

# Mesencephalic Hypertrophy Mimicking a Double Brainstem: A Case Report

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## Abstract

**Background:** Neuroimaging is pivotal in the evaluation of epilepsy. We present a case of incidental mesencephalic hypertrophy resembling a double brainstem in a patient with drug-dependent epilepsy. **Case Presentation:** A 16-year-old girl was hospitalized for generalized tonic-clonic status epilepticus following antiepileptic treatment discontinuation. Diagnosed with epilepsy at age 12, she had been prescribed levetiracetam (250 mg twice daily) but discontinued therapy for 4 months. Family history was unremarkable for similar conditions. Clinical examination revealed short stature, trisomy-like facies, bilateral hypoplasia of the fourth and fifth fingers, and pes planus. Mild cognitive impairment was noted, with no sensorimotor deficits. Contrast-enhanced brain computed tomography (CT) demonstrated a pseudo-double brainstem secondary to mesencephalic hypertrophy curving anteriorly between a pituitary adenoma and the pons. The patient was managed per institutional status epilepticus protocol, with no seizure recurrence. At discharge, lamotrigine (5 mg/kg/day) was initiated alongside counseling to improve treatment adherence. **Conclusions:** Epilepsy coexisting with congenital malformations necessitates comprehensive neuroimaging and genetic assessment. In resource-limited settings such as Madagascar, pedigree analysis and consistent follow-up are critical to optimizing care.

## Keywords

Epilepsy, Computed Tomography, Madagascar

## 1. Introduction

Epilepsy is a cerebral disorder affecting approximately 50 million individuals world-

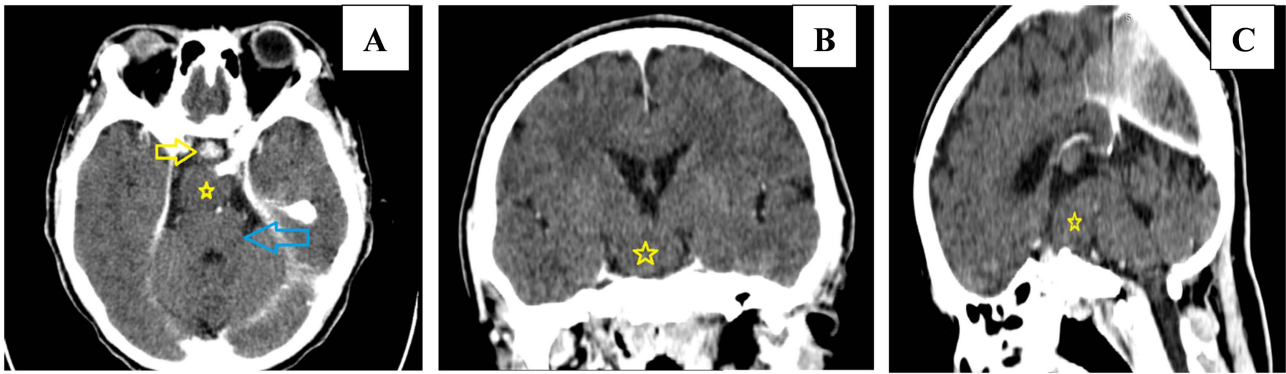
wide, with 2.4 million new cases reported annually [1]. Epilepsy in children may be attributable to structural abnormalities or to underlying genetic mutations. Neuroimaging holds a central role in the etiological evaluation of epilepsy, particularly in identifying structural causes, especially when surgical intervention is considered [2]. Access to magnetic resonance imaging (MRI) remains challenging for patients in Madagascar, necessitating frequent reliance on contrast-enhanced brain computed tomography (CT) for the detection of structural brain lesions [3]. We report a case of incidental mesencephalic hypertrophy mimicking a double brainstem, associated with drug-dependent epilepsy in a 16-year-old adolescent.

## 2. Case Report

A 16-year-old female adolescent was hospitalized for generalized tonic-clonic status epilepticus secondary to treatment discontinuation. Status epilepticus consisted of generalized tonic-clonic seizures lasting more than five minutes, occurring in three successive episodes within 30 minutes, without recovery of consciousness between seizures. The condition occurred in an afebrile context and in the absence of sleep deprivation. Seizures persisted despite levetiracetam therapy, with at least one generalized tonic-clonic seizure occurring weekly. There was no regular medical follow-up for epilepsy management. She had a documented history of epilepsy since the age of 12 years, previously managed with levetiracetam 250 mg administered twice daily, but had discontinued medication for four months prior to admission. She was born at term via uncomplicated vaginal delivery, with no history of neonatal resuscitation or neonatal jaundice. Her vaccination status was consistent with Madagascar's Expanded Program on Immunization. No significant maternal comorbidities were reported during pregnancy, and no familial history of similar conditions was identified through genealogical inquiry.

