SURGICAL SKIN TREATMENT OF A COMPLEX TUBEROUS SCLEROSIS: (AN EXCELLENT RESULT WITH ELECTRICAL SCALPEL)

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ABSTRACT Complex tuberous sclerosis is an autosomal phacomatosis dominant, characterized by the development of benign tumors with astrocytic hamartoma type in different organs. We represent the case of a male patient of 30 years old Followed since childhood for complex tuberous sclerosis. The patient benefited from surgical treatment for his facial lesions with an excellent post-operative evolution in only 3 months.

KEYWORDS Complex tuberous sclerosis, benign tumor, angiofibroma

Introduction

Complex tuberous sclerosis is a rare congenital condition that emphasizes the multiplicity of affected organs. Its prevalence is averagely estimated at 1/10,000 live births. it is an autosomal dominant phacomatosis characterized by the development of several astrocytic hamartomas in different organs. Clinical manifestations are usually observed before the age of 20 years. [1]

Observation

We report the case of a 30 years old male patient, followed since childhood for complex tuberous sclerosis with mental retardation.

The patient was referred to us for surgical management of his facial lesions.

The clinical examination objectified several angiofibromatous and nodular lesions in the face; concentrated in the chin and paralateronasal regions bilaterally (Figure 1), achromic stains at the trons, ungulate fibroids, and yellowish-brown skin patches in the lumbar region.

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The patient received a surgical removal of facial lesions with the electrical scalpel (Figure 2) with an excellent post-operative result in only 3 months allowing a good social insertion of the patient. (Figure 3)

Discusión

Complex tuberous sclerosis(CTS) is the most common neuroectodermosis after type 1 neurofibromatosis (Von Recklinghausen). It appears to be linked to disorders of migration, differentiation, and organization of neural tube cells. (2)

CTS is a congenital disease of autosomal dominant heredity, very high but incomplete penetration, and variable expressiveness. However, according to the authors, two-thirds of the cases are sporadic. His diagnosis remains clinical, based on the association, of at least two hamartomatous locations in the different target organs, including the retina, skin, brain, kidneys, and heart. (3)

Neuroradiological signs are a key component of diagnosis and are part of the diagnostic criteria for CTS. The clinical expression of CTS is characterized by its polymorphism and variability from one subject to another. The diagnosis can be obvious in the classical forms: epilepsy, mental retardation, skin signs. But often the expression is more discreet, and it is important to recognize (using strict clinical criteria) the signs of the disease that can confirm CTS.(4) The cutaneous manifestations are the most frequent, after neurological manifestations, and are represented mainly by achromic spots and angiofibroms. Achromic spots are detected in 90-98% of cases, but it is not disease-specific. They are common in the general population. They are most often present at birth or can occur during the first years of life(5).

