts patient questionnaires and holds focus groups. Data gathered is evaluated and used to promote and drive positive change within the organisation and the service we provide.

How you can help - call for contributions!

Following the release of the UK Rare Diseases Framework will be the government's publication of the action plans to deliver against each of the commitments. Now, more than ever before, BBS UK is committed to doing all we can to support with the delivery of the plans and help drive forward the government's work on rare disease.

QUESTIONNAIRE



BBS UK Membership Questionnaire

In order to achieve the aims set-out by the Framework we are continually looking at ways we can improve, primarily to ensure we remain focussed on the needs of our community. A vital part of this is ensuring that we listen to our members and use their feedback to direct our services effectively to reach the best possible outcome.

As part of our service review this year, we have created a questionnaire designed to assess how Bardet-Biedl syndrome affects those diagnosed and their quality of life. It also aims to assess how well those affected have been supported by the Charity where the gaps are and what they would like from BBS UK in the future. This questionnaire is available for all our members to complete and we really hope as many of you as possible will do so. We are so grateful for your continued support and cannot emphasise enough how valuable your suggestions and comments are for the development of the services we provide. The questionnaire can be completed online and accessed by the BBS UK website or via Survey Monkey: www.surveymonkey.co.uk/r/BBSUKSurvey.

Best Bloomin' Sunflower Competition

BBS UK are looking for the Best Bloomin' Sunflower of 2021 with a competition aimed at raising funds for the Charity.



To take part, all you need do is grow a sunflower from the BBS UK seed packs (that we will send you), and/or write an eight-line poem about sunflowers. Photos of your sunflowers and poems should be sent to us by 30th September 2021 to be entered into the competition of which we will choose two winners, one for the Best Bloomin' Sunflower of 2021 and one for the Best Bloomin' Sunflower Poem of 2021.

As part of the competition, we are also providing sunflower colouring-in pages and we would love to see your photos of these too! You can share via our Facebook group using the hashtag #bbsuksunflower or send to admin@bbsuk.org.uk.

We will provide regular updates of your plants' growth and will announce the winners with their sunflower photos (and the measurements) both on our website fundraising page and on Facebook.

Subject to demand, we will produce a set of notelets with the winning photo and poem and will make them available for purchase after October 2021.

Interested?

If you are interested in taking part, please email: christine.saxon@bbsuk.org.uk with your name and address and a pack will be sent to you.

Donations will be gratefully received. Suggested amount is £5.00 per entry. Donations can be made via Bank Transfer to the BBS UK bank account:

Sort code: 20-19-97
Account Number: 20368253
Reference: Sunflower

Or via the BBS UK Just Giving page: <https://www.justgiving.com/lmbbs>

Stocks are limited, so don't delay – and don't forget to share your pictures on our Facebook page and group using the hashtag #bbsuksunflower.

Five-A-Day: it's easier than you think!

Fruit and vegetables are packed with goodness. They contain essential vitamins, minerals, fibre and phytochemicals (naturally occurring plant substances) that are vital for good health. Research has shown that people who eat plenty of fruit and vegetables have a lower risk of developing many diseases, including high blood pressure, heart disease, stroke and some cancers.

Fruits and vegetables are naturally low in energy (calories) and their high water and high fibre content are beneficial in helping us maintain a healthier weight. Fibre bulks up food and also slows the rate at which foods are chewed and digested, which can help you feel fuller for longer.

How much fruit and vegetables should we eat?

Both the UK guidelines and the World Health Organization (WHO) recommend that all adults should eat at least five (80g) portions of different fruits and vegetables each day.

Children should also be encouraged to eat at least five portions a day. There is no recommended portion size for children. However, a useful guide to a portion is the amount that fits into your child's hand.

So, what is a portion?

