



Tanner (living with BBS) and Donna (Tanner's mother)

Understanding **BARDET-BIEDL SYNDROME (BBS)**

What you need to know about this rare genetic disease



Listen, Empower, Advocate,
and Drive change
for rare genetic diseases of obesity

TM

Rhythm®
PHARMACEUTICALS

What is Bardet-Biedl syndrome (BBS)?

Bardet-Biedl (pronounced BAR-day BEED-el) syndrome, also known as BBS, is a rare genetic disease of obesity

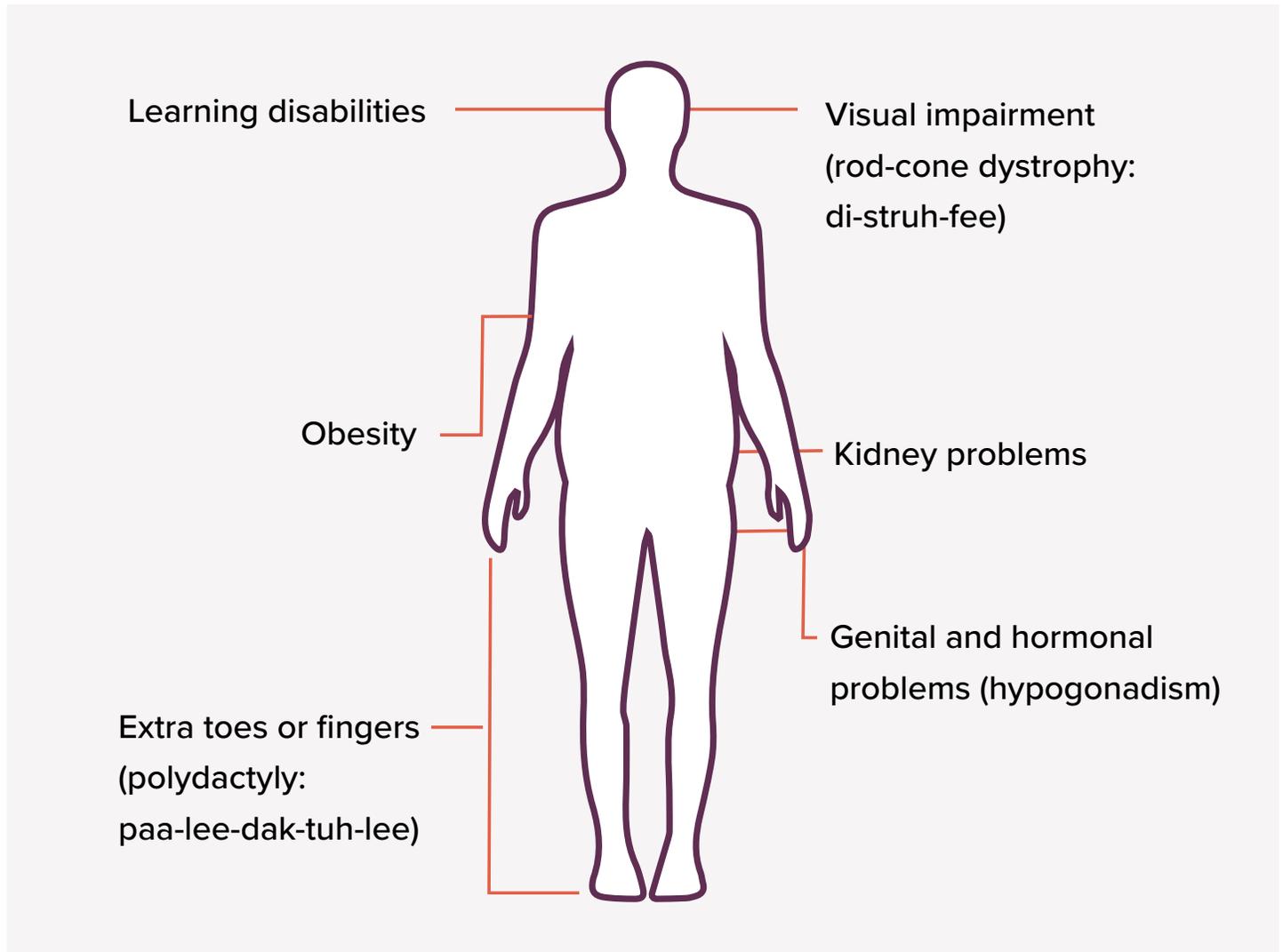
BBS has a wide range of symptoms, including early-onset obesity and hyperphagia (hai-pr-fay-jee-uh). Hyperphagia is insatiable hunger, also described as an intense, hard-to-control hunger. This brochure will help you learn more about these symptoms of BBS, why they occur, and how to better manage them for yourself or an individual you care for.

