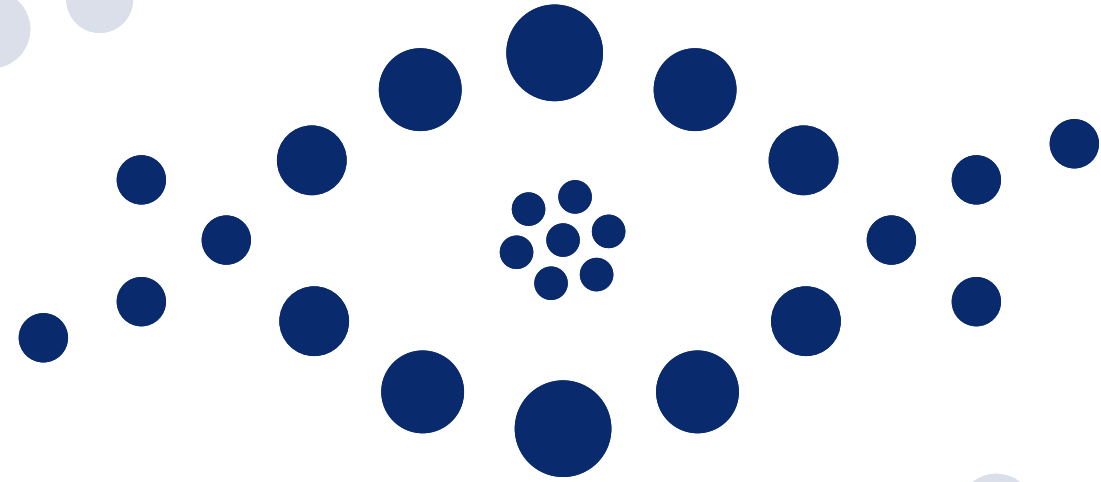
bbs uk

Bardet-Biedl Syndrome UK

Medical Information Booklet



The Origins of **Bardet-Biedl Syndrome**

The first formal description of Bardet-Biedl syndrome (BBS) appeared in the 19th century, in a paper by London ophthalmic surgeon, John Zachariah Laurence, and his house-surgeon, Robert Moon (1845–1914). Moon's father, William, had invented one of the earliest raised alphabets for the blind.

BBS remained largely unmentioned until 1920, when Georges Bardet discussed hypothalamic obesity in his MD thesis. In 1922, Artur Biedl independently described two siblings with retinitis pigmentosa and polydactyly, without referencing Laurence and Moon's work.

John Zachariah Laurence

In 1925, Solis-Cohen and Weiss rediscovered the Laurence-Moon paper and grouped these conditions together under Laurence-Moon-Biedl syndrome (LMBS). This remained the standard term until the 1980s.

Robert Moon

More recently, the condition has been divided based on clinical features into Laurence-Moon syndrome and Bardet-Biedl syndrome. Bardet-Biedl syndrome represents by far the majority of published cases and is now the more generally recognised term within the medical and scientific community.

Artur Biedl

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Acknowledgement

This booklet has been funded by NHS England and produced by Bardet-Biedl Syndrome UK (BBS UK), Registered Charity No. 1181244.

It is important to note that there are many symptoms and conditions listed within this booklet that may affect those who have BBS, but they will not necessarily affect everyone, and the severity of individual symptoms can vary greatly among those affected.

Our grateful thanks go to the clinicians from the BBS specialist clinics teams at Queen Elizabeth Hospital, Birmingham; Birmingham Children's Hospital; Great Ormond Street Hospital, London, and St Thomas' Hospital, London, for their contribution to this publication.

Our heartfelt thanks also go to the generous individuals living with BBS who contributed to this booklet; their guidance and personal perspective have been invaluable.

www.bbsuk.org.uk

> **There's no-one else who knows our child like we do, and we have to take this on board and make ourselves heard. We have to be an equal partner with medical, social and educational teams and believe that we're an equal partner. We have to use the resources available to us to give our children the confidence, courage and self-esteem to know that they have a great deal to contribute to this world.**
>
> Parent

Introduction

Bardet-Biedl syndrome is a rare, inherited disorder affecting approximately 1 in 100,000 babies born; many GPs, doctors and health professionals will not have come across BBS or have heard of the Syndrome before.

Features of the Syndrome include rod-cone dystrophy, a progressive eye disorder that leads to blindness, characterised by tunnel vision and night blindness; obesity; renal abnormalities; developmental delay; speech and language difficulties; extra fingers and/or toes and learning difficulties. Not all the features are always present in those diagnosed as having BBS and each one can vary in severity and appearance. The variability in presentation and severity of the symptoms, together with the rarity of the Syndrome, can lead to delayed diagnosis and a lack of adequate local health care.

This booklet is aimed at the medical and healthcare professionals involved in the care of BBS patients to promote a greater understanding of the Syndrome and the recommended care pathway. The information has been provided and checked by the BBS specialist clinics team and, where available, is supported by research and published articles. A bibliography of references and useful publications can be found at the back of the booklet. For further information, contact: **admin@bbsuk.org.uk.**

Diagnosis and Genetics

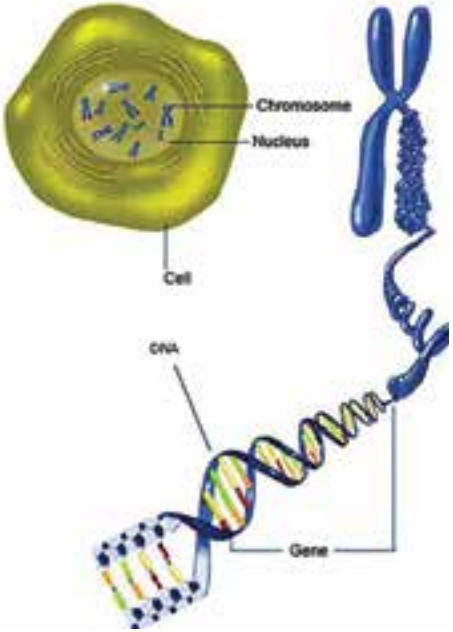
Diagnosis

Beales et al (1999 and 2001) suggest that the presence of four primary features or three primary features plus two secondary features is necessary for a clinical diagnosis of Bardet-Biedl syndrome.

