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

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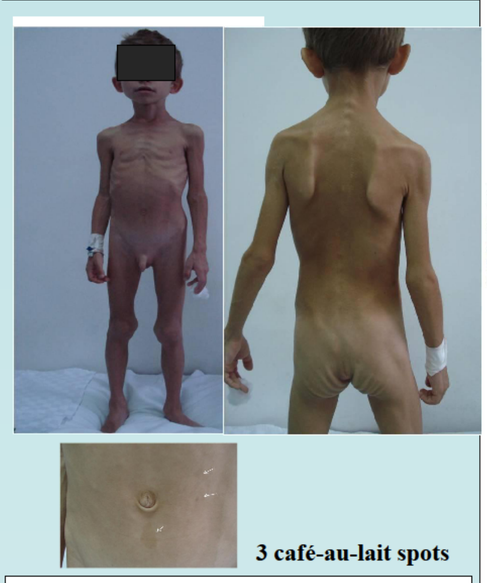
## *Background*

Synchronous, multiple central nervous system (CNS) tumours are usually rare in pediatric patients.
Neurofibromatosis type1 (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*

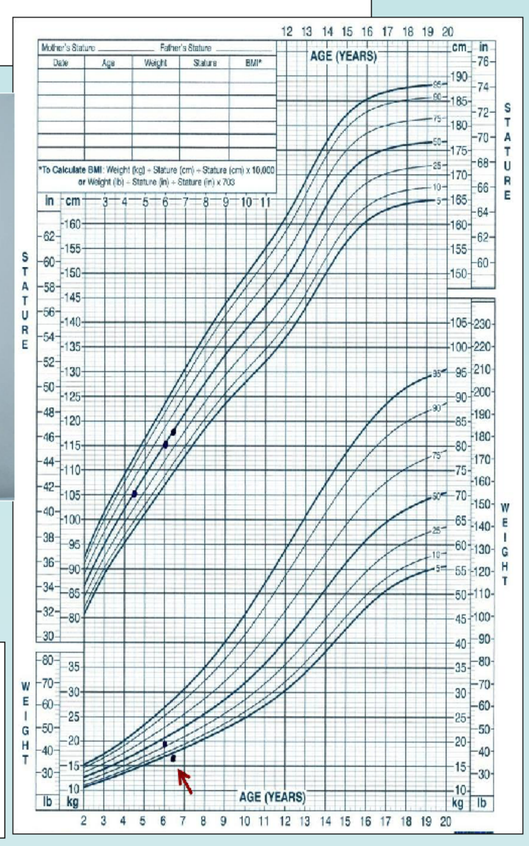
A 6.5-year-old boy was admitted to our hospital for severe emaciation. Profound fat and muscle wasting were the only prominent clinical features.
His growth rate was preserved despite his rapid dramatic weight loss (HT:118 cm, p.50; WT:16.5 kg, p.3; BMI: 11.9 kg/m2 <p. 0.4, T.J. Cole 1995). IGF1 value 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, nistagmus 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.

Patient's clinical features and growth chart



3 café-au-lait spots

Marked emaciation despite preserved liniar growth as the hallmark of the diencephalic syndrome

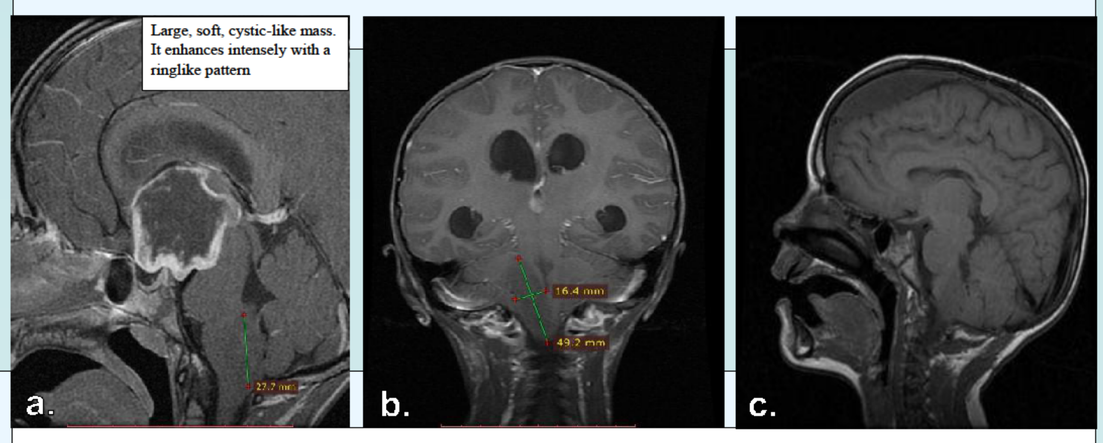



Esophageal endoscopy: exceedingly rigid esophagus
Esophageal manometry: hypertensive upper and lower esophageal sphincters with incomplete relaxations after wet swallows for the upper one.

Grade I Coma (GCS 12)

On the brain MRI, two concomitant large masses (5 cm each) were identified. The first mass had a diencephalic location (with wide hypothalamic involvement) while the second mass was located outside the optic pathway (extra-optic pathway tumors/extra-OPT) in the posterior fossa (with extension towards the brainstem and 4th ventricle).

Both proved to be pilocytic astrocytomas.
Pilocytic astrocytoma qualifies as "the tumor that is the exception to the rule"




Patient's brain MRI: a and b. before and c. after surgery

***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 EMACIATION (VP shunt- unishunt, 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*

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Misc 1
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