“ His whole world revolves around food. He is always thinking about it, always asking about it. ”
—Caregiver of a child living with BBS

Caregiver quotes have been edited for clarity and conciseness.

Signs and symptoms of BBS

The signs, symptoms, and severity of BBS can vary from person to person. This can make it hard for doctors to diagnose. The signs and symptoms of BBS may include:



Obesity is measured using what's known as body mass index, or BMI. BMI is a measure of body fat based on height and weight.

- In children, a BMI in the 95th percentile or higher is considered obesity
- In adults, a BMI of 30 or higher is considered obesity

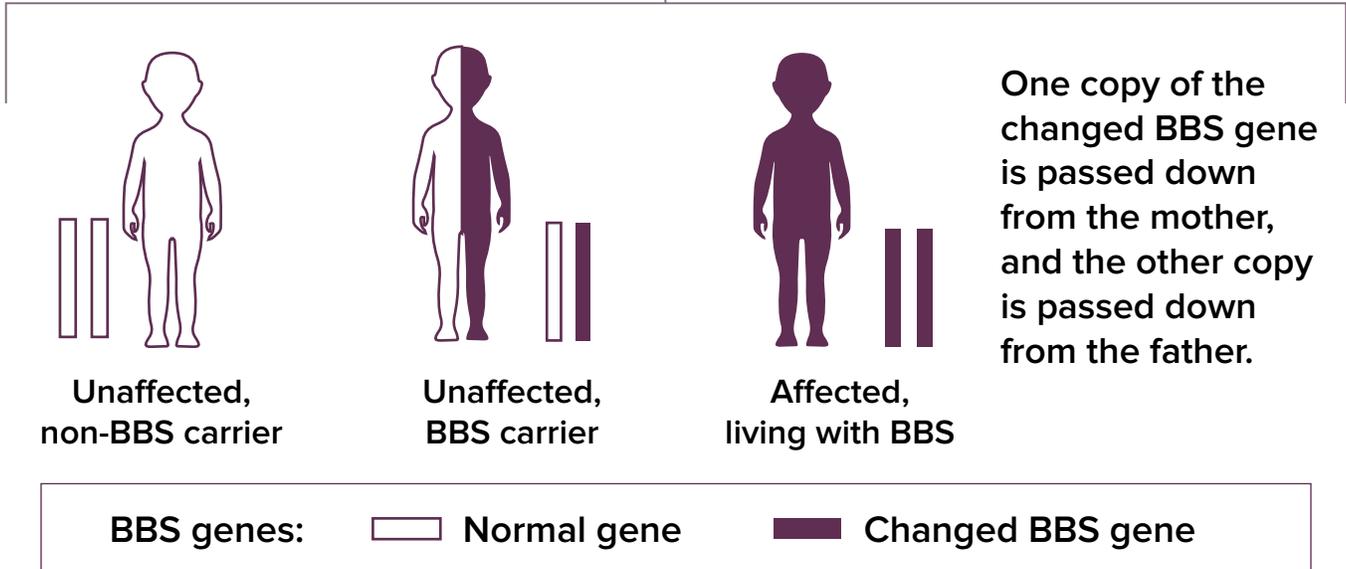
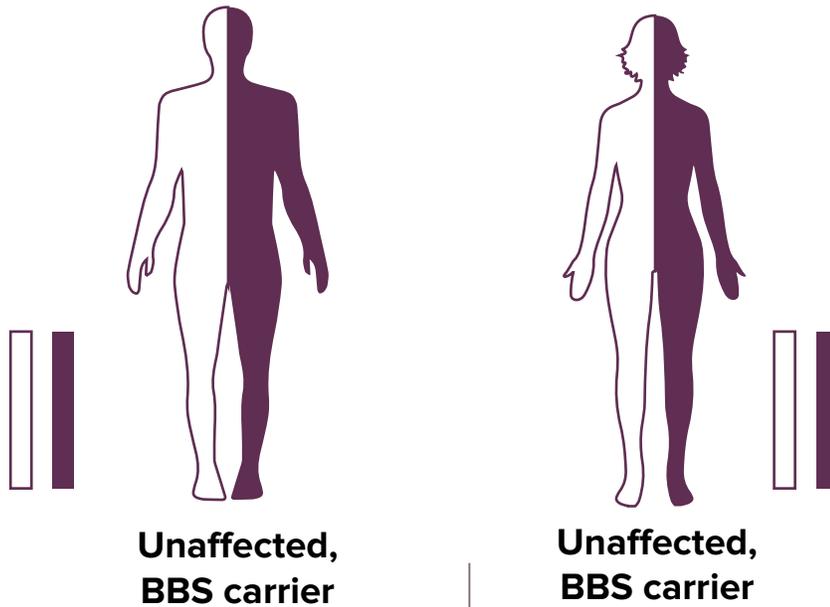
How is BBS inherited?

