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 **NTHL1 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 NTHL1 mutations.

Unlike other hereditary cancer syndromes, NTHL1 related cancer predisposition is inherited in a recessive manner, which means both copies of the NTHL1 gene in each cell have mutations in order to be at an elevated risk for cancer. Carriers of a recessive genetic condition have one working and one non-working copy of the gene. Individuals who have one *mutation* on NTHL1 (carriers), are not currently believed to have an increased cancer risk. Individuals with two *mutations* in the NTHL1 gene have an increased for developing colon polyps (adenomatous polyps) and colorectal cancer. There have been suggestions of other cancers, but further studies are needed to confirm this. Individuals with two mutations may benefit from increased and early coloscopy screening.

Understanding risk is an important tool in prevention and management of cancer. It is important to note that many individuals with NTHL1 mutations will never develop cancer over their lifetime. When one inherits an NTHL1 mutation, he/she is not inheriting cancer; rather they are inheriting a predisposition to cancer.

We strongly recommend that you and all persons in your family who are at risk for carrying the NTHL1 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 NTHL1 mutation. Also, reproductive partners of patients who are positive for NTHL1 gene mutation(s) may also consider genetic testing for NTHL1 to determine if their children are at risk for having two NTHL1 mutations.

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 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
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