Fruit

- One banana, orange, nectarine, apple or similar sized fruit
- Half a large grapefruit
- A slice of large fruit such as melon, papaya or pineapple
- Two satsumas, plums or similar sized fruit
- A handful of grapes, cherries or berries

- One heaped tablespoon of dried fruit e.g., raisins or sultanas
- Three heaped tablespoons of fruit salad or stewed fruit

Vegetables

- Three heaped tablespoons of vegetables (raw, cooked, frozen or tinned)
- Three heaped tablespoons of beans and pulses e.g., baked beans, kidney beans (only one portion of beans and pulses can count towards your five-a-day).

Remember that potatoes, yam, plantain and cassava do not count towards your five-a-day because they are starchy foods.

What about fruit/vegetable juices and smoothies?

A small glass (150ml) of unsweetened fruit juice, vegetable juice or smoothie can only count as one of your 'five-a-day'. Fruit juices and smoothies have a very similar calorie content to sugary, fizzy drinks like cola. Try drinking water instead and eating all of your 'five-a-day' instead.

Are frozen, canned and dried vegetables and fruit as healthy as fresh ones?

Although nothing beats the taste of fresh fruits and vegetables in season, frozen, canned or dry products that contain little or no added sugar, fat and salt are healthy alternatives. They are often harvested and packed at the height of the season when nutrients are at their peak, so they can provide us with nutritious options all year round.

So now for five-a-day!

It may sound daunting, so start by increasing by one extra portion. Once you've done this for a few weeks, add another portion, then increase again until you are regularly eating five or more each day.

Try dividing your five portions throughout the day. Why not try eating one or two portions with each of your meals and try making fruit or vegetables your first choice for a snack? Adding a portion at breakfast time can really help to get you off to a great start!

The following sample menu may give you some ideas:

Breakfast

- A bowl of cereal with a sliced banana.

Mid-morning snack

- Celery, cucumber, pepper or carrot sticks on their own or with two tablespoons of salsa or low-fat hummus, if you prefer.
- Need something quicker? Grab a piece of fruit instead!

Lunch

- Add a combination of sliced tomato, lettuce and cucumber to your usual sandwich.

Mid-afternoon snack

- Try ditching the biscuits and have a portion of fruit instead e.g., two

satsumas, an apple or a small pack of dried raisins.

Dinner

- Try having two portions of vegetables with your meal or add extra vegetables into your meal - they are easy to add to many dishes including chilli, bolognese or pasta bake.

More top tips

1. **Buy in season:** Opting for fresh seasonal produce means you will get the best value for money.
2. **Think of rainbows:** The easiest way to ensure a good mix of vitamins and minerals is to eat a variety of different coloured fruits and vegetables. So, whilst it's important to 'eat your greens', it's just as important to eat your reds, oranges, yellows, blues, purples and whites too. The following list may help you think of how to build this rainbow idea into your meals:
 - **Red:** raspberries, cherries, tomatoes and peppers
 - **Green:** cucumber, peas, apples and grapes
 - **Purple/Blue:** blackberries, blueberries, plums and aubergine
 - **Orange/Yellow:** apricots, grapefruit, carrots and swede
 - **White:** onions, cauliflower, parsnips and bananas
3. **Avoid adding butter, cream, oil, margarine and cheese sauces:** Even though fruit and vegetables have a low energy content, remember how you prepare or serve them will determine their final calorie content.
4. **Watch your intake of dried fruits:** Whilst dried fruits like apricots, prunes or raisins count towards your

five-a-day, once fruit is dried, it also becomes a concentrated source of sugar and calories. Try to keep to a maximum of one serving of dried fruit a day.

5. **Bulk up your meals:** try adding extra vegetables to stews and casseroles, to give extra flavour, texture and nutrition! Alternatively, try covering half of your plate at main meal-times with vegetables.
6. **Raw or cooked?:** Nutrients in fruit and vegetables are easily destroyed during cooking by heat, so, whenever possible, eat them raw. Try steaming, microwaving or poaching vegetables in preference to boiling. This allows you to retain the maximum amount of nutrients.
7. **Short on time?:** Buy pre-washed bagged salads or cut-up vegetables and fruit.
8. **Keep a stock of frozen or canned vegetables and fruit:** these all count to your '5-a-day' and can help you quickly rustle up a balanced meal!

