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Lhermitte - Duclos Disease with Syrinx: Case Report and Literature Review

Sirinks ile Birlikte Lhermitte - Duclos Hastalığı: Olgu Sunumu ve Literatürün Gözden Geçirilmesi

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ABSTRACT

BACKGROUND: Lhermitte-Duclos disease is typified by a hamartomatous lesion of the cerebellum. It is usually seen in females. The usual presentation is of raised intracranial pressure along with cerebellar signs.

CASE REPORT: We report an 18-year-old female patient who presented to us with history of headache and gait ataxia and was observed to have Lhermitte-Duclos disease with cervical cord syrinx on imaging.

DISCUSSION: To our knowledge this is the fifth case of Lhermitte-Duclos disease with syringomyelia in the pediatric age group. The treatment of this condition is decompression of the lesion. Malignant transformation never occurs and the prognosis is excellent.

KEYWORDS: Lhermitte-Duclos disease, T-2 shine through phenomenon, Tigroid appearance, Cerebellar tumors, Gangliocytoma, Hydrocephalus, Syringomyelia

ÖZ

ÖNBİLGİ: Lhermitte-Duclos hastalığı serebellumun hamartamoz lezyonu ile karekterizedir. Genellikle bayanlarda görülür. En sık prezentasyonu serebellar bulgularla birlikte intrakranyal basınç artışıdır.

OLGU SUNUMU: Başağrısı ve yürüme ataksisi hikayesiyle bize başvuran ve görüntülemesinde servikal kord siringomyeli ile birlikte Lhermitte-Duclos hastalığı olduğu gözlenen 18 yaşında bir bayan hasta sunuyoruz.

TARTIŞMA: Bildiğimiz kadarı ile bu hasta pediatrik yaş grubunda sirengomyeli ile birlikte Lhermitte-Duclos hastalığı olan beşinci vakadır. Bu durumun tedavisi lezyonun dekompresyonudur. Malign transformasyon hiçbir zaman olmaz ve prognoz mükemmeldir.

ANAHTAR SÖZCÜKLER: Lhermitte-Duclos hastalığı, T-2 shine through fenomeni, Tigroid görüntü, Serebellar tümörler, Gangliositoma, Hidrosefali, Sirengomyeli

INTRODUCTION

Lhermitte-Duclos disease is a congenital malformation in which a hamartoma arises in cerebellar cortex. Other descriptive names for this entity are dysplastic gangliocytoma, granule cell hypertrophy, hamartoma of the cerebellum, diffuse ganglioneuroma of the cerebellum, neurocytic blastoma, myelinated neurocytoma, granulomolecular hypertrophy, Purkinjeoma (5). The usual manifestation of this disorder is headache, vomiting, ataxia and visual disturbances (11).

CASE REPORT

An 18-year-old female reported to neurosurgical outpatient department with a 5 month history of global headache and one month history of gait ataxia. There was no history of vomiting or visual disturbances.

Her general physical and systemic examinations were normal. There were no neurocutaneous markers. She was conscious, coherent and alert. Fundi revealed early papilledema. She had ataxic gait and cerebellar signs on right side.

She was evaluated with MRI brain. It revealed a non-enhancing lesion involving the right cerebellar hemisphere and the vermis. The lesion had a characteristic gyriform pattern which was hypointense on TIW and hyperintense on T2W sequences giving it a classical tigroid appearance. Diffusion-weighted MRI showed increased signal intensity of the lesion. Magnetic resonance spectroscopy revealed high choline and high lactate peaks. The lesion was causing obstructive hydrocephalus. There was tonsillar herniation and syringomyelia of the cervical cord from C2 to C3 level (Figures,1A-C).

Patient underwent a left ventriculoperitoneal shunt followed by right suboccipital craniectomy, C1 arch excision and

