








Bardet-Biedl Syndrome



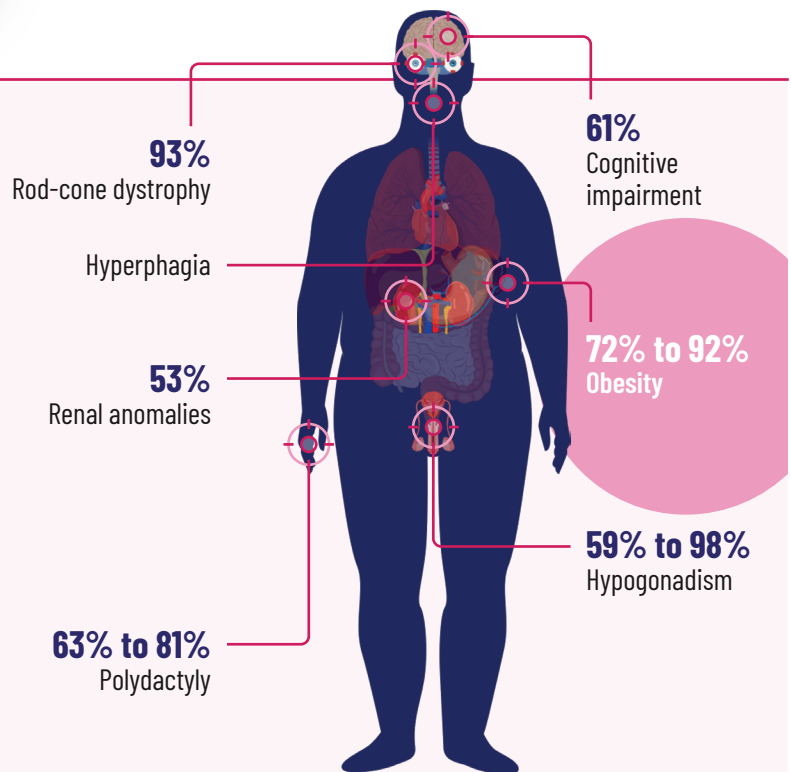
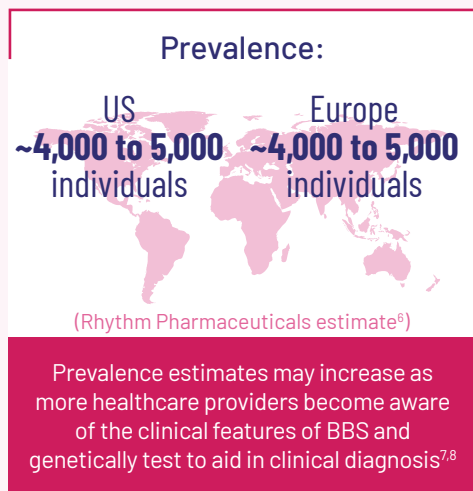
Solomon, living with BBS.

What is BBS?

Bardet-Biedl syndrome (BBS) is a rare genetic disease that presents with a variety of symptoms that evolve over time, including¹⁻³

-  Visual impairments
-  Early-onset, severe obesity
-  Hyperphagia (insatiable hunger)
-  Renal disease
-  Polydactyly
-  Cognitive impairment
-  Hypogonadism

Most Common Features of BBS^{4,5}



Percentages represent frequency of feature appearance among individuals diagnosed with BBS.

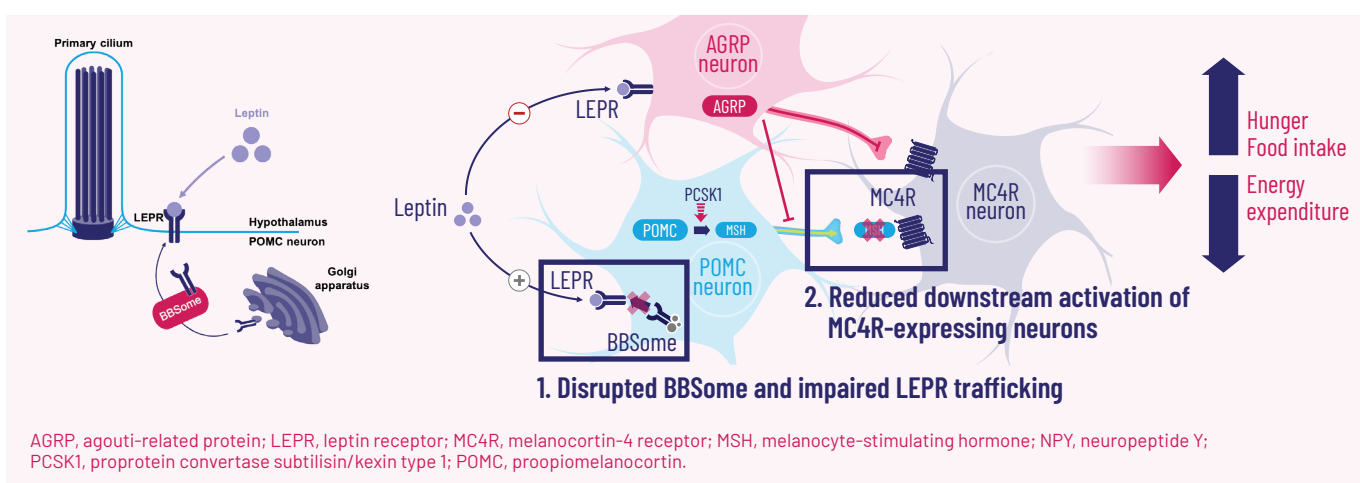
Bardet-Biedl Syndrome

More than 20 genes associated with BBS are involved in the melanocortin-4 receptor (MC4R) pathway^{1,2,4,9-12}