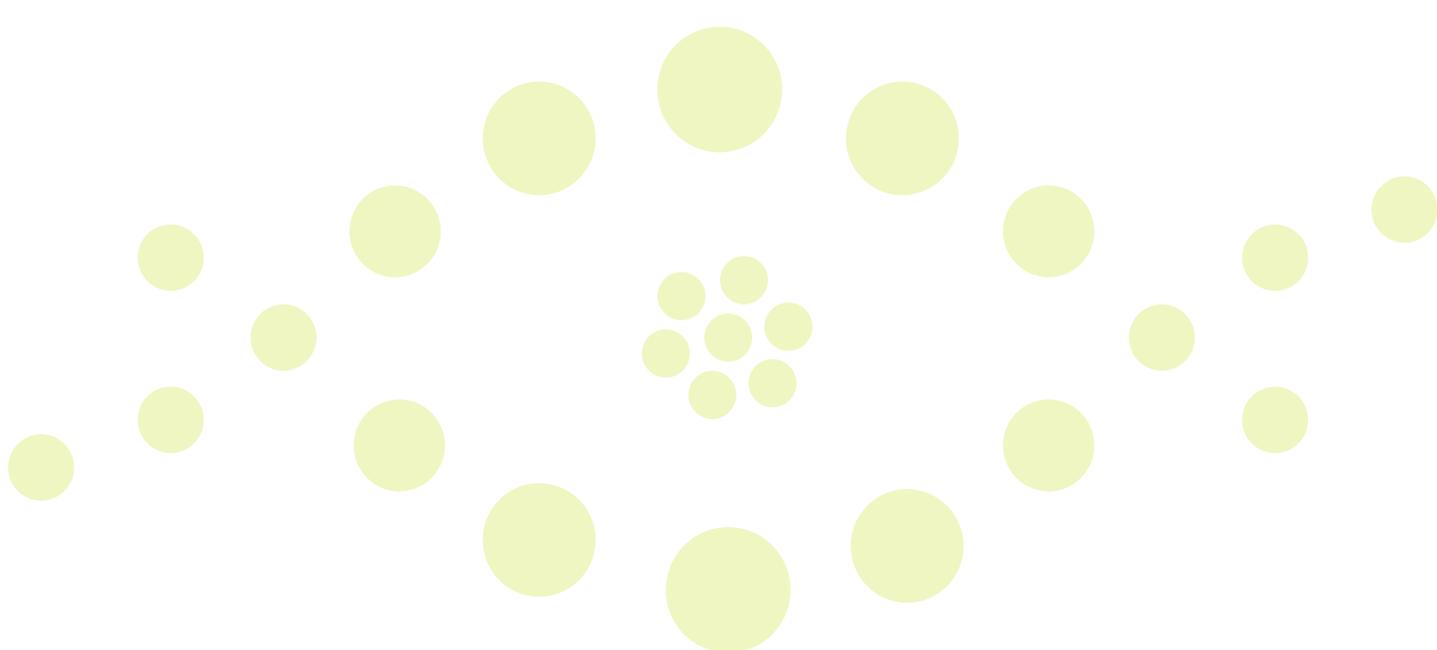
9. **Be adventurous:** Try one new fruit or vegetable every week/month – you'll be amazed at how good you find it tastes! Why not visit shops and markets that have foods from different parts of the world and then go home and experiment!

Please note: If you regularly see a specialist renal (kidney) doctor or dietitian - please ask them if you need to take care with the amount and types of fruits and vegetables that you can eat before you make any changes to your diet. Also, if you have impaired kidney function, it is best to avoid eating star fruit.

