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Case Report

L'hermitte–Duclos Disease In An Infant: A Case Report With Review of Literature

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Abstract

Jacques Jean Lhermitte a French neurologist and P. Duclos in 1920 first described a growth in the cerebellum gangliocytoma, as a rare hamartomata's lesion due to abnormal development of the cerebellum. Since then it has been called by many names Lhermitte-Duclos disease, dysplastic gangliocytoma of the cerebellum, benign hypertrophy of the cerebellar cortex, granular cell hypertrophy and Purkinjeoma. It is a characteristic radiological feature and there has been reported in over 200 cases worldwide in adolescents [1] and old people but a handful of cases are detected and reported in infants. We report a 6 month old male child whose parents reported a history of irritability and typical lion facies with normal mile stones and radiological features of L'hermitte–Duclos disease.

Introduction

Although the exact aetiology remains unknown, a germline mutation of phosphatase and tensing homologue on chromosome 10q23 is considered widely as the underlying defect [1]. The abnormal tissue involves the cerebellar cortex, and is usually confined to one hemisphere, occasionally extending to the vermis but only rarely extending to the contralateral hemisphere [2]. A number of conditions are associated and have been described [2]. It is Imperative to look for Cowden's disease in which case it is termed Lhermitte-Duclos-Cowden syndrome or COLD syndrome. Other associated disorders of cortical formation include, megalencephalic, polymicrogyria, macroglossia and leontiasis ossea-Lion Facies. The MRI features are described as the Leopard skin sign (also known as tigroid pattern or stripe sign) results from dark-spots or stripes (spared perivascular white matter) within bright demyelinated periventricular white matter on T2W images [3].

Case presentation

A 6 month old male was brought by his parents with a history of irritability for a few months. There was no history of vomiting the remainder of the cranial nerves were within normal limits. Motor and sensory examinations were normal.

No delay in mile stones and an unremarkable An Ophthalmological & fundoscopic examination was unremarkable. An Ct Brain followed by an MRI picked a low-density non enhancing stripped lesion.

A CT examination of the head demonstrated a low-density non enhancing lesion of the left cerebral hemisphere (Figure 1). No mass effect of the fourth ventricle. A CT Scan may show a non-specific hypoattenuating cerebellar mass with or without calcification [3].

An MRI examination of the brain was performed that showed an inhomogeneous mass in the left cerebellum with a striated foliar pattern, trigonid pattern or stripe sign No Hydrocephalus was noted. On the basis of the imaging studies a diagnosis of Lhermitte-Duclos disease was postulated. An important differential to be considered is an atypical glioma.

The Infant was planned for Shunt Insertion following evidence of obstructive hydrocephalus. He is on follow up and is well.

