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A CASE OF NEUROFIBROMATOSIS TYPE 1

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ABSTRACT

Neurofibromatosis (NF) is a term that has been applied to a variety of related syndromes, characterized by neuroectodermal tumors arising within multiple organs and autosomal-dominant inheritance. Neurofibromatosis type 1 (NF-1), known as well as Recklinghausen's disease, is the most common type of the disease accounting 90% of the cases. We present a case of 52-year-old men with NF-1. The disease started in childhood with the appearance of multiple hyperpigmented skin macules. At the age of 46 a lot of cutaneous tumors appeared and started growing bigger all over the body surface. Because of a vision problem due to an upper left eyelid neurofibroma, the patient came for a clinical examination at the age of 52 years.

Key words: Neurofibromatosis, Neurofibromatosis type I, Recklinghausen's disease

INTRODUCTION

Neurofibromatosis (NF) is a term that has been applied to a variety of related syndromes, characterized by neuroectodermal tumors arising within multiple organs and autosomal-dominant inheritance. At least 8 different clinical phenotypes of neurofibromatosis have been identified and are linked to at least two genetic disorders. Neurofibromatosis type I (NF-1) is the most common type of the disease accounting 90% of the cases, and is characterized by multiple café-au-lait spots and the occurrence of neurofibromas along peripheral nerves.

CASE REPORT

History

A 52-year-old man with Neurofibromatosis type I is presented. The disease started in childhood with the appearance of multiple hyperpigmented skin macules. At the age of 46 a lot of cutaneous tumors appeared and started to increase in size all over the body surface especially on the left eyelid. Due to his psychic condition (the patient is mentally retarded after encephalitis in childhood) he has not consulted a doctor. The growth of the fibroma on the upper eyelid of the left eye had caused visual difficulties, which made him seek care.

Physical examination

Dermatological status: hundreds of soft cutaneous neurofibromas, the largest amount being on the trunk and limbs, ranging from a few millimeters to several centimeters in diameter (fig 2), some of them pedunculated (fig 3); multiple café-au-lait spots with diameter > 1,5 cm (fig 4); axillary and inguinal freckling (fig 5). The mucous membranes were not affected.

Ophthalmological status: multiple cutaneous fibromas of different size on eyelids of both eyes (Fig. 6), without inflammation. There was a 1,5 cm fibroma that affected the edge of eyelid and spread approximately 1 cm to the eyelid margin on the lateral part of the upper eyelid of the left eye. The upper eyelid had partial secondary ptosis. Lisch's nodules on the iris of both eyes were without clinical visual involvement (fig 7).

Lab and imaging studies, histologic findings and consultations

The standard laboratory tests values were in the normal range. X-ray photography and CT are within the normal too. The neurologist did not detect alterations in the central and peripheral nervous system. According to the otologist the acoustic nerve has not been damaged. The histological result confirmed the diagnosis of Neurofibromatosis.

DIAGNOSIS

The diagnosis NF-1 was made according to the presence of four of the seven diagnostic criteria of the National Institute of Health Consensus Development Conference:

- Five or more café-au-lait spots larger than 5 mm in diameter in prepubertal patients; six or more café-au-lait spots larger than 15 mm in diameter in postpubertal patients
- Two or more neurofibromas of any type, or one plexiform neurofibroma
- Axillary or inguinal freckling
- Two or more Lisch's nodules

Therapy

Because of the restriction in the peripheral visual field of the left eye due to the large tumor on the upper eyelid,