A case of epidermal nevus syndrome with carotid malformation

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Epidermal Nevus Syndrome (ENS) is characterized by linear verrucous lesions of the skin and congenital anomalies including bone deformities, eye and central nervous system (CNS) malformations. We describe a case of ENS associated with an abnormality of the carotid artery which is considered a risk for stroke.

Key-Words: Epidermal nevus syndrome — neurocutaneous diseases — carotid malformation

Introduction

Solomon et al. defined the epidermal nevus syndrome (ENS) in 1968. This is a rare neurocutaneous disease involving ectoderm and mesoderm, marked by specific skin changes, namely macular or verrucous hyperpigmentation in clusters or strips, and congenital defects including bone, eye and CNS changes [5, 6]. CNS abnormalities may appear later in the course of the illness and are reported in most cases in literature. Two kinds of malformations have been considered: a) vascular malformations, such as carotid and basilar aneurysms, anterior cerebral artery aneurysms, arteriovenous malformations, cerebrovascular dysplasias, internal carotid occlusion, venous sinus abnormalities, leptomeningeval angiomias; b) abnormalities of the CNS, such as hemimacrocerephaly, micro- and macrogyria, porencephalic cyst, arachnoid cyst, astrocytoma, mixed glioma, gliomatosis cerebri[2, 3]. The neurological findings commonly reported are mental retardation, epilepsy, hemiparesis, tetraparesis, cranial nerve defects, hyperkineisias [2].

Case report

Our patient was a 24 year old man. Pregnancy, delivery and labour were said to have been normal. Medical history revealed a dermoid cyst of the left cornea operated on at the age of 2 months, lame gait since infancy and generalized convulsive sei-
enhancing lesion, without mass effect, in the right frontal lobe as well as in the lateral aspect of the right lenticular nucleus with associated slight enlargement of the frontal horn of the lateral ventricle.

The CT features resemble the sequela of an old ischemic lesion in the territory between anterior cerebral artery and superficial/deep territory of the middle cerebral artery (Fig. 2a-b). Carotid angiography disclosed coiling of the right internal carotid artery and a minimal lengthening of the carotid siphon (Fig. 3a-b).

Discussion

ENS is caused by an unknown congenital noxa which acts on ectoderm and mesoderm. So far there has been non absolute evidence of an hereditary factor, such as a dominant abnormal gene of very low penetrance [1, 3]. For this reason prevention is still not possible and the association of many different developmental defects of CNS are a diagnosis challenge for the neurologist. Our case presented a coiling of the internal carotid...