

## Bloom Syndrome: *BLM* Gene Deletion/Duplication

**Test Code:** DBLM1

**Turnaround time:** 2 weeks

**CPT Codes:** 81228 x1

### Condition Description

Bloom syndrome should be considered in the following individuals:

- An individual with unexplained, severe intrauterine growth retardation that persists into infancy and childhood
- An unusually small, well-proportioned individual with a sun-sensitive erythematous skin lesion in a "butterfly distribution" on the face
- An unusually small individual who develops cancer

Bloom syndrome is an autosomal recessive disorder characterized by severe prenatal and postnatal growth retardation and a sun-sensitive erythematous skin lesion that occurs most commonly on the face. Recurrent infections (otitis media and pneumonia), chronic pulmonary disease, and diabetes mellitus are common. Many affected individuals have learning disabilities. Males are infertile; females enter menopause prematurely. The most common cause of death is cancer (epithelial, hematopoietic, lymphoid, connective tissue, germ cell, nervous system, and kidney), which occurs at younger-than-usual ages.

The diagnosis of Bloom syndrome can be confirmed or ruled out by chromosome analysis of any cell type that can be cultured. The most commonly used cells are blood lymphocytes, but cultures of skin fibroblasts and amniocytes also can be studied. The cytogenetic features of Bloom syndrome cells in mitosis are increased numbers of chromatid gaps, breaks, and rearrangements and increased numbers of quadriradial configurations. A greatly increased frequency (e.g., greater than 50 per metaphase) of sister chromatid exchanges is demonstrable in Bloom syndrome cells exposed to bromodeoxyuridine (BrdU). *BLM* (15q26.1) is the only gene known to be mutated in Bloom syndrome.

Fewer than 300 cases of Bloom syndrome have been reported. Although very rare in all populations, Bloom syndrome is less rare in Ashkenazi Jews. Approximately 25% of persons in the Bloom Syndrome Registry have Ashkenazi Jewish ancestry. The carrier frequency in the Ashkenazi Jewish population is estimated to be approximately 1%.

For patients with suspected Bloom syndrome, sequence analysis is recommended as the first step in mutation identification. For patients in whom mutations are not identified by full gene sequencing, deletion/duplication analysis is appropriate.

References:

- GeneTests: <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gen&part=bloom>

### Genes

[BLM](#)

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Bloom syndrome in individuals who have tested negative for sequence analysis.

### Methodology

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

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: 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.

**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.

**Special Instructions**

Submit copies of diagnostic biochemical test results with the sample, if appropriate. Contact the laboratory if further information is needed.

Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside of EGL Genetics, please submit a copy of the sequencing report with the test requisition.

**Related Tests**

- Sequence analysis of the *BLM* gene is available and is required before deletion/duplication analysis.
- Prenatal testing is available to couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal specimen.