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Creutzfeldt - Jakob Disease – A Rare Case Report

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Introduction- Prion diseases are neurodegenerative diseases that have incubation period. Five prion diseases are recognized they are kuru, Creutzfeldt-Jakob disease (CJD), variant CJD, Gerstmann-straussler Scheinker syndrome (GSS) and fatal familial insomnia1. Among all these disease, CJD accounts for 90% of all prion disease. One case of CJD occurs among 1,000,000 populations per year. It is such a rare presentation. Dementia and myoclonus are the most common presenting condition of CJD. This is a rare case report of a patient who had a rare clinical presentation, and finally it was diagnosed to be a case of CJD.

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Creutzfeldt - Jakob Disease – A Rare Case Report

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I. Introduction

Prion diseases are neurodegenerative diseases that have incubation period. Five prion diseases are recognized they are kuru, Creutzfeldt-Jakob disease (CJD), variant CJD, Gerstmann-straussler Scheinker syndrome (GSS) and fatal familial insomnia[1]. Among all these disease, CJD accounts for 90% of all prion disease. One case of CJD occurs among 1,000,000 populations per year. It is such a rare presentation. Dementia and myoclonus are the most common presenting condition of CJD. This is a rare case report of a patient who had a rare clinical presentation, and finally it was diagnosed to be a case of CJD.

II. Case Report

40 years old male patient presented to the Emergency department with complaints of weakness of left side of body two and half months back which was diagnosed and treated a s CVA from some private hospital. Now patient presented with complaints of right side of body along with progressive deterioration of speech and orientation. Patient also had myoclonus. He is not a known diabetic, hypertensive, asthmatic. He was on any long term medication. When patient presented to us, he was taking treatment for CVA. On examination patient was disoriented, responding to pain. Tone all four limbs spasticity present. Power could not be assessed properly. Deep tendon reflex exaggerated in all the four limbs. Superficial reflexes were present. Sensory system, cerebellar signs and gait couldn't be assessed. Spine was normal. Other system examination was within normal limits. The patient was planned for routine blood and urine examination. In complete hemogram there was mild leucocytosis. Other routine tests were within normal limits.

Lumbar puncture and CSF study was found to be within normal limits, protein value in CSF was within normal limits. CSF study for abnormal proteins could not be done. As the condition of the patient was slowly deteriorating day by day, the patient was planned for MRI scan. MRI scan was showing features of increased signal intensity in putamen and caudate nucleus (Figure 1). EEG done in this patient was showing periodic sharp wave complex (Figure 2). Hence from these findings we came to a conclusion that there are chances suggestive of CJD in this patient.