BBS is passed down from both parents through genes

Genes are your body's blueprint. They determine how your body grows, develops, and functions. Traits such as eye and hair color, and the likelihood of getting certain conditions, are passed from parent to child through genes.



BBS can be passed from parent to child if both parents have variants, or changes, in a BBS gene.

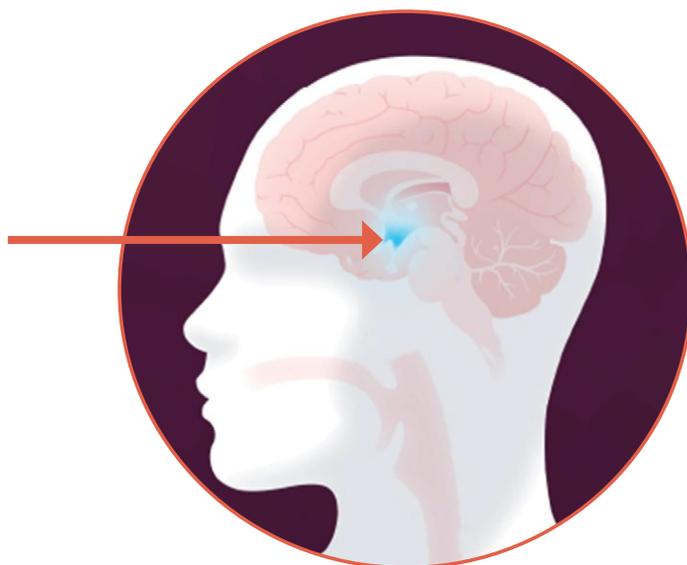


In people living with BBS, changes in BBS genes can stop a key part of the brain that controls hunger from working properly.

What causes intense, hard-to-control hunger and obesity in someone living with BBS?

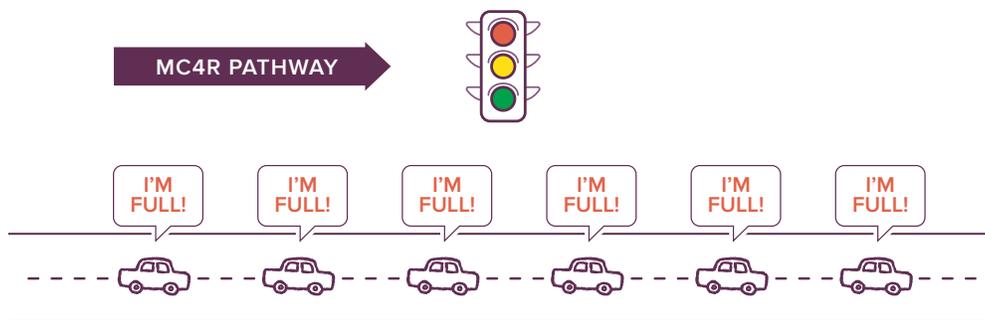
In someone living with BBS, the signals that turn hunger “off” don’t work correctly in the brain

The key part of the brain that communicates feeling hungry or full is called the hypothalamus.

