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.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

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
Specimen Requirements:
In EDTA (purple top) tube:
Infants (2 years): 3-5 ml
Older Children & Adults: 5-10 ml
Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA
Specimen Requirements:
In microtainer: 10 ug
Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.
Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

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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