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 **POLD1** 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 POLD1 mutations.

Individuals with *pathogenic mutations* in the POLD1 gene have Polymerase Proofreading-Associated Polyposis (PPAP) syndrome. PPAP is characterized by increased risks for colon cancer and adenomatous polyps. Patients with POLD1 mutations may also have an increased risk for endometrial cancer. The lifetime risk for cancer for men and women with POLD1 mutations is not fully understood, but is presumed to be high. Management guidelines for individuals with POLD1 mutations are evolving and final surveillance regimens should be considered in the context of patient preferences and new knowledge that may emerge. Current management recommendations for individuals identified to have a pathogenic mutation in the POLD1 gene may include early colonoscopies, education regarding early signs of uterine cancer, endometrial sampling, and possible total hysterectomy.

Understanding risk is an important tool in prevention and management of cancer. It is important to note that many individuals with POLD1 mutations will never develop cancer over their lifetime. When one inherits a POLD1 mutation, he/she is not inheriting cancer; rather they are inheriting a predisposition to cancer.

When an individual has one copy of the POLD1 gene mutation they have an increased risk for cancers as outlined above. Because everyone has two copies of the gene for POLD1, this means that a parent who carries a POLD1 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.

We strongly recommend that you and all persons in your family who are at risk for carrying the POLD1 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 POLD1 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 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,

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Genescreen

Jill Chisholm, APN
President
Genescreen