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An atypical presentation of Creutzfeldt - Jacob disease

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Abstract

Creutzfeldt-Jacob disease (CJD) is a rare prion transmitted disease which causes neurodegeneration and ultimately results in death within a very short span of time. In view of the very low prevalence of 1-1.6 per 1000000 population the diagnosis of the CJD is the challenge for the clinicians. The varied presentation makes it more vulnerable for misdiagnosis. We are reporting a case CJD due to its atypical presentation as it has presented with catatonia. The patient had visual disturbance to start with followed by depression, difficulty in walking and ultimately ended up to catatonia. The cases of CJD who presented with psychiatric manifestations are available in literature but presentation as catatonia is very rare in the literature.

Keywords: CJD, Prion, catatonia, psychiatric manifestation, depression

Introduction

Creutzfeldt-Jacob disease (CJD) is a rare prion transmitted disease which causes neurodegeneration and ultimately results in death within a very short span of time[1]. It usually affects the individuals in fifth or sixth decade of life. In view of the very low prevalence of 1-1.6 per 1000000 populations the diagnosis of the CJD is the challenge for the clinicians. The varied presentation makes it more vulnerable for misdiagnosis[2]. CJD is a rapidly progressive neurodegenerative disorder presenting with neurological, psychiatric or cognitive symptoms. It can be classified into four categories: sporadic, familial, iatrogenic and variant. Sporadic CJD (sCJD), occur worldwide symptoms. Sporadic CJD is the most common form and represents about 85% of all CJD cases[3]. Such a rare neurodegenerative disease and variable presentation makes it worth reported for the benefit of the scientific community. Consent was taken from the patient and their family members for its publication.

Case report

Mr. D, a 60 year old married male from urban background, graduate, a travelling salesman brought to the Department of Geriatric Mental Health, King George's Medical University, Lucknow with complaints of minimal oral intake, stiffness of whole body, maintaining a stooped posture, slurred and incomprehensible speech or most of the time no speech for last 2 weeks. He was admitted and provisional diagnosis of catatonia (cause?) was made. On further evaluation he had visual disturbances (could see but couldn't read

letters) and anxiety for last four months and complaints of double vision and fearfulness for last three months. He had forgetfulness, difficulty finding words, visual hallucinations, sadness, crying spells, involuntary movements of hands, difficulty in walking and stiffness in body for last two and half months. One month back he developed urinary incontinence and disorientation to time and place. For last two weeks he had incomprehensible speech no response on verbal commands maintaining unusual posture and would respond to painful stimuli only. The onset of illness was subacute with progressive course and no known precipitating factor. The total duration of illness was four months.

Patient started complaining of visual disturbances four months back. He was not able to read certain things though he could see them and was unable to use computer to write his accounts. His vision continued to deteriorate in spite of consultation and treatment from ophthalmologist. He had been anxious. Gradually, he developed diplopia and consequent increased severity of his anxiety. He became very apprehensive about his death. Over the next two-three weeks he became more apprehensive, sad and hopeless. He had crying spells and started saying that he would not survive and the family should not spend money on him. He became forgetful and also started having difficulty with language and communication. He had difficulty with balance while walking and became stooped & rigid. He developed involuntary movements in upper limbs which were writhing as well as jerky. He started experiencing visual hallucination. For last one month there was significant deterioration in his symptoms, also became incontinent for urine and stools became confused and couldn't tell time or recognize place, although continued to recognize his family members over the last two weeks before admission his speech became incomprehensible. He stopped responding totally to verbal commands. He used to stare at one place & respond to painful stimuli only. He had minimal oral intake.

Patient never had any psychiatric illness in the past. He never underwent major surgical procedure or received any transplants or transfusion. No family history of any psychiatric, neurological or metabolic disorders. He was a graduate, working for two decades as a Sales Officer. Job involved travelling all over India. He was not habitual but a social drinker, habitual meat eater. He married at the age of twenty two and had a stable marital life. He had stable, well-adjusted, social, fun-loving pre-morbid personality.

On examination his Vitals were within normal Limits. Patient was not responding to verbal commands and localizing pain. He had fixed gaze with little scanning of the environment with decreased blink rate. Bilateral pupils were equal in size and had sluggish reaction to light. He had choreo-athetoid movements in upper limbs with occasional myoclonic jerks. Plantar reflex was flexor; deep reflexes were 3+ bilaterally. There were catatonic symptoms in form of rigidity in all limbs, waxy flexibility and negativism. He was lying in bed, poorly communicative, not responding to verbal commands, only responding to painful stimuli and was not able to indicate about his bowel/bladder.

