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Dysplastic Gangliocytoma of the Cerebellum (Lhermitte-Duclos Disease) Associated with Cowden Syndrome: A Case Report and Literature Review

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Abstract | **Case Report**

Background: Lhermitte-Duclos disease (dysplastic gangliocytoma of the cerebellum) is an extremely rare, benign cerebellar lesion (WHO grade I) characterized by abnormal neuronal hypertrophy and cerebellar cortex disorganization. It is frequently linked to Cowden syndrome due to germline mutations in the *PTEN* tumor suppressor gene. ***Case Report:*** We report the case of a 49-year-old patient with a history of Cowden syndrome, desmoid tumor, and hyperthyroidism, who presented with a progressive cerebellar syndrome and intracranial hypertension. Brain CT and MRI revealed a characteristic left cerebellar hemispheric mass with the pathognomonic "tiger stripe" sign, obstructive hydrocephalus, and signs of cerebellar tonsillar herniation. ***Conclusion:*** Lhermitte-Duclos disease is a rare entity whose neuroimaging features are key to diagnosis. Recognition of this condition warrants systematic screening for Cowden syndrome and multidisciplinary follow-up.

Keywords: Lhermitte-Duclos disease, dysplastic gangliocytoma, Cowden syndrome, cerebellar syndrome, tiger stripe sign, PTEN tumor suppressor gene.

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INTRODUCTION

Initially described in 1920 by Jacques Lhermitte and P. Duclos, dysplastic gangliocytoma of the cerebellum (or Lhermitte-Duclos disease) is a rare pathology of uncertain etiology. Long considered a hamartoma, developmental anomaly, phakomatosis, or low-grade tumor, it is currently classified as WHO Grade I by the World Health Organization (WHO). Its pathogenesis is closely associated with Cowden syndrome within the framework of a hamartoma-neoplasia syndrome, resulting from a mutation in the *PTEN* gene located on chromosome 10. With an estimated incidence of 1 case per 2,500,000, this condition predominantly affects young adults in their third or fourth decade (mean age of 34 years), while pediatric cases remain exceptional. We report the clinical observation of a patient managed for this rare pathology, along with a review of the literature.

CASE PRESENTATION

A 49-year-old patient presented with the following history and clinical timeline:

- Followed for hyperthyroidism under Dimazol (thiamazole).
- History of surgery in 2017 for a desmoid tumor of the right iliac fossa (RIF).
- Followed in gastroenterology for Cowden syndrome.
- Followed in dermatology for cutaneous fibromatous lesions located on the face and forearm.

The patient presented for clinical evaluation of a progressively worsening cerebellar syndrome associated with a complete intracranial hypertension (ICH) syndrome. On initial clinical examination, the patient was conscious and hemodynamically stable. Neurological examination demonstrated normal gait and stance, no motor or sensory deficits, and no sphincter dysfunction.

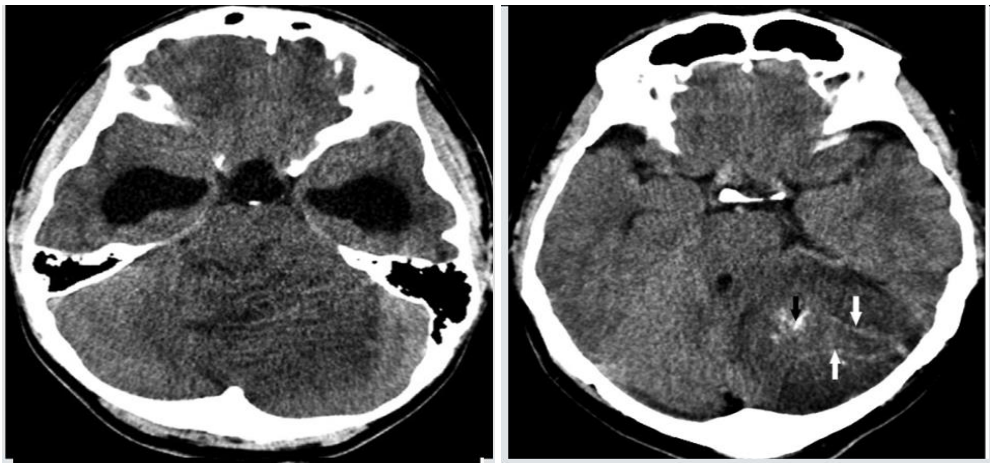
Medical Imaging and Diagnostic Workup

Given this neurological presentation, the imaging strategy relied on two key examinations:

1. Brain Computed Tomography (CT)

The initial radiological examination was a non-contrast brain CT scan. It demonstrated a hypodense, non-calcified cerebellar mass located in the left

