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CASE REPORT

A Neck Pain Alone Causing Death?

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Abstract:

Neck pain is pain in or around the spine beneath the head. It is a common symptom of many injuries and medical conditions which can be acute or chronic. Common causes of neck pain are increased physical strain, trauma, osteoarthritis, rheumatoid arthritis, meningitis, tumours of spine etc. Most cases of acute neck pain can resolve with or without treatment. But red flags which may signify serious pathologies such as myelopathy, atlantoaxial subluxation, meningitis, metastasis etc. should be promptly identified. A 29 year old male with complaints of neck pain was brought to the casualty of a local hospital in Thiruvananthapuram and expired after 6 hours due to cardiac arrest. The autopsy revealed a mass in the fourth ventricle. Cause of death was brain oedema following tumour in fourth ventricle.

Keywords: Neck pain; Tumour; Brain oedema; Mass in fourth ventricle.

Introduction:

Hamartoma lesions anywhere in the body are considered as rare entities and Lhermitte-Duclos Disease (LDD) or dysplastic gangliocytoma of cerebellum is an extremely rare hamartoma. It is a congenital malformation which is classified as a hamartoma. Lhermitte-Duclos disease arises in the cerebellum and is a unique tumour. Although it has characteristics of a benign neoplasm since categorized as a WHO grade 1 tumour histologically, it is more hamartomatous, containing dysplastic cells in an enlarged abnormal cerebellar folium.[1] There is no gender preference and whilst the condition is mostly diagnosed in the second and third decade of life with posterior fossa symptoms,[2] rare cases with childhood onset were also reported.[3] It has chances for recurrence otherwise no reports of malignant transformation have been found. Here a case is discussed which showed a different presentation.

Case report:

A 29 year old male with no previous comorbidities developed sudden onset of dizziness and vomiting. He went to a local hospital where he was diagnosed as benign paroxysmal positional vertigo and was conservatively managed. After three weeks, his symptoms persisted and developed neck pain. On examination, the patient had elevated blood pressure for which he was given antihypertensives and referred to an orthopaedician who advised conservative management.

Patient consulted another hospital where he was advised to take an MRI scan of his brain. The MRI scan of the brain showed an ill-defined parenchymal mass lesion measuring 4.5x3.5x5.5 cm involving inferior cerebellar vermis appearing heterogeneously hypointense on T1WI, hyperintense on T2 FLAIR sequence with a striated appearance. The lesion was protruding into and narrowing fourth ventricle causing obstructive hydrocephalus with inferior herniation of cerebellar tonsils along with diffuse effacement of cerebral sulcus and sylvian fissures. He collapsed after the MRI scan and despite all resuscitative efforts, the patient expired.

The body was brought for autopsy to the mortuary wing of Govt. Medical college, Thiruvananthapuram. On dissection, brain was congested, showed signs of raised intracranial tension and weighed 2241g. A soft mass of size 5x3.5x3 cm was present in the fourth ventricle arising from the left side of the cerebellum. The mass was sent for histopathological examination which revealed deranged laminar cellular organisation, loss of purkinje cell layer and infiltration of granular cell layer with dysplastic ganglion cells. It was diagnosed as dysplastic gangliocytoma of the cerebellum (otherwise known as Lhermitte-Duclos disease).

Discussion:

Lhermitte and Duclos first described the cerebellar dysplastic gangliocytoma in 1920. They reported on a 36-year-old man who suffered occipital headaches and diminished hearing on the left side that was progressive over 10 months. During the few weeks before his presentation he suffered paroxysmal vertigo with recurrent falls, gait ataxia, disorientation, and memory deficits. At the time of admission, he exhibited confusion, disorientation, dysarthria, nystagmus, and cerebellar ataxia. His condition worsened and he died.[2] Lhermitte - Duclos Disease is a very rare hamartomatous lesion. It is associated with Cowden's syndrome and is classfied as a phakomatoses/phacomatosis pigmentovascularis which includes rare disorders arising from embryonic ectoderm. These disorders have a cutaneous and neurological involvement, often with dysplasia of other organs. The typical presentation includes cerebellar ataxia, lower cranial nerve palsies, signs and symptoms of raised intracranial pressure and visual disturbances.[1]

