A NEUROFIBROMATOSIS CASE WITH DIFFUSE NEUROFIBROMAS AND OPTIC GLIOMAS

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SUMMARY

A 16-year-old male patient was admitted to our hospital with diffuse café-au-lait spots, multiple neurofibromas, and various osseous abnormalities. Radiologic examinations revealed multiple masses on the trace of optic nerves, mediastinum, paraaortic and parailiac region, and retroperitoneum. During the follow-up of the patient there was no problem of vision, but diminished tendon reflexes and progressive muscle weakness of the lower extremities appeared associated with the spinal compression by the neurofibromas. The case is presented because of optic gliomas in his orbitas and to emphasize the importance of radiology in the diagnosis of neurofibromatosis for better medical follow-up and early intervention.

Key words: *Neurofibromatosis type 1, neurofibromas, optic gliomas*

ÖZET

16 yaşındaki erkek hasta hastanemize diffuz café-au-lait lekeleri, multipl nörofibromlar ve çeşitli kemik anomalileri ile başvurdu. Radyolojik tetkiklerinde optik sinir trasesinde, mediastinum, paraaortik ve parailiak bölgelerde, retroperitoneumda çok sayıda kitleler izlendi. Nörofibromatozis tip 1 tanısı alan ve optik gliomları olan hastanın takiplerinde görmesinde bir problem ortaya çıkmadı, ancak alt ekstremitelerinde nörofibromların basısına bağlı progressif kas güçsüzlüğü ve derin tendon reflekslerinde azalma ortaya çıktı. Hasta, optik gliomları ve nörofibromatozis tip 1 tanısı alan bir hastada radyolojik incelemelerin erken tanıda ve takiplerdeki önemini belirtmek amacıyla sunuldu.

Anahtar sözcükler: *Nörofibromatozis tip 1, nörofibrom, optik glioma.*

Neurofibromatosis (NF-1) is a well-recognized problem in infancy and childhood that affects at least 1 in every 3000 people and exhibits great variability in its clinical manifestation (1-3). Approximately 25 % of patients with NF-1 develop complications which include plexiform neurofibromas, malignancies, congenital bone defects, scoliosis, and tumours of the central nervous system (1,4-7). Optic gliomas are found in 15% of patients with NF-1, and are mostly asymptomatic (8,9). In this article we report a case of NF-1 with optic gliomas without proptosis and diffuse neurofibromas at different localizations. We want to emphasize the importance of radiology in early diagnosis of NF-1 to allow the patient and the family to adjust to the diagnosis before the advent of severe complications, and to provide better follow-up and early intervention.

CASE REPORT

A 16- year-old male patient was admitted to our hospital with a history of growth retardation, subcutaneous tumors, hyperpigmented skin lesions, headache, back pain, and leg pain. He had no significance in family history and there was no consanguinity.

Physical examination revealed diffuse café-au - lait spots covering the trunk. Six of these spots were larger than 6 cm, more than 20 of them were larger than 1 cm, and numerous of them were smaller than 1 cm in diameter. At interscapular region a neurofibroma which was 6x5cm in diameter and multiple smaller neurofibromas in different localizations were present. A large mass, measuring 10 x 8cm, was palpated at the right hypocondrium. Thoracal scoliosis with a right sided curve was also recognizable. On ocular examination, the eyes and the lids looked externally normal and no proptosis was found. The measurements by Hertel exophthalmometer were 17mm on both sides. There was no facial asymmetry. The ocular movements were normal; there was not any ocular deviation. The visual acuity was 20/20 in both eyes. The pupils were symmetrical with normal reaction to light. The intraocular pressure was normal. Multiple iris hamartomas (Lisch nodules) were observed on slit lamp examination in both eyes. No choroideal hamartoma was detected and the optic nerve head was in normal appearance. Both fundi were normal. Other physical signs were within normal limits.

On laboratory investigations hemoglobin concentration, white blood cell count, urinalysis, serum electrolytes, hepatic and renal function tests were within normal limits. Echographic examination of the eyes revealed multiple well-limited and nonechogenic round lesions of different sizes. Orbital MRI examination showed multiple masses on the trace of optic nerves bilaterally (Figure 1). Cranial CT was normal. Abdominal ultrasonography and CT revealed a retroperitoneal solid mass, measuring 12x9x8 cm beneath the right kidney. Lower thoracic and lumbar spinal canal was filled with masses protruding into the paraspinal region through intervertebral foramina, causing erosion and malposition. MRI showed additional masses along paraaortic and parailiac regions, thoracic wall and posterior mediastinum (Figure 2).

The patient was operated because of the abdominal mass, since the malignancy risk was high. On pathological examination, the histologic sections of the retroperitoneal large mass and the paraaortic small masses showed features of neurofibromas confirming NF-1. Mitotic activity was not present in biopsy materials. During the follow-up of the patient, there was no visual problem, but progressive muscle weakness of the lower extremites and diminished tendon reflexes were noted. On neurosurgical consultation the neurological findings were found to be associated with the spinal compression by the neurofibromas. Total laminectomi of L1-L4, decompression and subtotal resections of the lumbar neurofibromas were performed. After surgical treatment, radiotherapy (35 Gy) was delivered since the results of surgery were insufficient. After radiotherapy the patient could walk with the support of parallel bar. Follow-up of the patient could not be done until then, since he did not appear for the controls.