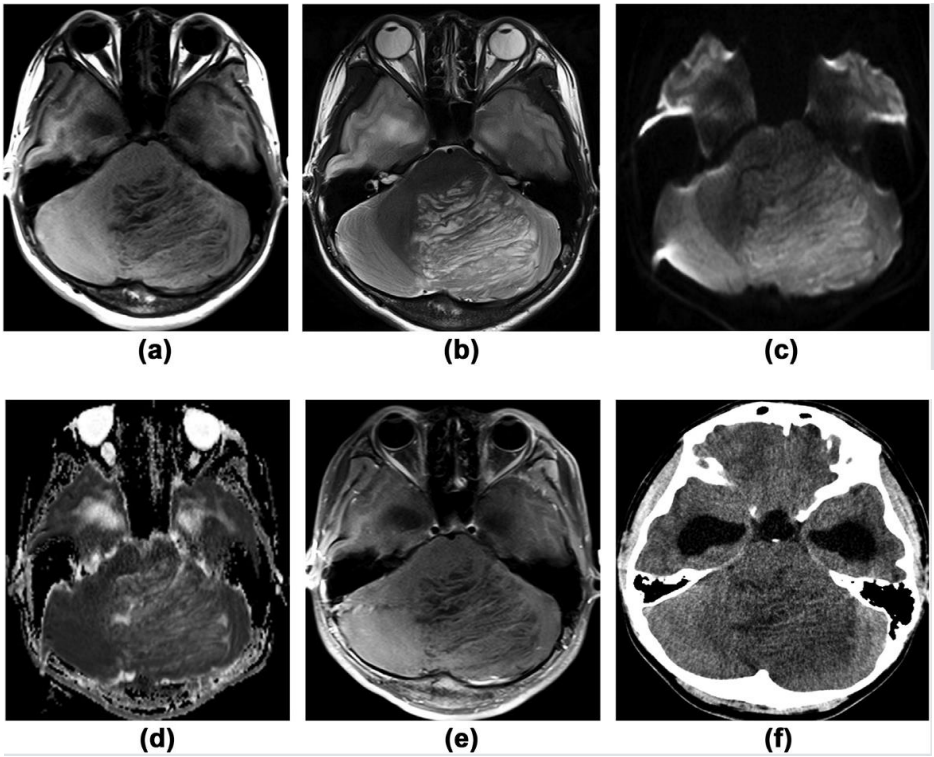
cerebellar hemisphere. This examination also identified major complications, including mass effect, obstructive hydrocephalus, and tonsillar herniation. Contrast administration showed no pathological enhancement, occasionally limited to contrast-enhancing veins traversing the tumor.

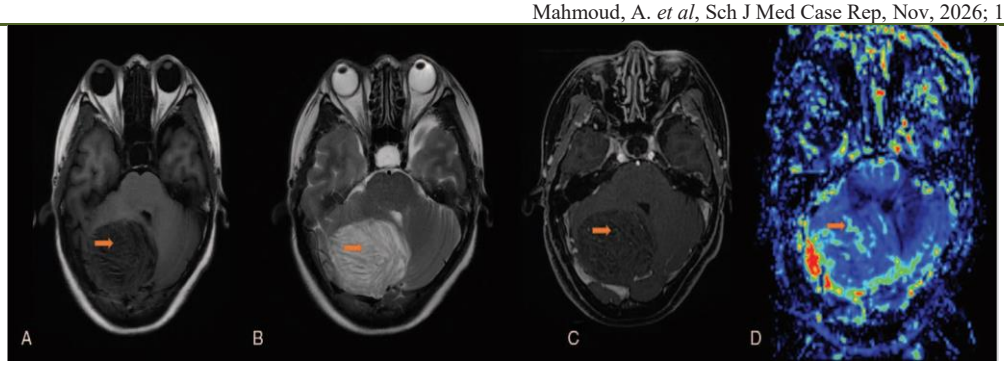


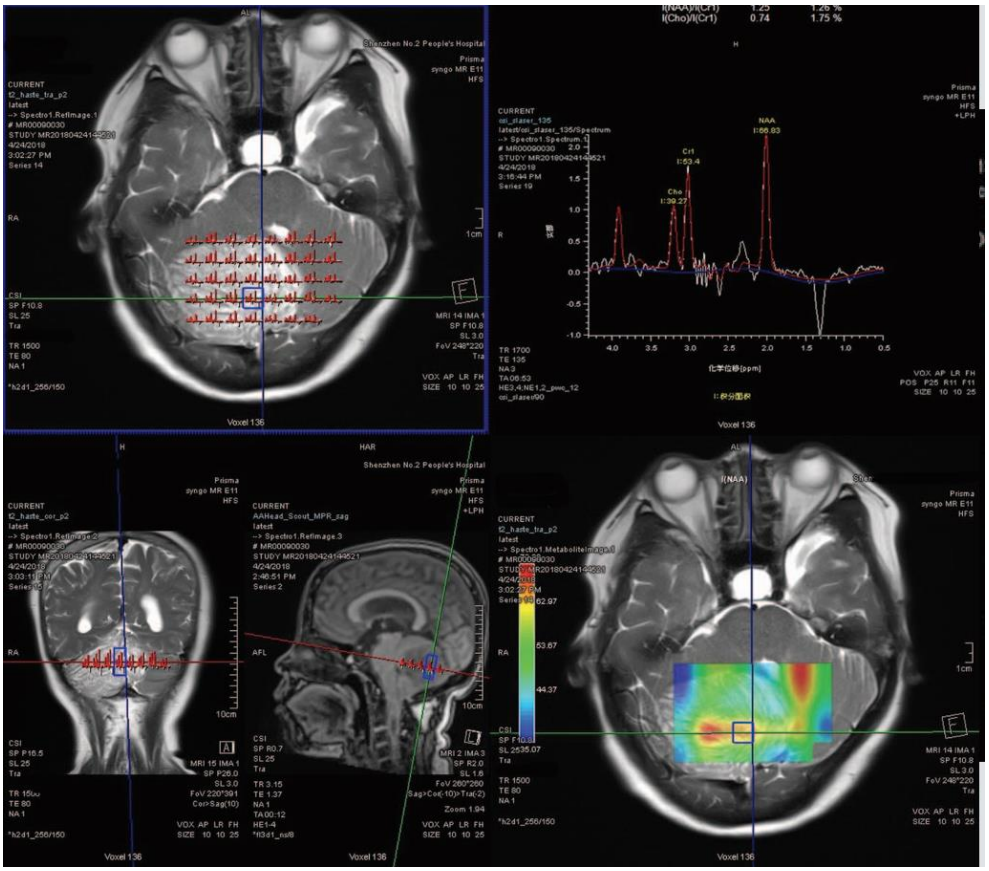
2. Brain Magnetic Resonance Imaging (MRI)

