

Bardet-Biedl Syndrome

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



Solomon, living with BBS



Visual impairments



Early-onset, severe obesity
Hyperphagia



Renal disease



Cognitive impairment



Polydactyly



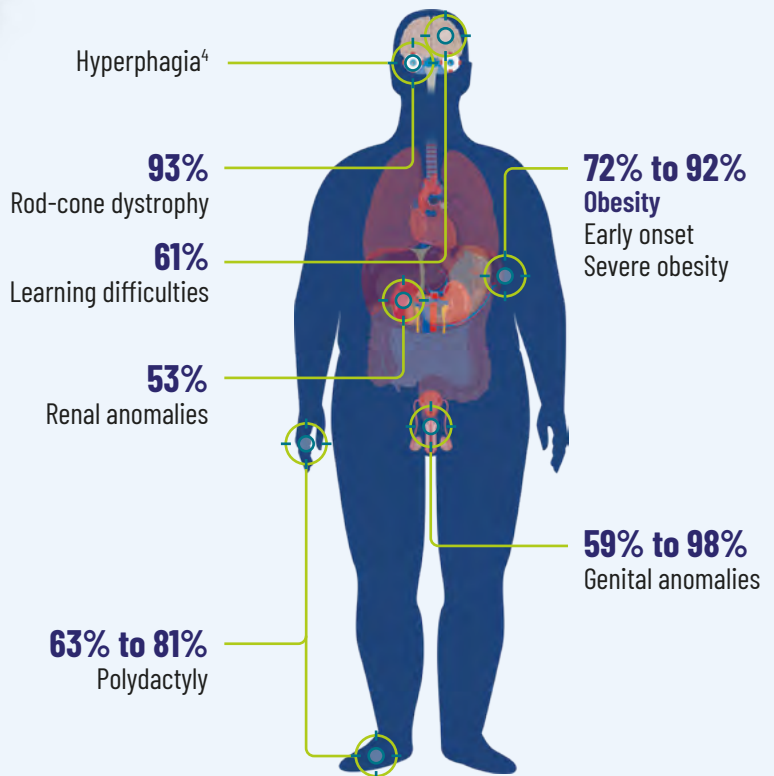
Genital anomalies

Common Clinical Features of BBS⁴

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.

A link and QR code for Prescribing Information and Adverse Event reporting information can be found on page 5