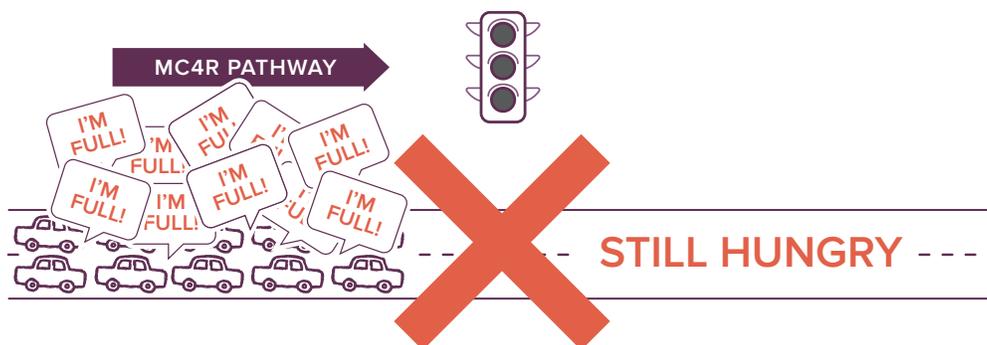


Think of the hypothalamus as a neighborhood within the brain

In this neighborhood, there are roads that allow cars to travel to and from the brain. One road is called the melanocortin-4 receptor, or MC4R, pathway. The cars on this road deliver messages between the body and the brain. These messages control hunger. On this road, there is a traffic light, or a BBS gene, that helps guide the cars to deliver their messages.



When the traffic light is working, the road is open, and the cars can deliver messages to the brain that the body is full.



When the traffic light is broken, it causes a traffic jam. Because of this, the cars have difficulty delivering messages to the brain that the body is full. Instead, the body believes that it is still hungry.

In people living with BBS, the traffic light is broken because of the change in their BBS gene. As a result, the MC4R pathway may not work properly. This can lead to intense, hard-to-control hunger and obesity.

How do you recognize intense, hard-to-control hunger in BBS?

Symptoms of intense, hard-to-control hunger can vary from person to person

Individuals living with BBS may experience one or more of the symptoms of intense, hard-to-control hunger. For some, these symptoms are constant. For others, they may happen from time to time.

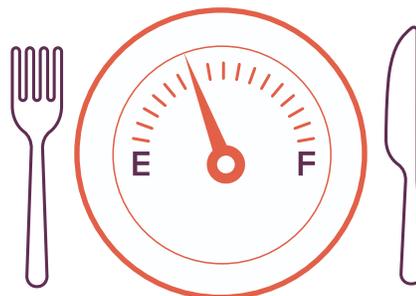
“ My daughter tells me,
‘I know you are going to feed me,
but it’s like I’m battling with my mind.’ ”
—Caregiver of a child living with BBS

Caregiver quotes have been edited for clarity and conciseness.

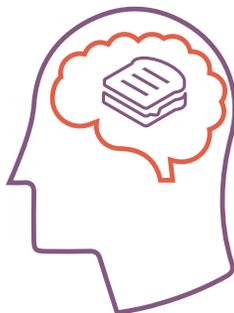
Symptoms of intense, hard-to-control hunger:



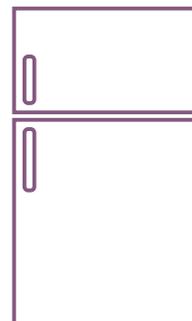
Taking a longer time
to feel full while eating



Feeling hungry again
right after a meal



Thinking about
food constantly



Food-seeking behavior
(sneaking or stealing food)

Intense, hard-to-control hunger can have a negative impact on daily life for people living with BBS.

What can make living with intense, hard-to-control hunger so challenging?

Having constant hunger can make daily life stressful

Intense, hard-to-control hunger can cause you or an individual you care for to think about food all the time and become very upset when food is unavailable.

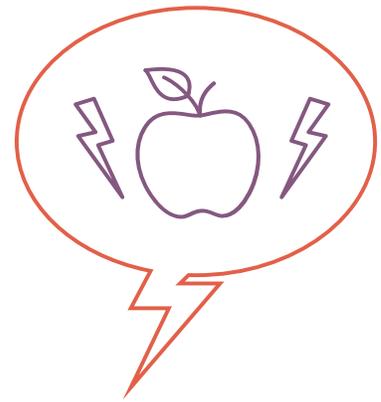
“ Lots of stress...lots of worry...lots of guilt too because you have to tell him ‘no’ a lot....We have to set strict boundaries for him, and he is not happy with that. ”

—Caregiver of a child living with BBS

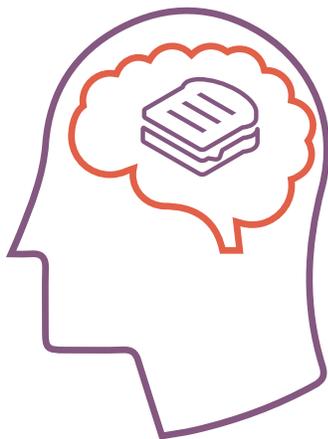
Caregiver quotes have been edited for clarity and conciseness.