Eight BBS proteins form a stable complex, the BBSome, which contributes to cilia development and function by trafficking intracellular proteins to ciliary membranes and potentially to other membrane compartments¹²

Variants in BBS genes disrupt the BBSome, resulting in ciliary defects and impaired signaling of receptors that regulate body weight, such as LEPR^{9,11,13,14}

This disrupts LEPR signaling, reducing activation of MC4R-expressing neurons, and can lead to hyperphagia and obesity^{9,11,13,14}



How is BBS diagnosed?

Diagnosis of BBS is based on clinical findings; genetic testing can help provide additional diagnostic information and can confirm a clinical diagnosis in nearly 80% of patients⁴

Consider the complete patient presentation and use your clinical judgment to diagnose BBS.^{1,2,5,15}



Most common features

- Rod-cone dystrophy
- Polydactyly
- Obesity
- Hypogonadism
- Renal anomalies
- Cognitive impairment
- Hyperphagia



Other features

- Speech delay or speech impairments
- Developmental delay
- Diabetes mellitus
- Dental anomalies
- Left ventricular hypertrophy or congenital heart disease
- Mild spasticity (especially lower limbs)
- Brachydactyly or syndactyly
- Strabismus, cataracts, or astigmatism
- Ataxia or poor coordination
- Anosmia or hyposmia
- Polyuria or polydipsia
- Hepatic fibrosis

There is no cure for BBS, and patients are treated and monitored based on individual symptoms¹⁴

● Obesity in BBS

- Obesity can begin in childhood and can increase in severity with age^{4,16}
- Obesity may have a detrimental impact on long-term health, due to its association with increased morbidity, social stigma, and reduced quality of life¹⁷
- Hyperphagia may contribute to obesity in patients with BBS^{5,18}
- Hyperphagia is generally characterized by the following^{5,19}:



Insatiable hunger

Heightened and prolonged hunger
 Longer time to reach satiation
 Shorter duration of satiety



Excessive drive to eat

Severe preoccupation with food
 In severe cases, stealing food, night eating, eating food from the trash



Distress and functional impairment due to denial of food

Mean Body Mass Index (BMI) of Patients With BBS by Age²⁰

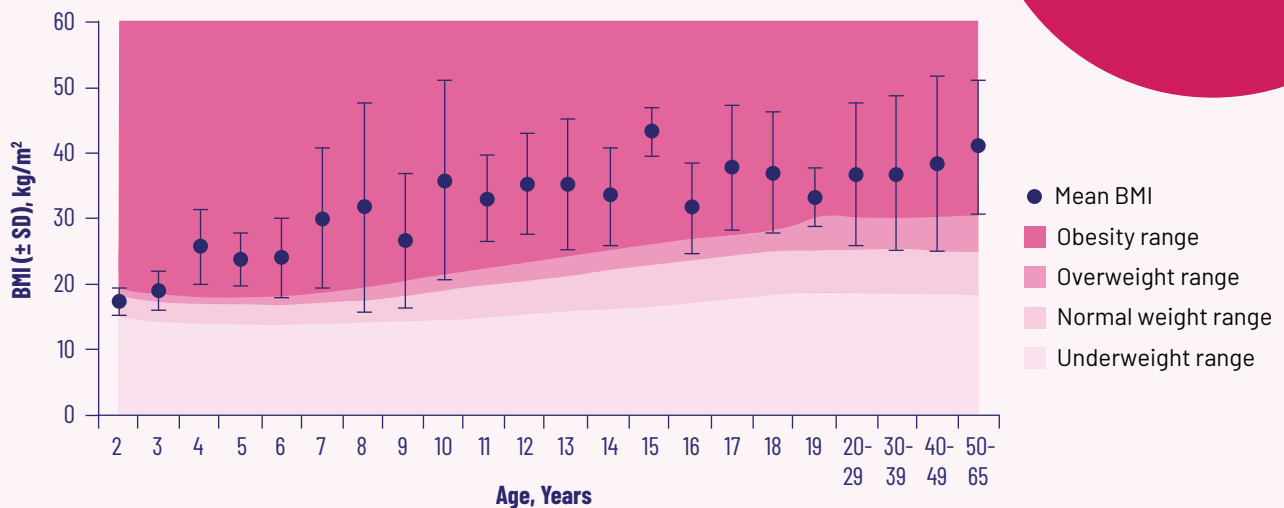


Figure adapted with permission from Marshfield Clinic Research Institute, the research division of Marshfield Clinic Health System.

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