Case report

Late onset malignant peripheral nerve sheath tumor in a patient with neurofibromatosis type I and lung metastasis : A case report

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ABSTRACT

Malignant peripheral nerve sheath tumors are one of the rare soft tissue sarcomas, which have not good prognosis despite of treatments.
In this paper we tend to report a malignant peripheral nerve sheath tumor in the legs of a 44-year-old patient with neurofibromatosis type I. The patient underwent surgical operation and chemotherapy, but unfortunately died due to lung metastatic involvement several years later.
Despite the poor prognosis of the disease, early diagnosis can reduce mortalityand morbidity

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Introduction

Neurofibromatosis type one (von Recklinghausen disease), an autosomal dominant genetic disorder with an incidence of one per 3000 cases [1]. Half of them are familial and others are as a result of mutations in the neurofibromin producing gene [2, 3]. The diagnosis is based on the NIH diagnostic criteria including two of these: 6 or more Café au lait spots with a diameter greater than 5 mm before puberty and greater than 15 mm in adults, 2 or more neurofibromas and with a plexiform neuroma, auxiliary or groin freckles, optic glioma, 2 or more Lysh nodules, characterized bone lesions like sphenoid dysplasia, thinning

of the cortex with or without arthritis and the also first degree relative involvement to the diagnostic criteria for neurofibromatosis type I [4, 5]. The risk of malignant peripheral nerve sheath tumor increase throughout the time in 2 percent of patients with neurofibromatosis type I. The most common malignancy of peripheral nerve sheath is neurofibrosarcoma [6, 7]. Malignant peripheral nerve sheath tumor (MPNST) accounts for about 5 to 10% of all soft tissue sarcomas that about 25 to 50 percent of them are detected in patients with neurofibromatosis type I [8, 9].

Case Presentation

A 44-year-old with von Recklinghausen syndrome referred to the oncology clinic with complained of nocturnal pain and swelling of the right posterior tibia. Neurofibromatosis was detected in this patient since he was 18-years old with signs and symptoms like Café au lait spots, multiple nodules of neurofibroma on the trunk and extremities, Lesch nodules of the eye, kyphoscoliosis, and the loss of hypertension, seizures and pseudoartrosis. Because of the recent problem a sonography conducted that revealed Solid masses of soft tissue with non-uniform echogenicity and containing of necrotic areas, with dimensions of approximately 80 x 50 x 51 mm with flat boarders in muscle tissue, suggesting low grade malignant tumor. Computed Tomography of the left leg showed a density with mild peripheral enhancement in the outer surface of fibula and no visible damage to the bone. The mass excised and sent to pathology. Results showed relative pleomorphic proliferation of spindle and oval cells with the proliferation of small size blood vessels within spindle cells. Cellular and nuclear polymorphism and mitotic index were at moderate level. The diagnosis was made based on these. Immunohistochemical staining was positive for S100 and CD99 and negative for SMA, which proposed neurofibroma with low potential malignancy. The mass was relapse again three months later. Again a biopsy was taken of tumors that showed a neoplasm with parenchymal nature containing of spindle cells with hayperchromic corn and little cytoplasm as a fulfilled cellular layer with perivascular accumulation of tumoral cells with frequent mitosis and also geographic necrotic areas **(Figure 1).** The left leg was dysfunctioned so

amputated above the knee. Chemotherapy included Gemsa and Taxoter administered but despite this, he died due to pulmonary metastasis two years later.

