Childhood microphthalmic neurofibromatosis

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We present an atypical case of neurofibromatosis marked by young age at onset, rapid invasiveness of the tumor, the presence of curious autonomic symptoms, cranial malformation and congenital microphthalmus, a combination that suggests a new syndrome, which might be called childhood microphthalmic neurofibromatosis.

Key-Words: Neurofibromatosis — Leschke syndrome — microphthalmus

Introduction

The term phakomatosis (φακός—birthmark) was coined by Van der Hoeve in 1923 to indicate two dysplasias with a tendency to blastoma formation: tuberous sclerosis and neurofibromatosis, already grouped together by Bielschowsky in 1919 [1]. Actually there are several phakomatoses and they are considered to be hereditary anomalies induced by disorders of the proliferative phase of the embryofetal development of the nervous system (they present alterations of tissues and organs of ectodermal origin such as skin, eyes and CNS, and sometimes tissues of meso- and endodermal origin are also involved). We may therefore consider two categories of phakomatosis: a) primary neuroectodermal phakomatosis, and b) meso-endo-dermal phakomatosis [2].

The main features of phakomatosis are: small spots or patches (phakos) on the skin or mucous membranes in any part of the body; localized tumor-like hyperplastic formations of various types (phakomata); different true tumors arising from undifferentiated embryonic cells, which subsequently become active and form blastomata (phakoblastomata) such as neuroblastomata; various congenital malformations.

The case we describe may be regarded as a type of neuroectodermal phakomatosis, termed neurofibromatosis or Von Recklinghausen disease after the author who identified it in 1882. This uncommon heredofamilial disease is characterized by patches of brown pigmentation on the skin (café-au-lait spots) and multiple tumoral formations affecting the CNS and PNS.

The neurological manifestations of von Recklinghausen disease consist of intracranial, intraspinal and peripheral nerve tumors and very complex symptoms such as convulsions, mental retardation, radicular pains, precocious puberty and skeletal abnormalities [4]. One of the most characteristic of the skeletal abnormalities is a unilateral defect of the posterior superior wall of the orbit [4] involving both wings of the sphenoid bone, the anterior clinoid process and occasionally the sella turcica and resulting in unilateral pulsating exophthalmos.

The highest frequency of this disease with the classic symptoms described above occurs in young adulthood.

Disorders of the autonomic nervous system have not been observed in neurofibromatosis, though they have in Leschke syndrome, another neuroectodermal phakomatosis, which lacks skin tumors and is considered to be a peculiar form of neurofibromatosis. The clinical features of Leschke syndrome, or congenital pigmentary dystrophy, are multiple pigmented cutaneous macules, somatic infantilism, dystrophy, mental retardation, metabolic (glucose metabolism) and endocrine disorders such as genital dystrophy, pituitary obesity
and adrenocortical hypofunction, abnormalities of the autonomic nervous system (ANS) [5].

Case report
This 13 year old boy was admitted to our depart-

ment because he could no longer walk or stand upright. According to his parents, these difficulties had begun a month before. His parents stated that the boy was born without pathological defects except for a right microphthalmus and a pendulous tumor on his left forearm and later another on his forehead. Both tumors were removed and proved on histological examination to be neurofibromata. The patient showed such remarkable hypersensitivity that his gait worsened when he was watched. Another disorder was dry vomiting in the morning daily. Physical examination revealed somatic infantilism with congenital right microphthalmus, short neck with abnormal position of the head, a cervicothoracic left convex scoliosis, valgus knees, a malformation of the feet with altered plantar arch. Other findings were a tissue imbibition, palmar hyperhidrosis, poor hair growth, facial vasomotor disturbances, numerous café-au-lait spots on the shoulders and frequent pinhead neoformations over the trunk. Neurological examination demonstrated bilateral nystagmus and slight protrusion of the left eyeball, muscle hypotonus and decreased tendon reflexes of the upper limbs, severe weakness of the lower limbs with decreased patellar reflexes, spastic muscular hypertonus, bilateral Babinski sign and paraparetic spastic gait, and remarkable ANS disturbances (vomiting, hypersensitivity). The boy