Meeting other families living with BBS is so important and was a wonderful added bonus of traveling to Marshfield. I feel my girls received the best care possible. We plan to return every year.”
- parent
Dear family and friends,

We are dedicated to improving the health and quality of life for individuals with Bardet-Biedl syndrome (BBS) here at the Center of Excellence for Bardet-Biedl Syndrome. To achieve that mission we have established a three-fold purpose:

1. Provide comprehensive and compassionate care to individuals attending the BBS Multispecialty Clinic at Marshfield Clinic Health System, and to be a resource for all individuals with BBS and their health care providers regardless of their location or need.

2. Maintain a comprehensive natural history registry called the Clinical Registry Investigating BBS (CRIBBS). This international registry offers families, clinicians and researchers unsurpassed knowledge on health patterns in BBS.

3. Promote research by collaboration with researchers across the world at universities, pharmaceutical companies and public organizations such as the National Institutes of Health to advance knowledge and promote translational research.

I hope that the information we provide for you in this packet will help you in your journey.

Sincerely,
Dr. Bob Haws
Director, Center of Excellence for BBS
WHAT IS BARDET-BIEDL SYNDROME?

BBS is a rare genetic disorder that affects many parts of the body. Common features of BBS include:

• Impaired vision
• Obesity
• Problems affecting urinary and genital organ function
• Chronic kidney disease
• Extra fingers and toes
• Problems with language, thinking, judgment and memory
• Endocrine disorders that may affect growth and development, metabolism, sexual function, reproduction and mood

BBS affects less than 3,000 individuals in the U.S. Because it is so rare, BBS may not be recognized early. This may delay the start of treatment and support for at-risk families.

In a given year, we need to take our child to numerous specialists. The BBS clinic gave us the opportunity to see specialists in a condensed amount of time, and the specialists offered new insights in our child's health.”
- parent

QUICK FACTS about Bardet-Biedl syndrome (BBS).

✓ BBS is a condition present from birth with no cure.
✓ BBS affects almost every organ system.
✓ BBS is inherited in an autosomal recessive manner (both parents passing one BBS gene to their child) and evolves slowly throughout the first decade of life, although there is a lot of variability.
✓ Those with BBS should have regular ophthalmology evaluations; monitoring of renal, liver, glucose, lipid and endocrine profiles; and weight and blood pressure measurements.
✓ A BBS diagnosis is based on four primary features, or it can be based on three primary features and two secondary features including:

PRIMARY FEATURES:
• Obesity
• Extra fingers/toes (polydactyly)
• Abnormalities of urinary tract
• Learning disabilities
• Rod-cone dystrophy
• Hypogonadism (which may include delayed puberty, genital abnormalities or abnormal endocrine testing of sexual hormones)

SECONDARY FEATURES:
• Speech delay/difficulties
• Strabismus, astigmatism, cataracts
• Short fingers/toes
• Developmental delay
• Excessive thirst/urination
• Awkward gait/imbalance
• Spasticity
• Diabetes mellitus
• Dental crowding, high arched palate
• Hepatic fibrosis
• Congenital heart disease
BBS specialty care at Marshfield Clinic Health System

A multispecialty clinic for children and adults with BBS is offered at Marshfield. There is no other clinic in the U.S. providing comprehensive, clinical evaluations and individualized treatment care recommendations for BBS. Families travel from around the world to participate in this clinic.

Each clinic is approximately three to four days and takes an innovative approach to improving care for patients with BBS. Our providers understand BBS and are interested in meeting the needs of patients and families it affects.