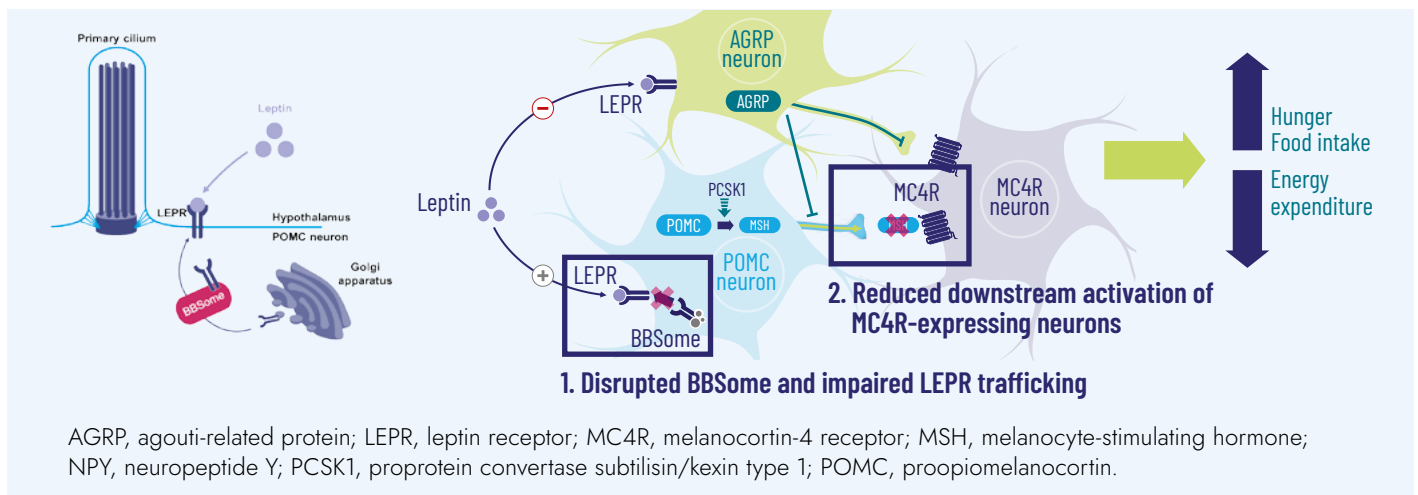
Bardet-Biedl Syndrome

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

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 symptoms¹

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Obesity in BBS

- Obesity can begin in childhood and can increase in severity with age⁴
- Obesity may have a detrimental impact on long-term health, due to its association with increased morbidity, social stigma, and reduced quality of life¹⁵
- Hyperphagia may contribute to obesity in patients with BBS^{16,17}
- Hyperphagia is generally characterized by the following:^{16,18}



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 bin)



Distress and functional impairment due to denial of food

Mean Body Mass Index (BMI) of Patients With BBS by Age¹⁹

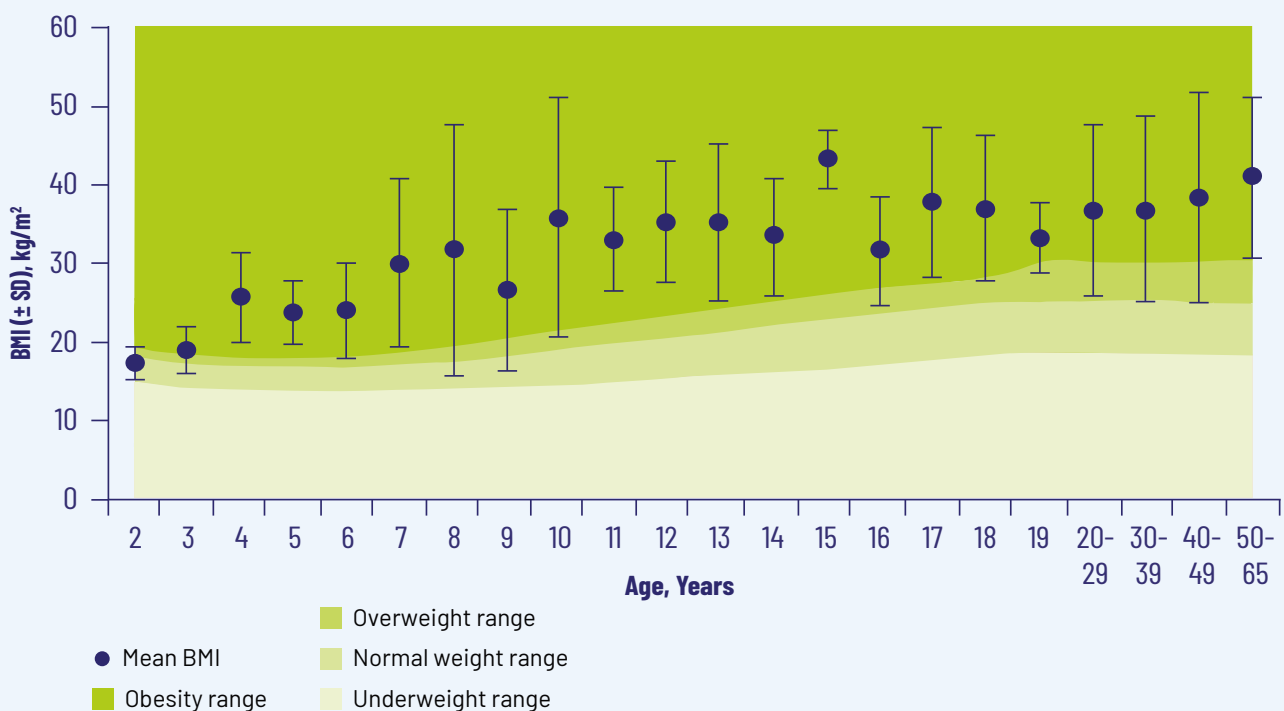


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