# COWDEN SYNDROME

## CLINICAL CASE

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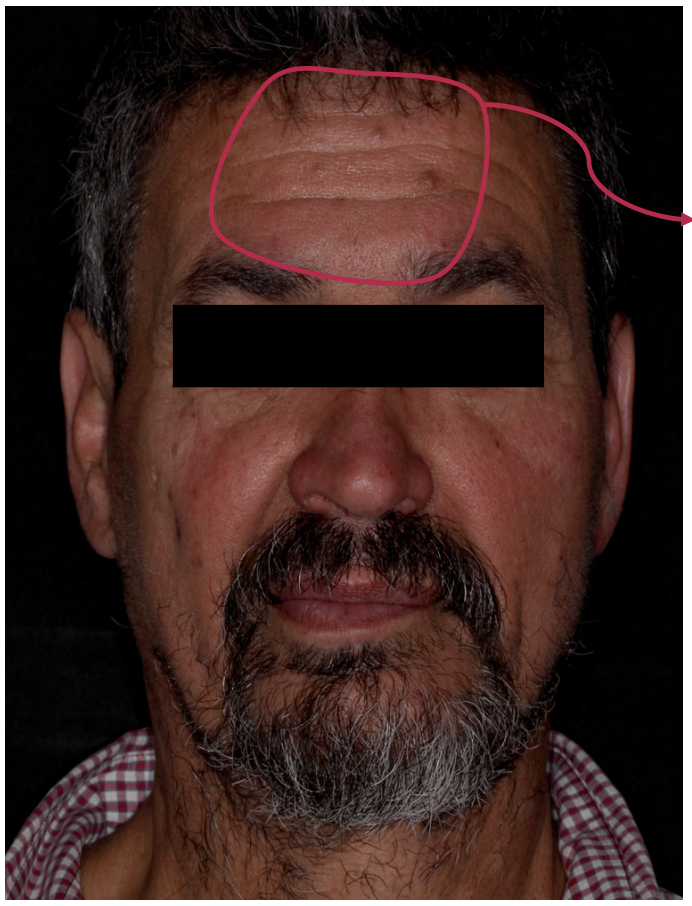
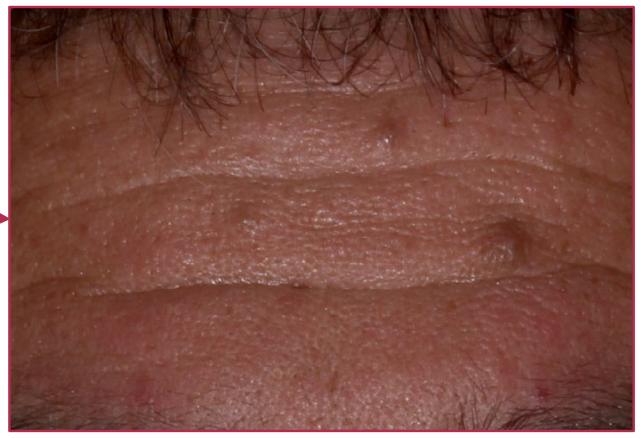
## | DESCRIPTION OF THE CLINICAL CASE

