

Using Genetic Testing to Identify Mutations in *BBS1* and *BBS2* as the Cause of Retinal Degeneration

Indications:

- Retinal degeneration, in conjunction with one other characteristic feature of Bardet-Biedl syndrome (BBS)
- Family history of BBS

Benefits:

- Genetic testing for BBS can:
 - identify BBS as the cause of retinal degeneration.
 - alert patients and physicians to the risk of other BBS-associated manifestations, such as renal malformations.
 - aid in genetic counseling for BBS.

Background:

- Retinal degeneration due to BBS typically presents in children at around age 8 years.¹
- BBS is also associated with postaxial polydactyly, syn- or brachydactyly, early-onset obesity, cardiac and renal abnormalities, mild to moderate learning disabilities, anosmia, hypogonadism in males, genitourinary malformation in females, dental anomalies, nephrogenic diabetes insipidus, diabetes mellitus, and hypertension.¹
- Renal malformations are a leading cause of morbidity and mortality in patients with BBS.^{1,2}
- BBS is typically inherited in an autosomal recessive manner.³
- Loss-of-function mutations in the genes *BBS1* or *BBS2* account for about 23% and 8%, respectively, of all cases of BBS.³⁻⁵

References: 1. Beales PL, et al (1999) J Med Genet 36:437–446. 2. O'Dea D, et al (1996) Am J Kidney Dis. 27:776-83. 3. Katsanis N (2004) Hum Mol Genet 13:R65–R71. 4. Mykytyn K, et al (2002) Nat Genet 31:435-8. 5. Nishimura DY, et al (2001) Hum Mol Genet 10:865-74.

Ordering Information: please see other side



Ordering Information for the *BBS1* and *BBS2* DNA Sequencing Tests

Indications for Testing

- Retinal degeneration, in conjunction with one other characteristic feature of Bardet-Biedl syndrome
- Family history of Bardet-Biedl syndrome

Ordering Information

Gene(s)	Test Code
<i>BBS1</i>	180101
<i>BBS2</i>	180102

Multi-Gene Panel*

<i>BBS1 + BBS2</i>	180199
--------------------	--------

* For the multi-gene panel, a summary report will be issued in addition to an abbreviated report for each individual gene.

Family Testing (single amplicon)

Family Testing is available for both genes. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit,
please contact Client Services at 1-866-647-0735 or visit us on the web at
www.correlagen.com.**