Pediatric BBS clinics may include:
- Child psychology
- Community resources
- Dental
- Dermatology
- Medical genetics
- Neuropsychology
- Nutrition
- Occupational and physical therapy
- Otolaryngology (Ear-Nose-Throat)
- Pediatric cardiology
- Pediatric endocrinology
- Pediatric gastroenterology
- Pediatric nephrology
- Pediatric neurology
- Pediatric ophthalmology
- Pediatric physical medicine and rehabilitation
- Pediatric speech pathology and audiology
- Sleep medicine
- Speech therapy

Adult BBS clinics may include:
- Cardiology
- Community resources
- Dental
- Dermatology
- Endocrinology
- Gastroenterology
- Gynecology
- Medical genetics
- Nephrology
- Neurology and sleep medicine
- Neuropsychology
- Nutrition
- Occupational and physical therapy
- Ophthalmology

Patients and families meet with a team of specialty providers, including a social worker, who helps identify local, state and national resources for BBS.

On the final day of the clinic, the BBS clinic director and clinical coordinator will meet with you to review the overall information and recommendations of the clinic.

Follow-up care

About three to four weeks after you attend the BBS clinic, a binder will be sent to you containing all physician notes and recommendations as well as contact information for each provider you were evaluated by. CD copies of all provider notes and radiology scans are sent to you, as well as the primary health care provider. A comprehensive care plan and recommendations are also sent outlining major points of the evaluations and specific individualized recommendations.

This complete summary with recommendations is vital to ongoing care. Many local providers that see you might not have another patient in their practice with this diagnosis. The additional information and recommendations assist them in providing optimal local care. Follow-ups are recommended with a primary care provider on an annual or bi-annual basis.
Hello everyone my name is Mikyla Mobley. I was diagnosed with BBS when I was about 15. I had already been diagnosed with retinitis pigmentosa (RP) when I was 13. Ever since I was a little girl, I knew that my vision was impaired. I never liked talking about my health problems, but in eighth grade I had to do a science project and I decided to do my presentation on my vision, to learn more about what was actually going on and to help my classmates understand. As part of my presentation, I brought some folding canes to class and let people try them. That helped people understand my situation.

Once I found out that I had RP, I was devastated because I thought that I was going to go blind the next day. I had no idea what to expect or how to handle the situation. I didn’t understand that my condition was degenerative and that no one knew for sure when I’d officially go blind. Once I understood that, I knew that I had to take one day at a time and that things would figure themselves out in the end.

The people that surround me have always told me that I was a happy person that didn’t seem like she had all these issues - that I didn’t show it. So, everyday I figured out a way to try to look at everything on the positive side and have more empathy for other people.

I’ve been participating in the BBS study since June of 2017. The study team has helped tremendously with reducing the risk of obesity-related diseases. Being part of the study gives me a sense of community, and helps me be positive about my future.

In the fall of 2018 I will be starting college at Edgewood College in Madison, Wisconsin. I hope to major in psychology and minor in criminal justice to pursue my dream of becoming a therapist and helping those in need in the community as well.

-Mikyla Mobley

Mikyla is a delightful young woman who loves to hang out with her friends, watch Netflix and sing, sing, sing."

- Colleen Dunahhee
We received Reed’s official BBS diagnosis at 10 months old. Our feelings about Reed’s diagnosis began with disbelief and denial, then progressed into guilt, worry, despair, envy, jealousy and then more guilt. Often we felt lonely and isolated from everyone - that no one else understood the feelings we had - the worry for our child about his future. Ultimately, we asked ourselves, “Why did Reed get this?”

Once the initial shock subsided, we searched for information regarding BBS and desperately to find other families like ours. Through our search, we found a blog about a family dealing with BBS, a BBS family association and a clinic specializing in the treatment and research for BBS. Learning from other families who have gone through similar experiences helped immensely. The advice and guidance shared about early intervention services, necessary medical providers and therapies, as well as the current and ongoing research felt like a blessing.

Looking back, my advice to BBS parents is to be proactive in finding and building the village for your child. Make connections with the BBS Family Association and Marshfield. You will learn so much from them, and to know you are not alone in this journey is a huge support and comfort. Take all the help offered and don’t be afraid
Appointment schedule
Depending on the patient’s specialty care needs, patients and families may have three to seven appointments with various specialty care providers each day of the BBS clinic. There is built in time for breaks between the appointments.

