TWO SYNCHRONOUS CENTRAL NERVOUS SYSTEM TUMOURS IN A CHILD WITH NEUROFIBROMATOSIS TYPE 1

Rodica Elena Cornean¹, Monica Scutariu², Gheorghe Ungureanu³, Dorin Farcau³, Ştefan Florian²

¹Paediatrics Clinic No. 2; ²Neurosurgery Clinic; ³Paediatrics Clinic No. 3, University of Medicine and Pharmacy "Iuliu Hațieganu” Cluj-Napoca, Romania.

**Background**

Synchronous, multiple central nervous system (CNS) tumours are usually rare in pediatric patients. Neurofibromatosis type 1 (NF1) is a rare genetic condition which is known to predispose to multiple tumours even such synchronous CNS tumours. The pilocytic astrocytomas (WHO grade I) are the main histological type of CNS tumours reported in NF1. Therefore, they are considered to be NF1-associated tumours.

**Case presentation**

A 6.5-year-old boy was admitted to our hospital for severe emaciation. Profound fat and muscle wasting were the only prominent clinical features.

His growth rate was preserved despite his rapid dramatic weight loss (HT: 118 cm, p.50; WT: 16.5 kg, p.3; BMI: 11.9 kg/m² <p. 0.4, T.J. Cole 1995), IGFI value was low (45 ng/ml). Physical exam showed 3 café-au-lait spots (0.5 cm) and mild scoliosis. No signs of high intracranial pressure (headaches, vomiting, seizures), ataxia, blurred vision, diplopia, nystagmus or signs of progressing into puberty were present. No cognitive deficits and no definite neurologic dysfunction were present. Serum electrolytes, adrenal and thyroid tests were normal. Tests for Crohn, celiac disease, HIV and other malignancies were negative as well.

**What a rare case...**

**Neurofibromatosis 1 with so few spots coexisted with 2 CNS large synchronous tumours (about 5cm each)**

Despite the different MRI aspect, both tumours were low grade pilocytic astrocytomas.

**4 brain surgeries in a patient with EMAClATION (VP shunt - mini shunt, approach to supratentorial tumour, posterior fossa tumour, bifrontal hygroma)**

Chemotherapy (Vincristine, Carboplatin) and radiotherapy for the remaining brainstem glioma.

**Conclusions**

The same histological type in both tumors pointed out towards NF1 as the underlying medical condition even when the NIH diagnostic criteria for NF1 (Bethesda, 1988) didn’t seem to be met at the chronological age of the patient.

In our case, the diencephalic syndrome (characterized by cachexia and preserved linear growth) was the revealing metabolic signature of the hypothalamic tumour regardless of the paucity of the accompanying symptoms while the results of the esophageal manometry pointed to the brainstem tumour.

Even if the presence of two synchronous CNS tumors is certainly a rare event, this is not the case anymore in patients with NF1 regardless of the severity of their skin involvement.

**References**