Peutz-Jeghers Syndrome

This is a type of gastrointestinal polyposis syndrome that is characterized by multiple GI hamartomatous polyps and mucocutaneous hyperpigmentation.

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Mechanism and Characteristics

Symptomatic Age: 10-30 Years

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Polyps usually begin developing in these patients within the first decade of life. Patients are often symptomatic between the ages of 10 and 30 years of age.

LKB1/STK11 Gene Mutation

LaKe-Bee-(1) Wand/ STaKe 11 Mutant

Around 50% of individuals with familial Peutz-Jeghers syndrome have a heterozygous loss of function mutation in the gene LKB1/STK11. The absence of this mutation does not preclude the presence of Peutz-Jeghers syndrome.

Autosomal Dominant

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Peutz-Jeghers Syndrome is a rare autosomal dominant syndrome.

Signs and Symptoms

Benign GI Hamartomatous Polyps

Bunny GI Hammerhead Polyps

The polyps consist of an excess accumulation of arborizing normal tissue such as mucosal glands, smooth muscle, and connective tissue. The polyps occur most frequently in the small intestine and can serve as a lead point for intussusception.

Mucocutaneous Hyperpigmentation

Mucocutaneous Hiker-pig with Hyperpigmentation

Dark blue to brown macules are present in the mouth (including buccal mucosa), lips, hands, and genitalia.

Positive Family History

Positive Family-portrait

A positive family history can aid in the diagnosis of Peutz-Jeghers syndrome.

Increased Risk of Colorectal Carcinoma

Up-arrow Risk of Colon-hat Car-gnome

There is an increased risk of colorectal cancer (CRC) and other malignancies. These carcinomas arise independently of the nonmalignant GI hamartomatous polyps. The overall cancer risk is around 50% in the 6th decade, thus, routine cancer surveillance should be performed.