bbs uk
Bardet-Biedl Syndrome UK

Introducing **Bardet-Biedl Syndrome**

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The aim of this booklet is to provide a basic summary of Bardet-Biedl Syndrome (BBS) and in doing so, perhaps answer the most commonly asked questions from those diagnosed with BBS, their parents/ carers and extended support network. More comprehensive information can be found in the BBS UK Medical Information Booklet, from which this information has been adapted and 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 is a rare inherited disorder affecting approximately 1 in 100,000 babies born. 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. Not everyone who has BBS will have all of these symptoms, and the effects will be different for everyone; some may have mild symptoms and others may have more severe symptoms.

22 BBS genes have been identified to date (2020) 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; 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 it and will have it throughout their life. People do not catch BBS, nor can they grow out of it.

For someone to be born with BBS, both their parents will usually be *carriers* of BBS. 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 all ethnic groups can be affected by genetic disorders, including BBS. As an inherited disorder, BBS is more common in communities where couples are blood relatives, for example, cousins. This is because family members, including extended family members, are more likely to share the same genes, including BBS genes where present.

Where BBS is confirmed genetically within a family (e.g. siblings) a simple carrier test is possible in at-risk adult relatives to help them make informed family planning decisions. The option of Prenatal testing and Pre-implantation Genetic Diagnosis (in some families) is available 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 of these symptoms, and the effects will be different for everyone; some may have mild symptoms and others may have more severe symptoms.

Rod-Cone Dystrophy

Rod-Cone Dystrophy is similar to, and sometimes diagnosed as, 'Retinitis Pigmentosa'.

Rod-Cone Dystrophy usually causes night blindness first, which tends to happen during primary school years. This is usually followed by a gradual loss of peripheral vision. The average age of registration of blindness is fifteen years.

Referrals should be made to a low vision clinic and organisations aimed at supporting the visually impaired. Mobility training and low vision aids including digital systems can improve independence and confidence.

Obesity

Many children who have BBS gain weight rapidly during their first year. The majority of adults who have BBS are obese which, untreated, can lead to multiple health problems, for example, diabetes and heart disease.

There is no single-treatment approach for obesity, but those with BBS can successfully manage their weight by following a healthy well-balanced diet and active lifestyle. Early referral to a registered dietitian is important, and attendance at a BBS clinic is strongly advised. This enables advice tailored to an individual patient's needs to be given by an expert clinical team.

Renal Abnormalities

Although BBS can affect the kidneys in a number of ways, serious kidney problems are rare. Many individuals who have BBS find they are often thirsty and pass urine frequently and in large amounts; this is usually because they are unable to concentrate their urine.

Minor kidney problems and high blood pressure are more common and, in general, if kidney function is normal at the age of twenty then 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.

Learning and Emotional Difficulties and Developmental Delay

Developmental delay is common; sitting, standing and walking may be delayed and speech and language development, general motor skills and fine motor skills may be affected. Children may also be affected by emotional immaturity and challenging behaviour.

Children and young people who have BBS often have some degree of learning disability but do well in mainstream education if good support and low vision aids are available. Learning difficulties and autism should be assessed to ensure appropriate help and support can be put in place. An Education, Health and Care Plan (EHCP) will ensure the child is fully supported. If a child or young person is having difficulties with their emotional or behavioural wellbeing, a referral to CAMHS (Child and Adolescent Mental Health Services) can be requested through a GP.

Depression and anxiety commonly affect adults with BBS and contact should be made with the GP for support with accessing appropriate local services.

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.

Extra Fingers/Toes

Extra fingers and/or toes are usually surgically removed within the first year of life; skin tags can be tied off at birth.

Additional Features

Bardet-Biedl Syndrome may also affect the heart, liver, joints, hearing, digestive system and respiratory system. Seizures and epilepsy are also reported as being more common in BBS than in the general population, however in most cases, stop before adulthood.

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

What treatments are available for Bardet-Biedl Syndrome?

There is currently no cure for Bardet-Biedl Syndrome, however research is providing hope of future treatments and therapies.

Individuals have access to specialised NHS multi-disciplinary clinics held in four centres in London and Birmingham. Bardet-Biedl Syndrome 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 of BBS.

Who are **BBS UK?**

Bardet-Biedl Syndrome UK is the only registered charity in the UK supporting those who have BBS, their families and carers. In addition to providing the BBS UK Clinics support service, the Charity produces information booklets and resources, twice-yearly newsletters, an annual Weekend Family Conference and a Conference Report. The Charity also organises events designed to reduce isolation and increase knowledge and understanding across its membership group.

If you would like further information about Bardet-Biedl Syndrome, BBS UK or BBS UK Clinics, please go to **www.bbsuk.org.uk** or email **admin@bbsuk.org.uk**

Further resources 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)
- **Who We Are and How We Can Help!**

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

Sign Post

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RNIB
www.rnib.org.uk
0303 123 9999

Guide Dogs
www.guidedogs.org.uk
0118 983 5555

VICTA
www.victa.org.uk
01908 240831

Look
www.look-uk.org
07464 351 958

Sense
www.sense.org.uk
0300 330 9250

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

The National Autistic Society
www.autism.org.uk
0808 800 4104

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

I Can Help
Information services that provide help and advice to parents and practitioners about speech, language and communication.
www.ican.org.uk
020 7843 2544

Talking Point
information for parents and carers to help their children develop communication skills.
www.talkingpoint.org.uk

Afasic: voice for life
Information and support about speech, language and communication needs.
www.afasic.org.uk
0300 666 9410

Dyspraxia Foundation
www.dyspraxiafoundation.org.uk
01462 454986

Makaton Information Website
www.makaton.org

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Bardet-Biedl Syndrome UK

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