An unusual case of sudden-onset psychiatric symptoms: a diagnostic challenge

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Background

- ✓ Preclinical studies have demonstrated that human chorionic gonadotropin (hCG) is implicated in the pathophysiology of several diseases of the central nervous system (CNS) such as Parkinson's and Alzheimer's diseases[1].
- ✓ Intracranial germ cell tumors (iGCTs) account for up to 15% of primary pediatric brain neoplasms and are believed to originate from primordial germ cells that failed to migrate to the gonads [2].
- ✓ Approximately 90% of iGCTs are diagnosed before the age of 20, most commonly in the suprasellar or pineal regions. Typical presentations include endocrine dysfunction, visual disturbances, headaches, and neuropsychiatric symptoms[3].

Neurological Examination

No focal deficits. Akathisia, motor impersistence and motor stereotypies.

Exams And Analysis Performed

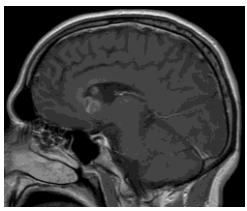
- ✓ Standard ECG
- ✓ Routine blood test
- ✓ Neuropshycological evaluation
- ✓ Endocrinological evaluation
- ✓ Standard EEG
- ✓ MRI with gadolinium
- ✓ Lombar puncture with isofocusing, cytologic exam and dosage of anti-encephalitis and anti-unknown antigen antibodies
- ✓ Pituitary hormones serum dosage
- ✓ Testicular echography
- ✓ Total-body CT with contrast agent
- ✓ PET-CT with 18-FDG
- ✓ β-hCG CSF dosage

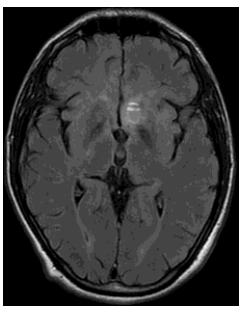
Case Description

We report the case of an 18-year-old Caucasian male who initially presented with progressive polyuria and polydipsia. Brain MRI and endocrine screening at onset were inconclusive, and he was misdiagnosed with stress-induced potomania. Over the subsequent two years, the patient developed progressive behavioral and cognitive changes, including disinhibition, hyperactivity, and loss of academic interest, ultimately leading to school dropout. He was diagnosed with ADHD-like symptoms and treated with antipsychotics, without clinical benefit. A repeated endocrinological evaluation prompted initiation of desmopressin for suspected central diabetes insipidus. Upon admission to our center, he underwent EEG, lumbar puncture, brain MRI, and hormonal assessments. Imaging revealed a lesion in the left caudate nucleus and periventricular white matter. Hormonal testing demonstrated elevated testosterone (12.79 ng/mL), suppressed LH (0.10 ng/mL) and FSH (0.48 ng/mL), and markedly increased β-hCG levels in both serum (55.0 mUI/mL) and CSF (426 mUI/mL). Follow-up neuroimaging documented additional cerebral lesions. Given the atypical location, multifocal presentation, and high hemorrhagic risk, a biopsy was not performed. Based on the clinical, radiological, and biochemical findings, a diagnosis of β-hCG–secreting non-germinomatous germ cell tumor was established. The patient subsequently received chemotherapy (one cycle of carboplatin/etoposide followed by three cycles of cisplatin/etoposide/ifosfamide) and was scheduled for radiotherapy.

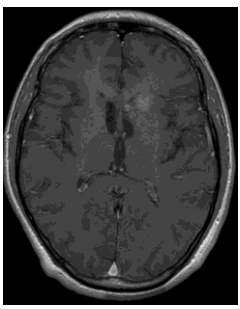
Axial DWI

Sagittal T1





Axial FLAIR



Axial T1

Conclusions

This case underscores the importance of considering intracranial germ cell tumors in adolescents presenting with psychiatric or behavioral symptoms, particularly when neuroimaging findings are inconclusive or do not fully account for the clinical picture. We hypothesize that the patient's neuropsychiatric manifestations were not primarily attributable to local structural damage caused by the tumor, but rather to the neuroactive effects of β-hCG on CNS function.

Bibliography

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