BLM gene

Associated Syndrome Name: *BLM*-associated cancer risk

BLM Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Colorectal	Elevated Risk

BLM gene Overview

BLM-associated cancer risk [1, 2, 3, 4]

- Individuals with mutations in only one copy of the *BLM* gene (a monoallelic mutation) may have a slightly increased risk of colorectal cancer.
- Some studies have observed an increased risk for colorectal cancer, particularly in the Ashkenazi Jewish population. However, other studies have not replicated these findings in the Ashkenazi Jewish population or other ancestries.
- Individuals with mutations in both copies of the *BLM* gene (biallelic mutations) have Bloom syndrome. Bloom syndrome is typically diagnosed in childhood and characterized by a wide range of symptoms, including but not limited to growth deficiency, immunodeficiency, ultraviolet sensitivity, and increased cancer risks. This individual does not have a diagnosis of Bloom syndrome, but may have relatives who are at risk for this condition. Please see the Information for Family Members section below for details.
- Currently there are no medical management guidelines for individuals with a single *BLM* mutation. However, this may change as we learn more, and therefore individuals with monoallelic *BLM* mutations may benefit from consultation with healthcare providers who have expertise in medical genetics and the care of individuals with hereditary cancer syndromes.

BLM gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Colorectal	To age 80[5]	Possibly elevated	2.8%

BLM Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Colorectal	Currently there are no medical management guidelines for colorectal cancer risk in individuals with a single *BLM* mutation.	NA	NA

Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the BLM gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts,

uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

In rare instances, an individual may inherit mutations in both copies of the *BLM* gene, leading to the condition Bloom syndrome. This condition typically presents in childhood and includes growth deficiency, immunodeficiency, ultraviolet sensitivity, and increased cancer risks.[2]

The children of this individual are at risk of inheriting Bloom syndrome only if the other parent is also a carrier of a *BLM* mutation. Carrier testing of the partner may be considered for reproductive decision-making and planning.[2]

References

1. de Voer RM, et al. Deleterious Germline *BLM* Mutations and the Risk for Early-onset Colorectal Cancer. Sci Rep. 2015 Sep 11;5:14060. PMID: 26358404.
2. Cleary SP, et al. Heterozygosity for the *BLM*(Ash) mutation and cancer risk. Cancer Res. 2003 Apr 15;63(8):1769-71. PMID: 12702560.
3. Laitman Y, et al. The risk for developing cancer in Israeli *ATM*, *BLM*, and *FANCC* heterozygous mutation carriers. Cancer Genet. 2016 Mar;209(3):70-4. PMID: 26778106.
4. Langer K, et al. Bloom Syndrome. 2023 Oct 12. In: Adam MP, et al., editors. GeneReviews® [Internet]. PMID: 20301572
5. SEER*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2025 Aug 12]. Available from https://seer.cancer.gov/explorer/.

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