Opportunities to network
Patients and families have opportunities to connect throughout the BBS clinic. Upon arrival at Marshfield, you will be met by staff members who will be participating in your care throughout your time at the clinic.

A dinner gathering hosted for patients and families on the first night provides an opportunity to share information and learn more about BBS resources. A luncheon for patients and families is also hosted during the week.

Insurance coverage
Marshfield contracts with many insurance plans and networks. Please check with your employer or insurance company to see if you have access to health care services with us. We will work with your health insurance company to complete prior authorization when it is required.

Self-referral
Patients and families may self-refer to the BBS clinic or primary care providers can make the referral.

For more information or to make a referral to the Bardet-Biedl syndrome clinic at Marshfield, visit www.marshfieldclinic.org/bbs, or call 715-389-3235 or fax 715-387-5240.

“
Our son has BBS, but BBS is not our son. Our son is Reed, a beautiful, witty and fun-loving boy who starts kindergarten this fall.”

- Kevin and Kat Smith

to ask for more. Also, know it is ok to cry, to question why, to look at other children with envy, to feel frustrated and angry and all the many other emotions you will feel. We have all done it, but believe me you will get through this.

That’s not to say we don’t have bad days still. If we’re honest, we all do. Our son has BBS, but BBS is not our son. Our son is Reed, a beautiful, witty and fun-loving boy who starts kindergarten this fall. We love every little bit of Reed with all our hearts and we always will.

- Kevin and Kat Smith

The onus has always been on us to find the best specialists and coordinate our child’s care. This BBS clinic coordinates the care allowing us to concentrate on better care for our child.”

- parent
Clinical Registry Investigating Bardet-Biedl Syndrome

The Clinical Registry Investigating Bardet-Biedl Syndrome (CRIBBS) is the largest worldwide registry examining the long-term health of individuals with BBS. The mission of CRIBBS is to promote research that will improve the health and quality of life of individuals affected by BBS.

The research team at Marshfield Clinic Research Institute collects health information of individuals with BBS so it can be used in many scientific endeavors. The success of CRIBBS reflects the time, energy and vision of individuals with BBS, caregivers, families, therapists, doctors and scientists. CRIBBS serves as a valuable tool to learn about BBS and its impact on the health of a person. Information gathered in CRIBBS is used to:

- Help families better understand the condition.
- Help health care providers provide timely preventative and comprehensive care.
- Foster scientific efforts in understanding this complex disorder.
- Learn how BBS impacts the life and health of individuals.

The information gathered is shared with the BBS community and in professional journals. We believe this knowledge will spark the interest of scientists, private industry and government agencies - fostering high quality care and the development of more effective treatment strategies for individuals with BBS.

To register for CRIBBS, please visit www.bbs-registry.org or call 1-877-594-3499.

“Building and maintaining CRIBBS is a top priority of the association. There’s nothing we could do to have a greater impact on research and treatment development.”

- Bardet-Biedl Syndrome Family Association President Tim Ogden
We define ourselves in the roles we have. I am a wife, mom, grandma, sister and a psychotherapist. I live in Madison, Wisconsin with my husband, Jim. We have been married for 32 years. We met in Boulder, Colorado when we were both students at the university there. I was completing a degree in psychology and he was finishing his Ph.D. in literature. I was 25 then.

It was shortly after we met that I got a diagnosis of retinitis pigmentosa. I was a single mom with a small child. It was a devastating time for me, but I was determined to remain independent. My brother had vision loss and serious medical issues, and I had grown up seeing how few options seemed open to him. My parents coddled him rather than pushing him to be more independent. I was determined to adapt and not have the same restrictions.

We have learned over the years that siblings with BBS may not have the same symptoms, and this is the case in my family. Of the three of us who were affected by BBS, one died before the age of one, and my brother died ten years ago. I did not develop the more serious medical problems that they had. Carrier genes for my three remaining siblings have not been ruled out.

