Peutz-Jeghers syndrome is a rare disorder, also known as hereditary inherited polyposis syndrome. This syndrome can affect individuals at any age, but generally first presents in children less than ten years of age. It is hereditary and thought to be due to a mutation in the gene 19p13.3. This gene is autosomal dominant in transmission, which means that it is likely to be seen from generation to generation in at least half of first-degree family members.

The most common manifestation of Peutz-Jegher's syndrome is the development of hyperpigmented macules, or spots, on the oral mucosa, lips, nose, hands, feet, and anogenital region. The spots range in color from dark brown to bluish-black, and are generally only a few millimeters in size. Because the oral lesions most commonly manifest before the age of two, most people are diagnosed with this condition at an early age. Another common sign of Peutz-Jegher's syndrome is the development of benign hamartomatous polyps throughout the body. The polyps are most likely to develop in the gastrointestinal tract beginning in childhood, but do not typically become symptomatic until adulthood. Symptoms range from abdominal pain and bloody stools, to intussusception, a life-threatening condition that can present as an obstruction of the small intestine. Polyps are generally benign, although some researchers theorize that portions of the polyps may become malignant over many years, due to an increased risk of malignancies in the same distribution as the polyps.

Patients with this syndrome are at increased risk for malignancies of the stomach, small intestine, colon, pancreas, ovaries, testicles, and breasts. Because of the increased risk of cancer, patients should receive a baseline endoscopy and colonoscopy by the age of eight and should be monitored every three years thereafter if polyps are present. Removal of the polyps may be indicated. Patients should receive annual physical and gynecologic examinations, including evaluation of the pigmented lesions, and perform monthly breast self-examinations. If cancer is suspected, further evaluation and treatment from an oncologist may be indicated. Patients are projected to have a slightly shorter life-span than the average adult due to the increased risk of cancer, and other first-degree family members should be screened at birth for the disease.