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My46 Trait Profile

Peutz-Jeghers syndrome

Peutz-Jeghers syndrome is a rare genetic disorder characterized by an increased chance to develop noncancerous growths in the stomach and intestine as well as certain types of cancer. It is caused by mutations in the STK11 gene which makes the serine/threonine kinase 11 protein.

Characteristics of Peutz-Jeghers syndrome

Most individuals with Peutz-Jeghers syndrome (PJS) develop many noncancerous growths called hamartomatous polyps in the stomach and intestines. Polyps may also develop in other areas of the body including in the airways, ureters (the tubes that connec the kidney to the bladder), and bladder. The polyps seen in PJS most often develop in childhood or adolescence. Although the polyps are nancancerous, they may cause bleeding, obstruction, and low blood count (i.e., anemia). The majority of affected children develop harmless dark spots or macules on the lips, around and inside the mouth, on the eyes, nostrils and anus, and sometimes on the fingers. Women with PJS may also develop cysts in the ovaries. PJS is considered a cancer predisposition syndrome since affected individuals are at increased risk for developing certain forms of cancer, including cancer of the breast, colon, rectum, thyroid, pancreas, ovary, cervix, uterus, and testis. Not all individuals with PJS develop cancer.

Diagnosis/Testing

Most individuals with PJS have a change or mutation in a gene called STK11. The STK11 gene is a tumor suppressor gene which means that it keeps cells from growing too fast. Mutations in the STK11 gene do not allow the STK11 protein to work normally and as a result, cells may grow uncontrollably. This uncontrolled growth is what can cause the tumors and cysts in affected individuals.

Management/Surveillance

Management of PJS includes monitoring the growth of polyps by endoscopic surveillance and enteroscopy (procedures used to examine the small intestine) and the removal of the polyps when they are large or causing health problems.

Individuals who have PJS or are at risk to have the condition and have not had genetic testing should follow specific surveillance recommendations with their healthcare provider. These vary depending upon an individuals age and include upper endoscopies, colonoscopies, breast examinations, transvaginal ultrasounds, pelvic exams with pap smear, special blood tests, and testicular exams at varying intervals.

It is very important that individuals with PJS be followed closely by their healthcare providers. Referral to a high-risk cancer center with experience in managing individuals with PJS is recommended for individuals with PJS and for those who are at risk of having PJS.

Mode of inheritance

PJS is inherited in an autosomal dominant pattern. This means inheriting one STK11 mutation is enough for an individual to be affected and show signs of PJS. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with PJS has a parent affected with PJS. If a parent also has PJS, the risk of having an affected child is 50% with each pregnancy. If a parent does not have PJS, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Peutz-Jeghers syndrome online support group
http://listserv.acor.org/scripts/wa-ACOR.exe?A0=PJS
Genetics Home Reference: Peutz-Jeghers syndrome
http://ghr.nlm.nih.gov/condition/peutz-jeghers-syndrome
The Johns Hopkins Guide for Patients and Families: Peutz-Jeghers Syndrome
http://www.karmanos.org/upload/docs/Patient%20and%20Visitors/pjs-book.pdf

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Created: 05/2013 **Created by:** Lakshmi Warrier, MS, CGC

Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC