

A CASE OF FACIAL ASYMMETRY

Neurofibromatosis type 1 with subcutaneous mass in occipital region, bilateral lisch nodules of iris and cafe- au- lait spots.

Köksal Yuca¹, Nebi Yılmaz², Adnan Çınal³, Ömer Etlik⁴, Muzaffer Kırış¹

Yüzüncü Yıl University, Medical School, Departments of Otorhinolaryngology¹, Neurosurgery², Ophthalmology³, and Radiology⁴, Van, Turkey

Neurofibromatosis type 1 (NF1) has been a well-documented and described disease process since 1882, when it was first described by Friedrich von Recklinghausen (1). NF1 is a multisystem hamartomatous disorder with protean expression of cutaneous, neurologic, skeletal, visceral, and ocular manifestations (2). Children and adolescents with NF1 have a high incidence of CNS anomalies (3). Lisch nodules are a component of NF1 and, when present, are included in the clinical diagnostic criteria for NF1 but as an isolated finding are not considered diagnostic. The nodules are also considered the most common ocular feature of NF1 (4). A 10-year-old male presented to our clinic with facial asymmetry and a painful, soft, occipital mass. According to the family of the patient, the facial asymmetry had occurred gradually over the past 7 year and the occipital mass had grown gradually since his birth. The patient was otherwise healthy, had no history of chronic disease, trauma or surgery. He was a full-term product of an uncomplicated pregnancy, labor, and delivery. There was no history of birthmarks, learning disabilities, or short stature. There was no family and siblings' history for neurofibromatosis type 1 and neurologic disorders. Physical examination showed soft, elastic and tender 8x9 cm mass in the occipital region. The hairy skin overlying the mass was intact without erythema. He had remarkable facial asymmetry involving his left upper lip, hard palate and cheek (Figure 1). Also there is a soft and tender nodule sized approximately 2x3 cm, over the zygomatic bone. Moreover diffuse cafe-au lait spots were present in his body and bilateral Lisch nodules were presented (2 right, 3 left) in the iris of the patient in ophthalmological examination (Figure 2).

There was a reddish-purple hemangiomatic lesion sized 4x5 cm over left clavicle. The remainder of the physical examination was normal. Routine laboratory values and the chest roentgenogram were normal. The cranial MRI of the patient clearly revealed a lesion in the region of the left occipital bone, with the classic appearance of a fusiform mass. The mass was hypointense on T1-weighted images and showed an overall increased heterogeneous signal on T2-weighted images. An excisional biopsy of the occipital mass was done. In histopathologic examination neurofibroma was revealed. The clinical diagnosis was NF1.

Diagnosis of NF1 is based on clinical criteria (2). Café-au-lait spots arise in 95% of patients with NF1. These flat areas of skin hyperpigmentation with rounded edges are seen in newborn babies with NF1, and their number and size increase during infancy. 70% of patients also have axillary or groin freckling (5). The incidence of head and neck involvement ranged from 1% to 22 in published literature (6). In our case diffuse cafe-au lait spots were present in his body with soft, elastic swelling of 8x9 cm size in the occipital region and remarkable facial asymmetry. In our patient no axillary or groin freckling was observed. Patients with NF1 tend to be short in stature with macrocephaly. Learning disabilities are present in 50% of patients, and some behavioural problems and speech difficulties have been described. There is also a increased risk of epilepsy and headaches.

In our case, macrocephaly, learning difficulties and epilepsy were not present. But he had an occipital ache due to a superficial mass in this region. About 95% of patients develop Lisch nodules- benign multiple melanotic hamartomas of the iris (5). In our

Correspondence: Köksal Yuca, MD,
Yüzüncü Yıl University, Faculty of Medicine, Department
of Otorhinolaryngology, 65200 Van/TURKEY
Tel: 904322150470-8 (1001), Fax:0904322166563
E-mail: koksalyuca@yahoo.com

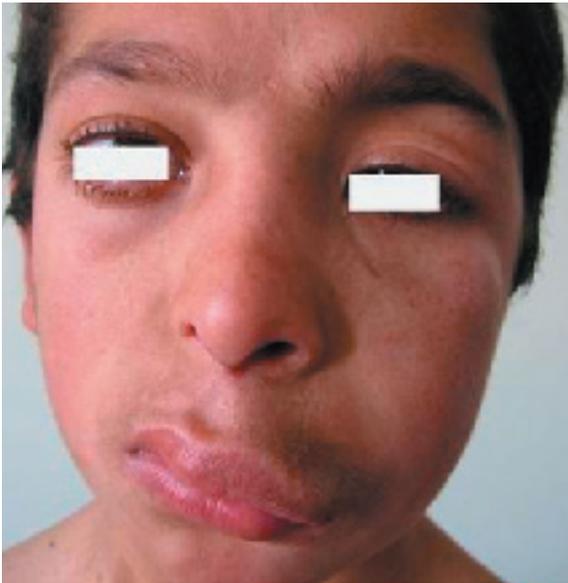


Figure 1. Facial appearance of the patient.

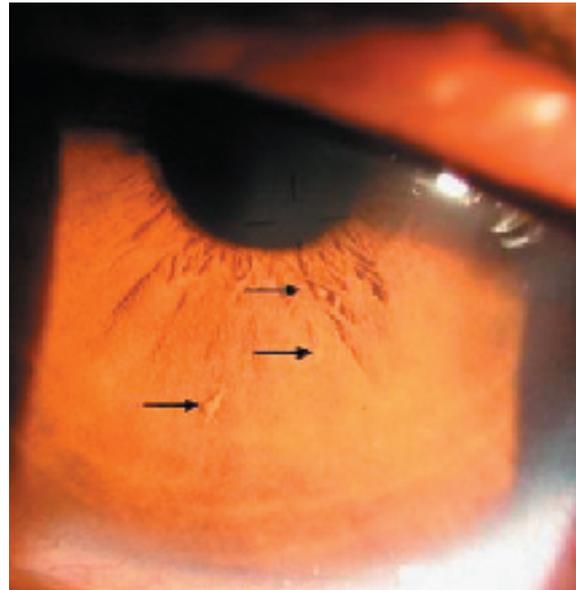


Figure 2. Lisch nodules in the left iris of the patient.

case bilateral Lisch nodules were revealed (two in the right and three in the left iris) by ophthalmological examination

Neurofibromas generally display intermediate to low signal on T1-weighted images and show a diffuse increase in signal on T2-weighted images. Heterogeneous appearance with degeneration and cystic cavitation can be seen in long-standing neurofibromas. “Ancient” neurofibromas refer to long-standing lesions with advanced degeneration exhibiting calcification, hyalinization, and cystic cavitation, findings that can be identified on imaging (7). In our case, an MRI clearly revealed a lesion in the region of the left occipital bone, with the classic appearance of a fusiform mass. The mass was hypointense on T1-weighted images and showed an overall increased heterogeneous signal on T2-weighted images .

Neurofibromas are one of the most common facial hamartomas. They consist mainly of Schwann cells, nerve fibers, and fibroblasts. Neurofibroma-infiltrated tissues are very vascular with wide-open capillaries that bleed copiously at surgery. The neurofibromas are not circumscribed or encapsulated and diffusely infiltrate tissues. Radical and definitive surgery is best delayed until the disease progression had slowed. In adults, a more aggressive surgical approach can be adopted to achieve a more permanent and definitive result (6). In our case surgical resection was recommended for occipital and zygomatic mass but refused by the patient’s family.

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