It usually affects patients aged 30-50 years, even though in rare

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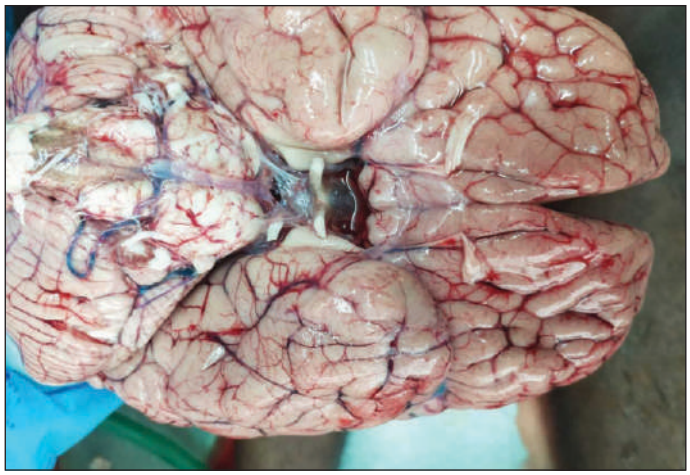
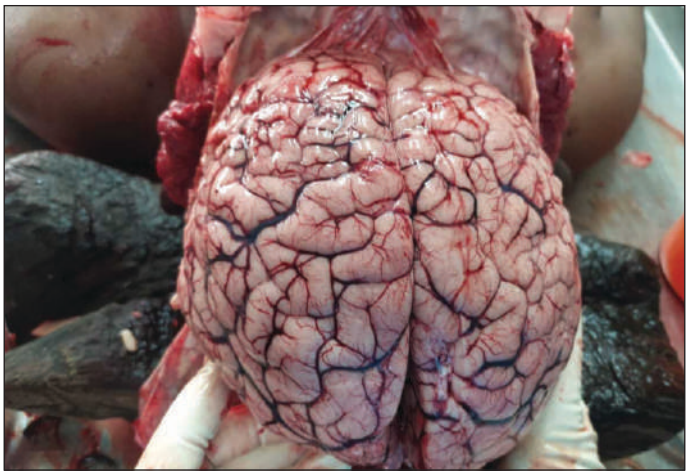
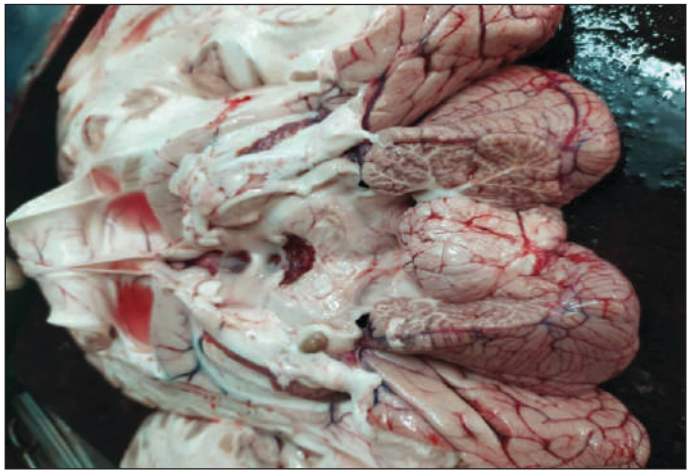
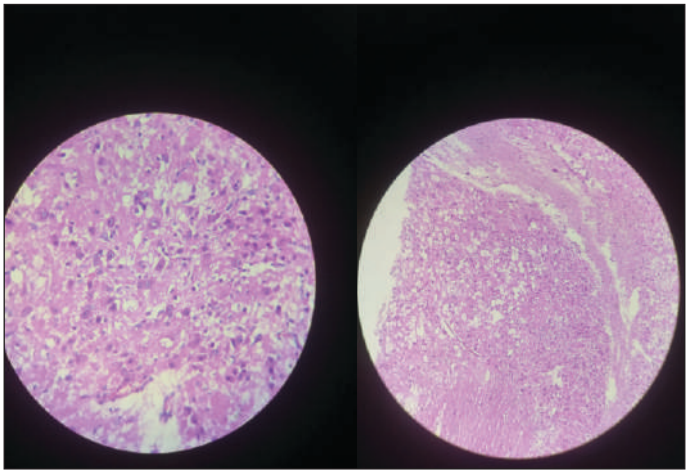

cases, it is found in infants and adults over 60 years of age. There is no known sex or race preference. There is difference of opinion regarding the cause of this disease: it can be hamartomatous, neoplastic, or congenital malformative origin. There can be associated malformations like macrocephaly, syringomyelia, polydactyly, multiple haemangiomas and mucocutaneous lesions as well as breast, thyroid and gastrointestinal malignancies. Hence, a genetic correlation between Lhermitte- Duclos disease and Cowden's syndrome has been postulated.[1,2,4,5]

Small tumours may be asymptomatic or only present with comparatively subtle cerebellar signs. Since the tumour grows very slowly, the onset of any symptoms is gradual. When the tumour increased in size, the signs and symptoms are due to raised intracranial pressure, obstructive hydrocephalus or cerebellar signs.[2]

Lhermitte- Duclos disease is a germline loss of the PTEN allele and the subsequent loss of the second allele leading to pathological growth of the granular cells along with additional mutations in the EGFR and SDHB-D. Due to the rarity of this disease, clinical diagnosis can be challenging. Imaging, however, is crucial, and the typical appearance of 'tiger stripes' on T2 MRI can be pathognomonic for Lhermitte- Duclos disease which is also known as corduroy or laminated appearance. The lesion is described as focal, well-circumscribed, isolated to a single cerebellar hemisphere with a characteristic gyri- form pattern and hypertrophy of the folia as the dysplastic ganglionic cells infiltrate and hypertrophy in the granular layer of the cerebellum. MRI scan is very sensitive in detecting enlarged folia with T2 showing the characteristic tigroid appearance, which is considered specific.[1,4-6]

The initial treatment is based on treating hydrocephalus. Surgical resection is almost curative in most cases with few cases of recurrence. Since there is a possible association to Cowden's syndrome, increased risk of other neoplasms should be expected. Therefore, a recommendation for further imaging or clinical assessment of possible tumours should be included.[7,8]

The significance of identifying such rare conditions is to widen the spectra of differential diagnosis for physicians and extend the investigations keeping in mind such unique diseases also. Thus a timely diagnosis may not only save a life but also prevent occurrences of medical negligence in the future.

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