My daughter’s family lives close by and my granddaughter, now 14, is very much part of our household. My family of origin was often secretive about the problems that the three affected siblings had, and I have made sure that my own family talks openly about BBS and vision loss and that it is not seen as something that limits choices. We do things differently, and differences are accepted and encouraged.

Adapting to vision loss is an ongoing lifestyle challenge. I am a licensed marriage and family therapist in private practice. I earned my master’s degree in 1987 before we left Boulder. I went back to school over the next few years for postgraduate coursework needed for my license. I found that even working in a clinic was challenging, and for many years I was not very upfront about my disappearing vision. Seven years ago I left group practice and am now a solo practitioner, sharing an office with another professional. I have a mixed practice of both men and women, seen individually or as couples. I see clients three days a week, and the other days are set aside for phone calls to clients, dealing with insurance companies or typing up clinic notes.

There is a lot of documenting in this field, but technology makes it all possible for someone with very little vision. WindowEyes and an iPhone keep my practice up and running. I communicate with my clients by voice text and email. Siri makes it easy to set up appointments and enter phone numbers. I'm still an avid reader through audiobooks, and as needed I make book referrals to my clients. I have office staff, just like any other practice. Jim is my silent business partner with HIPAA clearance. He is the “eyes”. We work around each other’s schedules so that I do not have to take many cabs to work. Besides my practice as a psychotherapist, I am now facilitating support groups for adults with BBS, and a book discussion group is also starting. I have worked as a psychotherapist for the last 25 years and I have no plans of retiring any time soon.

-Ellyn Hunter
I am 35 and have Bardet-Biedl syndrome. I live alone and work in Boston as a legislative aide for a state representative. I help constituents with issues, answer emails, work with state agencies and answer phones for the representative. I got my current job because I did and do a lot of lobbying on behalf of the blind, and the representative thought I would be a good advocate for his constituents. I use a screen reader called J.A.W.S. for Windows, which makes my computer talk.

I have a master’s degree from Columbia University in education. I went to University of Massachusetts-Amherst for my undergraduate degree. I am fluent in braille. I am successful as an independent adult because my parents pushed me to be independent and I got training in skills of blindness when I was in school while losing my vision. I now have light perception. I also have some medical challenges due to Bardet-Biedl syndrome.

I love to read, ride tandem bike, sing, cook and take walks with my guide dog Bodie, who is a yellow English lab. I belong to a book club in Boston and ride my tandem bike with one of my parents in western Massachusetts when I can.

Having a rare disease can be difficult at times. One thing that has helped me for most of my life is the idea of putting one foot in front of another and being persistent.

- Kristina Constant
Bardet-Biedl Syndrome Family Association

This nonprofit association is dedicated to improving the lives of individuals and families affected by BBS. The mission is to provide a community of support for individuals and families; information about BBS; and promote science and research to improve the lives of individuals with BBS. Visit www.bardetbiedl.org for more information.

Additional resources and information

Medical genetic testing providers for BBS include:

Prevention Genetics, Marshfield, Wisconsin
www.preventiongenetics.com

Invitae, San Francisco, California
www.invitae.com

Casey Eye Institute, Oregon Health and Science University, Portland
www.ohsu.edu

Athena Diagnostics, Worcester, Massachusetts
www.athenadiagnostics.com

Support groups

Marshfield sponsors support groups for parents of children with BBS and adults with BBS. These support groups include a combined men’s and women’s group and a women’s group. Additional support groups are planned for the future. A book club is also available to anyone in the BBS community including parents and spouses.

For more information, please contact Sonia Suda at 715-389-3235.
Marshfield Clinic Health System is committed to helping individuals with Bardet-Biedl syndrome live healthier lives. Your support allows scientists and the medical team at Marshfield to pioneer BBS research and care. If you are interested in supporting families living with BBS, please contact MCHS Foundation at giving@marshfieldclinic.org or by phone at 715-387-9249 or 1-800-858-5220.