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Case Report Open Access

Lhermitte Duclos Disease in the Absence of Cowdens - A Varied Presentation

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Abstract

Background: Lhermitte-Duclos is a rare hamartomatous lesion of the cerebellar cortex with both neoplastic and hamartomatous features. Also known as dysplastic cerebellar gangliocytoma, they usually occur in the setting of Cowden's syndrome, an autosomal dominant condition characterized by multiple hamartomas and neoplastic lesions in skin and internal organs. Although enlargement of the internal granular layer of the cerebellum is a consistent finding in our case, the background setting of Cowden's syndrome was not present in our case.

Case description: We present a unique case of LDD in the absence of Cowdens syndrome in a 58-year old gentleman who presented with cerebellar signs, a first to be reported in literature. Here we describe our clinical vignette.

Conclusion: Most of the cases of LDD, reported in literature, were associated with Cowdens disease, however our case is the first to present without Cowdens disease. To date no other similar cases have been reported in literature.

Keywords: Lhermitte-duclos disease; Dysplastic gangliocytoma; Cowden syndrome; Cerebellar lesion

Introduction

Lhermitte-Duclos is a rare hamartomatous lesion of the cerebellar cortex with both neoplastic and hamartomatous features. Also known as dysplastic cerebellar gangliocytoma, they usually occur in the setting of Cowden's syndrome, an autosomal dominant condition characterized by multiple hamartomas and neoplastic lesions in skin and internal organs. Although enlargement of the internal granular layer of the cerebellum is a consistent finding in our case, the background setting of Cowden's syndrome was not present in our case.

They can cause symptoms and signs of mass effect in the posterior fossa and lead to hydrocephalus, brain herniation, and death, if not treated.

Clinical and pathological diagnosis can sometimes be challenging and it is of extreme importance for the pathologist to be aware of the variable clinical and histopathological presentations of such a rare lesion particularly to differentiate it from the low-grade glial and neuronal tumors.

Accurate preoperative diagnosis of Lhermitte-Duclos disease can be made on the characteristic magnetic resonance imaging appearances. Pathological diagnosis of Lhermitte-Duclos disease can be extremely difficult in the absence of proper clinical information and the pathologist should be watchful for any irregularity in the internal granular layer in evaluating the cerebellar tissue, which is otherwise normal.

Case Report

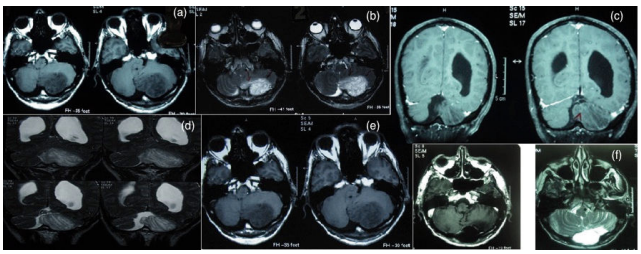

Figure 1: Preoperative and postoperative magnetic resonance imaging of the brain (MRI Brain) showing the following, preoperative TIW (a) series showing a well circumscribed, hypointense lesion in the left cerebellar hemisphere, hyperintense "tiger-striped" lesion on T2W (d) with no enhancement on contrast. (c) The last section of the figure shows a postoperative MRI showing post excision status, with T1 (e) and T2W (f) showing gross total excision of tumor.

A 58 year-old gentleman presented to our outpatient department with history of giddiness since 3 months, imbalance while walking since 2 months and holocranial headache since 2 months. The

headache was more in the morning and associated with blurring of vision at the peak of headache. On examination he had bilateral grade 1 papilledema and left side cerebellar signs with impaired tandem walking. Preoperative magnetic resonance imaging (MRI) of the brain showed a well circumscribed, hypointense lesion in the left cerebellar hemisphere, hyperintense "tiger-striped" lesion on T2W with no enhancement on contrast (Figure 1a-d). There was the typical non-enhancing gyriform pattern of enlargement of cerebellar folia. The characteristic imaging pattern obviated the need for any other differential diagnosis. He underwent a suboccipital craniotomy with gross total excision of the tumor with Ommayya reservoir placement (Figures 2-4). Postoperative MRI showed gross total excision of tumor and patient improved significantly with gross reduction in ataxia and giddiness (Figure 1e and 1f). Histopathological examination, proferred a diagnosis of Lhermitte Duclos disease (Figure 5).

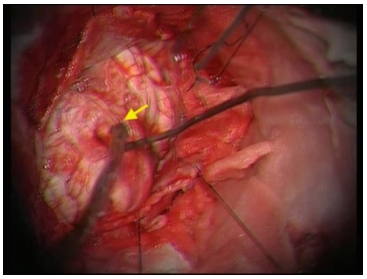

Figure 2: Intraoperative image (yellow arrow) showing diseased cerebellum.