Some have reported seeing these behaviors in themselves or an individual they care for

- Arguing with or manipulating people to get food
- Having tantrums to get food
- Taking extreme measures to get food



For someone living with BBS, intense, hard-to-control hunger can affect other aspects of life



- Trouble focusing at school or work because food is always top of mind
- Difficulty socializing due to a constant pursuit of food
- Psychological and emotional problems

Is intense, hard-to-control hunger seriously affecting you or an individual you care for? Complete the survey at the end of this brochure and share the results with your doctor.

Why is it important to manage hard-to-control hunger and obesity?

Intense, hard-to-control hunger can contribute to obesity in people living with BBS. If left unmanaged, obesity can lead to other serious health issues

“ Making changes can have a positive influence. It’s good to know it’s not his fault, but we shouldn’t do nothing about it. ”

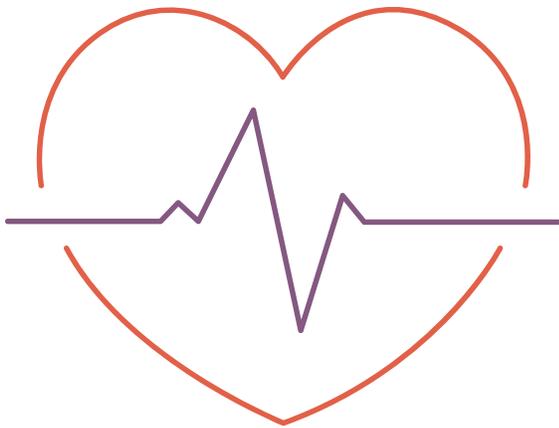
—Caregiver of a child living with BBS

Caregiver quotes have been edited for clarity and conciseness.

Addressing obesity in BBS can help avoid serious health issues

Obesity is a serious health problem for anyone. However, people living with BBS struggle with many health issues, all of which can be further complicated by obesity. That means it's even more important to manage it from the start to help lessen the chance of serious complications later in life.

Serious health problems related to obesity can include:



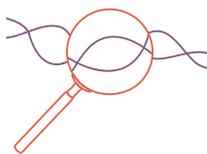
- Diabetes
- High blood pressure
- Heart-related issues (heart attack and stroke)
- Orthopedic issues (problems with bones and muscles)

Ask your doctor about ways you or an individual you care for can address intense, hard-to-control hunger and obesity.

What are some ways to manage hard-to-control hunger and obesity?

Helpful advice for managing obesity

Even though obesity in BBS is a matter of genetics, it is still important to eat a healthy diet low in sugars and high in vegetables, whole grains, and lean protein to help reduce the risk of diabetes and heart disease. Physical activity, such as taking a walk, is another simple way to help reduce your risk. As always, talk to your doctor about how diet and exercise can fit into your personal treatment plan. It can also be good to:



Actively seek out news and information about obesity in BBS to help give you a more complete understanding of the disease



Learn from the first-hand experiences of individuals living with BBS and their caregivers