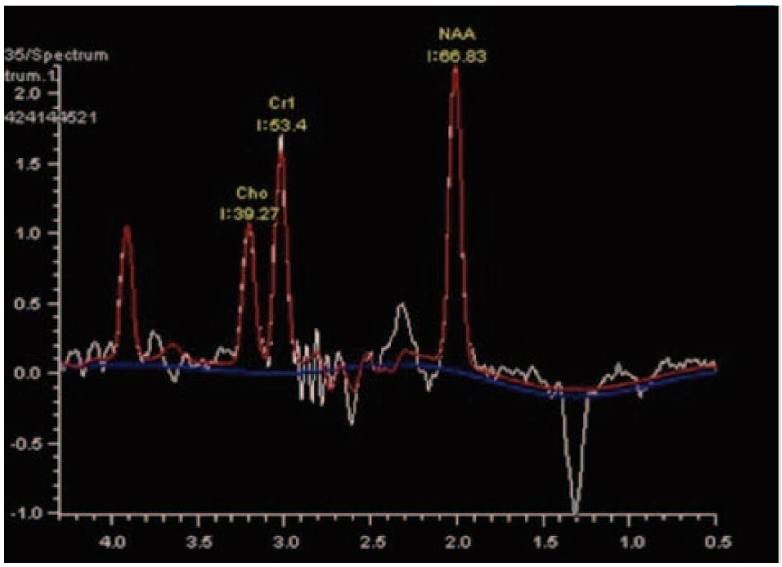
Brain MRI, the essential imaging modality to further characterize the lesion, revealed the following characteristics:

- **T1, T2, and FLAIR Sequences:** Characteristic striated, lamellar appearance formed by alternating, roughly parallel linear bands displaying low signal intensity on T1-weighted images and high signal intensity on T2-weighted images, corresponding to widened cerebellar folia.
- **Advanced Sequences:**
  - **Diffusion (DWI):** High signal intensity with variable ADC values (increased due to the T2 shine-through effect or decreased secondary to hypercellularity from dysplastic neurons).
  - **Perfusion (PWI):** Local elevation of rCBV related to proliferation of the cerebellar venous drainage system with dilated draining veins, without associated signs of malignancy or blood-brain barrier (BBB) breakdown.
  - **Spectroscopy (MRS):** Moderate decrease in Choline (Cho) and N-acetylaspartate (NAA) (reflecting loss of normal neuronal architecture), elevation of lactate (reflecting increased glycolysis), and absence of lipids (absence of tumor necrosis).




A
B
C
D


NAA
I:66.83
Cr1
I:53.4
Cho
I:39.27


NAA
I:66.83
Cr1
I:53.4
Cho
I:39.27

Pathology and Radio-Pathological Correlations

Histologically, the lesion corresponds to a benign hamartomatous lesion classified as WHO Grade I. Histopathological analysis highlights:

- Expansion and thickening of the cerebellar folia associated with cortical disorganization without a clear tumor margin.
- Involvement of cerebellar cortical layers with disappearance of Purkinje cells.
- Hypertrophy and proliferation of dysplastic ganglion cells (large cells with abundant cytoplasm and vesicular nuclei), thickening of the molecular layer, and white matter atrophy.

The histological striation of the folia (hypermyelination and lamellar disorganization) correlates directly with the "tiger stripe" pattern seen on MRI.

MANAGEMENT AND OUTCOME

Treatment relies on surgical resection. Complete resection is often difficult due to the gradual and poorly defined transition between healthy and pathological tissue. Recurrences are not uncommon following partial surgery and are managed according to clinical presentation with re-resection. The overall prognosis is generally favorable.

CONCLUSION

Dysplastic gangliocytoma of the cerebellum is a rare hamartomatous entity whose imaging characteristics allow for an easy diagnosis. Its identification in an adult mandates etiologic screening to search for an association with Cowden syndrome.

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