If you have any questions, contact your BBS clinic dietitian.

Sarah Flack

BBS Dietitian, Great Ormond Street Hospital for Children NHS Trust



Axovia Therapeutics



Professor Phil Beales has led the most comprehensive research and care efforts for Bardet-Biedl Syndrome (BBS) patients in the world and is now launching Axovia Therapeutics, alongside Dr. Victor Hernandez. The aim of Axovia Therapeutics is to advance gene therapy programs that have already shown early promise in treating some of the most severe symptoms of BBS, such as early onset blindness and severe obesity. Members of BBS UK will be familiar with the research of Professor Beales and Dr. Hernandez through attending the BBS UK Conference and reading the Conference Reports.



“Our goal is to build on the scientific discoveries and molecular diagnosis of Bardet-Biedl syndrome to accelerate the development of disease-transformative treatments for patients as early as possible,” said Philip Beales, CEO and co-founder of Axovia Therapeutics. “Our aim is to halt the progression of BBS so patients and families will experience dramatic improvements in their quality of life.”

The Axovia team is currently working to develop a gene therapy treatment for BBS1, which affects many organs of the body. The major impact of BBS includes early-onset retinal blindness, metabolic disease (including severe obesity and diabetes), intellectual disability often with autism, kidney failure and a range of abnormalities of the endocrine system.

Axovia is a majority-owned subsidiary of Jaguar Gene Therapy. Jaguar brings streamlined research and design, manufacturing and clinical operations to accelerate development and potential approval of therapies for severe genetic diseases. Axovia is backed by Deerfield Management, a healthcare-specific

investment firm, partially funded by University College London Technology Fund.

Professor Beales has long been at the forefront of research, discovery and clinical care of BBS. Discovered in 1866, BBS lacked diagnostic criteria until Professor Beales established them in 1999. A year later, he co-led the team that discovered the first BBS gene and determined that the cause lies with dysfunctional cilia. This work has driven research efforts for treatments for BBS and the broader disease group of ciliopathies. Alongside this, Professor Beales developed the specialised multi-disciplinary clinics service that has provided exemplary care for patients in the UK for over ten years.

To help answer any queries that you may have BBS UK have created a Question-and-Answer document which can be accessed via our website using the following link:
www.bbsuk.org.uk/news-events/

For more information about Axovia Therapeutics, go to:
www.axoviatherapeutics.com

BBS UK

IT Equipment Scheme – we are here to help!

We know that there is an increasing need for people to access services through means of digital technology. Whether it be accessing online education and healthcare, paying for services online, interacting with social media or simply talking to a friend or family member over a video call; now, more than ever before, technology is proving a vital tool for us all.

In view of this, BBS UK would like to encourage those members who have limited or no access to technology to get in touch! The charity team are ready and waiting to offer both practical and financial support to help with the purchase of devices such as laptops, tablets and computers which have become a necessity in terms of accessing education, your GP or Clinic service and providing opportunities to socialise (albeit virtually).



Scheme guidelines – your questions answered

1. Who can apply for help?

The scheme is open to all those who have BBS, from primary school age upwards; there is no upper age limit. Applications will be completed in the name of the person who needs the equipment however applications can be submitted by parents, carers, or healthcare professionals. Only one application per household may be submitted.

2. How do I apply?

Step 1 - If you have either limited or no access to a device such as a laptop, computer, or tablet then the first step is to contact Rebecca Perfect, the BBS UK Advice Worker, whose details are provided below. Rebecca will talk to you either by phone or by email and ask you some questions to try and establish your needs and understand what type of device would be most suitable.

Step 2 – Following the initial assessment, Rebecca will explore whether the equipment needed could be supplied by other external sources. Where appropriate, support will then be offered to help with the applications to such organisations.

Step 3 - If these options prove unsuccessful then BBS UK may be able to provide financial support to purchase a device.

