



COWDEN SYNDROME

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Cowden syndrome (Cowden's disease, multiple hamartoma syndrome) is a rare autosomal dominant disorder that causes benign tumor-like growths called hamartomas of the skin, mucosa, bones, genitourinary tract, gastrointestinal tract, eyes, and the CNS. The skin is affected in nearly 90-100% of cases of Cowden syndrome. This disorder is also associated with the development of various malignancies such as cancers of the breast, thyroid, and endometrium (lining of the uterus) which makes early detection crucial for treatment. Males and females inherit this disorder equally, however the incidence of malignancies vary among gender. Nearly 1 out of 250,000 people are affected and the onset of clinical manifestations ranges from birth to approximately 46 years of age.

The cause of Cowden syndrome is most commonly due to a mutation in the PTEN (phosphatase and tensin homolog) tumor suppressor gene which causes uncontrolled cell division that lead to the development of hamartomas and cancers. Approximately 20% of patients with Cowden disease have no identified genetic explanation.

The majority of patients present with benign mucocutaneous lesions that are characteristic of Cowden syndrome. The first include multiple trichilemmomas, which are benign tumors that appear as small flesh-colored papules that typically present around the eyes, nose, and mouth. Patients also commonly present with oral mucosal papillomatous papules, which appear as 1-3 mm white papules with a smooth surface in the mouth. These lesions often group together forming a cobblestone-like appearance. Acral keratoses are also another common presentation which presents as 1-4 mm wart-like papules on the back of the hands and feet. Palmoplantar keratoses which present as translucent, punctate wart-like growths that appear on the palms and soles may also be observed. Other less common skin lesions include lipomas, neuromas, xanthomas, and hemangiomas.

A complete medical history including a family history of malignancies should be performed on all patients. Furthermore, a thorough physical examination with yearly follow-up is recommended to detect any malignancies. Some components of essential lab work include a CBC, thyroid function tests, urinalysis, and liver function tests. Appropriate imaging studies should be performed to detect malignancies. A skin **biopsy** should be performed to diagnose skin lesions that are associated with Cowden syndrome.

Treatment with systemic retinoids has shown to control the skin lesions associated with Cowden syndrome. Topical treatment has shown to be of minimal benefit. Currently, treatment with rapamycin is under clinical trial and has shown promising outcomes in the regression of the skin manifestations of Cowden syndrome. **Chemical peels**, dermabrasion, and **laser** resurfacing can be of help with facial lesions. Shave excisions or surgeries should be done with care because of the complication of scarring and recurrence. Counseling patients regarding the importance of close follow-up and proper screening for malignancies is essential as many of the cancers are curable if identified early. Annual skin checks with a dermatologist are recommended along with proper consultations with other specialties.

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