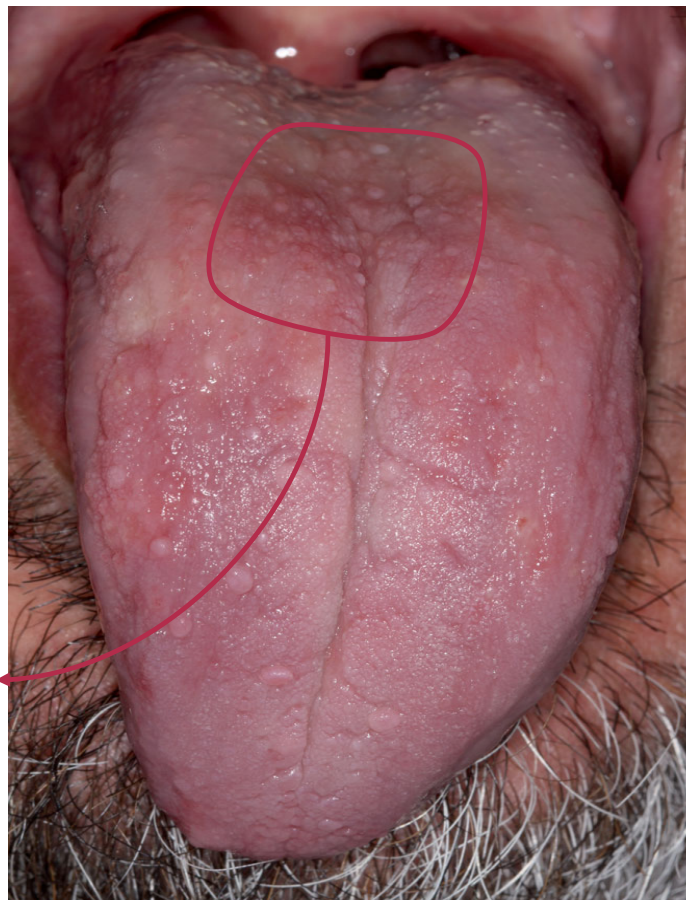
A 65-year-old male leucodermic patient, accompanied at the Clinic of Surgery and Oral Medicine of FMDUL, with indication for multiple exodontia and posterior removable prosthetic rehabilitation. Medicated with enalapril maleate/lercanidipine hydrochloride and simvastatin for the control of hypertension and hypercholesterolemia. History of sinusitis, malaria and intestinal polyposis.

Extraoral objective examination revealed multiple papular cutaneous lesions on the face. Intraorally, several asymptomatic papules on the dorsum of the tongue were also identified. The clinical condition was suggestive of Cowden syndrome or multiple hamartoma syndrome. The differential diagnosis of these lesions includes transient lingual papillitis, neurofibromatosis and multifocal epitelial hyperplasia (or Heck's disease), among other entities.

An excisional biopsy of one of the lesions of the lingual mucosa was performed, whose anatomopathological examination confirmed fibroepithelial hyperplasia.







**FIGURES 01, 02, 03 AND 04 |** Extra and intraoral photographs. Observation of multiple facial papular lesions. Identification, also, of several asymptomatic papules, with a diameter of approximately 2 mm, on the dorsal side of the tongue. [May 30, 2018]

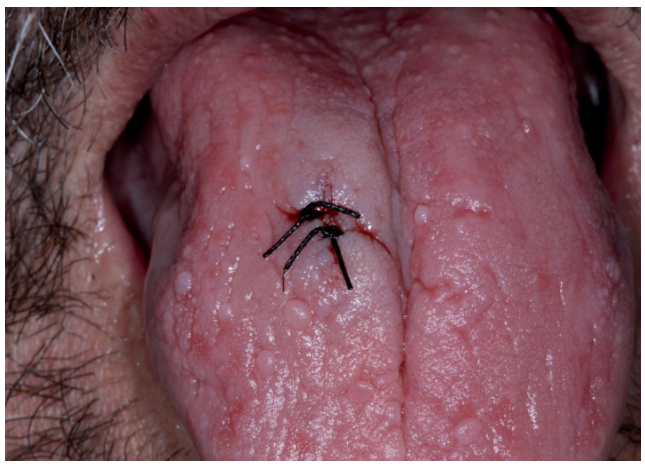


**FIGURE 05 |** Excisional biopsy of a lesion present on the dorsal side of the tongue. [April 02, 2019]

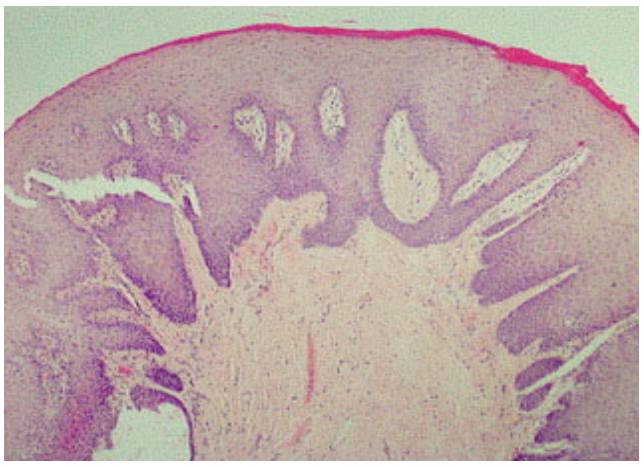


**FIGURE 06 |** Histopathological analysis (HE staining and magnification of ×4) – fibroepithelial hyperplasia.
**Dr. Saudade André** [April 16, 2019]