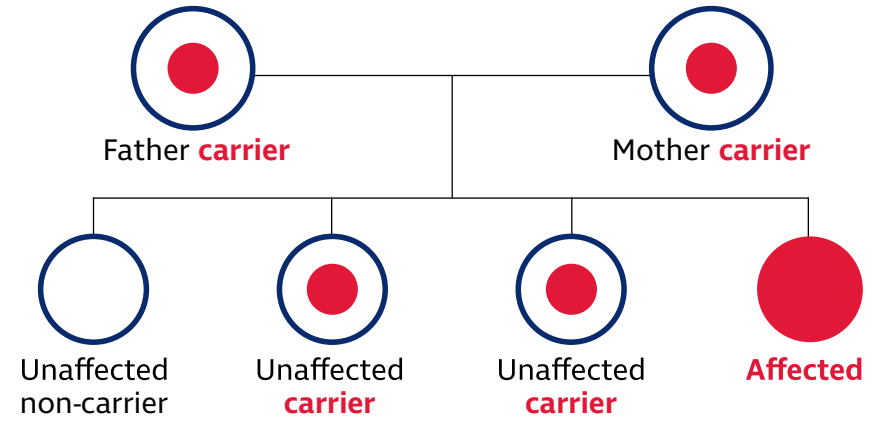
Primary Features	Secondary Features
Rod-cone dystrophy	Speech delay/disorder
Polydactyly	Developmental delay
Obesity	Brachydactyly
Learning disabilities	Polyuria/polydipsia
Hypogonadism in males	Ataxia
Renal anomalies	Poor co-ordination
	Diabetes mellitus
	Left ventricular hypertrophy
	Hepatic fibrosis
	Hypertonia
	Hearing loss

Genetics

In most cases of BBS, both parents carry a normal gene and a faulty, recessive gene. Although the parents have one copy of the faulty gene and are called carriers of the disease, they are unaffected by the presence of the faulty gene. For a recessive disease to occur, a child has to inherit two faulty copies of the gene, one from each parent. The child from each pregnancy has a 1 in 4 chance of being affected; if the child is not affected, then there is a 2 in 3 chance that they will be a carrier of the faulty gene for BBS.



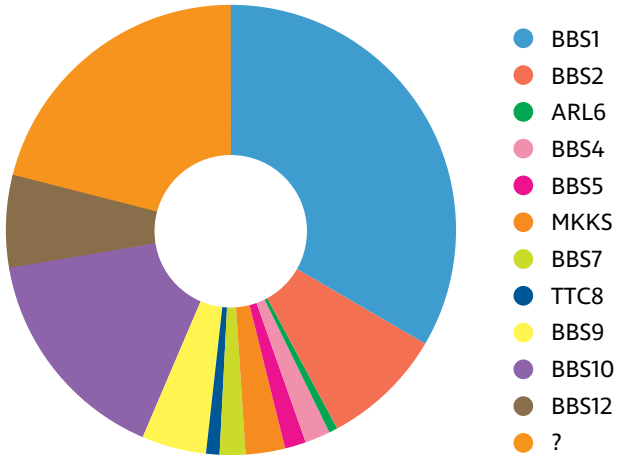
Autosomal Recessive Inheritance



Some BBS genes are more common than others, and there is variation in the frequency with which they are being carried, known or unknown, in the population, and across certain ethnic groups. For the most common BBS genes, BBS1 and BBS10, the frequency is estimated to be 1 in 250, whereas for a rarer gene such as BBS9, the frequency is closer to 1 in 820. The risk of encountering another carrier increases if couples are related.

Pie chart showing relative frequency of mutations in 12 BBS genes tested

(Courtesy of North Thames Regional Genetics Service)



To date, (2025) mutations in over 25 BBS genes have been identified in 80% of BBS patients. It is thought that there are still more genes to be discovered since not all patients have an identified mutation in any of these identified BBS genes. Some genes are more common than others; around half of patients in the UK have mutations in BBS1 and a quarter have mutations in BBS10. However, patients who carry mutations in the same BBS gene can display quite different symptoms of the condition: one might have extra digits at birth whereas another person with an identical mutation may not have extra digits at all. It is hoped that the comprehensive genetic testing currently available will improve our understanding about the progression of BBS in the future and whether any particular aspects in some forms of BBS may need closer monitoring.

Cilia

Although the mechanism that leads to BBS is still unclear, the main cause of the symptoms that feature in BBS is due to abnormally functioning or structured cilia, long thin, hair-like projections that stick out of the surface of a cell. There are two types of cilium, motile and non-motile cilia (also called primary or sensory cilia).

Motile cilia are present on cells where their movement helps to perform an essential function, for example, in the respiratory tract or the middle ear. These cilia can be affected in some people with BBS, causing respiratory difficulties or sensorineural deafness. More information can be found in the 'Additional Features' section of this booklet.

Non-motile cilia are found on nearly all cell types. Their main function is to receive signals from outside of the cell and transmit them to the inside, allowing neighbouring cells to communicate with each other; they play an important role in terms of smell, vision, touch, and temperature sensation. They are present in the retinal photoreceptor in the eye, and in kidney cells.

Mutations in BBS genes cause changes in the proteins that are needed for the correct functioning of cilia and for this reason, BBS has been categorised medically as a 'Ciliopathy'. The scientific community is trying to gain a better understanding of exactly what roles the BBS proteins play in cilia formation and function.

Carrier Testing

Where BBS is confirmed genetically, a simple carrier test is possible in at-risk adult relatives (e.g. siblings) to help determine their own risk of having affected children. Knowledge of the precise BBS mutations can also provide the basis for prenatal screening tests, should parents need to know in the early stages whether a pregnancy is affected.

> **During the early days of diagnosis, it is important to allow time to come to terms with all that has happened and to give yourself time and space to adjust.**
>
> Parent

Family Planning

Where both parents are carriers of a change in the same BBS gene, every pregnancy has a 1 in 4 risk of the child having Bardet-Biedl syndrome. If the genetic changes are known in the parents, there are several options available to them, should they wish to have more children.

Prenatal Testing

Couples who are already pregnant may consider prenatal testing (e.g. chorionic villus sampling or amniocentesis). It involves testing either the tissue or fluid around the baby in early to mid-pregnancy (11-16 weeks). There is a small risk to the pregnancy with prenatal testing; couples who wish to pursue this option should discuss it with their geneticist and obstetrician.

Pre-implantation Genetic Diagnosis (PGD)

Couples may be referred for consideration of PGD, subject to meeting certain criteria. Common requirements are that they do not already have a healthy child and do not smoke, though specific criteria may vary by region. Couples interested in this option should consult their GP or geneticist for guidance.

Couples who are not interested in either of these options, or do not know the genetic change that has caused BBS, may opt to have detailed scans throughout pregnancy to assess if there is any physical evidence that the baby is affected. This should be done in a specialist centre by an ultra-sonographer who is familiar with the features associated with BBS. Features of BBS may be difficult to pick up before 20 weeks gestation and, as already mentioned, may not be present in all babies with BBS.

Some couples may choose not to have any form of testing during a pregnancy and opt for either an examination of the baby after birth, or to test a blood sample taken from the umbilical cord after delivery. Ideally, it is best if discussions can be undertaken before a pregnancy occurs, to enable the wishes of the parents to be captured fully.