Clinical examination revealed dwarfism, a dysmorphic facial phenotype suggestive of trisomy, and bilateral hypoplasia of the fourth and fifth digits, accompanied by flat feet. Mild cognitive impairment was noted, with no sensorimotor deficits detected. Cardiac auscultation revealed a regular rhythm without murmurs or added sounds. Pulmonary examination was unremarkable, and no dermatologic abnormalities were observed.

Contrast-enhanced brain CT demonstrated mesencephalic hypertrophy curving anteriorly, simulating a double brainstem, localized between a pituitary adenoma (the lesion measured 13 mm in height and 10 mm in width, showed homogeneous enhancement with iodinated contrast, was free of hemorrhage, and exhibited suprasellar extension without bone thinning) and the pontine region (**Figure 1**). Hormonal profiling and genetic testing could not be performed due to resource limitations. The patient was managed according to our institutional protocol for status epilepticus, with no recurrence of seizures during hospitalization. Neurosurgical intervention was deemed unnecessary given the presence of the pituitary adenoma. At discharge, she was initiated on lamotrigine at 5 mg/kg/day, with reinforced adherence counseling and scheduled medical follow-up every six months.



**Figure 1.** Brain CT scan with iodinated contrast. (A) Axial section through the posterior fossa demonstrates a pseudo-double brainstem (yellow asterisk) situated between a pituitary adenoma and the pontine region. (B) Coronal reconstruction highlights the pseudo-double brainstem appearance (yellow asterisk). (C) Sagittal reconstruction reveals mesencephalic hypertrophy with anterior curvature (yellow asterisk).

### 3. Discussion

Our clinical case describes the incidental discovery of mesencephalic hypertrophy within a polymalformative syndrome associated with drug-resistant epilepsy in a 16-year-old adolescent. The coexistence of epilepsy and congenital malformations in this patient raises suspicion for an underlying genetic abnormality. The diagnostic algorithm proposed by the International League Against Epilepsy (ILAE) emphasizes the concurrent evaluation of epilepsy-related comorbidities and systematic etiological investigations, including genetic testing [4]. In this case, genetic mutation analysis could not be performed, and no familial cases of similar presentation were identified. This clinical scenario remains plausible, as a *de novo* mutation may occur in an individual.

Given the patient's clinical phenotype, a potential etiology could involve a mutation in the *FGFR3* gene, implicated in hypochondroplasia [5] or achondroplasia [6]. Prior reports associate *FGFR3* mutations with bilateral medial temporal lobe dysgenesis and epilepsy [5], underscoring the importance of epilepsy-protocol MRI for detecting structural abnormalities that may underlie seizure disorders [2] [4]. To our knowledge, no prior cases of epilepsy associated with mesencephalic hypertrophy, as observed in our patient, have been documented in the literature.

Epilepsy may also manifest as a presenting feature of pituitary adenomas, such as prolactinomas or lactosomatotropic adenomas with suprasellar extension or involvement of adjacent cerebral structures [7]. Although pituitary dwarfism cannot be definitively excluded in this patient due to the absence of pituitary hormone assessment, therapeutic management prioritized antiepileptic therapy. Surgical intervention is typically reserved for drug-resistant epilepsy under stringent clinical criteria [8]. In children, the differential diagnosis of midbrain tumors includes neoplastic, malformative, and developmental etiologies. While tectal gliomas are typically indolent low-grade astrocytomas, often harboring KRAS or BRAF alterations and occasionally progressing to high-grade gliomas, structural malformations such as anterior mesencephalic cap dysplasia (AMCD) in Joubert syndrome can mimic

mass-like lesions due to aberrant white matter tracts. Advanced MRI and diffusion tensor imaging, combined with molecular profiling, are essential to distinguish between these entities and avoid misdiagnosis [9] [10]. Lamotrigine was selected for its broad-spectrum efficacy and tolerability, with additional emphasis on patient education, adherence strategies, and lifestyle modifications. The management of drug-resistant epilepsy remains challenging in our Malagasy setting. The lack of universal health coverage, the absence of specialized epilepsy centers, and limited access to advanced investigations such as stereoelectroencephalography and brain MRI are significant barriers. Telemedicine could serve as a potential alternative for case discussion with international experts, provided that appropriate diagnostic workups are performed and legal frameworks are established to ensure the protection of medical records.

#### 4. Conclusion

The diagnostic approach to epilepsy requires adherence to well-defined steps, integrating etiological investigation and comorbidity assessment. In patients with clinical evidence of congenital malformations, neuroimaging and comprehensive genetic evaluation are critical. In resource-limited settings such as Madagascar, the implementation of genealogical mapping, regular multidisciplinary follow-up, and patient education remain pivotal to optimizing care for individuals with complex epilepsy syndromes.

#### Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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