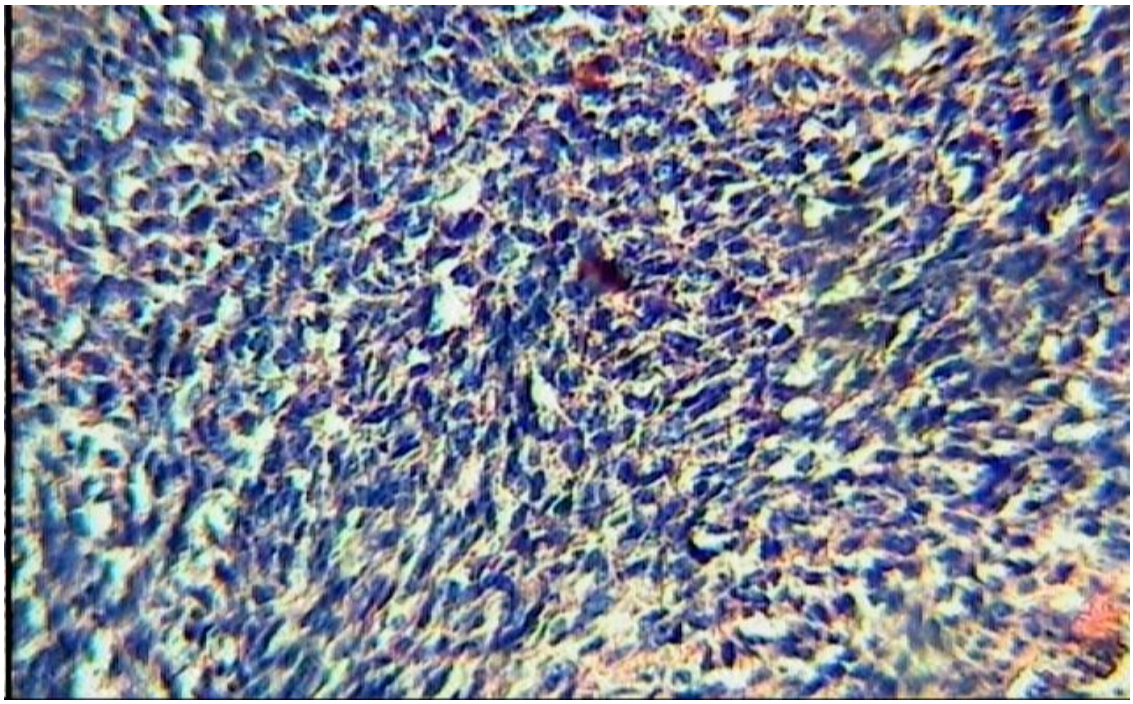

Figure 1: Parenchymal nature containing of spindle cells and little cytoplasm with perivascular accumulation of tumoral cells with frequent mitosis

.Discussion

Malignant peripheral nerve sheath tumor (malignant schwannoma, neurogenic sarcoma, Neurofibrosarcoma) comes to Schwann cells and are able to spread as peripheral nerves and hematogenous. Relationship between MPNST and neurofibromatosis has been determined as 40% of cases with MPNST have been observed in association with neurofibromatosis [10, 11]. Despite this cancer, other tumors, such as ampullary and duodenal carcinoid tumors, pancreatic adenocarcinoma and gastrointestinal tract stromal tumors are often seen in patients with NF1 [12, 13].

Malignant peripheral nerve sheath tumor (MPNST) occurs in rarely adolescents and most often in adults

between 20 and 50 years. Recent researches have shown a five year survival rate of about 42 to 57 years in these patients. This rate is 16 to 38 percent in neurofibromatosis patients [11, 14]. This difference is due to the less differentiation, high rate of metastasis and multifocality in neurofibromatosis. An important thing in our case was that; regard to the presentation of neurofibromatosis symptoms in adolescence, it is expected that malignancy would be developed more rapidly in patient, however, these malignant changes appeared after about 26 years as malignant peripheral nerve sheath tumor. Differential diagnosis and surgery in rapid progressive masses in type I neurofibromatosis like our case is so important [15].

Conclusion

Although malignant peripheral nerve sheath in patients with neurofibromatosis 1 have not a good prognosis, however recent progress in studies on the pathogenesis of this disease provide a new treatment strategy for specialists. According to the extensive rate of metastasis after disease that have an effect on its improvement, therefore exact physical examination and regular and continuous follow-up appears necessary.

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Competing interests

There are no competing interests.

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