Do you know this syndrome?*
Você conhece esta síndrome? *

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CASE REPORT

Seventy-year-old male patient, who, from the third decade of life, had asymptomatic dome-shaped papulæ, skin-colored or whitish, of non-uniform sizes, located at face, neck and upper trunk (Figure 1). In the axillæ and dorsum there were light brown or skin-colored pedicled papulæ – acrochordons (Figure 2). Morbid history: basocellular carcinomas treated by exeresis with safety margins. Smoker from 18 to 32 years of age, 10 cigarettes a day. History of “emphysema bubbles” and spontaneous pneumothorax 38 years ago. There were no other findings in history or clinical examination. Seven biopsies were performed, with the findings of two types of tumors at histopathological examination: fibrofolliculomas – a tumor with dilated follicle and proliferation of the epithelial wall, formation of strings of cells anastomosed in a soft stroma (Figure 3). The other tumor was a perifollicular fibroma – follicular epithelium within a prominent stroma (Figure 4). The axillary lesion was excised – histology of acrochordon.

Computarized tomography from the thyroid cleavage plan to the pelvic floor showed: cysts and multiple emphysema bubbles in both lungs, pleural calcification and a nodular lesion to the right – the side where pneumothorax is referred – with calcified content (Figure 5), and diverticular disease at the sigmoid. Laboratory tests without any particularities. Patient refused to undergo colonoscopy or contrast-

FIGURE 1: Multiple whitish or skin-colored papular lesions in the upper third of the body: head, neck and upper trunk.

FIGURE 2: Presence of acrochordons in the axillary region.
ed study of the intestine.

The patient reported that his father, three brothers and a sister also had similar lesions in the face, being the father and himself the most affected. He had 11 brothers and sisters, one had passed away because of disseminated non-pigmented skin tumors, which he could not specify. The affected sister was 80 and had similar lesions that had begun at 30 years of age (Figure 6). Anatomopathologic exams demonstrated perifollicular fibromas. There were no other clinical, laboratorial or imaging alterations. Management has been guidance and medical follow-up with emphasis on possible associated diseases.

COMMENTS

In 1977, dermatologist Birt, pathologist Hogg and intern physician Dubé described the syndrome that has their names, characterized by an eruption of face, neck and upper trunk tumors, with dominant autosomic inheritance.\(^1\) Lesions are fibrofolliculomas, trichodiscomas and acrochodons. Throughout the last 28 years, new and more frequent associations, histopathological features and gene mutations have been discovered.
These three tumors of the perifollicular connective tissue are clinically undistinguishable, present as smooth, skin-colored or pink papules and almost always are asymptomatic.2 Regarding histological aspects, there are two considerations: fibrofolliculoma is the most frequent tumor, according to available evidence; there is still controversy as to the existence of these three types of tumors, for there is a possibility that they are only different interpretations for the same lesion, according to the adopted histological slicing. Schulz and Hartschuh3 demonstrated that fibrofolliculomas and perifollicular fibromas represented the same lesion, albeit with different section plans. A horizontal section yields the interpretation of perifollicular fibroma, and a vertical section, fibrofolliculoma. Collins et al.4 demonstrated, by means of immunohistochemical techniques, that fibrofolliculoma and trichodiscoma share the same origin, and that morphologic differences are subtle.

Multiple familial trichodiscomas,5 multiple familial fibrofolliculomas,6 familial perifollicular fibromas,7 and the association of fibrofolliculoma, trichodiscoma and perifollicular fibroma8 have already been described.

Initial association with thyroid carcinoma has been described only in the original paper of Birt, Hogg and Dubé. More consistent associations are those with kidney neoplasia and spontaneous pneumothorax.9,10 A recent study has indicated a relative risk of nine times for kidney neoplasia and up to 50 times for spontaneous pneumothorax, which was influenced by a more advanced age of the patient.11 There are reports with association with type II multiple endocrine neoplasia,9 basocellular carcinoma,9 intestinal polips,5,7,9 emphysema bubbles,10 pulmonary cysts,10 parotid oncocytoma,12 connective tissue nevus, lipoma, angiolipoma and parotid adenoma.13 The patient described here has association which are already described in the literature: pneumothorax, cysts and emphysema bubbles, and basocellular carcinoma.

Mutations on gene 17p11.2 have been identified. They result in the production of an abnormal protein, folliculin, found in the skin, lungs and kidneys. This may explain the observed alterations.10 Treatment can be done with YAG laser, CO2 laser and electrocauterization, with irregular results.14 Genetic counseling and clinical control of the parents are essential.

REFERENCES