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AN UNSUAL CASE OF CREUTZFELD- JACOB DISEASE PRESENTING WITH CEREBELLAR ATAXIA-BROWNELL OPENHEIMER VARIANT.

Radiodiagnosis

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ABSTRACT

Creutzfeld -Jacob Disease (CJD) is a rare type of neurodegenerative disease caused due to prion disease and is rapidly progressive and fatal. It is usually suspected based on the typical presenting symptoms, most important symptom being rapidly progressive dementia. Cases with atypical presentation are usually missed due to low clinical suspicion.
We present a case report of a 64-year-old male who presented with worsening cerebellar ataxia and only mild dementia.MRI done to rule out any organic cause, revealed classic findings of CJD.Following this EEG was performed. MRI helped in clinching the diagnosis in this case of low clinical suspicion. This was an extremely rare variant of CJD-the brownell openheimer variant.[1]

KEYWORDS

Creutzfeld -Jacob Disease (CJD),prion disease, variant CJD (v CJD),brownell openheimer variant, sporadic CJD (s CJD).

INTRODUCTION:

Creutzfeld -Jacob Disease (CJD) belongs to the spectrum of spongiform encephalopathies and is a rare neurodegenerative disease with a fatal outcome .Considered as prion diseases, the sporadic CJD forms the largest entity in the spectrum [2]It is a challenging entity because it is rare, there is low clinical suspicion and lack of knowledge about its imaging appearance. Even more rare is the brownell openheimer variant which presents only with ataxia.[1]Here we report an unusual case of CJD in an elderly man presenting with cerebellar ataxia with no clinical suspicion of CJD but MRI showing the classic findings of the same.

CASE REPORT:

A 64 years male patient came with the chief complaint of recent onset ataxia and imbalance.. There was no history of any fever, alcoholism or HIV. Neurological examination confirmed the ataxia of cerebellar origin. There was mild dementia, which was attributed to his age. There were no other neurological signs and symptoms. No previous imaging . Blood investigations were within normal limits. The patient underwent MRI Brain to rule out any organic cause/cerebellar abnormality. Limited MRI brain was performed, as the patient was highly restless. MRI revealed few classic imaging findings :the patient had cortical and cerebellar atrophy.T2Weighted and DWI sequences revealed hyperintensity with restricted diffusivity in bilateral caudate and lentiform nucleus.Subtle hyperintensity with restriction was also noted in few cortical regions of parietal lobes (cortical ribbon sign). A curvilinear rim of restricted diffusivity was also noted in the posteromedial aspect of bilateral thalamus resembling the hockey stick/pulvinar sign. No space-occupying lesion was detected.

These imaging findings were quite typical of Creutzfeld Jacob Disease. As this was not suspected clinically, this case was discussed with the concerned neurologist. After this an EEG was performed which revealed generalized and synchronous periodic stereotypic sharp waves occurring at intervals of 0.5 -1 seconds. Based on CDC criteria, the patient was characterised as probable CJD (brownell openheimer variant) while he was alive.

Clinically the patient worsened as he developed rapidly progressive dementia and myoclonic jerks.

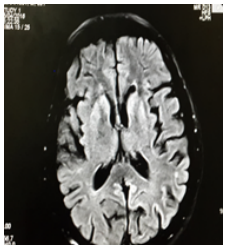

Figure1. MRI Brain.: FLAIR image reveals hyperintensity in bilateral caudate and putamen. A subtle hyperintensity is also noted along the cortex of left parietal region.

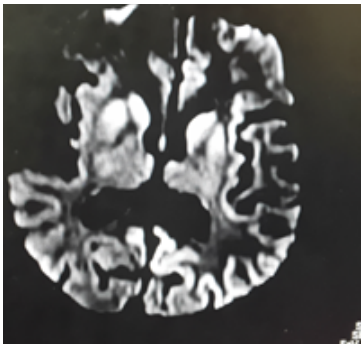

Figure 2: : DWI (Diffusion weighted Images) shows restriction in the bilateral basal ganglia. cortical areas of restricted diffusivity(cortical ribbon sign)

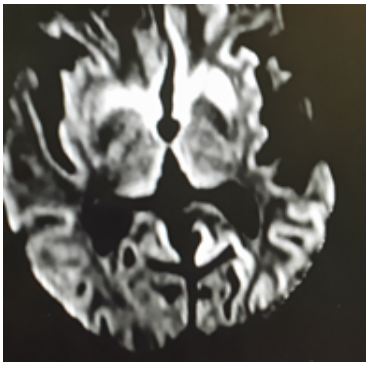

Figure 3: : DWI (Diffusion weighted Images) shows curvilinear areas of restriction in bilateral posteromedial thalamus resembling hockey stick and consistent with pulvinar sign.

