**Bardet-Biedl Syndrome: Deletion/Duplication Panel**

**Test Code:** MD132  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Bardet-Biedl syndrome (BBS) is a genetically heterogeneous disorder clinically characterized by the presence of photoreceptor dystrophy (rod-cone), postaxial polydactyly, truncal obesity, learning disabilities, hypogonadism in males, genital abnormalities in females, and renal abnormalities. A wide range of clinical variability may be observed and a variety of secondary features may also occur. BBS is most commonly inherited in an autosomal recessive manner.


### References:

- OMIM
- GeneReviews

### Genes

ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Bardet-Biedl syndrome.
- Carrier testing in adults with a family history of Bardet-Biedl syndrome.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region.

**Detection**

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

**Submit only 1 of the following specimen types**

**Type:** Whole Blood (EDTA)

**Specimen Requirements:**

EDTA (Purple Top)  
Infants and Young Children (2 years of age to 10 years old): 3-5 ml  
Older Children & Adults: 5-10 ml  
Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**Type:** DNA, Isolated

**Specimen Requirements:**

Microtainer  
3µg  
Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

**Special Instructions**

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.
Related Tests

- Bardet-Biedl Syndrome: Sequencing Panel
- Eye Disorders: Comprehensive Sequencing
- Eye Disorders: Deletion/Duplication Panel