Turban tumor syndrome – a case report

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Abstract

Familial cylindromatosis is a rare benign neoplasm with a dominant autosomal transmission, characterized by the existence of multiple cylindroma spread mainly on the scalp, neck or forehead, risking disfiguration and underlying malignancy. When the entire scalp is attained it is called "turban tumor".

We report a clinical case of a 74 year-old man, attending the emergency department with a condition compatible with respiratory infection. On physical examination, we detected multiple tumors on his face, scalp, back and both legs evolving for 30 years. We referred him to the Internal Medicine to evaluate his clinical condition. After the Dermatology and Plastic Surgery cooperation, it was possible to diagnose the existence of multiple cylindroma tumors. We also managed several other conditions diagnosed in the visit: arterial hypertension, diabetes mellitus and mixed dyslipidaemia. The authors would like to emphasize the scope and impact such disease can have on an individual's life. Due to its aspect, he isolated himself, allowing tumors to evolve and emerging diseases remaining undiagnosed. Although it is a rare nosological entity, it can become malignant and it deserves, also for this, our special attention to the patient and his family.

Key words: Familial cylindromatosis, turban tumor, Brooke--Spiegler syndrome

INTRODUCTION

Cylindroma is s benign neoplasm of skin annexes, rare, which can present itself on its own or as an association of multiple tumors. In 90% of cases it is located in the scalp, face and neck.[1] It reaches more frequently Caucasian individuals and of female gender.[2] It is a hereditary disease, of dominant autosomal transmission and incomplete penetrance. The family cylindromatosis gene (CYLD) is located on the chromosome 16q 12 – 13 and it has the same features as a tumor suppressor gene.[3,4]

The diagnosis was histological revealing an epithelial neoplasm, non-encapsulated, made up by lobes in the superficial dermis, surrounded by a thick hyaline sheath.[2] Cylindroma can appear in association with a trichoepithelioma and spiradenoma (Brooke – Spiegler syndrome) or the several tumors can be histologically identical (Poncet-Spiegler Syndrome).[3] Clinically, such tumors started at adult age (3th or 4th decade) growing slowly and increasing in size and number. When reaching practically all the extension of the scalp their presentation is as "turban tumor". The malignancy risk is low but exists and it is high in this case. Malignant cylindromas are very aggressive, metastazing preferentially in the lymphatic ganglia, viscera and bones. Such patients, therefore must be closely followed up to screening a malignant lesion. It is known that one same mutation can generate different histological differentiations, in the same family. It is also recommended the study of the family to exclude the malignant lesion. The main differential diagnoses are made with the basocellular carcinoma, cutaneous metastases and neurofibromatosis.[3,5]

The treatment relies mainly in the surgical excision of tumor lesions, for aesthetic reasons firstly, but it is also indicated in cases of sores, infection, suspicion of malignancy, functional change (auditive, for instances) or pain, less frequently.[2,6] The individual excision of lesions under 1 cm in diameter is recommended, with the purpose of improving the aesthetic presentation and reducing the risk of evolving towards a turban tumor. Local recurrence after individual excision is It is relatively frequent, and it might reach 40% in some courses.[2] In cases of turban tumor it is anticipated a full scalp exeresis, with a secondary reconstruction with a skin graft.[1]

As it reaches mainly the face and the scalp this is potentially a disabling disease, from a social point of view, therefore the importance of acting correctly and earlier, in a way that will provide individuals to have a family and social life as closer to normal as possible.

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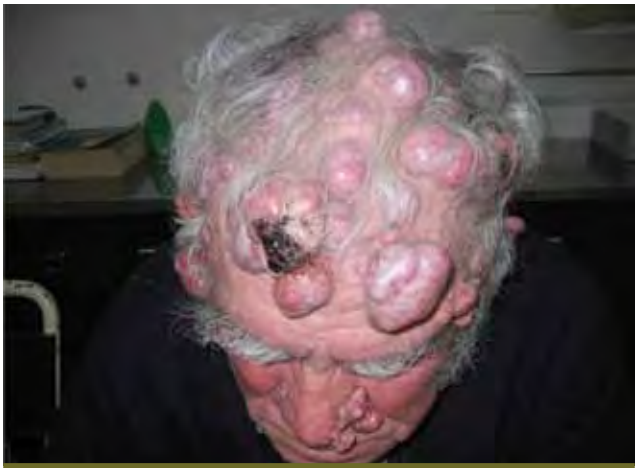
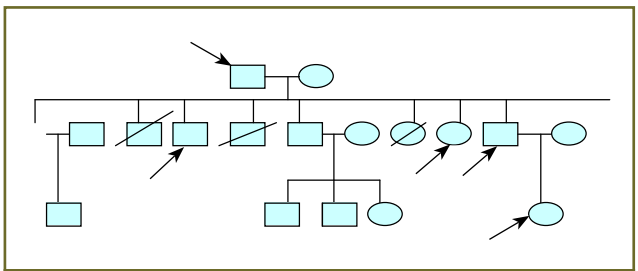


The patient's scalp (father).

FIG. 1



Genealogy tree.