Prospective parents who have Bardet-Biedl syndrome

Those who have BBS who wish to have children, may also have several options available to them, and should discuss their plans with a member of their medical team.

The children of a BBS affected parent would be carriers, but will not have the condition, provided that the other parent does not have any changes in the same gene. If the other parent is a carrier of a change in the same gene, then each child has a 50% chance of having BBS . If both parents have BBS caused by changes in the same gene, then all children will be affected with BBS.

> **No one should ever put limitations on what they think someone will be able to accomplish just because of a diagnosis of BBS.**
>
> **Patient**

Signpost:

Bardet-Biedl Syndrome UK
www.bbsuk.org.uk

Gene People
www.genepeople.org.uk
0800 987 8987

Genetic Alliance UK
www.geneticalliance.org.uk
0300 124 0441

Clinical Features and Presentation

There are many aspects to BBS and it is important to remember that there is great variation among those affected, even between siblings. The following section looks at each aspect in turn and includes recommended tests and assessments.

Eyes

Pigment changes, similar to those in retinitis pigmentosa, are often seen upon examination of the retina in individuals with BBS; the correct term is rod-cone dystrophy. The light-sensitive photoreceptor cells in the retina, the rods and cones, degenerate over time because of defective cilia.

The rods provide night vision and peripheral vision, and the affected individual will experience poor night vision and loss of peripheral vision as the rods degenerate. The cones provide colour vision and central vision, and as they degenerate, colour and detailed vision will be impaired.

The onset of rod-cone dystrophy is usually noticeable during primary school years and is initially experienced as difficulty with seeing at night or in poor light conditions. In some cases however, visual symptoms can be delayed into the late teens or beyond. As the retina degenerates and the condition progresses, the affected individual may lose some ability to see through the whole visual field. Loss of peripheral vision is frequently referred to as tunnel vision; as the visual fields `close in', the young person may begin to appear clumsy, especially at night-time. Functional vision will also be affected by changes in lighting and low lighting, dark evenings will make it much more difficult to use residual sight and daytime glare will affect central vision.

A study found rod-cone dystrophy present in almost all patients who have a genetic diagnosis of BBS. A significant number of people with BBS have retinal signs that are seen on examination of the retina, however in some, the retina can appear normal and electro-diagnostic testing is needed to provide evidence of retinopathy.

A study found that the average age at which symptoms or signs of visual loss are reported is twelve years of age, although night blindness was often noted earlier. The study also found that the average age of registration of blindness is approximately fifteen years. Other eye conditions associated with BBS include wobbly eyes (nystagmus), an irregularly shaped cornea (astigmatism), squints (strabismus), glaucoma and cataracts.

Treatment

Research is ongoing, regarding the possible effectiveness of gene therapy to treat rod-cone dystrophy in BBS. It is possible that there may be research projects over the next few years for a small number of BBS patients, to see whether the treatment is safe and beneficial.

In the meantime, correction of short or long sightedness and astigmatism and the provision of tinted glasses (for light sensitivity) can assist in maximising useable vision. Cataract surgery should be offered where appropriate, and where it will be of actual visual benefit. Low vision aids and mobility training can improve independence and confidence. Magnifying glasses, digital systems and voice systems may also be helpful.

Consideration should be given to educational planning, and if there are disrupted sleep patterns and difficulties with breathing at night (nocturnal apnoea), sleep studies should be considered. Referrals to local low-vision clinics and organisations assisting the visually impaired are recommended. Partial sight and blind registration (certificate of visual impairment) should be carried out in the first instance where any visual impairment is diagnosed.

Sign Post:

Retina UK
www.retinauk.org.uk
0300 111 4000

RNIB
www.rnib.org.uk
0303 123 9999

VICTA
www.victa.org.uk
01908 240 831

Look
www.look-uk.org
07464 351 958

Sense
www.sense.org.uk

Visual Assessment

At diagnosis:

- Visual acuity assessment and refraction
- Visual field testing
- Examination of the back of the eyes
- Electro-diagnostic testing
- OCT scanning

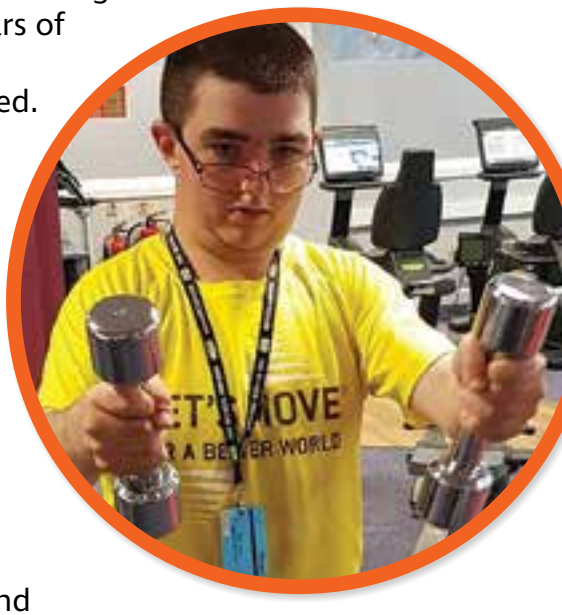
Follow-Up

Yearly eye examination:

- Visual acuity
- Visual field testing (where possible)
- Examination of the back of the eyes
- Electrodiagnostic testing if indicated
- Screening for cataract, glaucoma and diabetic retinopathy as appropriate
- General eye health check (by local optician between hospital appointments)

Obesity

Obesity is reported in 72% to 86% of patients. Weight issues usually begin in childhood and increase in severity with age. Children may exhibit rapid weight gain, with one third of babies developing obesity by their first birthday. Controlling rates of weight gain during the first years of life can be challenging, especially when crawling or walking is delayed. However, some children do not present with weight concerns until puberty and final adult height is achieved.



Most adults who have BBS, have a BMI greater than 30 which is often accompanied by other health issues, for example, high blood pressure, high cholesterol and type 2 diabetes.

Hyperphagia (excessive hunger) and food seeking behaviour are common, resulting in energy intake exceeding requirements, which makes dietary management challenging.

Treatment

BBS symptoms cover a wide spectrum and vary significantly between individuals. It is important that all patients have an individual assessment of their weight and dietary requirements.

Understandably, diet and weight concerns are a major source of stress for patients and their families. The knowledge that untreated obesity can lead to multiple health problems, for example, type 2 diabetes, coronary heart disease, liver disease and metabolic syndrome, adds to this stress.

Unfortunately, there is no single treatment approach for obesity in BBS, just as for obesity in general, but those with BBS can successfully lose weight. In younger children, care should focus on controlling the rate of weight gain, without compromising growth. A healthy well-balanced diet and active lifestyle approach is encouraged. Establishing routine meal and age-appropriate snack times and portion control strategies are important to manage the excessive appetite that can be associated with BBS. Very low-calorie diets are not required as this over restricts energy intake.