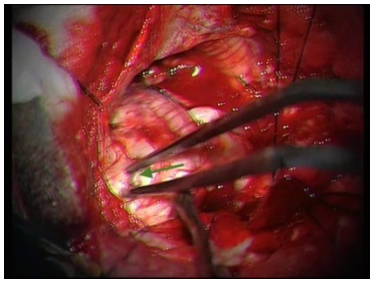

Figure 3: Intraoperative image showing (green arrow) resection of diseased cerebellum.

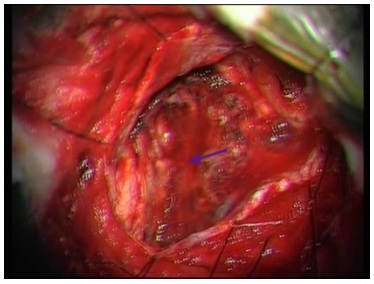

Figure 4: Intraoperative image showing post resection tumor bed (purple arrow).

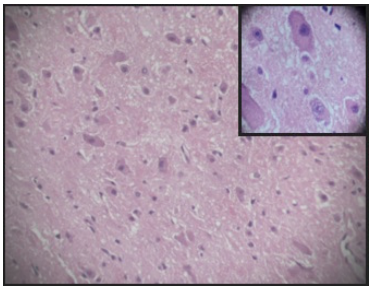

Figure 5: Broadened molecular layer with disappearance of Purkinje cells and progressive hypertrophy of the granular cells with loss of central white matter core of the cerebellar foliae.

Discussion

Dysplastic gangliocytoma (Lhermitte Duclos Disease), first described by Jacques Jean Lhermitte and P Duclos in 1920 [1-4], is a rare entity with only 220 cases reported in literature [5-9]. It usually affects the cerebellum and manifests in young adults though it can present from birth to sixth decade of life. The nature of this lesion remains unclear, however, hamartoma/hypertrophy accounts for most of the nomenclatures of this disease. Typically, patients present with headache, cerebellar dysfunction, occlusive hydrocephalus and cranial nerve palsies [3,4,9,10]. It is often associated with Cowden syndrome and is pathognomonic for this disease. It usually presents as a progressive lesion in the posterior fossa with surgical excision being the mainstay of treatment.

Cowden syndrome (Multiple hamartoma syndrome) is associated with mucocutaneous lesions and a strong association with breast, thyroid and endometrial cancers [2,4]. Various diagnostic criteria have been described for Cowdens (Table 1). These patients demonstrate a germline mutation in the PTEN gene. MRI brain has revolutionized the early detection and diagnosis of this entity. The T1W images typically show a hypointense, non-enhancing lesion with T2W images showing a high intensity lesion with a characteristic "tiger-striped" pattern [11,12]. Padberg et al. [13] described the association of LDD with CD in 1991. He stated that they both were a single phakomatosis and in the presence of LDD, the diagnostic criteria for CS are fulfilled if one other major manifestation of CS or three minor criteria are present.

The first successful surgery for LDD was performed in 1937 in a 34 year-old man who had experienced intermittent symptoms for 6 years. By 1955, only three patients had survived surgery for LDD. One third of the 90 patients diagnosed with LDD, before 1994, had died of the complications of their disease [14,15].

Most of the cases of LDD, reported in literature, were associated with Cowdens disease, however our case is the first to present without Cowdens disease. To date no other similar cases have been reported in literature.

Criteria for diagnosis of Cowden disease				
Mucocutaneous lesions	**Major**	**Minor**	**Working diagnosis (w/no family history)**	**Working diagnosis in a family w/ 1 person in whom cowden disease**
Facial trichilemmomas	Breast cancer	Noncancerous thyroid lesions	Mucocutaneous lesion alone ≥6 facial papules, of which ≥3 must be trichilemmomas, or cutaneous facial papules plus oral mucosal papillomatosis, or oral cutaneous papillomatosis and acral keratosis or ≥6 palmoplantar keratoses	Pathognomonic criteria 1 major criterion
Acral keratosis	Thyroid cancer	Cognitive delay (IQ≤75)	2 major criteria	2 minor criteria
Papillomatous papules	Macrocephaly	Gastrointestinal hamartomas	1 major and 3 minor criteria	History of Bannayan-Riley-Ruvalcaba syndrome
Mucosal lesions	Endometrial cancer	Breast fibrocystic diseasel	4 minor criteria	
		Lipomas, fibromas		
		Genitourinary tumors (renal cell cancer)		
		Genitourinary manifestations		
		Uterine fibroid tumors		

Table 1: Diagnostic criteria for Cowdens disease.

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