FIG. 2

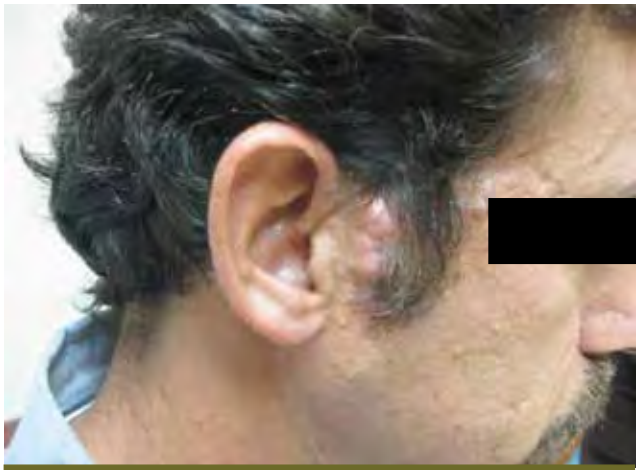
CASE REPORT

Male patient, 74-year-old, seen in the Emergency Service of Funchal Central Hospital (HCF) in February 2009, due to a bronchopulmonary infection, already medicated, as an outpatient with amoxicillin and clavulamic acid, without improvement. On the objective exam, it should be highlighted: BP 196/88 mmHg; cardiac auscultation: systolic blow degree II/VI in the aortic and mitral areas; pulmonary auscultation: inspiratory crepitations on the right pulmonary base. Multiple cutaneous tumors with diameters between 0.5 and 10 cm, often with a bumpy and vascularised surface, painless when moved, some with ulcers, answers others with signs of infection (*Fig. 1*). Most of these tumors were spreading in the face, scalp and dorso and the biggest one was located on upper pubic region. Personal background: the first "cysts" appeared (quote) at 30 years of age; their surgical removal on the scalp: in 1979. From that date onwards, he had been isolating himself at home due to the growing and appearance of an increased number of lesions. Before such condition he was given at the Emergency Service, levofloxacin for the respiratory infection and referred to the Internal Medicine clinic to study the clinical situation. He started therapy with Valsartan 160 mg per day.

In the appointment it was raised the family history (*Fig. 2*) and it was verified he had no knowledge of similar conditions in his family history. He had nine children, three of them deceased (he did not know the reason) and three (two male and one female), aged between 35 and 50 years old, who also had tumoral lesions similar to his own, although of smaller dimensions, limited to the face and scalp, since the third decade of life (*Fig. 3 and 4*). The reminder three children (two female and one male) were healthy. He had five grandchildren aged between 14 and 26 but only the youngest, daughter of one of the sick children, also had lesions in the face for the last two years.

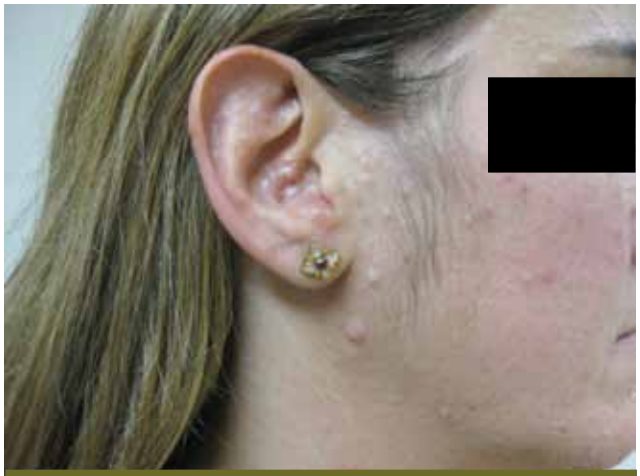
The patient was improving of an acute infectious intercurrence. He kept high blood pressure. He presented significantly depressed moods. The cooperation of Dermatology and Plastic Surgery was requested to the excision and histological diagnosis of some of the general lesions. General evaluation analyses were requested, electrocardiogram and echocardiogram. From the diagnosis additional tests carried out, the most relevant results were: fasting glucose 163 mg/dL; Hg A1C: 6.6%; urea: 139 mg/dL; creatinine: 2.3 mg/dL; total cholesterol: 255 mg/dL; HDL: 36 mg/dL; LDL: 178 mg/dL; triglycerides 348 m/dL. The echocardiogram has revealed "a slight concentric hypertrophy of the left ventricle (LV); fibro-calcifying mitral-aortic, with a slight valve insufficiency. Good LV function (shortening fraction – 48%)". To the medication previously started it was associated metformin 850 mg (1+0+1); amlodipine 5 mg/day, rosuvastatin 10 mg/day and paroxetine 20 mg/day.

Four tumors from the scalp and one from the upper pubic region were surgically removed and a secondary reconstruction of the scalp with tight skin grafts was carried out (*Fig. 5*). When the pathological anatomy assessment of the excised lesions was carried out, the outcome was a diagnosis of eccrine cylindroma (*Fig. 6*). Due to the total covering of the scalp, we can classify this as a turban tumor. Before such histological diagnosis, we asked to see the chil-



Face profile, right side (son).

