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A Case Report on Rapidly Progressive Sporadic Creutzfeldt-Jakob Disease

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Abstract | **Case Report**

Creutzfeldt-Jakob disease (CJD) is a rare CNS disorder which is notoriously known to be rapidly progressive. There are various types of CJD:
(a) Iatrogenic CJD: Caused due to duramater graft or human growth hormone from infected individuals
(b) Familial CJD: caused due to genetic mutations in PRNP gene located on chromosome 20
(c) Variant CJD: caused due to consumption of beef from cattle infected with Bovine Spongiform Encephalopathy
(d) Sporadic CJD: most commonly form type of CJD where cause remains unknown (85%)
The disease usually presents itself with rapidly progressive dementia, myoclonus, cerebellar ataxia and visual disturbances. This presentation is very similar to autoimmune encephalitis and steroid induced encephalitis and requires specific diagnostic tests. The gold standard for diagnosis is through brain biopsy which though helpful in autopsies is not a feasible option in living patients. An MRI showing cortical ribboning and EEG with periodic sharp wave complexes are usually seen. This is backed with Real Time Quaking Induced Conversion (RT-QUIC) assay which is specific to protease resistant prions scrapie isoform (Prpsc). The following case report highlights a classical presentation of sporadic CJD which was diagnosed in the absence of newer diagnostic methods like RT-QUIC and managed with symptomatic treatment.

Keywords: Sporadic Creutzfeldt-Jakob Disease, Prion Diseases, Rapidly Progressive Dementia, Cortical Ribboning, Myoclonus, RT-QUIC.

INTRODUCTION

CJD incidence is about 1/million/year with majority of the cases being reported from countries like China, USA and Japan. According to the National CJD Registry maintained by NIMHANS, Bengaluru only about 120 cases over last 40 years have been confirmed and recorded. There are several case reports and case series highlighting cases over south India and North India; however, one cannot deny that there has been massive under reporting.

With no definite cure for the disease, early diagnosis and symptomatic management is the main course of action. This can happen only when physicians are well versed in the classical presentation of the disease while also being able to confirm their diagnosis in the absence of gold standard testing facilities.

CASE REPORT

A 52-year-old male patient was admitted to the hospital with chief complaints of blurring of vision (x15 days), left upper limb weakness (x10 days) and decreased responsiveness (x3 days).

The blurring of vision was described bilateral, gradual and progressive. The weakness that had started in left upper limb in the form of not being able to hold objects had spread progressively to involve all the limbs. Finally, decreased responsiveness presented itself as not being able to recognize even the attenders and follow basic instructions.

The GCS score when the patient was brought to the hospital was E1V1M1. Based on these an MRI-Brain and an EEG were ordered. The EEG showed periodic sharp wave complexes at 1-2HZ with a slow background. MRI-BRAIN revealed symmetrical altered signal intensity appearing hyperintense on T2/FLAIR images in both basal ganglia and fronto-parietal cortex showing diffusion restriction. Since, RT-QUIC was unavailable, final diagnosis was obtained by ruling out all the possible differentials. A negative autoimmune encephalitis panel established the final diagnosis as CJD.

History further revealed that there were no similar complaints in the past in any of the family members. There was also no history of any invasive procedures and the diet followed was strictly vegetarian. This ruled out the various other types of CJD and the final diagnosis was modified to Sporadic CJD.

The patient was given symptomatic treatment and stabilized. However, the patient was discharged against medical advice by the family members.

DISCUSSION

This case highlights the typical presentation of a CJD case that starts with cerebellar ataxia, blurred vision and has progressive dementia. It is often associated with myoclonus and limb weakness.

In a country like India, with a population of over a billion, an average of at least a 1000 cases should be recorded as per the incidence rate on CJD across the world. The massive under reporting can be attributed to the lack of advanced diagnostic facilities and the rapid progression of the disease which gives very less time for diagnosing it.

The above case report shows an alternative solution to the problem of lack of gold standard diagnostic facilities to arrive at a final diagnosis which proves to be equally effective. It also tries to bridge the gap of under reporting to the small extent possible.

CONCLUSION

In resource-limited settings, where access to gold-standard tests is often restricted, heightened clinical suspicion and familiarity with classical presentations become essential for early identification. Given the uniformly fatal nature of the disease and lack of definitive treatment, timely diagnosis facilitates appropriate counselling, symptomatic management, and infection control measures. Furthermore, improving awareness and reporting mechanisms is crucial to address the significant underreporting of sporadic CJD in India and to better understand its true epidemiological burden.

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