



Introducing **Bardet-Biedl Syndrome**





Introducing **Bardet-Biedl Syndrome**

This booklet provides a clear and accessible introduction to Bardet-Biedl syndrome (BBS) for those diagnosed with BBS, their families, carers and wider support network. More detailed information can be found in the BBS UK Medical Information Booklet (accessed via QR code below), from which this information has been adapted, and where associated references can be found.

A list of useful organisations is included at the back of the booklet.



What is Bardet-Biedl syndrome?

Bardet-Biedl syndrome (BBS) is a rare inherited disorder, affecting approximately 1 in 100,000 people. It is a condition that can affect a number of different body systems to varying degrees. Symptoms may include visual impairment, obesity, kidney abnormalities, developmental delay, speech and language difficulties, extra fingers and/or toes, and learning difficulties. It is important to remember that not everyone who has BBS will have all these symptoms, and the effects will be different for everyone; some may have mild symptoms and others may have more severe symptoms.

Over 25 BBS genes have been identified to date (2026) and are named BBS1, BBS2, and so on; it is likely that there are still more genes to be identified. Individuals who have the same BBS gene are not necessarily affected in the same way, for example, one may have extra digits whereas another may not. There is still much to learn about BBS.



Why do I have BBS? Why does my child have BBS?

BBS is an inherited disorder. People are born with the syndrome and will have it throughout their lives. People do not catch BBS, nor can they grow out of it.

For someone to be born with BBS, both parents will usually be *carriers* of the same BBS gene. Where both parents are carriers of the same BBS gene, there is a 1 in 4 chance with *every* pregnancy of passing on both copies of the gene to the baby, resulting in the baby having BBS.

Families from all communities and ethnic backgrounds can be affected by genetic conditions, including BBS. As an inherited disorder, BBS is more common in communities where parents are related to each other, for example, cousins. This is because relatives are more likely to share the same genetic changes, increasing the chance that both parents carry the same altered BBS gene.

Where BBS is confirmed genetically within a family, carrier testing may be available for at-risk adult relatives to help support informed family planning decisions. Prenatal testing and pre-implantation genetic diagnosis (PGD) may also be options for some families where the BBS gene has been identified.





What are the symptoms of BBS?

As explained in the introduction, it is very important to remember that not everyone who has BBS will have all these symptoms, and the effects will be different for everyone; some may have mild symptoms and others may have more severe symptoms.

Eyes

The eye condition seen in BBS is called rod-cone dystrophy. It is similar to, and sometimes diagnosed as 'retinitis pigmentosa', which is a broad term used to describe a range of genetic eye conditions.

Rod-cone dystrophy often begins with difficulty seeing in low light or at night, usually during the primary school years. Over time, this is typically followed by a gradual loss of peripheral (side) vision. In some individuals, visual symptoms may progress more slowly, with significant visual changes occurring later in the teenage years or beyond.

As vision narrows and central vision becomes more affected, many young people reach the threshold for registration as severely sight impaired (blind) during their teenage years, with an average registration age of around fifteen years.

Research is ongoing into possible future treatments, including gene therapy. In the meantime, it is important to prioritise eye health and make the best use of remaining vision. Referral to organisations that support people with visual impairment and to a low vision clinic is recommended. Mobility training and low vision aids, including digital systems, can help improve independence and confidence.



Obesity

Many children with BBS gain weight rapidly during their first year. In adulthood, maintaining a healthy weight can remain difficult, and many people with BBS live with obesity, which may increase the risk of health problems such as diabetes and heart disease.

Hyperphagia (excessive hunger) is common in BBS and can make weight control more difficult. There is no single treatment for obesity; however, with the right support, progress is possible through a healthy, well-balanced diet and active lifestyle. Early referral to a registered dietitian is important, and attendance at a specialist BBS clinic is strongly advised. This enables advice tailored to an individual's needs to be provided by an expert clinical team.

Research into treatments for obesity and excessive hunger in BBS is ongoing, and new treatments may be discussed within specialist BBS clinics where appropriate.

Extra Fingers/Toes

Extra fingers and/or toes may be present at birth in some children with BBS. These are usually surgically removed within the first year of life, and small skin tags can often be tied off shortly after birth.

Renal (Kidneys)

Although BBS can affect the kidneys in several ways, severe kidney problems are uncommon. Minor kidney problems and high blood pressure are more common and, in general, if kidney function is normal at the age of twenty, it is unlikely to get worse in later life. In those who experience end-stage kidney failure, transplantation or dialysis is offered; kidney failure is not sudden and so there is usually plenty of time to plan.

Some people with BBS may pass urine very frequently, produce large amounts of urine, or need to pass urine during the night. Others may pass urine less often, but in larger amounts. Passing urine regularly, even if there is no strong urge to go, can help protect bladder and kidney health.



Development, Learning, Social and Emotional Wellbeing

Developmental delay is common in children and young people with BBS. Sitting, standing and walking may happen later than usual, and speech and language, movement and hand skills may also be affected. Some children may find it hard to manage their feelings (emotional dysregulation) and may show challenging behaviour. Many children and young people have learning or communication difficulties, and some may have autism spectrum differences.

