



Dear Family Member,

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

Individuals with a mutation in the *PTEN* gene have an increased risk for breast, endometrial, colorectal, thyroid, kidney, and melanoma. In addition to an increased cancer risk, those with a *PTEN* mutation generally develop non-cancerous (benign) tumors throughout the body known as hamartomas. Patients with *PTEN* mutations often have a wide variety of other, non-malignant features of the condition (i.e. large head size, colorectal polyps, brain tumors, and distinctive skin findings). Developmental delay and/or autism spectrum disorders may also be present in some families.

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

When an individual has one copy of the *PTEN* 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 *PTEN*, this means that a parent who carries an *PTEN* 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 *PTEN* 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 *PTEN* 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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