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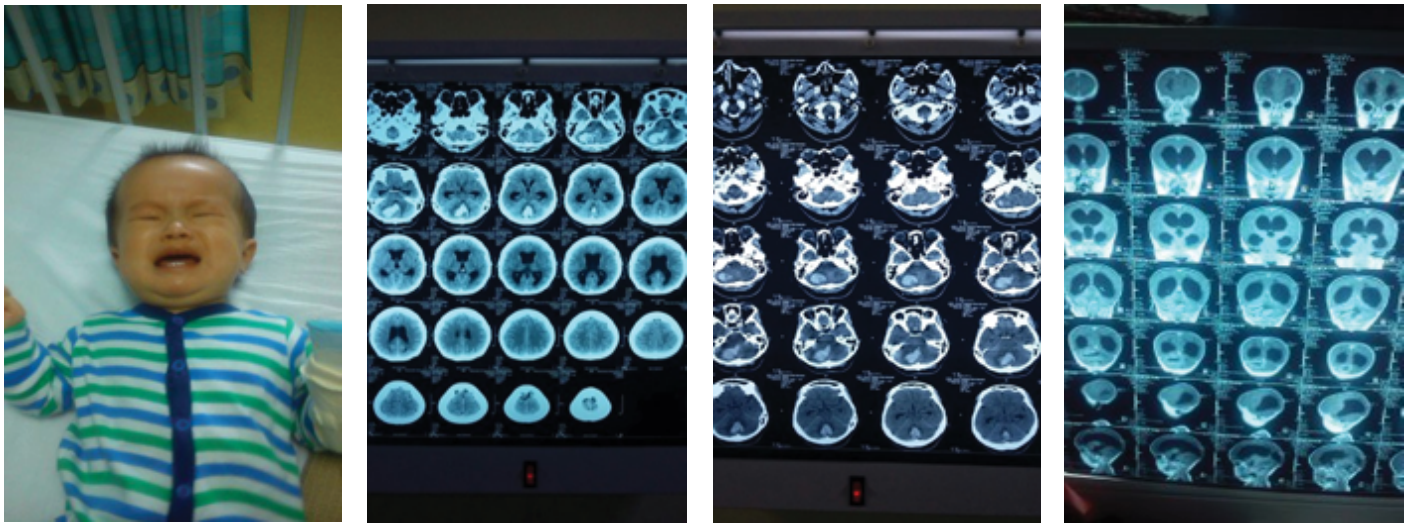

Figure 1. *A 6 month male infant, On T2-weighted images the lesions present with a well circumscribed high signal intensity and an unique striated pattern with isointense bands within the area of hyper intensity, indicating the structures of widened gyri and consequently displaced sulci of the cerebellar cortex.*

Discussion

Lhermitte and Duclos were the first to describe an abnormality in the cerebellum of locally enlarged areas that containing abnormal ganglion cells in the granular layer, with a thickened and hyper myelinated molecular layer and associated loss of Purkinje cells [1]. This disease goes by several names including Lhermitte-Duclos disease, granular cell hypertrophy, dysplastic gangliocytoma benign hypertrophy of the cerebellar cortex, and Purkinjeoma. The basic lesion may be secondary to a disturbance affecting the differentiation and migration of granule cell precursors [3]. The abnormality is hemispheric and unilateral. It occurs sporadically but also in a familial form have been reported [4]. This lesion has a slight female preponderance in the third or fourth decade [5] The lesion may be asymptomatic, if the lesion is small and it not unusual that some cases are discovered only at autopsy. The association of polydactyly and hemangiomas [6] is in favour of the dysplastic origin theory, other theories of origin include hamartomatous, neoplastic, or congenital malformative origin. Lhermitte-Duclos disease has a germline loss of one PTEN allele with the loss of the remaining PTEN allele at some point, is serves as a trigger for abnormal growth of the granule cells [7,8].

Lhermitte-Duclos disease and Cowden syndrome a component of single spectrum best classified as a single phacomatosis [9]. The red flags are symptoms of increased intracranial pressure, such as headaches, nausea and vomiting, papilloedema, mental disturbances and loss of consciousness indicate progressive mass effect of the growing tumor, an indication for an urgent decompression by sub-occipital based surgical approach [9].

The Clinical evidence and the close association of dysplastic cerebellar gangliocytoma with multiple hamartoma-neoplasia spectra (Cowden syndrome) [4,9,10] favor a hamartomata's origin. The presence of skin growth and oral lesions must alert the treating clinician to Cowden syndrome complex that has autosomal dominant inheritance and is characterized by macrocephaly, central nervous system lesions meningioma and gliomas [9,11] a variety of mucocutaneous lesions, and increased frequency of hamartomas and neoplasia in the breast, thyroid, colon, genitourinary organs. The neoplasms are of ectodermal, mesodermal, and endodermal origin. Tuberous sclerosis is a differential that is indicative of the presence of dysplastic cerebellar lesions it is characterized by cortical tubers, subependymal nodules. Cerebellar astrocytoma are enhancing cystic tumors an important differential that ought to be sought for cystic lesions.