It is important to educate with dietary strategies and encourage activity to control weight gain in childhood, so these behaviours can then be continued into adulthood. A multi-disciplinary approach is needed and should include dietary advice, behavioural management, and exercise. Early referral to a registered dietitian with experience of BBS is important, and attendance at the multi-disciplinary BBS clinics is strongly recommended.
Research continues into therapies to target obesity in BBS. The latest research developments are discussed at the BBS UK annual conference and can also be accessed via the BBS UK website. There have been widely publicised developments in the drug treatment of obesity with clinical trials ongoing and treatments becoming available. For any medication there will be criteria that need to be met for this to be available on prescription, and monitoring required to ensure safe use. Patients and families will be updated on any available drug treatment within their BBS clinic appointment and via BBS UK.

Surgical management approaches for obesity are not recommended as these procedures do not address hyperphagia. The associated risks and benefits of bariatric surgery options need to be considered for a person living with BBS. It is important to note that specialist dietary advice and support is needed post-surgery, and compliance with these dietary restrictions is imperative.

Patients and families are encouraged to discuss the latest treatment options for obesity and hyperphagia with their clinic team at their next BBS appointment.

Polydactyly and Brachydactyly

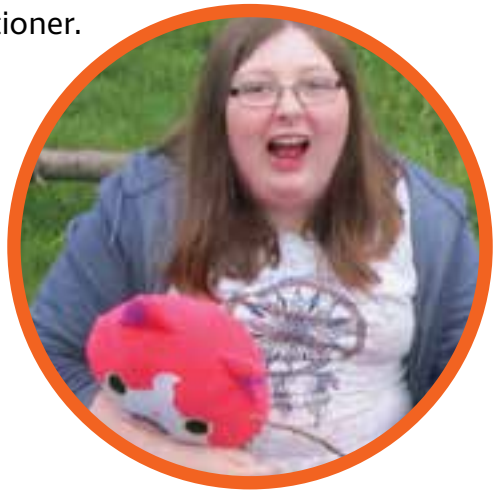
Most BBS patients (70% to 84%) are reported to have extra digits (polydactyly), most commonly appearing next to the little finger or toe; presentation ranges from a single skin tag to a fully formed digit on all four limbs.

Shortness of the fingers and toes (brachydactyly) is common, affecting 46%-100% of patients and is more frequently found in the feet than in the hands. When present in the hands, it may affect dexterity and the ability to use keyboards or Braille.

Webbing (syndactyly) is less frequently seen and is usually partial, and generally confined to the feet.

Treatment:

Skin tags can be tied off at birth after documentation by a medical practitioner. Fully formed digits are often non-functional and should be removed, where possible, within the first year of life by either an orthopaedic or plastic surgeon. Bony malformation in wide feet can lead to ill-fitting shoes and it is important to seek podiatric and, orthotic advice and specialist fitting.



Renal

BBS can affect the kidneys in a number of ways, although significant kidney problems and kidney failure are seen in only a small proportion of patients. Nevertheless, it is important to monitor all patients regularly.

Most patients with BBS do not have any serious kidney involvement. More than half of patients with BBS have kidneys that are a slightly abnormal shape or size, or they may have kidney cysts. However, these abnormalities seen on a kidney ultrasound scan hardly ever get worse or cause any problems. When kidney problems do occur, they usually cause mildly impaired kidney function, protein in the urine and high blood pressure. For these reasons blood and urine tests and ultrasound scanning of the kidneys should be a routine aspect of renal management in BBS.

A minority of patients with BBS experience more severe kidney problems that may lead to a significant reduction in kidney function or kidney failure, requiring dialysis or a kidney transplant. Fortunately, this is uncommon. A review of kidney involvement in 219 BBS patients seen in the specialised BBS clinics in the UK noted that only 12 (6%) of these patients had developed kidney failure requiring dialysis treatment or kidney transplantation. It was also noted that 5 out of the 12 patients with kidney failure developed it in the first year of life, and these were all patients that were born with severe abnormalities of their kidneys. The remaining 7 developed kidney failure later in childhood, probably reflecting less severe congenital abnormalities of the kidney.

The encouraging news from this review is that patients who do not have any evidence of kidney abnormalities in early childhood are very unlikely to develop severe kidney problems later in life. It is important to understand however, that with advancing age, other factors, such as obesity, diabetes, hardening of the arteries and high blood pressure may also contribute to a decline in kidney function.

Those patients who do develop kidney failure seem to do well with dialysis and kidney transplantation, so even if kidney failure does develop there are very good treatments to support patients.

Other kidney problems may occur in BBS, including a reduced capacity to concentrate the urine. This leads to higher-than-average urine output and sometimes excessive thirst, although few patients notice this. Infections of the bladder (cystitis) and, occasionally, of the kidneys themselves can also occur, and are treated in the conventional way with antibiotics.

Clinicians working in the BBS specialised clinics have also recognised that it is quite common for patients to hold onto their urine, and pass large quantities just a few times a day, whilst others wake up to pass urine frequently through the night. Holding onto urine for most of the day can lead to stretching of the bladder and cause problems to develop later in life. Patients can avoid this by ensuring they empty their bladder frequently, for example, by going for a wee when they wake, after mealtimes, and before bed, whether they feel the urge to go or not.

High blood pressure may be evident in patients with kidney disease of any type, and in BBS it is often present and should therefore be regularly checked and treated if necessary. High blood pressure can also put further strain on the kidneys, so in BBS patients, where there is known kidney involvement, it is particularly important to monitor and treat this.

> **When I was 16, I went to the West of England College in Exeter. At the college I learnt how to be independent, doing such things as cooking, cleaning, and shopping. I also completed courses in further education, including an NVQ in administration. I also did work experience placements. I love living independently and being able to see my friends and go where I want to. I feel that I have achieved a lot so far in my life, and I look forward to enjoying life to the full.**
>
> Patient

Obesity is a very common problem in people with BBS and excessive body weight can put additional strain on the kidneys and make high blood pressure worse. This can exacerbate any underlying kidney problems or can even occasionally cause kidney damage on its own. The reasons for managing weight in BBS therefore extends to protecting the kidneys from damage.

Renal Assessment

At diagnosis:

- Check symptoms of anaemia, polyuria and polydipsia
- Baseline blood pressure assessment: 24-hour blood pressure monitoring, if there are concerns and if possible
- Measure plasma creatinine, urea, electrolytes, GFR
- Bladder and renal ultrasound examination

Follow-Up:

- Yearly for symptoms, baseline blood pressure
- Yearly early morning urine analysis for albumin creatinine ratio and dipstick testing for microscopic haematuria
- Yearly monitoring of plasma creatinine, urea and electrolytes, GFR

Sign Post

National Kidney Federation
www.kidney.org.uk
0800 169 09 36

Kidney Care UK
www.kidneycareuk.org
0808 801 00 00

PKD Charity
www.pkdcharity.org.uk
0300 111 1234

Development, Learning, Social and Emotional Wellbeing

BBS is a condition that has a wide spectrum and so each person will be affected differently. Some children and adults will have few developmental or learning needs, whereas others may need ongoing support throughout their lives. Like all children, those with BBS will generally continue to progress and learn throughout their childhood. Each individual child will have their own strengths and personality and follow their own unique path in learning and development.