3. Is there a deadline to contact you for help?

There is no deadline; we welcome applications submitted at any time of the year.

4. What happens if the equipment is/becomes faulty?

The equipment will belong to you and ongoing maintenance/repair will be your responsibility. Any problems that arise with equipment after delivery must be resolved between you and the supplier. BBS UK will be unable to resolve technical issues with the equipment itself.

5. Will BBS UK send the funds direct to the applicant?

No, once funding has been allocated to your application, you will be authorised to place an order for the agreed equipment with the supplier and to instruct the supplier to send the invoice directly to BBS UK for payment. Alternatively BBS UK may purchase the equipment on your behalf.

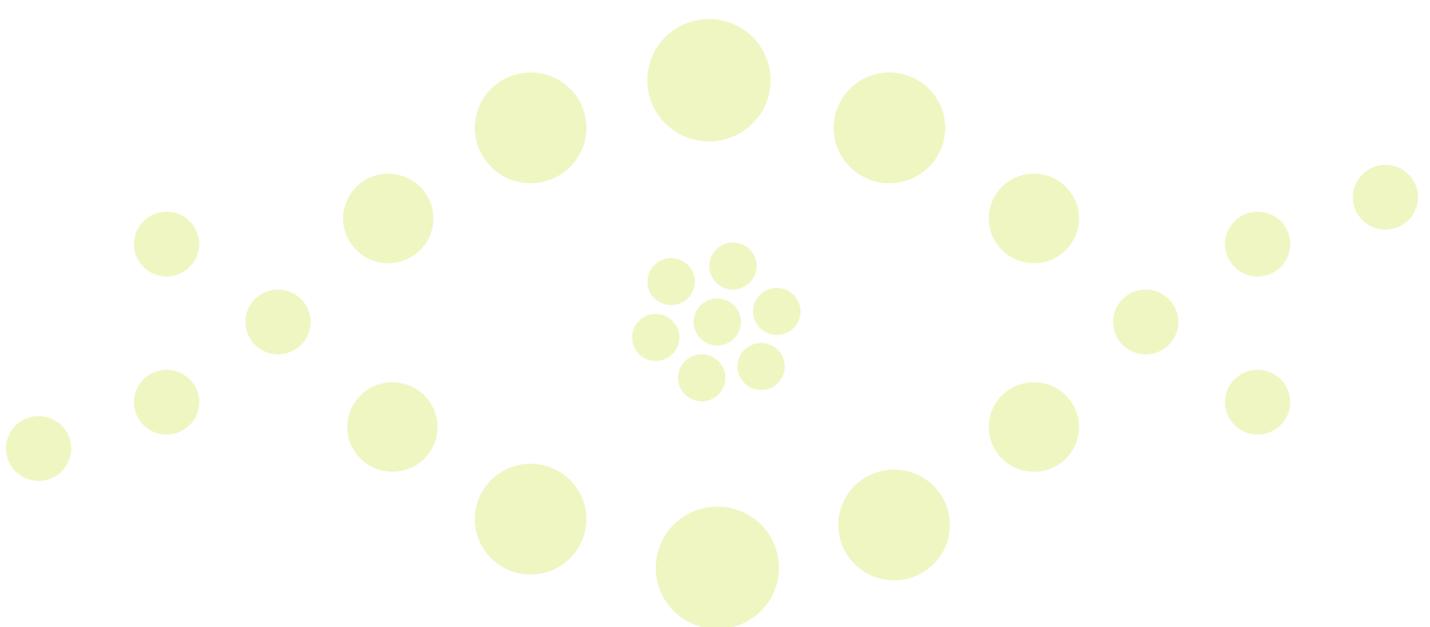
If you are interested then please get in touch!

Rebecca Perfect

Rebecca.perfect@bbsuk.org.uk

Mobile: 07421 455649

Mon-Thurs 10:30am – 2:30pm



Sarah wins a legal challenge for the visually impaired!

