

# Peutz-Jeghers Syndrome

Subjects: Genetics & Heredity

Contributor: Rita Xu

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.

Keywords: genetic conditions

---

## 1. Introduction

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.

## 2. Frequency

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

## 3. 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.

### The Gene Associated with Peutz-Jeghers Syndrome

- STK11

## 4. 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.

## 5. 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

---

## References

1. Beggs AD, Latchford AR, Vasen HF, Moslein G, Alonso A, Aretz S, Bertario L, Blanco I, Bülow S, Burn J, Capella G, Colas C, Friedl W, Møller P, Hes FJ, Järvinen H, Mecklin JP, Nagengast FM, Parc Y, Phillips RK, Hyer W, Ponz de Leon M, Renkonen-Sinisalo L, Sampson JR, Stormorken A, Tejpar S, Thomas HJ, Wijnen JT, Clark SK, Hodgson SV. Peutz-Jeghers syndrome: a systematic review and recommendations for management. *Gut*. 2010 Jul;59(7):975-86. doi:10.1136/gut.2009.198499.
2. Brinster DR, Raper SE. Synchronous colon and pancreatic cancers in a patient with Peutz-Jeghers syndrome: report of a case and review of the literature. *Surgery*. 2004 Mar;135(3):352-4.
3. Hearle N, Schumacher V, Menko FH, Olschwang S, Boardman LA, Gille JJ, Keller JJ, Westerman AM, Scott RJ, Lim W, Trimpathy JD, Giardiello FM, Gruber SB, Offerhaus GJ, de Rooij FW, Wilson JH, Hansmann A, Moslein G, Royer-Pokora B, Vogel T, Phillips RK, Spigelman AD, Houlston RS. Frequency and spectrum of cancers in the Peutz-Jeghers syndrome. *Clin Cancer Res*. 2006 May 15;12(10):3209-15.
4. Hernan I, Roig I, Martin B, Gamundi MJ, Martinez-Gimeno M, Carballo M. De novo germline mutation in the serine-threonine kinase STK11/LKB1 gene associated with Peutz-Jeghers syndrome. *Clin Genet*. 2004 Jul;66(1):58-62.
5. Latchford AR, Phillips RK. Gastrointestinal polyps and cancer in Peutz-Jeghers syndrome: clinical aspects. *Fam Cancer*. 2011 Sep;10(3):455-61. doi:10.1007/s10689-011-9442-1. Review.
6. McGarrity TJ, Amos CI, Baker MJ. Peutz-Jeghers Syndrome. 2001 Feb 23 [updated 2016 Jul 14]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1266/>
7. McGarrity TJ, Kulin HE, Zaino RJ. Peutz-Jeghers syndrome. *Am J Gastroenterol*. 2000 Mar;95(3):596-604. Review.
8. Merg A, Lynch HT, Lynch JF, Howe JR. Hereditary colorectal cancer-part II. *Curr Probl Surg*. 2005 May;42(5):267-333. Review.
9. van Lier MG, Wagner A, Mathus-Vliegen EM, Kuipers EJ, Steyerberg EW, van Leerdam ME. High cancer risk in Peutz-Jeghers syndrome: a systematic review and surveillance recommendations. *Am J Gastroenterol*. 2010 Jun;105(6):1258-64; author reply 1265. doi: 10.1038/ajg.2009.725.

---

Retrieved from <https://encyclopedia.pub/entry/history/show/11872>