



Dear Family Member,

A member of your family has been identified as having a genetic mutation in the POT1 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 POT1 mutations.

*Pathogenic mutations* in the POT1 gene has recently been identified as a susceptibility gene for familial, adult-onset cutaneous melanoma (CM). While gene-specific risks are not yet established, individuals with a *pathogenic* variant in POT1 appear to develop melanoma at an earlier age compared to sporadic cases. There is emerging data to suggest that POT1 may be associated with glioma and cardiac angiosarcoma and therefore this considered a “preliminary evidence” gene as the association between the gene and the specific condition has not been completely established.

Understanding risk is an important tool in prevention and management of cancer. It is important to note that many individuals with POT1 mutations will never develop cancer over their lifetime. When one inherits a POT1 mutation, he/she is not inheriting cancer; rather they are inheriting a predisposition to cancer.

When an individual has one copy of the POT1 gene mutation they have an increased risk for cancers as outlined above. Because everyone has two copies of the gene for POT1, this means that a parent who carries a POT1 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 POT1 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 POT1 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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