RENAL ANOMALIES EARLY IN LIFE? IT COULD BE BARDET-BIEDL SYNDROME (BBS)

Nephrologists play a key role is diagnosing BBS, a rare genetic disease of obesity.

- **More than 50%** of individuals living with BBS will develop renal anomalies ranging from cystic tubular disease, dysplastic renal disease, and FSGS to concentrating defects[1]
- Renal disease is often detectable before the age of 10, often within the first year of life, in individuals with BBS[1]
- Renal disease is one of the main causes of morbidity and mortality in BBS[1]
- Obesity is common in BBS and can complicate management of comorbidities such as diabetes, renal impairment, and hypertension[2-5]

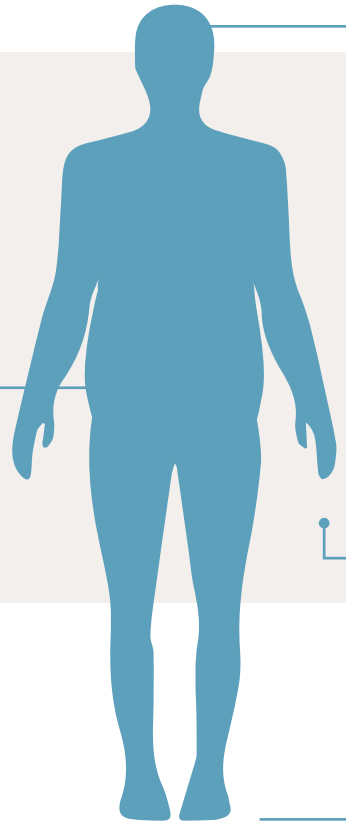
Think BBS if your patients with renal anomalies early in life have other common features.

Common features[5-7]

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%)

FSGS=focal segmental glomerulosclerosis.

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 renal anomalies early in life, 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[2,8]

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 et al. *J Am Soc Nephrol*. 2017;28(3):963-970. **2.** Forsythe E, Beales PL. *Eur J Hum Genet*. 2013;21(1):8-13. **3.** Pomeroy J et al. *Pediatr Obes*. 2021. doi:10.1111/ijpo.12703. **4.** Forsythe E et al. *Clin Genet*. 2015;87(4):343-349. **5.** Forsythe E et al. *Front Pediatr*. 2018. doi:10.3389/fped.2018.00023. **6.** Pigeyre M et al. *Clin Sci* (Lond). 2016;130(12):943-986. **7.** Sherafat-Kazemzadeh R et al. *Pediatr Obes*. 2013;8(5):e64-e67. **8.** Beales PL et al. *J Med Genet*. 1999;36(6):437-446.

Rhythm® PHARMACEUTICALS

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