ORIGINAL RESEARCH PAPER
Volume - 11 | Issue - 06 | June - 2022 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A RARE CASE OF CREUTZFELDT-JACOB DISEASE PRESENTING AS PSYCHOSIS

General Medicine

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ABSTRACT

Creutzfeldt-Jacob Disease is a rare neurodegenerative, prion disease characterised by rapidly progressive dementia with incidence one per million per year and mortality 100%. It is classified as Sporadic, genetic or familial ,Iatrogenic & variant. Most common clinical manifestations are rapidly progressive dementia, visual disturbances, myoclonus, gait disorder and behavioural disturbances. Disease is diagnosed with elevated CSF 14-3-3 protein,Tau and specific MRI abnormalities. We encountered a patient with CJD who had presented with only Psychosis. Therefore we describe this as unusual case of CJD & the consecutive MRI Brain findings.

KEYWORDS

Creutzfeldt-Jacob disease, Psychosis, CSF 14-3-3, Periodic sharp wave complexes

INTRODUCTION

Creutzfeldt-Jacob disease is most common prion disease. It is subclassified as Sporadic (sCJD),Genetic or Familial (gCJD),iatrogenic (iCJD) and variant (vCJD). sCJD accounts for 85-90% of all cases. Most patients develop disease between 50 to 80 years. Main known genetic risk factor is homozygosity at codon 129 on the PRNP gene. gCJD accounts for 10-15% of cases of CJD. It presents earlier (30-50 years). iCJD is reported from variety of transplants of nervous system-containing tissues including corneal grafts, reuse of contaminated intracerebral electrodes or neurosurgical equipments and human pituitary derived growth hormone. vCJD form was reported with food consumption from animal species to humans.

Rapidly progressive dementia,Focal neurological deficits and myoclonus are classical manifestations of CJD. The earliest symptoms may be vague and constitutional (insomnia,anorexia,fatigue) or psychiatric (depression,anxiety). Cognitive impairment (memory, concentration, aphasia) focal neurological deficits (hemianopia,focal weakness, ataxia) and psychiatric abnormalities (hallucinations and delusions) develop later. Myoclonus is present in late stage of disease. The neurological status later deteriorate to akinetic-mutism and then death within months of onset of symptoms. Other forms of CJD include Heidenhain Variant has primary visual hallucinations due to significant involvement of occipital cortex. Prominent cerebellar involvement is seen in Brownell-Oppenheimer Variant.

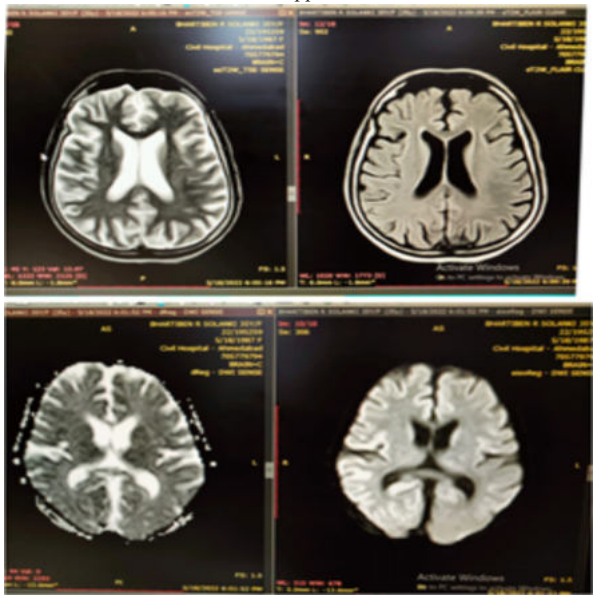

Figure 1: MRI Brain Of CJD Patient

Sources: Radiology department of B.J.M.C

CSF cell count and glucose level are typically normal, where protein level may be mildly elevated. Elevated levels of CSF 14-3-3 protein, Neuron specific enolase, S100 protein and Tau protein may support diagnosis of CJD in proper clinical settings. The real time quaking induced conversion (RT-QuIC) assay has improved diagnostic accuracy of CSF testing. Presence of periodic sharp wave complexes in Electroencephalography strongly supports diagnosis of sCJD. MRI Brain shows increased signals on DWI sequence especially in cortical ribbon, the basal ganglia & less commonly in thalamus. Neuropathological examination reveals typical spongiform changes.

CASE REPORT

A 35 years old female was brought to medical OPD with history of behavioural abnormalities, occasional irrelevant talk initially and later development of fluctuating orientation, poor self care and defecation & urination in the clothes for last 3 months for which patient was admitted in psychiatric hospital where Antipsychotics (Haloperidol, Risperidone & Olanzapine) were given but symptoms don't get relieved. After which patient was given 3 cycles of Electroconvulsive therapy but symptoms gradually worsened. The personal and family psychiatric history were unremarkable. There was no history of any toxin exposure or any drugs intake.There was a recent decline in personal & social functioning. Patient was vitally stable. At neurological assesment, the patient was conscious but confused, not oriented towards time, place and person. Patient had impaired speech in form of progressive mutism, difficulty in walking and lead pipe rigidity in all four extremities. Deep tendon reflexes were grade 2 in all limbs and planter responses were flexor bilaterally. She had disorganised thinking and judgment and sleeping tendency. According to the clinical picture, negative psychopathological history, rapid progression of memory impairment and worsening of psychiatric symptoms, an organic cause of this condition was hypothesised. Laboratory tests including CBC, RFT, LFT, Electrolytes, Urine routine & microscopy, Thyroid function tests, Random blood sugar, B12 level, ABGA were unremarkable. Blood culture, HIV, RPR, special tests for autoimmune encephalitis, vasculitides and malignancy were negative. ECG,Chest X-Ray,Ultrasonography of Abdomen & Pelvis were normal. Lumbar puncture was done and CSF routine & microscopy study revealed normal cell counts (4/hpf) and sugar (68 mg/dL). CSF protein was slightly elevated (72 mg/dL). MRI Brain was done which showed Gyral diffusion restriction on DWI with subtle hyperintensity in T2/FLAIR sequence were noted in bilateral cerebral hemisphere and bilateral corpus striatum suggestive of Creutzfeldt-Jacob disease. EEG showed periodic, synchronous , biphasic sharp wave complexes superimposed on a slow background rhythm. CSF Tau & CSF 14-3-3 were sent & reports showed elevation. Our patient was diagnosed as Creutzfeldt-Jacob disease by clinical presentation of rapidly progressive dementia, EEG, CSF analysis & MRI Brain. Patient was treated with supportive care.

CONCLUSIONS

In clinical practice,CJD should not be neglected as a differential diagnosis in adults and elderly with negative psychiatric history for recent onset and rapidly progressive psychosis. MRI Brain, CSF study and EEG should be done for patients who presents with the first episode of progressive psychosis of an uncertain origin, and the possibility of CJD should be considered.

DECLARATION OF PATIENT CONSENT

The authors certify that they have obtained all appropriate patient consent forms. In the form patient has given her consent for her images and other clinical information to be reported in journal. The patient understand that her name and initials will not be published and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

Financial Support/ Sponsorship: NIL

Conflict Of Interests

There are no conflict of interests.

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