Mucocutaneous Manifestations of Cowden Syndrome: Role of an Oral Diagnostician

Ahammad Kandari a, DDS; Michelle Springer b, MS, CGC; Pallavi Parashar c, DDS, FRCD(C)

a University of Alberta Faculty of Medicine and Dentistry, Oral Medicine graduate student, b University of Colorado Cancer Center, c University of Alberta Faculty of Medicine and Dentistry

Background:
Cowden Syndrome, also referred to as Multiple Hamartoma Syndrome or PTEN Hamartoma Tumor syndrome (PHTS), is an autosomal dominant disorder with a broad clinical spectrum and wide degree of penetrance. This disorder is rare and causes an increased predisposition to the development of numerous malignancies, including, but not limited to, breast, thyroid and endometrial cancers. Other common findings include macrocephaly and benign hamartomas and neoplasms of the skin, thyroid and GI tract/colon. The oral manifestations have also been well documented and are present in nearly all affected individuals by the third decade of life. Oral Diagnosticians can aid in the diagnosis of Cowden Syndrome.

Case Summary:
A 31-year-old female presented for a routine dental evaluation. The patient was noted to have multifocal papules affecting the gingiva, tongue and buccal mucosa. She reported a history of a thyroid tumor and a family history of breast and uterine cancer. She also presented with multiple papular skin lesions. Based on the review of the medical history, family history and oral mucosal findings, a diagnosis of Cowden syndrome was considered. The patient was referred to her family physician and a genetic counselor, where the diagnosis of Cowden syndrome was confirmed. The patient was referred to her family physician and a genetic counselor, where the diagnosis of Cowden syndrome was confirmed through genetic testing and the detection of a mutation in the PTEN gene. Additional clinical findings included macrocephaly, trichilemmomas and thyroid goiter. Her sibling who met the clinical criteria for Cowden syndrome was subsequently tested for the familial PTEN mutation, and a confirmed diagnosis of Cowden syndrome was also rendered.

Conclusions:
The NCCN 2021 guidelines list multifocal or extensive oral papules as one of the criteria in the diagnosis of Cowden Syndrome. Previously this was listed as a major criteria, however, the literature available on mucocutaneous lesions is not adequate to accurately specify the number or extent of mucocutaneous lesions required to be a major criterion for CS/PHTS. While Cowden syndrome is rare, it is believed to be highly underdiagnosed as the skin and oral manifestations are often under-appreciated and may be missed. We report a case of a patient with classic mucocutaneous manifestations of Cowden syndrome where the Oral Diagnostician played a crucial role in the confirmed diagnosis. Early diagnosis of patients affected with Cowden syndrome can facilitate early screening, detection and management of benign and malignant neoplasms.

References:
NCCN Br, Ov, Panc v1.2021