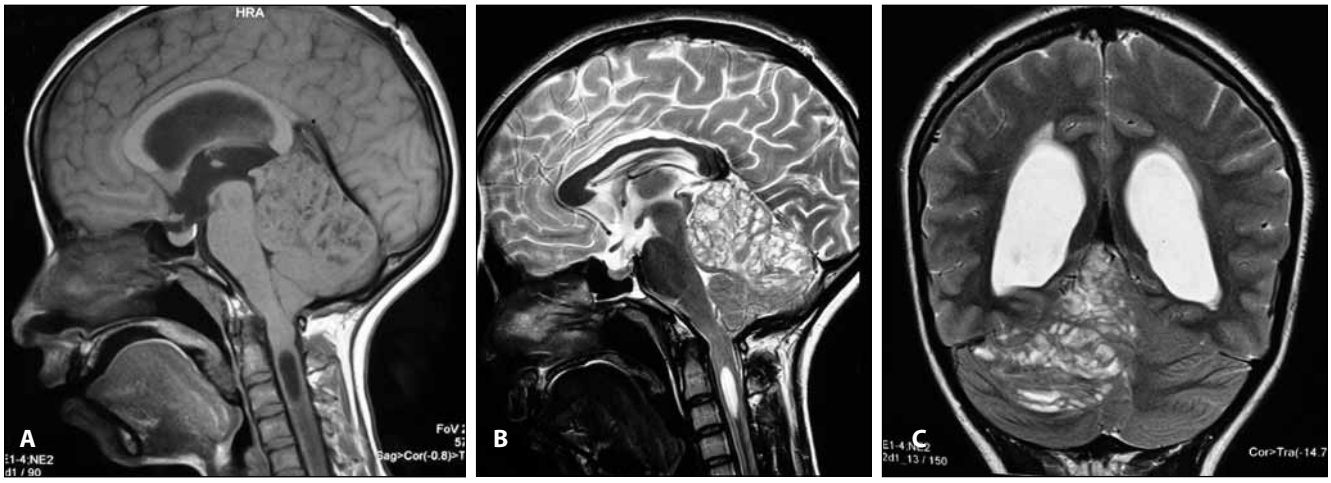

Figure 1: A) T1-W sagittal **B)** T2-Wsagittal **C)** T2-W coronal show the typical tigroid lesion involving the right cerebellar hemisphere. Also seen are the hydrocephalus and cervical cord syrinx.

decompression of the tumor. At surgery the occipital bone was normal, the cerebellum was diffusely enlarged, and the tumor was avascular with no definite plane of cleavage. A subtotal decompression of the tumor was done. Duraplasty up to the C1 level that was thought to take care of the cervical cord syrinx also was done. The patient had an uneventful postoperative recovery. Patient is now on 2 months follow-up and is doing well. There is no headache or vomiting. Her gait is normal and fundoscopic changes have also normalized.

Histopathological examination revealed replacement of granular cell layer by cells which were mainly ganglionic cells and few well differentiated neuron cells. Axons were in parallel stalks within the deep molecular zone and in perpendicular array superficially in relation to pial surface. The features were consistent with dysplastic gangliocytoma of cerebellum (Figure 2).

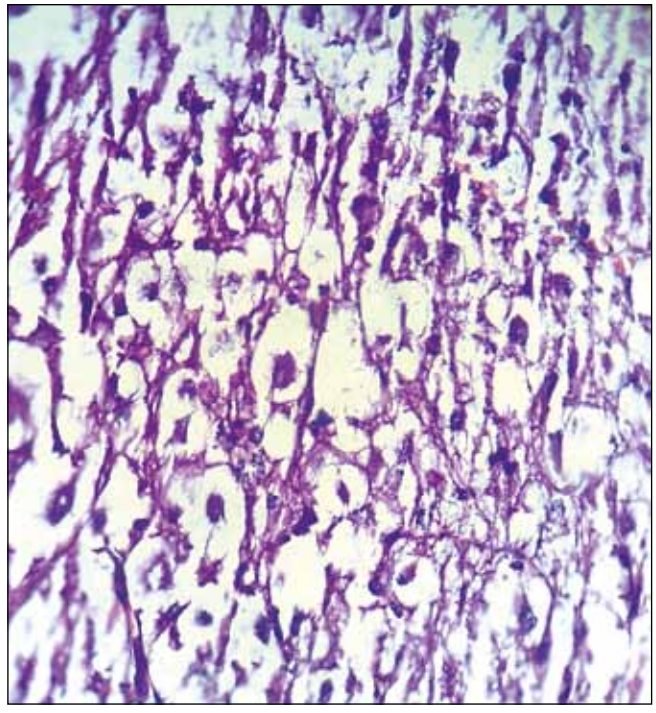

Figure 2: Large dysplastic neuronal cells with ganglion cell like morphology in dysplastic ganglioglioma of cerebellum (H&E; x 40).

DISCUSSION

Lhermitte-Duclos disease is a hamartomatous overgrowth of cerebellar ganglion cells (5). The first case was reported by Lhermitte and Duclos in 1920 (7). Patients usually present with headache, vomiting and cerebellar signs (11). The duration of symptoms varies from months to years and most patients are females and the usual age range is 9-37 years (14).

