Amelanotic leptomeningeal melanoblastosis. Case report

Allegranza A.****

* Dipartimento di Neurologia
** Dipartimento di Nueroradiologia
*** Dipartimento di Neurochirurgia
**** Dipartimento di Neuropatologia
Istituto Nazionale Neurologico "C. Besta", Milano

Leptomeningeal melanoblastosis is a rare phakomatosis; the amelanotic variant has not till now been described. In this paper we report the case of a young man suffering from amelanotic leptomeningeal melanoblastosis manifested as medullary syndrome and secondary intracranial hypertension. The diagnosis of leptomeningeal melanoblastosis was hypothesized on the basis of CSF and neuroradiological findings and it was finally confirmed by the histopathology.

Wey Words: Leptomeningeal melanoblastosis — S-100 protein

Introduction

Neurocutaneous melanosis (NM) is a rare phakomatosis described by Rokitanski [2] in 1861, and identified as a specific congenital syndrome by Van Bogaert [11] and Touraine[10]. Since that time many variants and incomplete forms have been reported, stressing mainly the predisposition to malignant evolution of the neurological lesions [6, 3]. We describe one patient suffering from NM manifested as amelanotic malignant leptomeningeal melanoblastosis. To the best of our knowledge, this amelanotic variant has been recognized only by Reed [8] in an autopsy case.

Case report

An 18 year old man was first seen by us due to a 2-month history of intracranial hypertension syndrome. Delivery was normal, but at one year of age, but in the following years he occasionally required a little support in order to walk. Two months before admission to our hospital, the patient complained of nuchal headache, vomiting, blurring in the left eye and sciatica. He was unable to stand up.

On physical examination, the patient appeared well, alert and oriented. A giant congenital pigmented hairy nevus in the skin of the left buttock and many other pigmented nevi on back and thighs were noted. The nevi appeared regular and were unchanged from birth, as the patient's mother reported. Slight nuchal rigidity, bilateral Lasègue at 30° and left cranial nerve VI palsy were found. On fundus oculi examination bilateral papilledema (3 diopters) was also noted. Visual acuity was 8/10 in the right eye and 2/10 in the left. Legs were hypotonic and amyotrophic. Marked paraparesis (3/5) was evident, knee jerks were brisk and ankle jerks were absent. Babinski sign was bilaterally evoked. A sensory level from T4 to
T9 and from T12 to L2, except for the saddle area and feet, was found. Spontaneous micturition was difficult. Routine laboratory analysis, chest and skull X-ray films and enhanced brain CT scan were within normal limits. A radiological study of the spine was performed. X-ray films of the spine revealed kyphoscoliosis with major curvature in T8-T9 segments and increased interpediculate distance at T4-T6. Myelography revealed characteristic features of arachnoiditis from T4 to T10, complete block at this level and a maldevelopmental enlargement of the dural sac at T4-T6 level with thinning of the thoracic cord.

CSF total proteins were 214 mg%. Upon cytological examination, 35 out of 40 cells found were atypical: large epitheliomorphic with multiple nuclei and nucleoli, suggesting malignancy, but without specific features to identify their origin (Fig. 1). Bacteria, mycetes and mycobacteria did not grow on CSF cultures.

Magnetic resonance (MR) of the spine showed serpentine vascular images outlining the anterior and posterior surfaces of the conus and selective spinal angiography showed a tumor stain at thoraco lumbar level (Fig. 2). One month after admission to our hospital the patient became paraplegic and anesthetic from T4 to sacral level. Bladder catheter was necessary. Headache recurred despite high-dose dexamethasone (16 mg/day i.m.). At operation a T12-L2 laminectomy exposed a firm gray subdural tissue which incorporated pathological vessels enveloped the conus medullaris and cauda equina. An histological examination small nests of epitheliomorphic cells lying in a delicate connective network similar to those found in CSF were detected. Lillie and Schmorl's methods for melanin were negative. S-100 protein was detectable in many cells (Fig. 3), leading to a diagnosis of amelanotic melanoblastosis of the leptomeninges. Electromicroscope examination was not possible for technical reason (material fixed in formalin).

3 weeks later marked hydrocephalus developed; ventricular Omaya reservoir was applied for transient relief of intracranial hypertension. Cytological examination of ventricular fluid samples confirmed the presence of atypical cells. After chemotherapy (decarbazine 400mg i.v./day for 5 days) a ventriculoperitoneal shunt was implanted and headache remitted. The patient refused radiotherapy and he died at home 8 months from the onset of the disease.

Fig. 1. CSF cytological smear: epitheliomorphic cells with large cytoplasm, one of them with a large nucleolus (arrow), no melanin granules are present (M.G.G.X. 800).