Genetic Counseling Service
75 Claremont Rd Suite 305
Bernardsville, NJ 07924

Dear Family Member,

A member of your family has been identified as having a genetic mutation in the BLM gene. Mutations in this gene are known to be associated with an increased risk for certain cancers. This means that other family members are at risk to carry this same gene mutation and hence at an increased risk for certain cancers. This letter has been written to summarize information regarding BLM mutations.

Individuals that are positive for a single BLM mutation may have an increased chance to develop female breast cancer, as well as an increased chance to develop colorectal cancer. However, exact cancer risk estimates are not yet available. A slightly increased risk for pancreatic cancer has also been reported. An increased risk for additional cancers has been reported by some studies, but these risks are not well defined.

Understanding risk is an important tool in prevention and management of cancer. It is important to note that many individuals with BLM mutations will never develop cancer over their lifetime. When one inherits an BLM mutation, he/she is not inheriting cancer; rather they are inheriting a predisposition to cancer.

At this time there are no established guidelines or recommendations for increased screening based solely on the presence of a BLM mutation. Overall cancer risk assessment incorporates multiple factors, including personal medical history, family history, and available genetic information. The available information regarding hereditary cancer susceptibility genes is constantly evolving and more clinically relevant BLM data is likely to become available in the future.

When an individual has one copy of the BLM gene mutation, they have an increased risk for breast and possibly other cancers as outlined above. Because everyone has two copies of the gene for BLM, this means that a parent who carries an BLM mutation has a 50% chance of passing the mutation onto each of their children. It also means that if a person carries such a mutation, their siblings have a 50% chance to carry the same mutation. Extended family members may also be at risk for having the same mutation. If a person does not carry the mutation, they cannot pass it onto their children. It should be noted that very rarely an individual may have two BLM mutations, one inherited from their mother, and one inherited from their father. If a person has two BLM mutations (one inherited from each parent) they have a condition known as Bloom syndrome. This is a rare, chromosomal instability condition for which symptoms include growth deficiency, sun-sensitive skin rash, infertility, and an increased risk of malignancy.

We strongly recommend that you and all persons in your family who are at risk for carrying the BLM gene mutation consider scheduling an appointment for cancer genetic counseling to discuss the benefits and limitations of genetic testing. To pursue cancer genetic testing, it is helpful if you have a copy of your family member's genetic testing report indicating that they have an BLM mutation.

GeneScreen is available to discuss this information in more detail with you and coordinate appropriate testing. Please visit the following website to submit your request for genetic

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counseling: https://hipaa.jotform.com/200926739003048. You can also contact Genescreen directly at 908-766-2800 to schedule a remote consultation with a genetic counselor.

Sincerely,

Stephanie Nunziato-Virga, MS, LGC
Senior Oncology Genetic Counselor
Genescreen

Jill Chisholm, APN
President
Genescreen