The association of Lhermitte-Duclos disease with Cowden's disease was first described by Padberg et al. in 1961(12). The clinical features of Cowden's disease include facial trichilemmomas, oral papillomas, thyroid ,breast, colon, endometrial and ovarian tumors (14). Both conditions are associated with mutations in PTEN tumor suppressor gene located on chromosome 10. We screened our patient for markers of Cowden's disease, and the result was negative. There are case reports where both conditions coexist (3) but isolated cases of Lhermitte-Duclos disease is more common (13,18). A screen for Cowden's disease in every case of LDD is a must as it may lead to an early diagnosis of skin, thyroid, breast, colonic, endometrial and ovarian carcinomas (14).

MRI reveals the thickened cerebellar folia as having a characteristic gyriform pattern ,the gyri being hypointense on TIW and hyperintense on T2W sequences (tigroid appearance) (9,15). The mass may be calcified (14). Mittal et al. (10) have reported a case of medulloblastoma which had few features resembling Lhermitte-Duclos disease on MRI. However

patchy enhancement was seen on contrast images and the mass showed restricted diffusion on diffusion-weighted/ apparent diffusion coefficient images (ADC) images. Hydrocephalus in LDD may be secondary to the 4th ventricle compression. In a series by Robinson et al., 4 out of 5 cases had hydrocephalus and these patients needed CSF diversion (14). Diffusion-weighted MRI sequence in LDD shows increased signal intensity due to a T2 shine through effect rather than restricted diffusion as reflected by lack of hypointensity on apparent diffusion coefficient (18). The lesion is usually unilateral involving predominantly one of the cerebellar hemispheres, preferably the left (20). The lesion is classically non-enhancing, however, enhancement has also been reported in few cases (16). Magnetic resonance spectroscopy reveals high choline peaks because of demyelination and also high lactate peaks secondary to anaerobic glycolysis. N-acetyl aspartate (NAA) levels are low (18). Thomas et al. demonstrated the characteristic deep running veins between the folia on SWI (susceptibility weighted imaging), thought to be the cause for vascular enhancement. Also demonstrated was increase in relative cerebral volume, relative cerebral blood flow and mean transit time in the lesion on perfusion scans (18). Tonsillar herniation may be associated and in few cases associated syringomyelia has also been reported (8,18). Syringomyelia in posterior fossa lesions is supposed to be secondary to cerebrospinal fluid block at foramen magnum level and obex resulting in cranio-spinal pressure dissociation (4). There was tonsillar herniation in our patient and we speculate this as the cause of cervical cord syrinx. Out of the previously reported cases of LDD with syrinx only one belongs to pediatric age group (18). Our patient was an 18-year-old female who is the second reported case of LDD with syrinx in pediatric age group.

The lesion in view of its slow growing nature may cause thinning of the occipital bone (14). In our patient the bone was normal, which can be explained by the fact that our patient had a short, 5-month history of symptoms.

Left untreated the outcome is poor because of the progressive growth of the tumor causing brain stem compression (1). The treatment of this condition is decompression of the lesion. Total excision usually is not possible as there is poor plane of cleavage between the lesion and the normal cerebellar tissue. We also could not get a clear plane around the tumor as it was infiltrative in nature, and we could thus do subtotal excision only. This subtotal excision of the tumor has been reported to be the cause of recurrence (6) and repeated decompression of the lesion has been reported after 11 years (2).

The lesion is a low grade WHO - grade 1 tumor. Histopathological examination reveals widening of molecular layer with abnormal myelination, loss of Purkinje cell layer and hypertrophy of granular cell layer. Also salient is the atrophy of folial white matter (19). Malignant transformation is very rare (17) and prognosis is excellent (14).

CONCLUSION

Lhermitte-Duclos disease is characterized by dysplastic gangliocytoma of the cerebellum. A screen for Cowden's syndrome should be done in every patient. Usual presentation is raised intracranial pressure.MRI imaging is characteristic and it may also reveal tonsillar herniation and syringomyelia. Prognosis is excellent and surgical decompression is the treatment of choice and patient need to be kept on follow up as long term recurrence can occur.

ABBREVIATIONS

LDD- Lhermitte-Duclos disease, **MRI-** Magnetic resonance imaging, **H&E-** Haematoxylin and Eosin.

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