

## **Peutz-Jeghers syndrome**

*Reviewed April 2006*

### **What is Peutz-Jeghers syndrome?**

Peutz-Jeghers syndrome is characterized by the development of 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 medical 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.

### **How common is Peutz-Jeghers syndrome?**

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

### **What genes are related to Peutz-Jeghers syndrome?**

Mutations in the *STK11* gene cause 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 one copy of this gene alters the structure or function of the STK11 protein, disrupting its ability to restrain cell division. Researchers suggest that an additional gene mutation, either in the second copy of the *STK11* gene or in another gene, can occur in certain cells during a person's lifetime. This combination of genetic changes may trigger the formation of noncancerous polyps and cancerous tumors in people with Peutz-Jeghers syndrome.

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

### **Related Gene(s)**

Changes in this gene are associated with Peutz-Jeghers syndrome.

- **STK11**

## **How do people inherit Peutz-Jeghers syndrome?**

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 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 mutations in the *STK11* gene.

## **Where can I find information about diagnosis or management of Peutz-Jeghers syndrome?**

These resources address the diagnosis or management of Peutz-Jeghers syndrome and may include treatment providers.

- Gene Review: Peutz-Jeghers Syndrome (<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=pjs>)
- Gene Tests: Peutz-Jeghers Syndrome ([http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical\\_disease\\_id/21338](http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/21338))
- MedlinePlus Encyclopedia: Peutz-Jeghers Syndrome (<http://www.nlm.nih.gov/medlineplus/ency/article/000244.htm>)

You might also find information on the diagnosis or management of Peutz-Jeghers syndrome in Educational resources (<http://ghr.nlm.nih.gov/condition/peutz-jeghers-syndrome/show/Educational+resources>) and Patient support (<http://ghr.nlm.nih.gov/condition/peutz-jeghers-syndrome/show/Patient+support>).

To locate a healthcare provider, see How can I find a genetics professional in my area? (<http://ghr.nlm.nih.gov/handbook/consult/findingprofessional>) in the Handbook.

## **Where can I find additional information about Peutz-Jeghers syndrome?**

You may find the following resources about Peutz-Jeghers syndrome helpful. These materials are written for the general public.

- **MedlinePlus - Health information**
  - Encyclopedia: Peutz-Jeghers Syndrome (<http://www.nlm.nih.gov/medlineplus/ency/article/000244.htm>)
  - Health Topic: Cancer (<http://www.nlm.nih.gov/medlineplus/cancer.html>)
  - Health Topic: Colonic Polyps (<http://www.nlm.nih.gov/medlineplus/>)

colonicpolyps.html)

- Genetic and Rare Diseases Information Center - Information about genetic conditions and rare diseases (<http://rarediseases.info.nih.gov/GARD/Disease.aspx?PageID=4&DiseaseID=7378>)
- Additional NIH Resources - National Institutes of Health  
National Cancer Institute: Genetics of Breast and Ovarian Cancer (PDQ)  
(<http://www.cancer.gov/cancertopics/pdq/genetics/breast-and-ovarian/HealthProfessional/page2>)
- **Educational resources - Information pages**
  - Familial Cancer Syndromes (2009): Peutz-Jeghers Syndrome  
(<http://www.ncbi.nlm.nih.gov/books/NBK1826/>)
  - Madisons Foundation ([http://www.madisonsfoundation.org/index.php?option=com\\_mpower&task=disease&diseaseID=375](http://www.madisonsfoundation.org/index.php?option=com_mpower&task=disease&diseaseID=375))
  - Merck Manual Home Edition for Patients and Caregivers: Colorectal polyps  
([http://www.merckmanuals.com/home/digestive\\_disorders/tumors\\_of\\_the\\_digestive\\_system/colorectal\\_polyps.html](http://www.merckmanuals.com/home/digestive_disorders/tumors_of_the_digestive_system/colorectal_polyps.html))
  - New York Online Access to Health (NOAH) (<http://www.noah-health.org/en/genetic/conditions/pjs.html>)
  - Orphanet ([http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=2869](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2869))
- **Patient support - For patients and families**
  - National Organization for Rare Disorders (<http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/149/viewAbstract>)
  - Resource list from the University of Kansas Medical Center  
(<http://www.kumc.edu/gec/support/cancer.html>)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Gene Reviews - Clinical summary (<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=pjs>)
- Gene Tests - DNA tests ordered by healthcare professionals  
([http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical\\_disease\\_id/21338](http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/21338))
- ClinicalTrials.gov - Linking patients to medical research (<http://clinicaltrials.gov/ct2/results?cond=%22peutz-jeghers%20syndrome%22>)
- PubMed - Recent literature ([http://www.ncbi.nlm.nih.gov/pubmed?term=\(Peutz-Jeghers%20Syndrome%5BMAJR%5D\)%20AND%20\(Peutz-Jeghers%20syndrome%5B](http://www.ncbi.nlm.nih.gov/pubmed?term=(Peutz-Jeghers%20Syndrome%5BMAJR%5D)%20AND%20(Peutz-Jeghers%20syndrome%5B)

