

Peutz-Jeghers syndrome

Identity

Note syndrome associating mucocutaneous melanotic pigmentation, intestinal polyposis, and an increased risk of cancers

Inheritance Autosomal dominant with a high penetrance; frequency is about $3.5/10^5$ newborns; 1/3 to 1/2 of cases are new mutations.

Clinics

Phenotype and clinics **Skin** numerous brown or bleuish mucocutaneous macules (melanin spots) , especially around the orifices (mouth, including the buccal mucosa, eyes, nostrils, anus, genitalia), on the hands, ... ; they tend to deseappear with age; (at puberty or in adulthood); Note: in patients with isolated mucocutaneous melanotic pigmentation (without polyps), the cancer risk is lower, and the genetic defect looks different.

Gastrointestinal tract (GI tract): polyps of amartomatous origin (with a characteristic arborization of nonstriated muscles) may be found in any portion of the GI tract with varying frequencies: from 95% to 15%: in the small bowel, jejunum, ileum, large intestin, rectum, stomach, and the duodenum; risk of intussusception, which may be cause of death; onset for symptoms occurs from the firstyear of life to elderness (median age10-25 years, somewhat earlier in male patients).; polyps of other organs can occur.

Neoplastic risk **Tumors** develop, with a relative risk of 10-20, and a cumulative risk of more than 90% between ages 15 and 64; mean interval between the diagnosis of Peutz-Jeghers syndrome and the diagnosis of cancer is about 20 yrs..Cancers at risk are:

small intestin: 500 fold increase,
stomach:200 fold,
pancreas: 100,

[colon](#): 85,

esophagus: 60,

[ovary](#): 30, and the benign sex cord tumor with annular tubules

[uterus](#), [breast](#), [lung](#): 15 to 20

Treatment surveillance with endoscopic (GI tract) and gynecologic regular screenings, surgery when necessary

Genes involved and Proteins

Gene Name [STK11](#)

Location 19p13.3

Note mutations in STK11 is found in about 70 % of cases of Peutz-Jeghers syndrome; there is genetic heterogeneity, and yet undiscovered gene(s) may also be responsible for the disease

DNA/RNA

Description 10 exons

Protein

Function serine/threonine protein kinase

Mutations

Germinal most mutations in Peutz-Jeghers syndrome are null alleles; they are dispersed through the entire gene

Somatic many of the polyps that develop in Peutz-Jeghers syndrome show loss of heterozygosity;

External links

[OMIM](#) [175200](#)

[Orphanet](#) [Peutz-Jeghers syndrome](#)

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