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An Overview on Peutz-Jeghers Syndrome

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Opinion

Peutz-Jeghers syndrome is characterised by the formation of noncancerous hamartomatous polyps in the gastrointestinal tract (especially the stomach and intestines) as well as a significantly higher chance of developing specific cancers. Small, dark-colored spots on the lips, around and within the mouth, near the eyes and nose, and around the anus are common in children with Peutz-Jeghers syndrome. These patches can appear on the hands and feet as well. They usually occur in childhood and diminish as the person grows older. Furthermore, during childhood or adolescence, most patients with Peutz-Jeghers syndrome have numerous polyps in the stomach and intestines. Recurrent intestinal blockages, prolonged bleeding, and abdominal pain are all symptoms of polyps.

People with Peutz-Jeghers syndrome are at an increased risk of acquiring cancer throughout their lives. The most usually reported malignancies are cancers of the gastrointestinal system, pancreas, cervix, ovary, and breast. Most cases of Peutz-Jeghers syndrome are caused by mutations in the *STK11* gene (also known as LKB1). The *STK11* gene is a tumour suppressor gene, which means it generally blocks cells from dividing and developing too quickly or uncontrollably. A mutation in this gene causes the *STK11* protein to lose its capacity to control cell division by altering its structure or function. People with Peutz-Jeghers syndrome develop noncancerous polyps and cancerous tumours as a result of unregulated cell proliferation.

The *STK11* gene is not mutated in a tiny fraction of persons with Peutz-Jeghers syndrome. The cause of the disease is unknown in many situations. The autosomal dominant inheritance patterns for Peutz-Jeghers syndrome means that one copy of the mutated gene in each cell is enough to raise the chance of producing noncancerous polyps and cancerous tumours. Affected people inherit a mutation in the *STK11* gene from one of their parents in around half of the instances. The remaining occurrences occur in people who have no family history of Peutz-Jeghers syndrome. New (de novo) mutations in the *STK11* gene appear to be the cause of these occurrences.

Peutz-Jeghers syndrome (PJS) is a genetic disorder that increases the chance of hamartomatous polyps in the digestive tract, as well as cancers of the breast, colon, and rectum, pancreas, stomach, testicles, ovaries, lung, cervix, and other types listed below. People with PJS may have a lifetime risk of cancer of up to 93 percent if they do not receive proper medical care. A hamartoma is a benign (noncancerous) tumour that develops from a proliferation of normal-appearing tissue. Hamartomatous polyps in the small and large intestine are common in PJS, and they can cause bleeding or other issues such intestinal blockage.

The development of pigmented spots on the skin and in the mouth, known as mucocutaneous hyperpigmentation, might be one of the first indications of PJS in children. Dark blue or dark brown freckling is common in people with PJS, especially around the mouth and on the lips, fingers, and toes. Freckles emerge in childhood and diminish with age, thus they are rarely noticeable in an adult with PJS who has recently been diagnosed with cancer. The development of gastrointestinal hamartomatous polyps, which can cause bleeding and obstructions, is another symptom of PJS. When gastrointestinal problems first develop, the typical age is ten years old.

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