GENETICS AND GENODERMATOSES

COWDEN SYNDROME: A RARE GENODERMATOSIS

Da Sandoval Clavijo [1] - N Plaza Arteaga [2] - Ca Rodriguez Zakzuk [3]

Hospital Universitario Centro Dermatológico Federico Lleras Acosta. Fundación Universitaria Sanitas, Dermatology Department, Bogota, Colombia [1] - Hospital Universitario Centro Dermatológico Federico Lleras Acosta., Dermatology Department, Bogota, Colombia [2] - Fundación Universitaria Sanitas, Dermatology Department, Bogota, Colombia [3]

Background: Cowden syndrome is a rare genodermatosis caused by mutations in the tumor suppressor gene PTEN, characterized by formation of hamartomatous neoplasms in different organs and increased incidence of malignant breast and thyroid tumors. Early diagnosis is difficult due to its clinical heterogeneity and sporadic incidence. Cutaneous lesions are a common finding, being sometimes the first clue to guide a proper and early diagnosis. Mucocutaneous features include trichilemomas, acral keratosis and papular lesions on lips and oral mucosa. The most common extracutaneous manifestations include macrocephaly, benign thyroid disorders, benign breast disease and different types of polyps on gastrointestinal tract.

Observation: A 36-year-old female patient with asymptomatic lesions on her face, oral cavity, chest, palms and soles since the age of 5. She had a medical history of Hashimoto's thyroiditis, a surgical history of thyroid lobectomy at age of 10 and a thyroidectomy 5 years later for benign thyroid nodule. Family history presented her father with the similar features. Physical examination revealed a 63 cm cephalic perimeter, multiple normochromic papules on forehead, nose, cheeks, lips and oral mucosa, also hyperkeratotic papules on hands dorsum and left thenar region. A skin biopsy taken from a lesion on the right malar region revealed fibrofolliculoma. Additionally, an endoscopic capsule was performed and showed papillomatosis associated to esophageal, gastric and colonic polyposis; biopsy of one of the polyps reported tubular adenoma. Brain NMR revealed nonspecific supratentorial and subcortical punctate hyperintensities and genetic testing confirmed a pathogenic variant for the PTEN gene.

Key message: Cowden's disease is an uncommon genodermatosis that involves multiple organs in which mucocutaneous lesions are most of the times the first symptom evidenced. Therefore, early recognition of skin lesions may help in diagnosing underlying malignancies and also provide a comprehensive follow-up care for patients.