DISCUSSION

A classic case of CJD typically presents with rapidly progressive dementia, myoclonus, pyramidal/extrapyramidal symptoms and akinetic mutism. EEG findings usually reveal periodic sharp wave complexes .The clinical picture may not always be typical in all cases.

The brownell openheimer is an extremely rare variant of CJD presenting only with ataxia as seen in our case. Our patient did not have akinetic mutism or myoclonus which may not be present in all cases.[4] MRI Imaging plays a crucial role in the diagnosis of CJD especially in such rare variants where there is low/no clinical suspicion of CJD.

This rare entity is thus a diagnostic dilemma in the initial stages .It later develops all the classical symptoms of CJD.The CSF analysis of such patients may not show elevated 14-3-3 proteins.

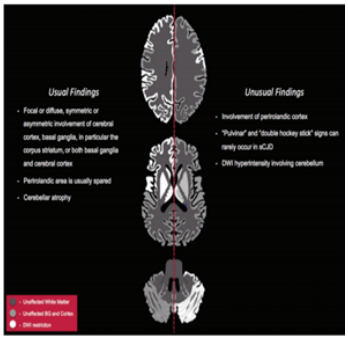

Figure 4.This figure illustrates the typical findings of CJD on MRI.[3]

Our case also showed curvilinear area of restricted diffusivity on DWI in bilateral posteromedial thalamus known as the pulvinar/hockey stick sign. Initially this sign was believed to be seen only in variant CJD. However later it was stated that it can also be seen in sporadic CJD including the rare brownell openheimer variant as seen in our case.[3,5,6]

Differential diagnosis

Predominant Cerebral Cortex Involvement

Severe Hypoxic Ischemic Encephalopathy.— The earliest finding at MR imaging is restricted diffusivity on DWI mimicking a pattern of sCJD and in severe cases can also affect the basal ganglia, hippocampus, and cerebral cortex. Involvement of perirolandic cortex and the typical clinical setting distinguishes it from CJD.[3]

Infectious Disease (Encephalitis).—Herpes simplex virus is the most common agent and typically reveals restricted diffusivity in the anterior and medial aspects of the temporal lobe, frontal lobe, cingulate gyrus and insular cortex. The febrile status and CSF picture easily differentiate from sporadic CJD.

Postictal State.—Transient and reversible signal intensity changes include gyral swelling, FLAIR/T2-weighted imaging hyperintensity restricted diffusion and enhancement[7].The reversible changes and the clinical presentation help in diagnosis Mitochondrial disorders like MELAS can have some imaging similarities .However clinical history removes the confusion.

Abnormalities of basal ganglia includes conditions like extra pontine myelinolysis,Epstein barr encephalitis,wilsons,wernikes encephalopathy and many others. However they have different clinical presentations and can be differentiated on imaging [3]

All cases of CJD including the brownell openheimer variant has poor prognosis and has a fatal outcome.

CONCLUSION

CJD, a rare but fatal neurodegenerative disease is often a diagnostic challenge. Our case was a rare brownell openheimer variant of CJD with an atypical presentation, where MRI helped in clinching the diagnosis in spite of low clinical suspicion. Though incurable early diagnosis is crucial to differentiate from other treatable conditions.

TEACHING POINTS.

1. The pulvinar hockey stick sign is usually noted in variant CJD, however may also be seen in sporadic CJD as seen in our case.[5,6]
2. Although a rare entity, brownell openheimer variant must be kept in mind as a possibility in such atypical cases. Hence, it is essential to know about its imaging appearance to suggest the diagnosis of CJD.

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