This section will talk about the range of difficulties that can occur for children and adults with BBS. This does not mean that each person will experience all of these difficulties. It will be important however for parents, carers, school staff and others around an individual with BBS to understand their strengths and needs in order to help them achieve their potential, to be themselves, and to enjoy life together as part of a family.

Learning and academic ability

Children with BBS show a wide range of developmental and learning abilities. Studies to date show that most children with BBS have some degree of learning needs and may benefit from additional support at school. Children with BBS often perform below the age-expected range on cognitive assessments, but there is variation, and some children perform within the average range.

A number of children will meet the diagnostic criteria for a learning disability, and previous research estimates of the rate vary widely - between 25-66%. A recent study, in a clinical sample of children with BBS in the London BBS clinics service, found that 9 out of 21 children who were assessed (41%) met diagnostic criteria for a learning disability (Blumer et al., in press). The exact rate is difficult to confirm due to the study having a small sample size, and not including all children in the service (meaning we cannot be sure how representative it is).

A learning disability means that someone shows consistent difficulty with learning new information, understanding concepts and ideas (low intellectual abilities), as well as difficulty with everyday activities – for example carrying out chores, personal hygiene and staying safe in the community. It affects people for their whole lives. A learning disability is diagnosed by a health professional (usually a paediatrician or a psychologist). A learning disability is different from 'learning difficulties', a term that is used to describe specific difficulties with learning that can be for a range of reasons (but do not meet the criteria for a diagnosis of a learning disability).

Whilst children with a learning disability learn and develop throughout their childhood, they do so more slowly than other children of their age, and will require support with some tasks which other people their age may be able to do independently. Someone with a mild learning disability may be able to complete most tasks independently (and only require support with complex tasks like filling in forms or managing financial decisions), whereas someone with a more severe learning disability may require support with daily living. Most people with BBS who have a learning disability are at the mild-moderate end of the spectrum.

Clinical assessments, undertaken within the BBS clinics service, have shown an additional pattern of weakness with motor skills, as well as a shorter attention span for people with BBS. These areas may also impact on their learning and participation. It is important that people working with children with BBS allow the child enough time

to interpret and understand information and provide answers. This will allow children to show the best of their abilities.

Children with BBS show a wide range of learning abilities and support needs. Many children with BBS attend mainstream schools and some attend specialist schools, depending on their needs. Some children are very academically able and will go on to university education.

Visual needs and learning

Regardless of the school setting that a child or young person is in, if they have any visual needs, these will influence how they need to access learning in the school setting. Regular vision assessments and timely support from vision specialists is needed; as the young person's visual needs change over time, so the support they receive will need to be adapted. As these visual changes occur, consideration of learning new skills will be important especially as the young person becomes more independent. Information about supporting children and young people in a learning environment can be found in the BBS UK Booklet for Schools and Colleges.

Social communication and autism spectrum

It is known that children with BBS often show differences in how they communicate and understand social situations. Children may also behave in a way that appears obsessive and rigid. Recent studies suggest that around 70%-80% of children with BBS will show autistic tendencies, with an overall estimate of 34% diagnosed as being on the autism spectrum. Therefore, social communication needs will be important to consider during childhood, adolescence and into adulthood.

Emotional and behavioural needs

As can be expected for children with learning, language, visual and social communication needs, children with BBS are more likely to show emotional and behavioural challenges. For young children with BBS, it is important to be mindful of their emotional wellbeing and consider what may be behind any behavioural challenges they show. Typical behaviour management strategies may not be effective and specialist support from local health services may be required. Setting clear boundaries around food and eating at an early stage may help to prevent future difficulties in this area.

Towards the later years of primary school, some children with BBS may show an increasing awareness of their needs which can lead to the feeling of being different, particularly in the school setting. At this age, young people may experience difficulties with friendships and bullying which, alongside weight issues, can result in low self-esteem. Helping children and young people to recognise and appreciate their own strengths, as well as developing their interests, can protect against such difficulties.

As the young person gets older, they and their family often experience fears and worries, relating to the daily management of any behaviour difficulties, education, living an independent life and

relationships. Fitting in socially is very important and the teenage years are when relationships and sexuality are important issues that young people may explore. This stage of development involves the young person exploring their identity as being separate from their parents, and so they may want to spend more time away from parents. As with the general population, hormonal changes can also contribute to developing emotional difficulties, with people feeling lonely and unhappy.

In adulthood, anxiety is commonly reported, which can significantly impact on the individual's ability to cope with their condition. Common concerns include worrying about health and medical needs, not feeling in control of emotions, apathy, and finding employment. Individuals with autism or autistic traits may have difficulty understanding other people's emotions and difficulty reading social cues, which makes it harder to relate to others, leading to social isolation. A combination of differences in social abilities, alongside changes to daily routines when young people leave education, can lead to feelings of anxiety. People may struggle with uncertainties including the loss of routine and relationships as well as increased unpredictability about the future. Independence is an issue for some, but not for others. Some people will find employment, whereas others find themselves understandably frustrated about the limitations that can come with BBS. Specialist support will continue to be needed for adults with visual, learning and social communication needs.

Low mood, panic attacks, obsessions and compulsions, anger and poor emotional control also commonly affect adults and young people with BBS. It is unclear whether this is a part of the Syndrome or an indirect result of it; most likely, it is a combination of both. A very small number of patients experience delusional thoughts and some develop periods of psychosis. If this is suspected, prompt referral for a psychiatric opinion and therapy is warranted.

Support and management

For young children of pre-school or school age, an Education, Health and Care Plan (EHCP) will ensure the child is fully supported. Parents or carers should contact the school or pre-school and request an assessment be commenced. The school or pre-school's SENCo (Special Educational Needs Co-ordinator) will co-ordinate the process and arrange for educational psychologist and sensory team involvement. Possible interventions for children would include:

- Assistance with school integration
- Behaviour support
- Speech and language therapy
- Qualified Teacher of Children with Visual Impairment
- Family support
- Management of symptoms
- Sensory support
- Exploring emotional needs
- Occupational therapy

Due to the high level of learning needs and social communication differences in children with BBS, it is important for children to access assessments if concerns are identified. It is important that needs are identified during childhood and adolescence. Assessments can identify whether children meet the criteria for a learning disability or autism diagnosis, in order to access further support.

