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 **STK11 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 STK11 mutations.

Gene mutations in the STK11 gene are associated with Peutz-Jeghers syndrome (PJS). PJS is characterized by gastrointestinal (GI) polyposis, dark-colored spots (mucocutaneous hyperpigmentations), and increased risk of several types of cancer (colorectal, gastric, pancreatic, breast, and ovarian). PJS-type hamartomatous polyps occur most commonly in the small intestine, but can also be found in the large bowel, stomach, renal pelvis, bronchus, gallbladder, nasal passages, and ureter. GI polyps can result in chronic intestinal obstruction, intussusception, bleeding, and anemia, requiring surgical intervention. Mucocutaneous hyperpigmentations present in childhood appear as dark blue to dark brown spots often occurring around the mouth, nostrils, perianal area, and fingers. Males with PJS can develop tumors of the testes, which, if untreated, can lead to excess growth of breast tissue, short stature, and advanced skeletal age. Females with PJS are at risk for sex cord tumors with annular tubules, and a rare aggressive form of cervical cancer. Approximately 45% of affected individuals have no family history of PJS.

Understanding risk is an important tool in prevention and management of cancer. It is important to note that many individuals with STK11 mutations will never develop cancer over their lifetime. When one inherits an STK11 mutation, he/she is not inheriting cancer; rather they are inheriting a predisposition to cancer.

When an individual has one copy of the STK11 gene mutation they have an increased risk for cancer as outlined above. Because everyone has two copies of the gene for STK11, this means that a parent who carries an STK11 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 STK11 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 STK11 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
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