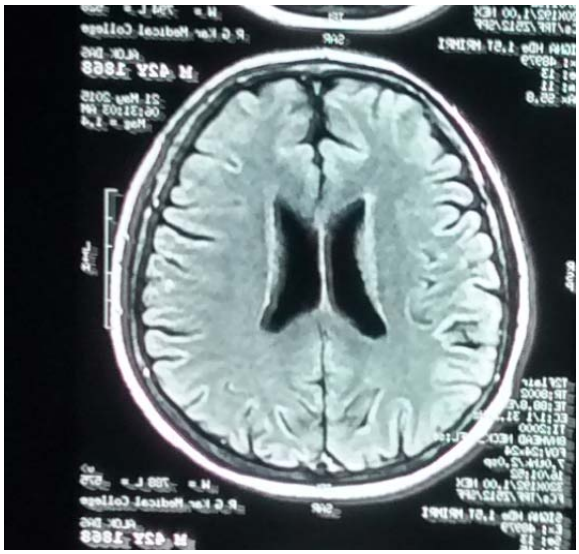

Figure 1 : Increased signal intensity in putamen and caudate nucleus

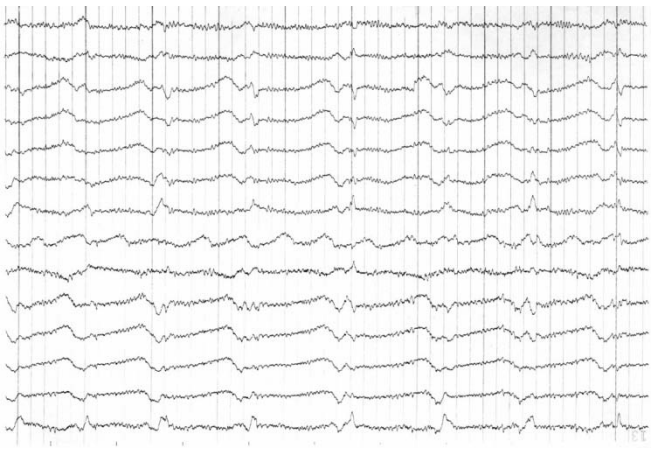

Figure 2 : EEG showing periodic sharp wave complex

III. Discussion

Rapidly progressive mental deterioration and myoclonus are the classical presentation for CJD. But in this patient, the presentation was totally confusing. The patient was having left sided weakness as the initial presentation. This was totally misleading as the earlier physician started suspecting it to be a CVA. He also started treating the patient for CVA. But slowly the general condition of the patient was deteriorating day by day. The patient was then having features of myoclonus and mental deterioration later. This is said to be a different presentation of CJD.

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Mental deterioration may be manifest as dementia, behavioural abnormalities involving higher cortical functions. With progression of age dementia becomes dominant in most patients and can advance rapidly.

Myoclonus especially provoked by startle is present in more than 90% of the patients with CJD. So whenever a patient presents with complaints of dementia and myoclonus one should have a strong suspicion of CJD. There are various subtypes of CJD described[2].

1. MM1 and MV1 variant accounts for 70% of CJD
2. VV2 ataxic variant accounts for 15% or less
3. MV2 accounts for 9 %
4. MM2 thalamic variant.

The various other differential diagnosis that we have to keep in our mind when we are suspecting CJD are parkinsonism, autoimmune disorders including Para neoplastic syndromes, sarcoidosis, infections including viral and post viral encephalitis, malignancy, toxic and metabolic encephalopathy, cerebro vascular disease, psychiatric disease. Keeping all the differential diagnosis in mind, the patient all routine and necessary investigations was done. The gold standard investigation for the diagnosis of CJD remains to be brain biopsy. Also MRI scan and EEG waves can aid in the diagnosis. All investigations clearly ruled out other possible differential diagnosis of CJD.

The MRI findings in a case with CJD will usually have abnormally increased T2 and flair signal intensity in the Globus palladius, thalamus, cerebral and cerebellar cortex[3]. EEG will provide supportive evidence but cannot be taken as confirmatory test for CJD. In this case report MRI findings suggestive of CJD was elicited also, EEG showed classical spike wave pattern. Though it is said that detection of CSF protein 14-3-3 considered as adjunctive rather than absolute test for CJD. Hence from the literature available, MRI and EEG alone was made as a tool for diagnosing CJD in this patient, as brain biopsy and CSF protein detection was very expensive could not be done.

The diagnostic criteria for CJD as described by centres of disease control and prevention outline the following criteria for probable CJD[4]

1. Progressive dementia
2. At least two of the following four features. Myoclonus, visual or cerebellar disturbance, pyramidal/extrapyramidal dysfunction, akinetic mutism
3. Atypical EEG/ CSF assay for protein 14-3-3/ MRI abnormalities in the caudate nucleus and putamen
4. Routine investigations should not suggest any other diagnosis.

In our case, all the conditions as pointed out above were present; hence the diagnosis of CJD was made.

After coming to the diagnosis of CJD, the patient was given supportive therapy. As there is no effective treatment for the prion disease, death usually occurs[5]. In this patient the condition was very rapidly progressive. The patient after the onset of initial symptom expired in a time period of two months.

IV. Conclusion

CJD is a very rare disease mean age of onset is fifth to sixth decade. Rapidly progressive mental deterioration with dementia is said to be the classical presentation of CJD. Though brain biopsy is the gold standard investigation of choice, as there is no proper treatment modality available for CJD, CDC has formulated the criteria for diagnosing CJD. When we are suspecting a diagnosis of CJD it is very important to keep in mind other common differential diagnosis. There is no treatment for CJD; death is the ultimate result for this disease.

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