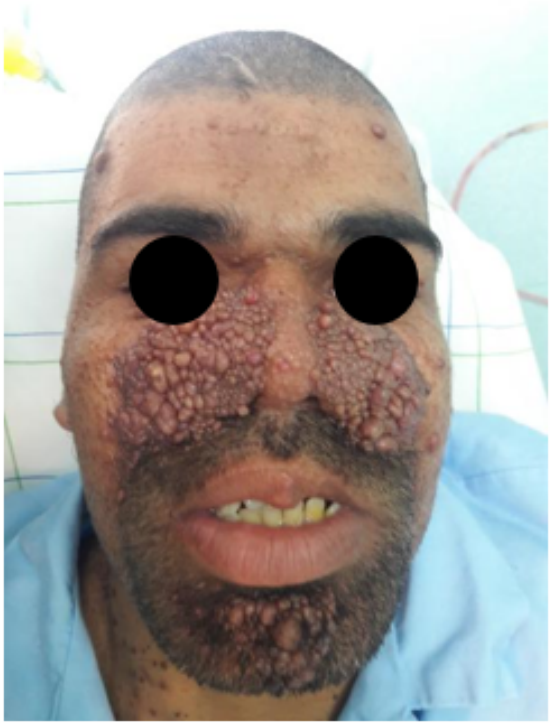

Figure 1 Paralateronasal and chin angiofibromatous nodular lesions

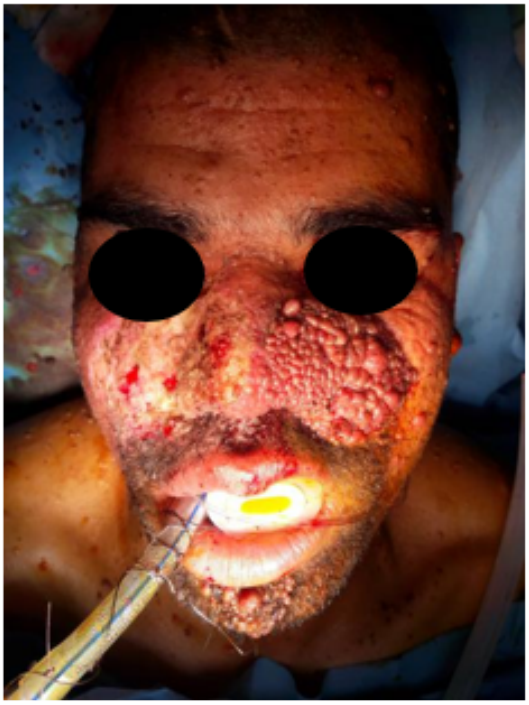

Figure 2 Peroperative image after resection of lesions in the right side

However, angiofibromas are pathognomonic of CTS and are found in 80 to 90% of cases. They are represented as the globular elevation of a smooth and shiny surface, 1-4 mm in diameter, grouped by tens or by hundreds. They are distributed bilaterally and symmetrically at the mediofacial level, mainly at the level of the nasogeneal furrows, and can extend towards the forehead and chin. (2) Patches of grief, raised yellowish-brown patches at the sacrolombar region, occur after the age of ten and are seen in 30-50% of cases. Periungual fibroids (Koenen) can also be observed which are pathognomonic. They are found in 20% of patients, more frequently in the feet [6]. Finally, there are coffee milk stains and dental enamel anomalies.

The kidney is reached in 60% of cases in subjects with CTS. This damage is most often revealed during the assessment, angiomyolipoma is the most frequently encountered tumor. It is a benign tumour whose lesions are often multiple, bilateral and may be the cause of hematuria with abdominal pain [7]

Cardiac involvement is manifested by the presence of rhabdomyosarcomas. If rhabdomyosarcomas are often asymptomatic, they can be responsible, if they are large, for obstruction or leakage of the valve leading to heart failure, with clinical manifestation of anasarks in the neonatal period of dreadful prognosis. Intramural tumours can manifest in a variety of rhythm disorders. They can also simulate cardiomyopathies(8)

Patients with CTS also have a high risk of arterial aneurysms due to loss of elastic fibers in the arterial wall [9]. Other ocular, pulmonary, and bony locations may be observed. The ocular involvement is described as hamartoma or retinal phacome [9]. Lung involvement due to lymphangioleiomyomatosis is rare

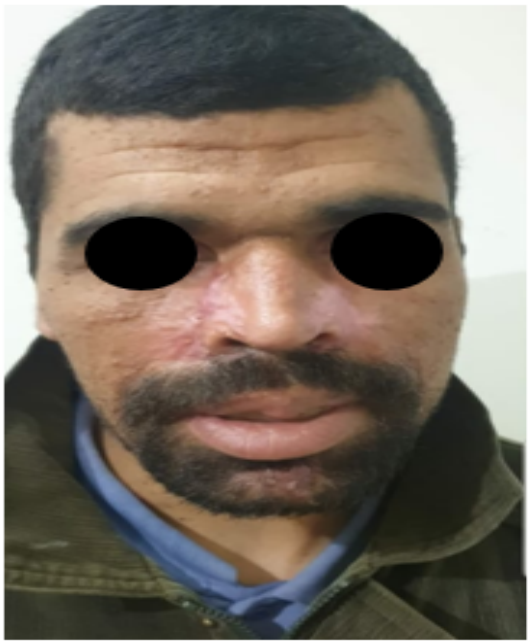

Figure 3 Postoperative evolution after 3 months

and occurs almost exclusively in adult women [10]. Bone lesions often correspond to cystic lesions and are usually asymptomatic [10].

The neuro-radiological diagnosis of CTS has come a long way since the advent of CT. Performed before and after contrast injection, it shows abnormalities 95% of the time. At CT, cortical tubers and abnormalities of the underlying white substance are present between 66% and 71%. They appear as hypodense cortical lesions, which do not take contrast. MRI has superior sensitivity for their detection. In MRI, tubers are better individualized in the form of T2 hypersignals and T1 hyposignals; ranges of hypomyelination of the white substance are often objective, subepidural nodules being objectified only in T1.(11)

Treatment is symptomatic, aimed at improving the quality of life to allow a better social reintegration. Antiepileptic treatment is often necessary to control seizures and should be adapted to the types of seizures and epileptic syndrome. [12]. Children with mental retardation or autism disorder require special neurocognitive and behavioural management. Cosmetic surgical treatment may be proposed for certain skin lesions as was the case for our patient. Patients should receive regular multidisciplinary follow-up.

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Conflict of interest

There are no conflicts of interest to declare by any of the authors of this study

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