

# Bardet-Biedl Syndrome Evaluation

## Identifying Bardet-Biedl Syndrome in infants with postaxial polydactyly

### What is Bardet-Biedl Syndrome (BBS)?

- BBS is a pleiotropic disorder characterized by rod-cone dystrophy, postaxial polydactyly, obesity, hypogenitalism, renal abnormalities, and learning disabilities, as well as a range of secondary symptoms.<sup>1</sup>
- Diagnosis of BBS is currently based on the presence of at least four characteristic features and occurs, on average, at age nine.<sup>1</sup>
- BBS has been linked to mutations in any one of several different genes, with mutations in *BBS1*, *BBS2*, or *BBS10* accounting for about half of all cases.<sup>2,3</sup>
- Inheritance of BBS is predominantly autosomal recessive and, in some cases, digenic.<sup>2</sup>
- Genetic testing for BBS-associated mutations can detect BBS at any age.
- Genetic testing is indicated in infants with postaxial polydactyly.

### Why genetic testing?

Genetic testing for mutations associated with Bardet-Biedl Syndrome can

- Confirm a diagnosis reached on the basis of clinical criteria.
- Allow diagnosis of BBS in infancy, enabling parents to recognize and understand their child's problems, such as behavioral and learning disabilities, and to seek appropriate help early.
- Aid in genetic counseling.
- Facilitate diagnosis in family members.

### Indications for testing:

- Postaxial polydactyly in infants\*
- Diagnosis of BBS based on clinical criteria
- Family history of BBS

\*In individuals of African descent, postaxial polydactyly is more common and often non-syndromic.<sup>4</sup>

**References:** 1. Beales PL et al (1999) J Med Genet 36:437–446. 2. Katsanis N (2004) Hum Mol Genet 13:R65–R71. 3. Stoetzel C et al (2006) Nat Genet 38:521-4. 4. Castilla EE et al (1996) Am J Med Genet 65:295-303.

For a brief review on Bardet-Biedl Syndrome, please visit [www.athenadiagnostics.com](http://www.athenadiagnostics.com).

## Bardet-Biedl Syndrome Evaluation

<b>Typical Presentation:</b>	At least four of the following symptoms: retinal degeneration, postaxial polydactyly, obesity, hypogenitalism, renal abnormalities, learning disabilities
<b>Indications for Testing:</b>	<ul style="list-style-type: none"><li>• Postaxial polydactyly in infants</li><li>• Diagnosis of BBS based on clinical criteria</li><li>• Family history of BBS</li></ul>
<b><u>Test Details</u></b>	
<b>Test Code:</b>	887
<b>Test Includes</b>	<i>BBS1</i> (Bardet-Biedl Syndrome) DNA Sequencing Test, # 871 <i>BBS2</i> (Bardet-Biedl Syndrome) DNA Sequencing Test, # 872 <i>BBS10</i> (Bardet-Biedl Syndrome) DNA Sequencing Test, # 886
<b>Test Turnaround:</b>	14 to 21 days
<b><u>Technical Information</u></b>	
<b>Methodology:</b>	Polymerase Chain Reaction (PCR), DNA sequencing of entire protein coding region of gene
<b>CPT Code(s):</b>	83891 (1); 83898 (xx); 83904 (xx); 83912 (x)
<b><u>Shipping Considerations</u></b>	
<b>Specimen Type:</b>	Whole blood
<b>Volume:</b>	10 mL (pediatric minimum: 2 mL)
<b>Collection Tube:</b>	Yellow or lavender top
<b>Stability:</b>	Hemolysis may compromise DNA recovery and integrity after 48 hrs
<b>Storage Conditions:</b>	For short periods (until shipped) at 4°C
<b>Shipping Conditions:</b>	Overnight at room temperature (specimen arrival must be less than 24 hrs after collection); ship Monday through Thursday only

Call Athena Diagnostics' Customer Service Representatives to order the Bardet-Biedl Syndrome Evaluation (Test #8xx) at (800) 394-4493 x2.