For adults, isolation can lead to or exacerbate anxiety and depression and it is vital that appropriate support is in place. If there are concerns with an adult's mental health or emotional wellbeing, they may benefit from accessing psychological or psychiatric support. Referrals for support can be made through the GP.

For adults with BBS who also have a learning disability and/or autism, there are specialist NHS services to support with mental health, tailored for people with learning disabilities. Referrals to these services can also be made through the GP. There are many ways for adults to access support:

- Ask GP to arrange a social services assessment and occupational therapy assessment.
- Ask GP or Social Worker about local support groups for the disabled or visually impaired.
- Ask GP about counselling or psychological intervention if more support is needed for low mood or anxiety.
- Talk to staff at a local disability centre or Social Worker about activities in the area.
- Contact BBS UK for advice or support. It may help to talk to others in the same situation.
- Exercise is vitally important for emotional and physical wellbeing.
- Local Disabilities Team or Social Worker will be able to help with claiming benefits, arranging support workers, carers or respite, and linking individuals into local services in the area.

Signposting for people with BBS

Mind
www.mind.org.uk
0300 123 3393

National Autistic Society
www.autism.org.uk

MENCAP
www.mencap.org.uk

Scope
www.scope.org.uk
0800 800 3333

The Challenging Behaviour Foundation
www.challengingbehaviour.org.uk
0300 666 0126

Endocrine

Male Reproductive System

Hypogonadism (lack of production of sex hormones) is common amongst males, affecting up to 30% of BBS patients; a small, buried penis with reduced volume testes is common. 10% of male patients with BBS have undescended testes at birth. In most cases this is due to a partial failure of the hypothalamus and/or pituitary gland with low or inappropriately normal luteinizing hormone (LH) and follicle stimulating hormone (FSH), with low circulating testosterone levels. In a small number of individuals, there may be primary testicular failure. There is usually enough testosterone production to permit normal pubertal development. The timing of male puberty is often slightly delayed but with normal progression and secondary sexual characteristics. Puberty can be a particularly stressful time for individuals with BBS, regardless of their sex, and, where possible, referral to a counsellor with experience in this field can be of help.

Adult male patients with hypogonadism may be prescribed testosterone replacement by an endocrinologist. This may take the form of either injections or daily gel preparation. While this may help with sexual function and libido, an important benefit is likely to be in the preservation of bone mineral density and reducing the risk of fractures. Although affected males may be infertile, several have fathered children.

Female Reproductive System

Hypogonadism is much less frequent in women, however, structural anomalies of the female genitourinary tract are documented, including vaginal atresia (abnormally closed or absent vagina), hydrometrocolpos (collection of watery fluid in the uterus and vagina), ectopic urethra (urethra that terminates somewhere other than the bladder), hypoplasia (underdevelopment) of the uterus, ovaries and fallopian tubes, and septate vagina (vagina that is

divided). Large ovarian cysts have also been found in pre-pubertal girls with BBS. Girls with BBS tend to go through puberty at the normal time, and although there is little evidence to suggest secondary hypogonadism (contrasting with men), polycystic ovary syndrome (PCOS) may be present in up to 20% of women with BBS. This can lead to problems with menstrual irregularity, acne, and excessive hair growth, notably on the face. For those patients who are considering fertility, polycystic ovary syndrome needs to be considered should there be a delay in the onset of menstruation or with conception. Several females with BBS have given birth successfully to healthy children.

Thyroid function

Thyroid function tests are normal in most patients with BBS, however, approximately 25% of patients may have thyroid test abnormalities. Most commonly, this is an underactive thyroid gland (hypothyroidism) which can cause tiredness, lack of energy, feeling cold, constipation and difficulty in losing weight. In the majority of cases, this is a very mild abnormality (often termed 'sub-clinical hypothyroidism') and may not require any treatment. In some individuals however, thyroid hormone replacement treatment is needed. Patients with BBS should have an annual blood test to measure thyroid function.

Sign post:

Patients affected by any of these issues should speak to their GP for a referral to an endocrinologist and/or counsellor where appropriate.

Patients of the BBS multi-disciplinary clinics should speak with their endocrinologist or BBS clinical psychologist.

Diabetes

The risk of diabetes associated with BBS increases with age (as it does in people without BBS). As such, accurate estimates of the risk of diabetes are difficult to make, but depending on age, it is seen in 6% to 48% of people living with BBS.

It rarely presents before adolescence and in this age group occurs in those with the highest BMIs. A higher proportion of patients may have abnormalities of glucose tolerance and regular screening for the development of diabetes is recommended.

The underlying BBS ciliopathy and high BMI contribute increased risk factors for diabetes mellitus. This can be controlled with combinations of diet and medicine including metformin and insulin. Newer therapies, for example, GLP-1 analogues, DPP-IV inhibitors and SGLT2 inhibitors are also potentially useful to try to optimise glycaemic control. Regular diabetes assessments are important and should be carried out at least annually. Preventable diabetic retinopathy can lead to rapid deterioration of vision already compromised by the rod-cone dystrophy.

Alongside monitoring for complications of diabetes, it is important to control blood pressure adequately and all patients with diabetes over the age of 40 should be considered for lipid lowering therapy. Both measures are aimed at decreasing long-term cardiovascular risk.

Endocrine Assessment

At diagnosis:

- Measure height and weight- calculate BMI
- Hormone levels: including testosterone, gonadotropins FSH and LH (over 8 years in girls and 9 years in boys)
- Pelvic ultrasound examination (females)
- Liver chemistry (including AST, GGT and ALT)
- Fasting lipid profile, including triglycerides
- Fasting plasma glucose (FPG)
- Glycated haemoglobin
- Thyroid function tests (including TSH and free T4)

Follow-Up: (at least annual, but may be more frequent)

- Measurement of weight, height, and calculation of BMI
- Annual thyroid function test (including TSH and free T4)
- Liver chemistry (including AST and ALT)
- Fasting lipid profile, including triglycerides
- Fasting plasma glucose or glycated haemoglobin

Sign post:

Diabetes UK
www.diabetes.org.uk
0345 123 2399

Additional Features

Speech, Language and Communication

Children with BBS may present with speech, language and communication differences, and development of these skills can be delayed. Though the literature is limited, it is documented that there is a wide range of communication abilities within the BBS population, varying from typical communication skills to more significant difficulties. BBS children may present with one, or a variety of needs relating to their speech, language and communication which are discussed below:

Understanding of language

'Receptive language skills' describes how children learn to develop their understanding of words, phrases and sentences from verbal information, and alternative communication approaches such as gesture and written word. BBS children vary in the development of their receptive language skills; some may find it difficult to follow information or instructions and develop their understanding of new words, and some may find it difficult to follow the narrative of a story or understand questions. Receptive language differences can mean that children find it difficult to engage in classroom tasks and access the curriculum.

