CT and MRI findings in Lhermitte-Duclos disease

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Abstract

Lhermitte-Duclos disease (LDD) is a rare cerebellar lesion of uncertain origin. It is linked to an autosomal- dominant phakomatosis known as Cowden's disease in 40% of patients. The MRI features of LDD are almost unique and can be considered diagnostic. We report on a patient who presented with the typical MRI features of the above disease, and discuss the imaging features, pathology and genetics.

Introduction

Lhermitte-Duclos disease (LDD) or dysplastic gangliocytoma is a rare slow-growing benign lesion of the cerebellum. Only 150 cases have been reported.[1] Its precise aetiology is uncertain. It is postulated that this disease may be a hamartoma, a developmental anomaly, a manifestation of a phakomatosis or a low-grade neoplasm. Recent published data suggest that LDD is a hamartomatous lesion related to a phakomatosis, rather than a neoplasm.[2]

LDD is often asymptomatic in early life, but usually becomes clinically apparent in the third and fourth decades. Reported cases have presented from birth to 74 years old.[1] There is no gender bias. The clinical manifestations are usually related to posterior fossa mass-effect and resultant secondary obstructive hydrocephalus. The most frequent complaints are headaches and ataxia due to intracranial hypertension. This occurs in 70% of patients. Cerebellar signs and symptoms are present in 40 - 50%. One-third of patients have associated cranial nerve palsies and long tract dysfunction. Visual problems, neck stiffness, vertigo and subarachnoid haemorrhage are less commonly noted presenting features.[1]

Case

A 62-year-old male patient presented in a delirious state to the casualty department. He had complained of severe headaches for the 4 days prior to his admission. There was no associated nausea and vomiting or obvious gait disturbance. On examination no papilloedema was noted. In view of his symptoms, a computed tomography (CT) scan (Siemens Somatom Sensation 16, Erlangen, Germany) of the brain was performed. CT revealed a large hypo dense lesion involving the posterior aspect of the right cerebellar hemisphere and the vermis (Fig. 1). It had a geographic shape, was well-defined, with no calcification. Compression and distortion of the 4th ventricle with consequent supratentorial hydrocephalus was noted.

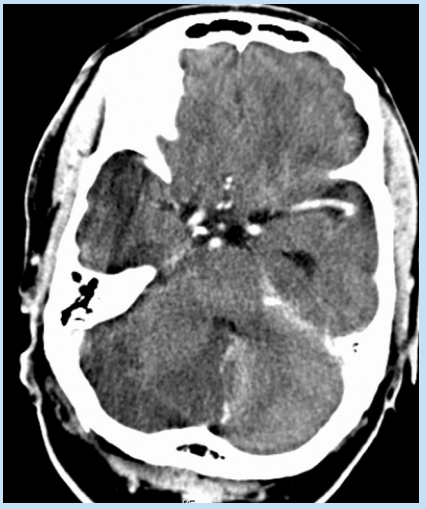

Fig. 1. Contrast-enhanced CT scan demonstrates a large non-enhancing intra-axial mass in the right cerebellum with a large temporal horn of left lateral ventricle.

Magnetic resonance imaging (MRI) (1,5T Siemens Symphony Maestro, Erlangen, Germany) revealed a well-marginated infratentorial mass in the right cerebellar hemisphere. Preservation and enlargement of the gyral pattern was noted (Figs 2 and 3). No enhancement was seen after administration of intravenous contrast.

Discussion

The definitive diagnosis of LDD is histopathological, but MR imaging can preoperatively characterise the disease sufficiently well. The abnormality seen in LDD is related to abnormal development of the cerebellar cortex. Usually, the normal cerebellar cortex consists of an inner gran-

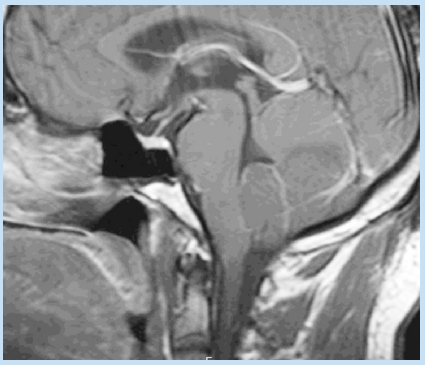

Fig. 2. Contrast-enhanced parasagittal coronal T1-weighted image reveals a hypo-intense mass with bands isodense to normal cerebellar tissue. A prominent vein is also noted draining into the straight sinus.