With the right support and low vision aids, children and young people can fully engage and thrive in their learning environment. An Education, Health and Care Plan (EHCP) can help make sure they get the support they need. If there are concerns about emotional or behavioural wellbeing, support can be accessed through school or a GP, who may refer to appropriate local services, including CAMHS (Child and Adolescent Mental Health Services) where needed.

Adults with BBS may experience learning, communication and emotional difficulties, and some may be affected by anxiety or low mood, particularly if they are worried about their health, feel isolated, or find changes in routine challenging. Support is available through a GP, local authority services, and BBS UK.

Endocrine (Hormones)

BBS can affect how the body produces and uses hormones. Hormones help control growth, puberty, weight, energy levels and blood sugar.



Some children and young people with BBS may experience changes in the timing of puberty, and some adults may have hormone-related conditions. Thyroid problems, which can affect energy levels, weight and overall wellbeing, are also more common in people with BBS.

Some people with BBS may develop diabetes, particularly in adulthood, and regular health checks are important. Hormone-related conditions can usually be managed with the right medical support. Monitoring and advice are provided through GPs and specialist BBS clinics where needed.

Respiratory (Breathing and Chest)

BBS can affect breathing and chest health in some people. Asthma and chest infections appear to be more common in people with BBS, particularly during childhood.

Sleep and breathing problems, such as obstructive sleep apnoea, can also occur in both children and adults. These may cause snoring, poor sleep, or feeling very tired during the day.

Breathing and sleep problems can usually be managed with the right medical support. Anyone with concerns about breathing, sleep, or frequent chest infections should speak to their GP or BBS clinic team.



Speech and Language Difficulties

It is very common for first words to emerge late in children with BBS. Difficulties with speech and language may range from mild to severe, and there can be problems across a range of communication skills or just in one area. A referral to speech and language therapy should be made at the first sign of difficulties.

Additional Features

BBS may affect other parts of the body and may also be linked to some health conditions, including:

- The digestive system
- The heart
- The liver
- The joints
- Hearing (usually caused by glue ear in childhood)
- Seizures and epilepsy

More information can be found in the BBS UK Medical Information Booklet.



What treatments are available for BBS?

There is currently no cure for BBS. However, research has led to the development of a targeted treatment that can help manage hyperphagia (excessive hunger) and obesity in some individuals with BBS. Access to this treatment is through specialist NHS BBS clinics and is determined in line with current NICE guidance.

Ongoing research, including gene therapy studies, continues to explore additional potential treatments.

Once diagnosed, regular monitoring can be put in place to support overall health and identify medical issues at an early stage. Individuals have access to specialised NHS multi-disciplinary clinics (for children and adults) held in four centres in London and Birmingham. Bardet-Biedl Syndrome UK (BBS UK) provides patient support, advocacy and facilitation services on behalf of the NHS. At each clinic, individuals see consultants and therapists who specialise in the different aspects of the syndrome and who have experience in managing BBS.

Who are BBS UK?

Bardet-Biedl Syndrome UK (BBS UK) is the only registered charity in the UK supporting those who have Bardet-Biedl syndrome, their families, carers, and interested professionals. Services include:

- Advice Service
- Clinics Support Service
- Annual Conference
- Information booklets
- Newsletters and Conference Report

If you would like to access our support services or would like further information about BBS, BBS UK and the support available, please go to www.bbsuk.org.uk or email admin@bbsuk.org.uk

Further resources are available to download from www.bbsuk.org.uk:

- BBS UK Medical Information Booklet
- BBS UK Booklet for Schools and Colleges
- My Life, My Future!
(transition handbook for young people)
- How Can We Help?



Signpost

BBS UK

Information, advice, clinic support and community support for people with BBS and their families
www.bbsuk.org.uk
admin@bbsuk.org.uk

RNIB

Support, advice and information for people with vision impairment
www.rnib.org.uk
0303 123 9999

Guide Dogs

Support with mobility, independence and confidence for people with vision impairment
www.guidedogs.org.uk
0118 983 5555

LOOK

Support for children and young people with vision impairment and their families
www.look-uk.org
07464 351 958

VICTA

Support for children and young people who are blind or partially sighted, and their families
www.victa.org.uk
01908 240 831

National Kidney Federation

Information and support for people with kidney conditions
www.kidney.org.uk
0800 169 09 36

National Autistic Society

Information and support for autistic people and their families
www.autism.org.uk
0808 800 4104

Diabetes UK

Support and information for people living with diabetes
www.diabetes.org.uk
0345 123 2399

Speech and Language UK

Information and support for speech, language and communication needs
www.speechandlanguage.org.uk
020 7843 2510

Mind

Information and support for mental health and emotional wellbeing
www.mind.org.uk
0300 123 3393



Acknowledgement

This booklet was produced by BBS UK and is based on the information contained in the BBS UK Medical Information Booklet. We would like to extend our grateful thanks to the clinicians from the BBS specialist clinics teams for their contribution to this publication.

This booklet is available in alternative formats, for more information, email admin@bbsuk.org.uk.



Bardet-Biedl Syndrome UK

Registered address:

43 Balton Way, Dovercourt
Harwich, Essex, CO12 4UP

Registered Charity 1181244

www.bbsuk.org.uk

admin@bbsuk.org.uk

Published date: April 2026

Review date: April 2029



YouTube