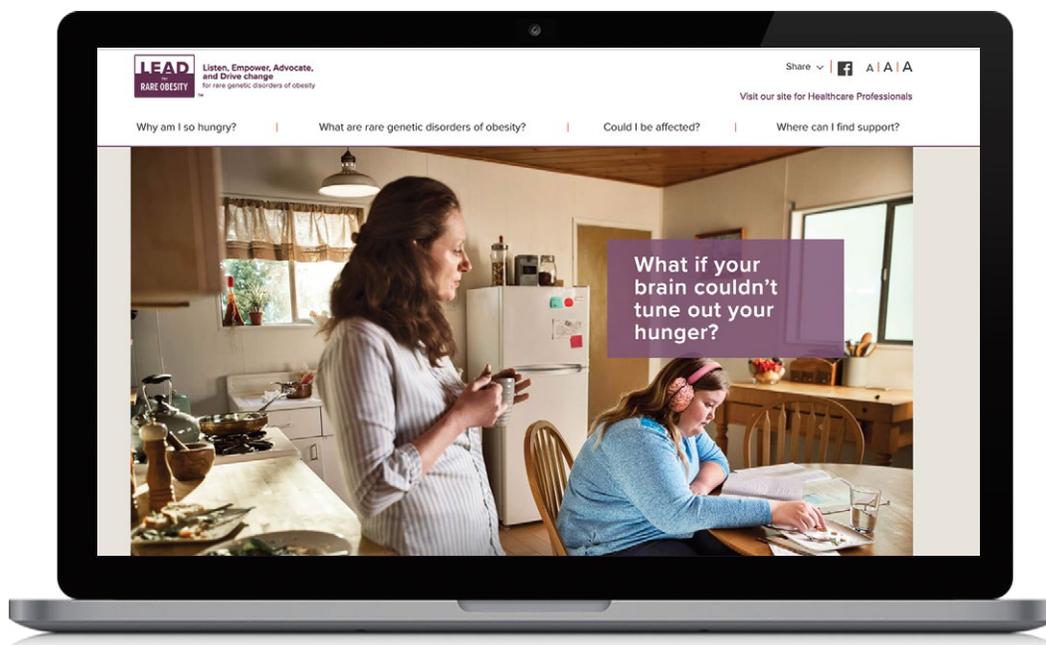
Find more information on rare genetic diseases of obesity, including BBS

LEAD for Rare Obesity is a resource created and sponsored by Rhythm Pharmaceuticals to **L**isten, **E**mpower, **A**dvocate, and **D**rive change for people affected by rare genetic diseases of obesity.



Scan to explore

Explore the **LEAD for Rare Obesity** website for more information, helpful resources, and videos about what it's like to live with and manage BBS. You can also sign up to receive updates and stories from the **rare genetic diseases of obesity community**.



Visit LeadForRareObesity.com

Looking for support during your BBS journey?

Email us for helpful information and educational resources

At Rhythm Pharmaceuticals, our patient support team can provide personalized educational resources to support you or an individual you care for living with BBS. If you're interested in more information, send us an email today.



Contact us today

patientsupport@rhythmtx.com



**Listen, Empower, Advocate,
and Drive change**
for rare genetic diseases of obesity

TM

Could hard-to-control hunger (hyperphagia) be affecting the individual you care for?

If the individual you care for is living with BBS, it's important to understand the effect intense, hard-to-control hunger may be having on their life. Complete the survey below and share the results with their doctor.

Does the individual you care for:	Yes	No
Constantly argue when food is taken away?		
Manipulate others into giving them more food?		
Throw severe tantrums when food is withheld?		
Have trouble focusing at school or work because of hunger?		
Have trouble participating in recreational activities because of hunger?		
Find that hunger impacts their relationships with family or friends?		
Exhibit unusual behavior because of their preoccupation with food?		
Wake up asking or searching for food during the night?		
Sneak or steal food no matter how much they've eaten?		
Constantly ask for more food, no matter how much they've just eaten?		
Seem preoccupied with food or talk about food a lot?		

Share the results of this survey with their doctor. Tell the doctor about any symptoms of intense, hard-to-control hunger that the individual you care for is experiencing.

Could hard-to-control hunger (hyperphagia) be affecting you?

If you are living with BBS, it's important to understand the effect intense, hard-to-control hunger may be having on your life. Complete the survey below and share the results with your doctor.

Do you:	Yes	No
Ever feel stressed out due to hunger, or worry about food?		
Manipulate others into giving you more food after you've already eaten?		
Have trouble focusing at school or work because of hunger?		
Exhibit behavior others find unusual because of a preoccupation with food?		
Wake up asking or searching for food in the night?		
Sneak or steal food even after you've eaten?		
Find that hunger impacts your ability to participate in recreational activities?		
Find that hunger impacts your relationships with family or friends?		
Feel hungry after you just ate, no matter how much you've eaten?		
Think about food a lot, to the point of being preoccupied?		

Share the results of this survey with your doctor. Tell your doctor about any symptoms of intense, hard-to-control hunger that you're experiencing.