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Abstract. **Germline mutations** of the tumor suppressor gene LKB1/STK11 are responsible for the **Peutz-Jeghers syndrome** (PJS), an autosomal-dominant disorder characterized by mucocutaneous pigmentation, hamartomatous polyps, and an increased risk of associated malignancies. In this study, we assessed the presence of **pathogenic mutations** in the ...

Cited by: 91 **Author:** Hamid Mehenni, Nicoletta Resta, Gin...

Publish Year: 2007

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Nov 30, 2010 · **BACKGROUND:** Peutz-Jeghers **syndrome** (PJS) is a rare autosomal dominantly inherited disease characterized by gastrointestinal hamartomatous polyposis and mucocutaneous pigmentation. The genetic predisposition for PJS has been shown to be associated with **germline mutations** in the STK11/LKB1 tumor suppressor gene.

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Basic Study
Detection and **analysis of** common pathogenic germline mutations of Peutz-Jeghers syndrome

Guo-Li Gu, Zhi Zhang, Yu-Hui Zhang, Peng-Fei Yu, Zhi-Wei Dong, Hai-Rui Yang, Ying Yuan

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Author: Hamid Mehenni, Nicoletta Resta, Geneva...

Publish Year: 2007

[Peutz-Jeghers Syndrome | Cancer Genetics Web](http://www.cancerindex.org/geneweb/Peutz_Jeghers.html)

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BACKGROUND: Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant inherited disorder characterized by gastrointestinal (GI) hamartomatous polyps, mucocutaneous hyperpigmentation, and an increased risk of cancer. **Mutations** in the serine-threonine kinase 11 gene (SKT11) are the major cause of PJS.

[Peutz-Jeghers Syndrome - GeneReviews® - NCBI Bookshelf](https://www.ncbi.nlm.nih.gov/books/NBK1266)

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Feb 23, 2001 · Diagnosis/testing. The diagnosis of Peutz-Jeghers syndrome is based on clinical findings. Identification of a heterozygous pathogenic variant in STK11 by molecular genetic testing confirms the diagnosis and allows for family studies.

Cited by: 337

Author: Thomas J McGarrity, Howard E Kulin, Ric...

Publish Year: 2000

[LKB1 exonic and whole gene deletions are a common cause ...](https://www.ncbi.nlm.nih.gov/pubmed/16648371)

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