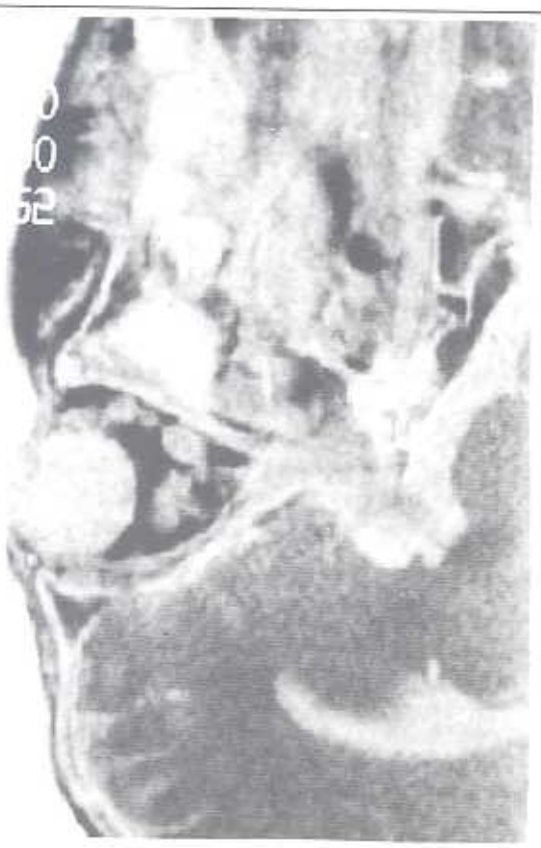

Figure 1: Orbital MRI of the patient showing optic gliomas.

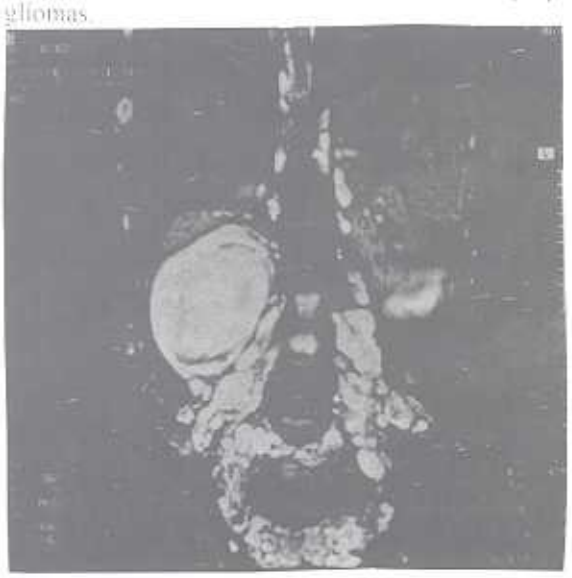

Figure 2: MRI of the patient showing diffuse neurofibromas and a retroperitoneal huge mass in the abdominal region.

DISCUSSION

On physical examination, our patient had multiple café-au-lait spots, neurofibromas, osseous lesions, and Lisch nodules which were very definitive findings for the diagnosis of NF-1 depending on National Institutes of Health Consensus Development Conference criteria (10).

One of the characteristic finding of NF-1 is the development of benign or malign tumors, including peripheral neurofibromas, plexiform neurofibromas, gliomas of the optic tract, other low grade gliomas, pheochromocytomas, sarcomas and carcinomas (1-3). In our patient when large mass palpated at the right hypochondrium was confirmed with USG and CT, operation was done since the risk for NF-1 related malignancy is about 8 % (11,12). According to pathological examination that large mass was accepted as neurofibroma without any malign feature.

Since many complications may occur in NF-1, we evaluated the patient with appropriate imaging techniques. Even though he had no complaints about his eyes, he had optic gliomas in his orbits not causing proptosis and loss of vision. Optic gliomas account for only 2% to 5% of all brain tumors in childhood, but as many as 70% of the cases are associated with NF-1 (13-15). Although biopsy is the only way to confirm the diagnosis of optic pathway gliomas, biopsy was not done to our patient since this procedure might further compromise vision in as many as 75% of patients, and in individuals with NF-1, the CT or MRI scan is highly reliable in assessing the clinical diagnosis of optic gliomas (16,17). An effective therapy for these tumors has not been found yet, and careful screening of the patients with no symptoms is recommended since no therapy

would be given to these patients (8,16). Our patient had no visual symptoms or tumor progression in control orbital MRI scan during the follow-up, so he did not need any therapy for optic gliomas.

Abdominal CT and MRI of our patient demonstrated diffuse neurofibromas at admission. His lower thoracic and lumbar spinal canal was filled with neurofibromas protruding into the paraspinal region through intervertebral foramina, causing erosion and malposition. During the follow-up, he had severe neurologic findings associated with the spinal compression by the neurofibromas in which surgical treatment and radiotherapy were necessary as reported in the literature (3,4).

Although the age at which neurofibromatosis is diagnosed is not predictive of the possible serious consequences of this disorder, early diagnosis is very important, because it allows the child and the parents to adjust to the diagnosis before the advent of serious complications, and provides the appropriate medical follow-up and early intervention. So we believe that once the diagnosis of NF-1 is established or suspected in a child, every system must be evaluated carefully even though the patient has no complaint or specific physical finding. A pediatrician, radiologist, neurologist, orthopedic surgeon, geneticist, ophthalmologist, neurosurgeon, plastic surgeon, psychologist, and physcotherapist must work together for better medical follow-up and early intervention.

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