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Case Report

An Unusual Presentation of Primitive Neuroectodermal Tumor (PNET) with Meninges and Bone Marrow Infiltration: A Case Report

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Abstract

We present the clinical case of a young man with sudden neurological and hematological symptoms. The analyses revealed meningeal and bone infiltration of cells compatible with primitive neuroectodermal tumor without a mass at the imaging.

Introduction

Primitive Neuroectodermal Tumor (PNET) is a rare, aggressive variant of small round cell malignant embryonal neoplasm derivate from the neural crest cells [1,2]. PNETs can be subdivided into central or peripheral (pPNET) types. They occur more frequently in adolescents and young adults with a slight male predominance and are characterized typically by the presence of brain mass on imaging. They represent about 1% of the brain tumors in this population group. The pPNETs arise anywhere in the bone and soft tissues outside the central and sympathetic nervous system. The overall 5 years survival rate is around 53% [1,2]. Standard therapeutic approach includes surgery, radiotherapy or chemotherapy [3].

Here, we present a case of young adult patient with the diagnosis of pPNET with bone marrow and cerebrospinal fluid infiltration as the only disease presentation.

Case Presentation

A 40 year old male patient complained of headache, mental confusion and diplopia for some months. After a syncope episode, he was admitted to the hospital: the Magnetic Resonance Imaging (MRI) and the Computed Tomography (CT) scan of the brain have been performed, as well as CT scan of lung and abdomen. CT scan and MRI were both negative for space occupying brain lesions and any parenchymal tumor. The patient has been evaluated by the ophthalmologic who diagnosed right papilledema. Subsequently, the lumbar puncture revealed an increased intracranial pressure without the presence of neoplastic cell in liquor. Therefore, the diagnosis of idiopathic intracranial hypertension was performed. The patient received symptomatic treatment and was discharged from the hospital in a good clinical condition.

After six months from the first episode of epileptic seizures, a new one appeared: a new MRI was repeated. This time, a MRI showed an increased dilatation of brain ventricles and alteration of signal in the spinal cord, suspicious for neoplastic infiltration (Figure 1).

In addition, at the blood tests, thrombocytopenia grade G3 was seen. The patient repeated the bone marrow biopsy. Immunohistochemical analysis confirmed infiltration of tumor cells, showing positive expression of Neuron Specific Enolase (NSE), CD56, GFAP, S-100 and Synaptophysin (Syn). Moreover, expression of CD99, TTF1, CAM5.2, cytokeratin 7, cytokeratin 20, and chromogranin, OCT4, PLAP, MEL COCKTAIL and PDL1 were negative. The same cells were present at the liquor examination. The 18-fluorodesossiglucose Positron Emission Tomography (18-FDG PET) confirmed an increased metabolism in the marrow site, without any other localization. Considering these findings, the diagnosis of PNET with meninges and bone marrow infiltration was done.

The histological examination and the therapeutic approach were confirmed by the team of the reference institute for rare cancer in Italy.

The patient started with chemotherapy (VAC schedule containing vincristine, cyclophosphamide and doxorubicin). During the first

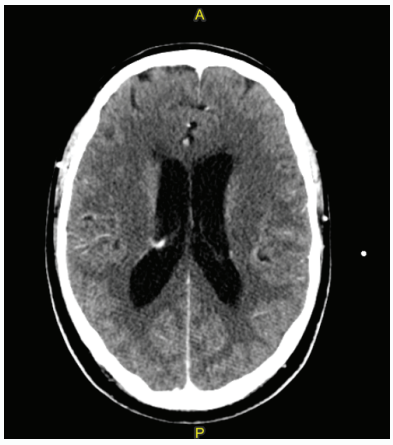

Figure 1: Dilatation of brain ventricles, absence of brain mass.

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cycle (due to thrombocytopenia) have been administered vincristine and cyclophosphamide only, from the second cycle, normalization of platelets permitted us to continue with all the three drugs, in total for 6 cycles.

The post treatment 18-FDG PET scan showed complete metabolic response; bone marrow biopsy and lumbar puncture were negative for neoplastic cells with a residual hydrocephalus at the CT scan. The patient had occasionally motorial epileptic seizures and headache with sight alteration due to intracranial hypertension caused by hydrocephalus, and, therefore, a ventriculo peritoneal shunting had been performed with important bettering the quality of life of our patient. He started the regular 3-months' follow up.

After 6 months, the patient suffered from a new epileptic seizure with persistent unconsciousness. At the CT scan no haemorrhagic events or expansive lesions were observed but there was an increase of dilatation of brain ventricles. No blood test no abnormalities. At the liquor exam, the presence of neoplastic cells were diagnosed, consistent with PNET cells at the diagnosis. A symptomatic therapy was administered in order to reduce intracranial hypertension, though without benefit. The patient died after couple of days.

Discussion

According to the literature, the most common presentation of PNET is a single or multiple brain masses [3-5]. In our case, no primary brain tumor has been found, therefore surgical or radiotherapy approach were excluded. Our patient had an unusual presentation: an infiltration of the bone marrow and cerebrospinal liquor dissemination without any brain mass. Moreover, any tumor mass in the abdominal, pelvic region or in the thoraco-pulmonary area were found in our patient.

The treatment of the pPNET remains unclear because of the lack of evidence and rarity of the condition. pPNET belongs to the group of the Ewing sarcoma, so following the guidelines, we chose a schedule combination with doxorubicin, cyclophosphamide and vincristine (VAC schedule) [6]. Our patient reached a pathological complete clinical and radiological response.

The best prognosis was observed in patient affected by PNET treated with surgery followed by adjuvant chemotherapy and radiotherapy [7].

Remain unclear whatever the residual intracranial hypertension after the chemotherapy was related to the PNET residual disease or was an independent condition.

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