Sarah Leadbetter has Bardet-Biedl Syndrome and is identified as clinically extremely vulnerable from COVID-19. Earlier this year when she received a letter from the government advising her to shield, she was unfortunately unable to read it as she is registered blind. Sarah had to ask her mother to read it which made her feel less independent and denied of her privacy. Sarah argued this was discrimination and contacted a lawyer who agreed her human rights and government standards had been breached.



As a result of this, Sarah and her legal representatives were granted a judicial review in the High Court. They were supported by the Equality and Human Rights Commission (EHRC). In advance of the Court hearing the Department of Health and Social Care (DHSC) made a number of agreements which included reconsidering how it communicates with people who are shielding, investigating how it can keep a better record of preferences and bringing in any changes within four months. Sarah is very proud of the results of her legal challenge and

is pleased that the government have made some promising commitments to make improvements with communication and access to information for the visually impaired. Well done, Sarah!

Introducing Slimming World BBS UK!

Bardet-Biedl Syndrome UK have gone into partnership with Slimming World on Referral to offer free 12 week membership to adults who have BBS and would like to lose weight. There are a limited number of places available which will be allocated on a first come, first served basis, to those with a BMI greater than 30, with priority given to those with a BMI greater than 40. Slimming World is currently running via Zoom however face-to-face local groups will soon begin to open up around the country. To be eligible it is important that you have a local group that you can access weekly. It is also important that if you live with family members, that they support your application, in particular the person who cooks the majority of meals, if this is not you.



In addition to the weekly Slimming World meetings, we aim to meet up as a BBS UK Slimming World group on a monthly basis via Zoom to share tips and to support each other. All BBS UK members who are already Slimming World Members can join this Zoom meeting, alongside those taking part in the BBS UK on Referral Scheme.

To access the Scheme or to add your name to the BBS UK Slimming World Zoom group, contact your BBS Clinic Patient Liaison Officer as follows:

Angela Scudder: 07591 206788
angela.scudder@bbsuk.org.uk

Amy Clapp: 07591 206787
amy.clapp@bbsuk.org.uk

Knitted by Nannies



Knitting Nanny Sandra Dale

Those of you who are Facebook members may be familiar with 'Knitted by Nannies', a Facebook shop that has been set up to sell lovingly hand-knitted toys to raise funds for BBS UK. The two 'Knitting Nannies' are Sandra Dale and Janet Wakelin and over the past few years they have knitted somewhere in the region of 80 toys, raising more than £1,500 for BBS UK.

Sandra and Janet started knitting the toys for a children's raffle held during our conference weekend, however with last year's event being cancelled, they needed a way of finding homes for their knitted creations whilst continuing to raise funds for their favourite charity... and so 'Knitted by Nannies' was born! Sandra and Janet donate all materials so that 100% of the money raised goes to BBS UK and since

setting up their dedicated JustGiving page eight months ago, they have sold more than 30 toys, raising over £550.

There is something very special about each and every toy created by our Knitting Nannies; they each have a unique character, quirky colours and clothes, special finishing touches and are filled with so much love. To see what is currently available, go to www.facebook.com/knittingnans

We are so grateful to Sandra and Janet for supporting BBS UK in this way. Not only are they raising much needed funds for the Charity, but they are putting so many smiles on faces too – keep up the good work, we cannot wait to see what comes out of the Knitting Nannies Workshop next!



Knitting Nanny Janet Wakelin

New Trustees for BBS UK

BBS UK would like to give a very warm welcome to BBS UK's newest trustees, Dr Shehla Mohammed and Liam Wilkie who have been co-opted to the Board.

Dr Shehla Mohammed



Many of you will know Dr Shehla Mohammed who is Clinical Lead for the BBS specialised service at St Thomas' Hospital, London. Dr Mohammed has been the Head of

Department of one of the largest Regional Genetics Service at Guys Hospital for 20 years and is keen to bring her managerial and strategic planning skills to complement the key work of the BBS UK Board of Trustees.