TIAB%5D)%20AND%20english%5Bla%5D%20AND%20human%5Bmh%5D%20AND%20%22last%20720%20days%22%5Bdp%5D)

- OMIM - Genetic disorder catalog (<http://omim.org/entry/175200>)

## **What other names do people use for Peutz-Jeghers syndrome?**

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

For more information about naming genetic conditions, see the Genetics Home Reference Condition Naming Guidelines (<http://ghr.nlm.nih.gov/ConditionNameGuide>) and How are genetic conditions and genes named? (<http://ghr.nlm.nih.gov/handbook/mutationsanddisorders/naming>) in the Handbook.

## **What if I still have specific questions about Peutz-Jeghers syndrome?**

Ask the Genetic and Rare Diseases Information Center (<http://rarediseases.info.nih.gov/GARD/>).

## **What glossary definitions help with understanding Peutz-Jeghers syndrome?**

anus ; autosomal ; autosomal dominant ; bowel obstruction ; cancer ; cell ; cell division ; chronic ; cutaneous ; gastrointestinal ; gene ; hamartoma ; hamartomatous ; intestine ; mutation ; new mutation ; obstruction ; ovary ; pancreas ; pigmentation ; polyp ; polyposis ; prevalence ; protein ; stomach ; syndrome ; tumor ; tumor suppressor gene

You may find definitions for these and many other terms in the Genetics Home Reference Glossary (<http://ghr.nlm.nih.gov/glossary>).

## **References**










- 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. (<http://www.ncbi.nlm.nih.gov/pubmed/14976486?dopt=Abstract>)
- de Jong MM, Nolte IM, te Meerman GJ, van der Graaf WT, Oosterwijk JC, Kleibeuker JH, Schaapveld M, de Vries EG. Genes other than BRCA1 and BRCA2 involved in breast cancer susceptibility. *J Med Genet*. 2002 Apr;39(4):225-42. Review. (<http://www.ncbi.nlm.nih.gov/pubmed/11950848?dopt=Abstract>)

- Gene Review: Peutz-Jeghers Syndrome (<http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=pjs>)
- Hearle N, Schumacher V, Menko FH, Olschwang S, Boardman LA, Gille JJ, Keller JJ, Westerman AM, Scott RJ, Lim W, Trimbath JD, Giardiello FM, Gruber SB, Offerhaus GJ, de Rooij FW, Wilson JH, Hansmann A, Möslein 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. (<http://www.ncbi.nlm.nih.gov/pubmed/16707622?dopt=Abstract>)
- 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. (<http://www.ncbi.nlm.nih.gov/pubmed/15200509?dopt=Abstract>)
- McGarrity TJ, Kulin HE, Zaino RJ. Peutz-Jeghers syndrome. Am J Gastroenterol. 2000 Mar;95(3):596-604. Review. (<http://www.ncbi.nlm.nih.gov/pubmed/10710046?dopt=Abstract>)
- Merg A, Lynch HT, Lynch JF, Howe JR. Hereditary colorectal cancer-part II. Curr Probl Surg. 2005 May;42(5):267-333. Review. (<http://www.ncbi.nlm.nih.gov/pubmed/15900295?dopt=Abstract>)

The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult with a qualified healthcare professional. See How can I find a genetics professional in my area? (<http://ghr.nlm.nih.gov/handbook/consult/findingprofessional>) in the Handbook.

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