If Clinical symptoms range from persistent headache, Ataxia, Ocullovestibular signs visual disturbances like scotomas and occlusive hydrocephalus [12] and sudden death due to herniation, hemangioblastoma should be high on the differential list.

The Histologic features of LDD include the thickening of outer molecular layer that contain hyper myelinated axons with but no Purkinje dendrites [13] There is also a loss of Purkinje cell bodies as is the hypertrophy of involved areas that brings about a growth and subsequent mass effect. In short there is a loss of normal cerebellar cortical cell layers with dysplastic hypertrophied ganglion cells leading to expansion of the granule layer and increased myelination in the molecular layer causing it to widen.

Conclusions

Because of the high incidence of systemic cancer in patients with Cowden disease, it is important for the treating Clinicians to recognize the association between L'hermitte–Duclos disease and Cowdens syndrome and the need for appropriate screening and surveillance in Cowdens syndrome complex in order to refer affected patients for appropriate management.

Declaration of Consent

Appropriate patient consent forms were obtained, every effort was taken to conceal patient identity.

Conflict of Interest

None

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References

1. L'hermitte J, Duclos P. On the diffuse ganglioneuroma of the cerebellar cortex. Bull Assoc Fr Etud Cancer. 1920;9:99–107.
2. Kulkantrakorn K, Awwad EE, Levy B, et al. MRI in Lhermitte-Duclos disease. Neurology. 1997;48(3):725-731.
3. Klisch J, Juengling F, Spreer J, et al. Lhermitte-Duclos disease: assessment with MR imaging, positron emission tomography, single-photon emission CT, and MR spectroscopy. AJNR Am J Neuroradiol. 2001;22(5):824-830.
4. Albrecht S, Haber RM, Goodman JC, Duvic M. Cowden syndrome and Lhermitte-Duclos disease. Cancer. 1992;70(4):869-876.
5. Roski RA, Roessmann U, Spetzler RF, Kaufman B, Nulsen FE. Clinical and pathological study of dysplastic gangliocytoma. J Neurosurg. 1981;55:318-321
6. Ambler M, Pagacar S, Sidman R. Lhermitte-Duclos disease (granule cell hypertrophy of the cerebellum). Pathological analysis of the first familial cases. J Neuropathol Exp Neurol. 1969;28:622-647
7. Zhou XP, Marsh DJ, Morrison CD, et al. Germline inactivation of PTEN and dysregulation of the phosphoinositol-3-kinase/Akt pathway cause human Lhermitte-Duclos disease in adults. Am J Hum Genet 2003;73:1191–1198.
8. Maehama T, Dixon JE. The tumor suppressor, PTEN/MMAC1, dephosphorylates the lipid second messenger, phosphatidylinositol 3,4,5-trisphosphate. Journal of Biol Chem. 1998;273:13375–13378.
9. Robinson S, Cohen AR. Cowden disease and Lhermitte-Duclos disease: characterization of a new phakomatosis. Neurosurgery. 2000;46(2):371–83.
10. Robinson S, Cohen AR. Cowden disease and Lhermitte-Duclos disease: characterization of a new phakomatosis. Neurosurgery 2000;46:371–383.
11. Padberg GW, Schot JDL, Vielvoye GJ, et al. Lhermitte-Duclos disease and Cowden disease: a single phakomatosis. Ann Neural. 1991;29:517-523.
12. Vinchon M, Blond S, Lejeune JP, et al. Association of Lhermitte-Duclos and Cowden disease: report of a new case and review of the literature. J Neurol Neurosurg Psychiatry. 1994;57:699–704.
13. Milbouw G, Born JD, Martin D, et al. Clinical and radiological aspects of dysplastic gangliocytoma (Lhermitte-Duclos disease): a report of two cases with review of the literature.Neurosurg. 1988;22:124–8.
14. Adams JH, Corsellis JAN, Duchen LW, eds. Greenfield's neuropathology, 4th ed. New York: Wiley, 1984