Use of language

'Expressive language skills' describes how children start to communicate their wants, needs, thoughts, feelings and reactions. BBS children vary in their expressive language skills; for some, they may need more time to use new words. They may find it difficult to tell a story, respond to questions or use grammatical markers. Others may use alternative communication methods such as visuals, signing or communication devices.

Social communication

The social use of language describes how we start to understand how to successfully interact with others in various environments. This may be by showing interest in others, taking turns and engaging in shared play activities, using facial expression, gesture and non-spoken communication. BBS children can present with social communication differences as well as possible sensory needs. Some children are neurodivergent and require adults around them to adapt their communication and environment, to maximise their opportunities to thrive and best support their needs.

Speech

Speech sound development describes how children acquire different speech sounds as they grow. Some BBS children may present with a speech delay, and some may not demonstrate early babble. Some BBS children may present with less typical speech patterns such as a speech sound disorder or Childhood Apraxia of Speech (CAS). Some children can present with differences in their resonance, prosody (rhythm and melody) and voice pitch.

Treatment

A referral to a local speech and language therapy service should be made if there are concerns regarding a child's speech, language or communication development. An assessment of the child's communication can then be completed, and specialist advice and treatment can be provided as early as possible.

Hearing differences, if present, can contribute to a child's speech and language development. Children with fluctuating glue ear or a mild hearing loss can find it very difficult to acquire speech sounds in isolation and within language. Regular hearing tests with ensure that children are monitored regularly, and any active management can be implemented early.

Useful Links:

Speech and Language UK
www.speechandlanguage.org.uk
020 7843 2510

Afasic
www.afasic.org.uk
0300 666 9410

> **Of course the two things which inspire me the most to carry on with strength and determination are my children, two bright, funny, intelligent, kind and loving young people who have enriched our lives and made us proud parents.**
>
> Parent

Seizures/Epilepsy

The prevalence of seizures and epilepsy in BBS is reported as being much higher than previously recognised and significantly higher than in the general population, however the reason behind this is still unclear. In the majority of cases, seizures stop before adulthood.

Seizures can affect people in different ways including a loss of awareness, unusual sensations and jerking and shaking of the body. Seizures can occur when awake or asleep and sometimes there are triggers such as tiredness.

Patients or parents should seek urgent medical help if they suspect they or their child has had a seizure. A referral to a neurologist would be appropriate for investigations and tests.

Sign post:

Epilepsy Action
www.epilepsy.org.uk
0808 800 5050

Epilepsy Society
www.epilepsysociety.org.uk
01494 601 400

Neurological Abnormalities

Ataxia is the term for a group of disorders that affect co-ordination, balance and speech and is reported in a significant number of individuals with BBS. Hypertonia, a condition marked by an abnormal increase in muscle tension and a reduced ability of a muscle to stretch, is also commonly reported, as is low muscle tone.

Some limited brain MRI studies have shown that some patients with BBS have smaller, unusually formed hippocampi and that there is a tendency towards decreased overall white matter. However, it is not clear if or how this affects neurocognition. A number of patients with BBS report difficulties with memory function.

People with BBS may have an impaired sense of smell (anosmia) due to changes in the region of the brain responsible for processing smell, called the olfactory bulb.

Hearing

Conductive hearing loss, almost always caused by glue ear, affects many children with BBS. This often resolves spontaneously or, with treatment, by adulthood. A minority of patients with BBS have sensorineural deafness.

Treatment

Glue ear management: myringotomy tubes and/or hearing aids.

Assessment

At diagnosis:

- Audiogram, audiometry, tympanogram
- Auditory evoked potentials

Follow-up:

- Yearly examination: audiometry
- Detect glue ear (acute and chronic otitis media) which can lead to conductive hearing loss

Digestive System

A survey undertaken by BBS UK indicates that gastrointestinal problems are a common issue in BBS and this is backed up by anecdotal evidence from patients attending the BBS multi-disciplinary clinics. Commonly reported symptoms include constipation, diarrhoea, bloating and cramping and further research is needed in this area. Individuals who are experiencing difficulties should consult their GP who may wish to refer for investigations or dietetic support. Patients attending BBS clinics should discuss their symptoms with a member of their clinic team.

Hirschsprung's disease occurs more frequently in Bardet-Biedl syndrome than in the general population, but the incidence is unknown. The condition is present at birth (congenital) as a result of missing nerve cells in the muscles of the baby's colon. A new-born who has Hirschsprung's disease usually cannot have a bowel movement in the days after birth. In mild cases, the condition might not be detected until later in childhood or rarely as an adult, presenting as a swollen abdomen, poor appetite and constipation that does not respond to any of the usual treatments.

Orthopaedic Abnormalities

Examination and early identification of scoliosis (curvature of the spine), genu valga (knock-knee), genu vara (bow-leg) and pes planus (flat-foot) will allow for early intervention/treatment and may avoid unnecessary discomfort.

Physiotherapy and orthotic referral should be considered; early orthotic intervention and supportive footwear and/or insoles will ease discomfort and protect feet against further damage.

Liver

Fibrocystic liver disease (problem with the biliary system) and fibrotic liver disease (scar tissue build-up) have been reported in a very small number of cases of BBS. Co-existent metabolic dysfunction-associated steatotic liver disease (MASLD) can evolve over many years, driven by obesity and type 2 diabetes. It can manifest within a spectrum of disease that ranges from simple fat accumulation that has few, if any clinical consequences, to inflammation (metabolic-associated steatohepatitis: MASH) and in some cases scarring that can lead to liver cirrhosis. It is progressive in some, but not all, patients. Managing the condition relies on staging the severity of steatotic liver disease (SLD) and treating the conditions that predispose to it, namely reducing weight and optimising blood sugar control in patients with diabetes.

Liver Assessment

- Measurement of plasma ALT, AST and GGT concentration
- Liver ultrasonography

Cardiovascular

Hypertension (high blood pressure) is a common feature of Bardet-Biedl syndrome and often presents in childhood or early adulthood. It may occur as a result of other features contributing to metabolic syndrome in BBS, in particular obesity. It is imperative that hypertension is treated, since it can contribute to renal deterioration and prematurely stiffening of blood vessels, predisposing to cardiovascular disease, such as a stroke or heart attack. Both lifestyle modification and pharmacological intervention are encouraged.

Congenital heart disease (present from birth) has been reported in a minority of BBS patients. Dextrocardia (heart points to the right instead of the left), and occasionally complete situs inversus (where organs are positioned opposite to normal position) have been reported but these are rare.

Cardio Assessment

- Blood pressure monitoring
- Auscultation
- ECG
- Echocardiography

Joints

Many patients with BBS report pain in weight-bearing joints, probably because of osteoarthritis. However, a small proportion also have abnormally hypermobile or lax joints, the cause of which is unknown, but may be associated with hypotonia (decreased muscle tone).