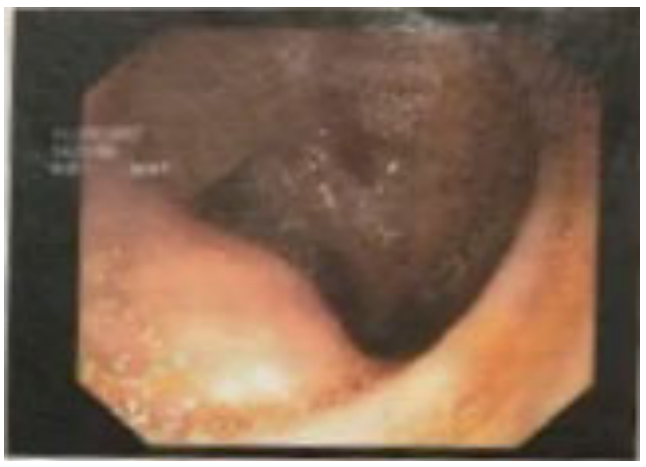


**FIGURE 07 |** Colonoscopy (image of the cecum) – diverticulosis of the colon.
**Dr. Joana Torres** [September 11, 2017]

## | DISCUSSION

Cowden syndrome or multiple hamartoma syndrome is a rare autosomal dominant genetic condition[01-04], with alteration of the PTEN gene, which is found on chromosome 10.[01, 04-05] Its prevalence is only 1:200 000 cases[01, 04-05] and affects, generally, leucodermic females.[03]

It is characterized by the presence of several mucocutaneous hamartomatous lesions of ectodermal, mesodermal and endodermal origin.[04, 06]

The clinical manifestations involve several organs and systems of the human body, including the gastrointestinal and genitourinary systems, the breast and the thyroid, where the development of malignant neoplasias can occur.[01-05]

The pathognomonic criteria include multiple skin (trichilemmomas) and oral papules (fibromas), as well as palmar or plantar acral keratosis.[06]

**HOW TO DO THE DIAGNOSIS?**

A) Presence of **1 pathognomonic criteria**;
B) Presence of 2 major criteria, one of them being Lhermitte-Duclos disease or macrocephaly;
C) Presence of 1 major criteria and 3 minor criteria;
D) Presence of 4 minor criteria.

| PATHOGNOMONIC CRITERIA | MAJOR CRITERIA | MINOR CRITERIA |
|---|---|---|
| 1. 6 or more facial papules, of which 3 or more must be trichilemmomas; | 1. Breast carcinoma; | 1. Other thyroid lesions; |
| **2. Cutaneous facial papules and oral mucosal papillomatosis;** | 2. Thyroid carcinoma (nonmedullary); | 2. Gastrointestinal hamartomas; |
| 3. Oral mucosal papillomatosis and acral keratosis; | 3. Macrocephaly (97th percentile or more); | 3. Mental retardation (IQ of 75 or less); |
| 4. 6 or more lesions of palmoplantar keratosis. | 4. Lhermitte-Duclos disease; | 4. Lipomas; |
| | 5. Endometrial carcinoma. | 5. Fibromas; |
| | | 6. Fibrocystic breast disease; |
| | | 7. Genitourinary tumours or malformations. |

**TABLE 01 |** Diagnostic criteria (*International Cowden Consortium: Operational Diagnostic Criteria 2000*).

## | CONCLUSION

**COWDEN SYNDROME** is a relatively unknown entity in the medical community and, therefore, underdiagnosed in the population.
Patients have an increased risk of malignant neoplasias in various organs and systems.
**DENTIST** plays a fundamental role in the diagnosis of oral manifestations of the disease.