Peutz-Jeghers syndrome

Description

Peutz-Jeghers syndrome is characterized by the development of noncancerous growths called hamartomatous polyps in the gastrointestinal tract (particularly the stomach and intestines) and a greatly increased risk of developing certain types of cancer.

Children with Peutz-Jeghers syndrome often develop small, dark-colored spots on the lips, around and inside the mouth, near the eyes and nostrils, and around the anus. These spots may also occur on the hands and feet. They appear during childhood and often fade as the person gets older. In addition, most people with Peutz-Jeghers syndrome develop multiple polyps in the stomach and intestines during childhood or adolescence. Polyps can cause health problems such as recurrent bowel obstructions, chronic bleeding, and abdominal pain.

People with Peutz-Jeghers syndrome have a high risk of developing cancer during their lifetimes. Cancers of the gastrointestinal tract, pancreas, cervix, ovary, and breast are among the most commonly reported tumors.

Frequency

The prevalence of this condition is uncertain; estimates range from 1 in 25,000 to 300,000 individuals.

Causes

Mutations in the *STK11* gene (also known as *LKB1*) cause most cases of Peutz-Jeghers syndrome. The *STK11* gene is a tumor suppressor gene, which means that it normally prevents cells from growing and dividing too rapidly or in an uncontrolled way. A mutation in this gene alters the structure or function of the STK11 protein, disrupting its ability to restrain cell division. The resulting uncontrolled cell growth leads to the formation of noncancerous polyps and cancerous tumors in people with Peutz-Jeghers syndrome.

A small percentage of people with Peutz-Jeghers syndrome do not have mutations in the *STK11* gene. In these cases, the cause of the disorder is unknown.

Learn more about the gene associated with Peutz-Jeghers syndrome

- STK11

Inheritance

Peutz-Jeghers syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to increase the risk of developing noncancerous polyps and cancerous tumors. In about half of all cases, an affected person inherits a mutation in the *STK11* gene from one affected parent. The remaining cases occur in people with no history of Peutz-Jeghers syndrome in their family. These cases appear to result from new (de novo) mutations in the *STK11* gene.

Other Names for This Condition

- Intestinal polyposis-cutaneous pigmentation syndrome
- Lentiginosis, perioral
- Periorificial lentiginosis syndrome
- Peutz-Jeghers polyposis
- PJS
- Polyposis, hamartomatous intestinal
- Polyposis, intestinal, II
- Polyps-and-spots syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Peutz-Jeghers syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0031269/)

Genetic and Rare Diseases Information Center

- Peutz-Jeghers syndrome (https://rarediseases.info.nih.gov/diseases/7378/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Peutz-Jeghers syndrome%22)

Catalog of Genes and Diseases from OMIM

- PEUTZ-JEGHERS SYNDROME; PJS (https://omim.org/entry/175200)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Peutz-Jeghers+Syndrome%5BMAJR%5D%29+AND+%28Peutz-Jeghers+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

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