Dental

Some people with BBS have unusually short tooth roots, especially of the front lower teeth. Crowding of the teeth necessitating extraction is common. Many patients have a high-arched palate and occasionally enamel dysplasia (developmental defect of the enamel) is evident. These abnormalities can increase susceptibility to tooth decay and loss, and regular dental supervision is advisable.

Respiratory System

Asthma commonly affects individuals with BBS with prevalence reported at around 21%. Recurrent chest infections are also more common in people with BBS, especially in childhood. This may be associated with dysfunction of the respiratory cilia which line the airways.

Obstructive sleep apnoea (OSA) is common and related to excess weight, particularly around the neck. This may be compounded by low muscle tone and soft palate dyscoordination. All patients should be questioned about their quality of sleep and the presence of daytime somnolence (excess sleepiness). Carers and parents should be asked about snoring and episodes of stopping breathing while sleeping.

Evidence suggesting obstructive sleep apnoea should lead to referral for sleep studies and lung function assessments. It is important to treat obstructive sleep apnoea as it is a risk factor for medical problems later in life, especially cardiovascular diseases such as heart attacks and strokes. Many BBS patients report that treatment (e.g., nocturnal CPAP devices) results in major benefits such as increased activity, weight loss and lower blood pressure.

If there are concerns about respiratory difficulties, we recommend a referral to a respiratory specialist who can decide on the most appropriate next steps.

Parents and carers

For parents and carers of children with BBS, the diagnosis brings with it many questions and worries about what this means for them and their family. This can understandably cause stress and anxiety, and adjusting emotionally can take time. Having a child with BBS can be hugely rewarding and frequently takes people's lives in unexpected directions. It can be helpful to take the journey one step at a time, take pleasure in the joyful moments and seek support when it is needed.

BBS is a complicated and lifelong condition that impacts in many ways, and so parents and carers can find it stressful to coordinate with the many health and support services involved.

The child or adult with BBS may have significant needs that may impact on parents, carers and their family. Siblings frequently have a large role in supporting people with BBS during childhood and into their adult lives. Therefore, of paramount importance is the need for parents, siblings and other family carers to receive support as well.

Key support is likely to come from family members and friends who may also need to learn about BBS. Parents and carers often find it useful to link with others who are in a similar situation. At times, parents and carers may struggle with their own mental health and so additional input may be warranted. There are many ways parents and carers can seek support:

- Ask GP to arrange a social services assessment and occupational therapy assessment.
- Ask GP about individual or couples counselling if more support is needed for low mood or anxiety or conflict arising in the family.
- Talk to staff at a local children's centre or child development centre about activities in the area for parents and carers.
- Contact BBS UK, it can sometimes help to talk to other parents and carers in the same situation.

- Keeping physically active and healthy is important for emotional and physical wellbeing.
- Parents and carers should try to find some time for themselves, it is important to have 'me' time, whether alone, with friends or each other. Relationships often suffer when caring for someone with a health condition and may benefit from support.
- It tends to be easier to cope with stresses when we have enough sleep, and so this can be important for caregivers to prioritise where possible.

The local Children with Disabilities Team or Social Worker will be able to help with claiming benefits, arranging support workers, carers or respite, and linking families with local services in the area

Signposting for carers

Contact
www.contact.org.uk
0808 808 3555

Listening Ear
Telephone support for parent carers looking for emotional support
www.contact.org.uk/help-for-families/listening-ear/

Carers UK
www.carersuk.org.uk
020 7378 4999

GingerBread
Support for single parents
www.gingerbread.org.uk
0808 802 0925

Family Fund
www.familyfund.org.uk
01904 550055

> **We sometimes need to find a little time for ourselves. This normally involves a bike ride, a swim, a run or simply spending time with friends. This time is important because it makes us function better as parents and my advice to any parents of newly-diagnosed Bardet-Biedl children, is to allow yourselves this time too, in whatever form it takes.**
> Parent

BBS Specialist Clinics

BBS UK, together with Great Ormond Street Children's Hospital, Guys and St Thomas' Hospital in London, and Birmingham Children's Hospital and Queen Elizabeth Hospital in Birmingham, are commissioned by NHS England to provide multi-disciplinary clinics for BBS patients. At each clinic, patients are seen by an ophthalmologist (eyes), nephrologist (kidneys), dietitian, clinical psychologist, geneticist, speech and language therapist and endocrinologist (hormones). The aim is to ensure patients receive specialised, expert attention and management with a focus on diagnosis, early intervention, and good health management. Telemedicine clinics are also available across the four centres and are offered in between face-to-face appointments, where appropriate.

BBS UK is an integral member of this valued multi-disciplinary team (MDT) and provides information and support to patients and their families and carers before, during, and after clinics. Our charity's pre-clinic involvement ensures that patients, their families and carers are well informed about the Syndrome and services, and feel well supported to attend. BBS UK Patient Liaison Officers attend each clinic, offering emotional and practical support to patients and their families. Post clinic, they provide ongoing support and liaise between patient and hospital where required and generally provide an ongoing point of contact.

It is recommended that all patients with a diagnosis of BBS should be referred to one of these centres.

Further information, including a link for referrals can be found on the BBS UK website:
www.bbsuk.org.uk/bbs-clinics-service
Or contact: **admin@bbsuk.org.uk**

> **Over the years, I have had the privilege of meeting many children, young people, and adults with BBS, and I am constantly impressed by their resilience and determination. Within our community, we have seen so much achievement, across business, sport, the workplace, music, art, and academia—with the right support in place, there are no limits to what can be achieved.**
>
> BBS UK Staff Member

Bardet-Biedl Syndrome UK

BBS UK is the only charity in the UK providing information and support to those diagnosed with BBS, their families and carers, from diagnosis and beyond. The Charity is a lifeline to over 700 individuals, their families and carers, providing essential support, information, and advocacy services. BBS UK also focusses on raising awareness of the Syndrome amongst medical professionals and wider society, and supporting the medical and scientific community with research.

> **I got in touch with two wonderful people from the BBS charity. They were a huge source of support to me and put me in contact with even more families. Sometimes I think the hardest thing for people is thinking that you are alone and that is where the Charity comes into its own.**
>
> Parent

BBS UK provides a Clinic Support Service and an Advice Service, offering invaluable support and advocacy to individuals living with Bardet-Biedl Syndrome (BBS) and their families. The Charity's annual conference brings members together to learn about the latest research and treatments while connecting with others who share similar experiences. Additionally, BBS UK maintains a website and an active Facebook page, fostering a supportive community which self-supports, with members often helping each other through shared challenges.
For more information, visit **www.bbsuk.org.uk** or email **admin@bbsuk.org.uk.**

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bbs uk
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