FIG. 3



Face profile, right side (daughter).

FIG. 4

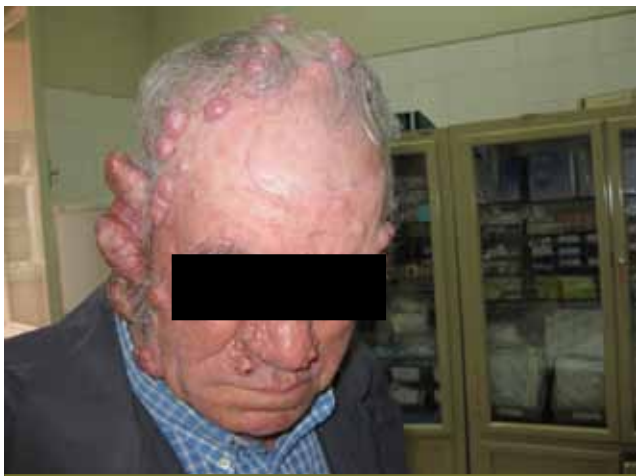
dren and grandchildren, to follow up and screening the malignant lesion. The genetic study of the family was not carried out because it is a rare disease, there are not in our hospital the technical resources to do so. We did not get financial support to perform the study outside our institution, once the result does not indicate the phenotype found, it does not predict the transmission to descendants and it would not change a bit the procedure to be carried out.

At present, the patient awaits new surgical intervention to remove the biggest tumors in the face and the scalp. His condition has improved both clinically and analytically. He has been followed up every six months in Internal Medicine and Plastic Surgery appointments.

The children and granddaughter were also referred to Plastic Surgery to evaluate the possibility of surgical indication, at the moment, and followed up to trace any malignant lesion.

DISCUSSION AND CONCLUSION

Data in the international literature about family cylindromatosis are scarce. As mentioned before, this is a rare nosological condition of dominant autosomal transmission but with incomplete penetrance, reason why there is such clinical heterogeneity, even within the same family. It predominantly affects the female gender, above the third decade of life.[2] Around 90% of tumoral lesions are located in the head and neck and have a profile of slow-growing.[1] Conversely, in the family presented by the authors, those affected

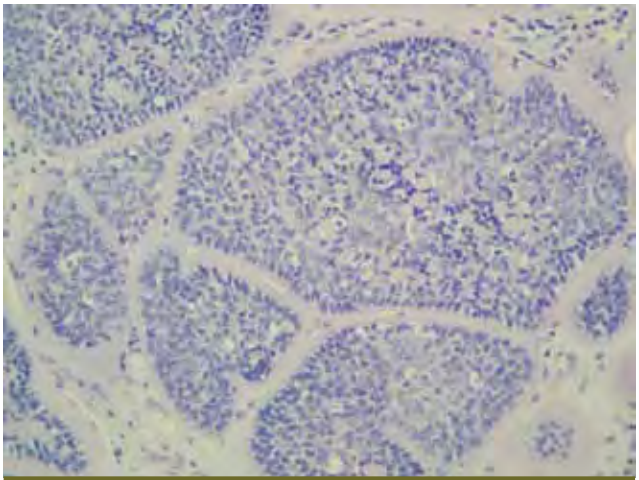


Skin graft (after surgical intervention).

FIG. 5

individuals are male (three cases), with two living women (daughters) having no manifestation of the disease. The onset age, in all of them, was in the third or fourth decade of life and lesions started in fact through the face and scalp. Only the father with more years of evolution has developed the disease in the wider extension, with tumors in other corporal areas and all over the scalp (turban tumor), a condition with a higher risk of malignancy.[5]

The authors point out the peculiarity of this case, residing in the fact this patient had lived secluded in his home for about three decades. Therefore, tumoral lesions have been increasing in number and size and



Histology of excised tumors (cylindroma).

FIG. 6

he was silently acquiring other medical pathologies, already committing the kidneys, kept without a diagnosis and consequently without therapy guidance. Sometimes the patients look for us for another reason that we must observe them as a whole.

The authors end this work, coming to the conclusion that this condition although rare, deserves our attention. The diagnosis is histological and the follow-up is not restricted to the patient. It is compulsory to follow up the family.[5]

The genetic study would enable to document the existing mutation, which would be relevant, mainly from an academic point of view however as the authors did not have available the technological and financial means to do it, it was not possible to carry out such evaluation. In any way, the outcome would not change the procedure to be followed, even because the same mutation can originate different histological differentiations, in the same family.[5] On the other hand, preconception advice can be made even without the mutation documentation, once that in spite of a dominant autosomal transmission, the penetrance is incomplete. [4] Therefore it is not possible to know which will be the percentage of affected descendents, and even less which will be the sick individuals. Additionally, the diagnosis does not depend on the mutation in question but only of the tumor clinical evolution. There is no documented associating a certain mutation to the risk of malignancy. [4,5] The family careful follow-up will enable us to trace any forms, still in their initial stages and treat them promptly and adequately, with the purpose of reducing the evolution towards a turban tumor and consequently of malignancy, a risk that although low it is a real one. ■

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