Investigations and management

IV lorazepam was given to manage catatonia without any improvement. I.V. fluids with MVI were given to improve hydration and nutrition. All routine hematological parameters (including Complete Blood Count, Blood Sugar, Renal Function Test, Liver Function Test, Electrolytes, Serum B12, Folate & Thyroid Function tests) were within normal limits except serum protein which was 5.6mg/dl. VDRL was negative, HIV was non- reactive and CSF for viral markers was also negative. The patient continued to deteriorate in the ward. He

developed *startle myoclonus* on 3rd day onward. He also had multiple seizures in the ward 3rd day onward (There was no history of seizures prior to admission). EEG test was performed which revealed periodic sharp waves. MRI scan was done which shows cortical ribbon sign.

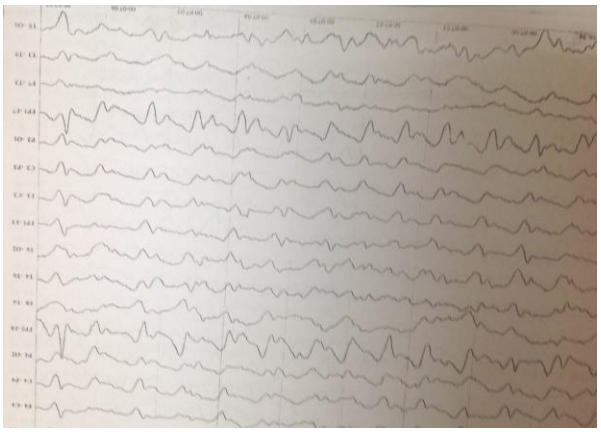

Fig. : 1 EEG showing periodic triphasic sharp waves.

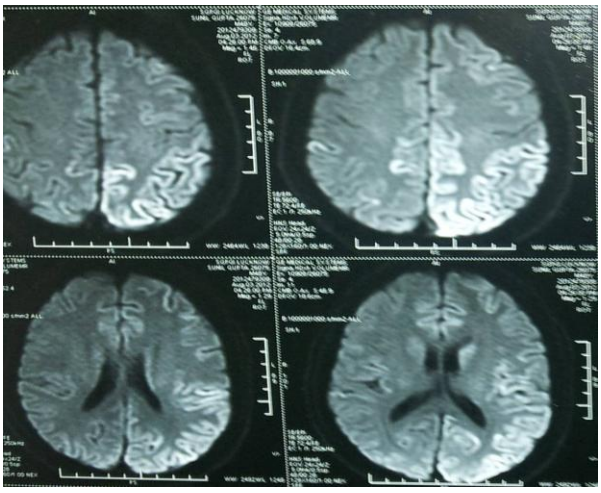

Fig. : 2 Diffusion-Weighted Images showing cortical ribbon sign and hyperintense caudate nucleus.

On the basis of clinical presentation, EEG and MRI findings, diagnosis of Dementia in Creutzfeldt - Jakob disease was made.

Discussion

Catatonia is a state of apparent unresponsiveness to external stimuli in a person who is apparently awake. Catatonia is not a diagnosis. Rather, catatonia is a descriptive term for a presentation observed in a wide variety of disorders, medical or psychiatric illness. Most common psychiatric causes of catatonia include depression, mania, schizophrenia, dissociative disorder and neurolept malignant syndrome. Medical causes of catatonia comprise of brain tumors, stroke, encephalitis, parkinson's disease, metabolic disease, renal disease and toxins. The case reported is a case of creutzfeldt-Jacob disease which can be considered to be a form of encephalopathy caused due to prion. It is one of Human Transmissible Spongiform Encephalopathies (TSEs). Hence as other encephalopathies it may also cause catatonia.

TSEs are neurodegenerative diseases, rapidly progressive, always fatal affect humans and animals. These have long incubation periods. Brain, spinal cord, and adjacent tissues are considered infectious. Patients usually present with dementia, visual problems, or cerebellar dysfunction[4]. Subsequent neurologic signs include myoclonus, tremors, and rigidity. Neurologic signs tend to deteriorate very rapidly ultimately result into akinetic mutism. Rigidity, myoclonus, and characteristic periodic complexes on EEG, favors diagnosis of CJD[5]. The present case had visual disturbance to start with followed by psychiatric symptoms in the form anxiety and depression. He had difficulty in walking suggestive of ataxia, rigidity and incomprehensible speech. He had periodic triphasic sharp waves in EEG and ribbon sign in MRI brain favoring the diagnosis of CJD. The disease progressed very rapidly within four months and reached to unresponsive state. CJD has no cure available till today and patient has to die invariably. Such a rapid progression is hallmark of CJD. The catching point in the present case is if a patient seeks consultation with a psychiatrist having sensory especially visual impairment and neurological

symptoms along with psychiatric manifestations, psychiatrists must be vigilant enough to go for initial investigations such as EEG and MRI to rule out diseases such as CJD.

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