RETINITIS PIGMENTOSA?
IT COULD BE BARDET-BIEDL SYNDROME (BBS)

Ophthalmologists and retinal specialists play a key role in diagnosing BBS, a rare genetic disease of obesity.

- **More than 90%** of individuals living with BBS display rod-cone dystrophy, which presents as atypical retinitis pigmentosa with early macular involvement[1]
- Rod-cone dystrophy is the most common feature of BBS and often leads to a diagnosis[1]
- Symptoms generally appear in the first decade of life and often lead to legal blindness by teens/early adulthood[1]

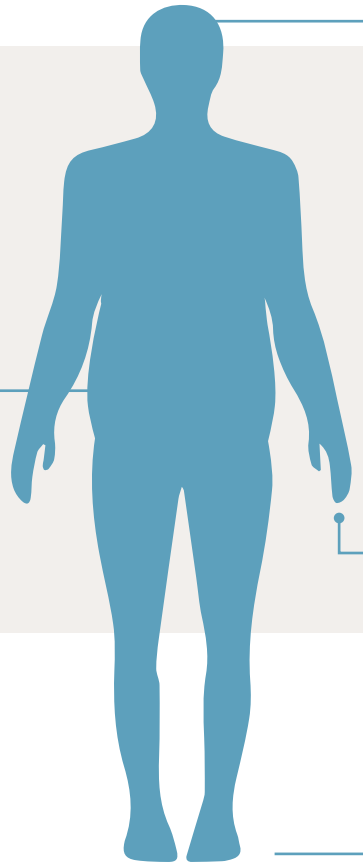
Think BBS if your patients with retinitis pigmentosa have other common features.

Common features[2-4]

Obesity (72%-92%)
- typically early onset by age 5

Hyperphagia (insatiable hunger)
- preoccupation with food
- excessive food-seeking behavior

Renal anomalies (53%)



Visual impairment (93%)
- typical onset: 5-10 years old

Cognitive impairment (>50%)
- learning difficulties
- speech delay
- developmental delay

Postaxial polydactyly (63%-81%)

Learn more about the most common clinical features of BBS ›

RARE Genetic Diseases of OBESITY

YOU CAN PLAY A KEY ROLE IN THE DIAGNOSIS AND MANAGEMENT OF BBS

If your patients have retinitis pigmentosa, along with other common features, they may have BBS. If you suspect BBS, contact your Rhythm Territory Manager to discuss additional education and resources to support diagnosis and management.

BBS can be diagnosed based on clinical features; genetic testing can help[1,5]

You can order test kits through the Uncovering Rare Obesity® program–the only no-charge,* comprehensive genetic testing program for rare genetic diseases of obesity, including BBS.

Learn more about diagnosis ›

A Rhythm Territory Manager is here to support you and your BBS multidisciplinary care team ›

*Rhythm Pharmaceuticals covers the cost of the test and supplies sample collection kits. Patients are responsible for any office visit, sample collection, or other costs.

References: 1. Forsythe E, Beales PL. *Eur J Hum Genet*. 2013;21(1):8-13. **2.** Forsythe E et al. *Front Pediatr*. 2018. doi:10.3389/fped.2018.00023. **3.** Pigeyre M et al. *Clin Sci* (Lond). 2016;130(12):943-986. **4.** Sherafat-Kazemzadeh R et al. *Pediatr Obes*. 2013;8(5):e64-e67. **5.** Beales PL et al. *J Med Genet*. 1999;36(6):437-446.

Rhythm® PHARMACEUTICALS

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