

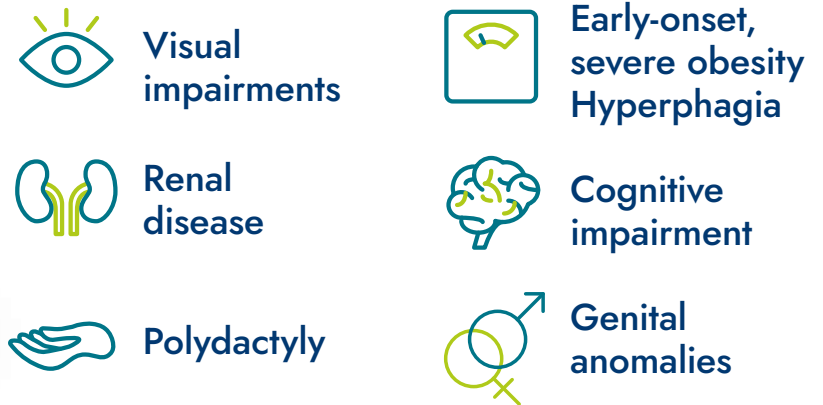
Bardet-Biedl Syndrome



Solomon, living with BBS

What is BBS?

Bardet-Biedl syndrome (BBS) is a rare ciliopathy, resulting from genetic variants within the BBS family of genes. This heterogeneous genetic disease presents with a variety of symptoms that evolve over time, including.¹⁻³

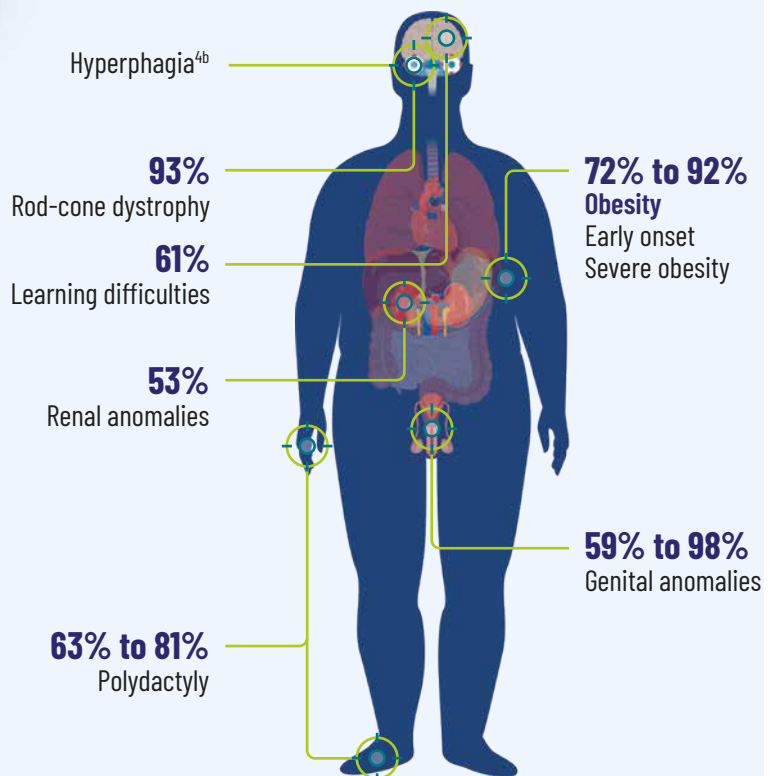


Primary Clinical Features of BBS^{4a}

Prevalence:



Prevalence estimates may increase as more healthcare providers become aware of the clinical features of BBS and genetically test to aid in clinical diagnosis^{6,7}



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

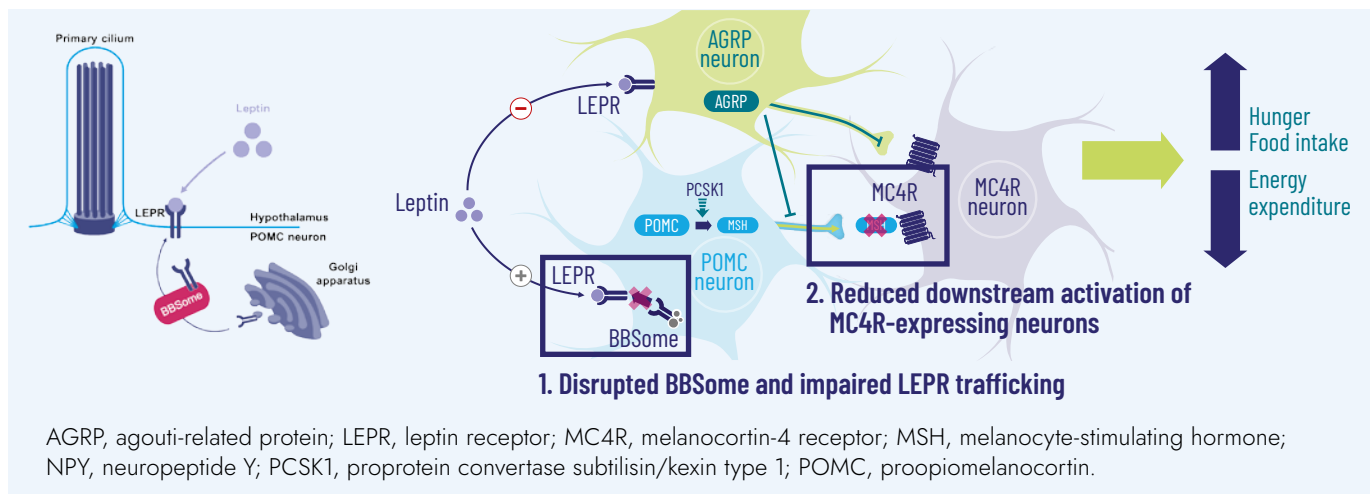
Bardet-Biedl Syndrome

More than 20 genes associated with BBS are involved in the melanocortin-4 receptor (MC4R) pathway.^{1,2,4,8-11}