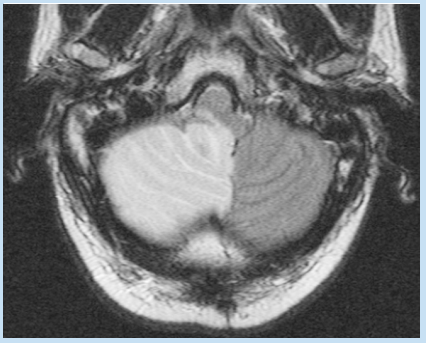

Fig. 3. Axial T2-weighted image shows the classic findings of a right hemi-cerebellar lesion with laminated hypertrophic folia and alternating curvilinear bands of high and low signal intensity. Similar findings were noted on the fluid attenuated inversion recovery (FLAIR) images.

ular layer, an outer molecular layer and an intervening Purkinje cell layer. In LDD there are abnormal ganglion cells in the granular layer, thickening and hypermyelination of the molecular layer and loss of the middle Purkinje cell bodies.[3]

On CT, LDD is usually seen as a well-defined isodense- to hypodense mass in the posterior cranial fossa with no contrast enhancement. Alternating bands of density that are usually seen on MRI may be seen on high-resolution fine-slice CT through the posterior fossa. Calcifications may be present. Depending on the size, mass effect with displacement of the 4th ventricle and obstructive hydrocephalus may be present

The histopathology of LDD forms the basis of the MRI findings. The demonstration of the typically striated, laminated or 'tiger-striped' appearance involving the cerebellar tissue is unique. Focal or diffuse cerebellar engorgement with prominent cerebellar folia and mass-effect are noted. This is caused by the close apposition of thickened cerebellar folia that have lost their secondary arborisation, resulting in alternating bands of tissue intensities. The atrophic white matter, the adjacent layers of abnormal ganglionic neurons and the innermost part of the molecular layers have prolonged signal (hypointense on T1 and hyperintense on T2 and FLAIR). The outer part of the molecular layer of adjacent folia and the intervening leptomeningeal space are iso-intense to normal cerebellar tissue both on T1 and T2 sequences. Lack of MR contrast enhancement is emphasised as an important MR diagnostic criterion of the disease but according to Spaargaren *et al.*[3] contrast enhancement has been reported. This is postulated to be due to venous proliferation that may also explain the large draining veins that are seen on angiography and as flow voids on MRI.

LDD is the only lesion besides cerebellar infarction that respects the normal cerebellar convolution markings despite causing enlargement. It is the abovementioned features together with the history that are pathognomonic. The only other lesion that may produce similar findings is cerebellar infarction, but the clinical presentation is usually very different.

MRI is excellent in defining the limits of the lesion to accomplish the most radical excision during surgery.

There is an association between Lhermitte-Duclos and Cowden's disease. This association has been noted in 40% of cases.[4] Cowden's disease is an autosomal-dominant condition with a prevalence of 1 in 200 000. It is due to a mutation of the PTEN gene that is located on chromosome 10q 22-23. This gene is related to tumour suppression. Mutation promotes proliferation and invasion of cells and failure of normal apoptosis. Cowden's disease is characterised by multiple hamartomas and neoplasms affecting the skin, thyroid, breast, and genitourinary and gastrointestinal tracts. The skin lesions are present in 90% of patients. The mucocutaneous manifestations form the pathognomonic basis for diagnosing Cowden's disease and consist of the following features: trichilemmomas, acral keratoses, as well as mucinous fibromas and oral papillomas.[1,4]

Despite the benign nature, surgical excision is the treatment of choice, although some centres may prefer conservative management in asymptomatic cases. Wide excision is necessary due to the risk of recurrence. However this is difficult because of ill-defined macroscopic margins. Radiation therapy is ineffective.

References

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