Dr Mohammed is a strong advocate for patient focused services and helped to set up the National Highly Specialist Service for Bardet-Biedl Syndrome over 11 years ago. Dr Mohammed works with a number of support groups and has also set up and run dedicated multi-disciplinary clinics for other rare diseases, including Cockayne Syndrome in partnership with the family support group Amy and Friends.

Dr Mohammed has experience working with a number of external organisations, committees and working groups as a member and Chair, influencing policy development for clinical and laboratory services. Shehla is currently a member of the Highly Specialist Technology Committee of NICE, which considers new products for the treatment of rare and ultra-rare diseases.

In her spare time, Dr Mohammed enjoys travel, gardening and kick-boxing and is a mature student of the cello.

Liam Wilkie

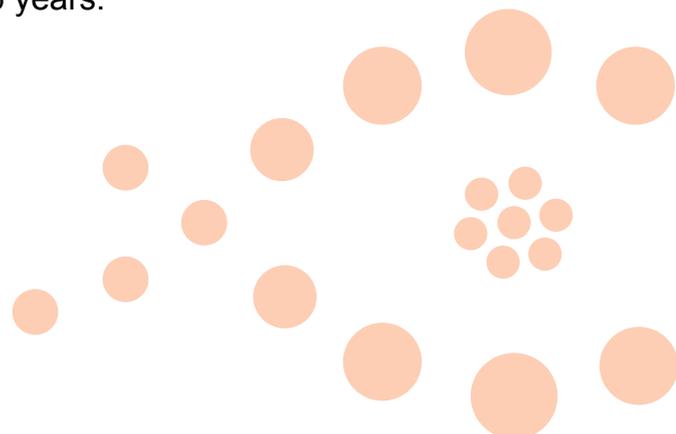


Liam Wilkie has been a strong advocate of BBS UK, raising awareness and funds alongside the Wilkie Warriors in Scotland and beyond. Married to Ashley, together they have two children, Caitlin and Niamh. The Wilkie

Warriors had an incredible fundraising year not long after their daughter, Caitlin was diagnosed with BBS, raising over £80K for the Charity.

Liam is a contracts manager for a medium sized construction company in Scotland. Alongside utilising his business, contract and management skills to benefit the BBS UK Board and Charity, Liam is also looking to raise greater awareness in Scotland to improve understanding and create pathways that will hopefully lead to increased funding and fundraising. Liam credits his community and network of friends for the incredible results to date, both with raising awareness and funds locally and is looking forward to taking this further afield.

In his spare time, Liam enjoys keeping active outdoors with running and cycling, he also plays drums in a local band which has been a long-term hobby over the past 16 years.



Contact Details

Office Manager

Liz Loughery

07421 312 731

liz.loughery@bbsuk.org.uk

admin@bbsuk.org.uk

Mon-Thur, 9.00am - 4.30pm

Fundraising Officer

Christine Saxon

07421 453896

christine.saxon@bbsuk.org.uk

fundraising@bbsuk.org.uk

10 hours per week

Advice Worker

Rebecca Perfect

07421 455649

rebecca.perfect@bbsuk.org.uk

Mon-Thurs, 10:30am – 2:30pm

Service Manager

Tonia Hymers

07591 206680

tonia.hymers@bbsuk.org.uk

Mon-Thurs, 8.30am - 4.00pm

Patient Liaison Officer:

Birmingham BBS Clinics Service

Amy Clapp

07591 206787

amy.clapp@bbsuk.org.uk

Mon-Thurs, 9.00am - 4.30pm

Patient Liaison Officer:

London BBS Clinics Service

Angela Scudder

07591 206788

angela.scudder@bbsuk.org.uk

Mon-Fri, 9.00am - 3.00pm



bbsuk
Bardet-Biedl Syndrome UK
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