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 compartments.¹¹

Variants in BBS genes disrupt the BBSome, resulting in ciliary defects and impaired signaling of receptors that regulate body weight, such as LEPR.^{8,10,12,13}

This disrupts LEPR signaling, reducing activation of MC4R-expressing neurons, and can lead to hyperphagia and obesity.^{8,10,12,13}



How is BBS diagnosed?

Diagnosis of BBS is based on clinical findings; diagnosis confirmed by genetic testing⁴

The following criteria have been used to help diagnose BBS. According to these criteria, diagnosis is based on the presence of a combination of features.^{1,14}



Primary features

- Rod-cone dystrophy
- Polydactyly
- Obesity
- Genital anomalies
- Renal anomalies
- Learning difficulties



Secondary 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 specific therapy for BBS, and patients are treated and monitored based on individual symptoms^{1,4}

Bardet-Biedl Syndrome

Obesity in BBS

- Obesity can begin in childhood and can increase in severity with age^{4,15}
- Obesity may have a detrimental impact on long-term health, due to its association with increased morbidity, social stigma, and reduced quality of life¹⁶
- Hyperphagia may contribute to obesity in patients with BBS^{17,18}
- Hyperphagia is generally characterized by the following:^{17,19}



Insatiable hunger

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



Excessive drive to eat

Severe preoccupation with food
Persistent food-seeking behaviors (eg, 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

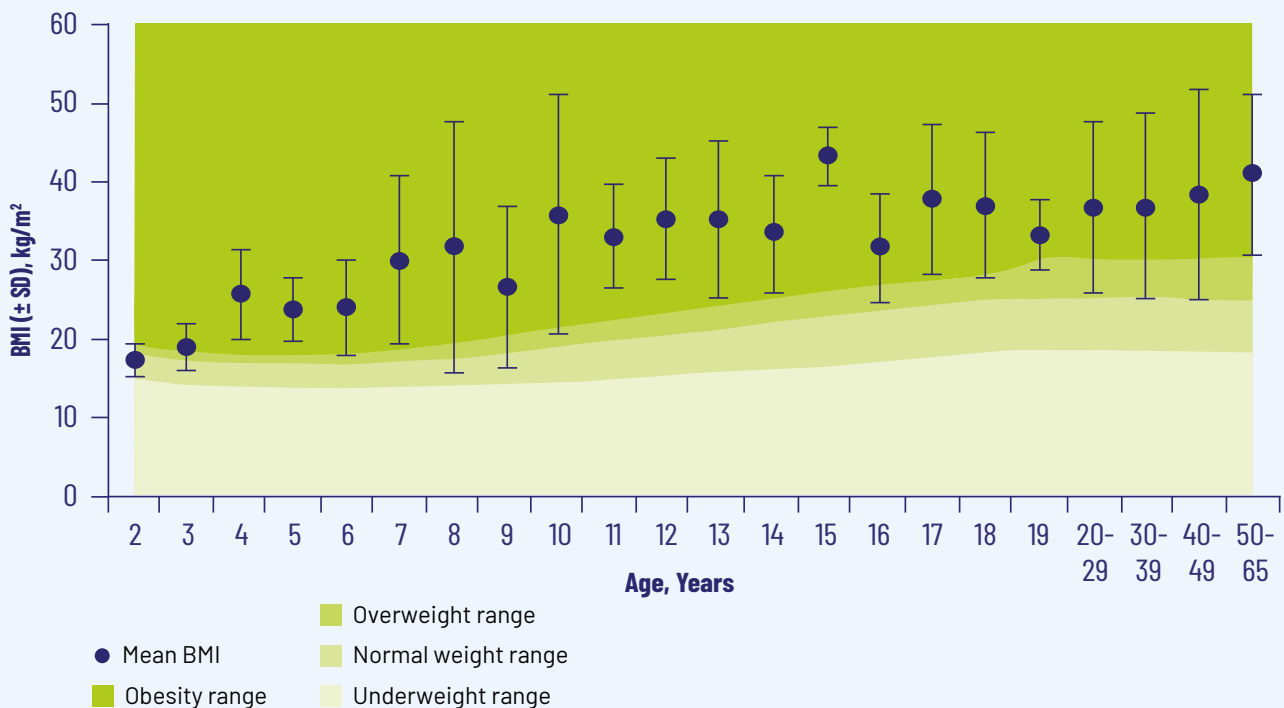


Figure adapted with permission from Marshfield Clinic Research Institute, the research division of Marshfield Clinic Health System.

Bardet-Biedl Syndrome

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