CASE REPORT

Creutzfeldt- Jackob disease: A Case Report with MRI (DWI) Findings

Hari Om Joshi[1], Tong Dan[2], Ting Tang Yuan[3], Ajit Thapa[4], Jyotsna Shakya[5]

[1] Consultant Radiologist, Department of Radiology, Bhaktapur Hospital, Bhaktapur, Nepal
[2] Department of Radiology, The First Hospital of Jilin University, Changchun, Jilin, China
[3] Department of Radiology, The First Hospital of Jilin University, Changchun, Jilin, China
[4] Department of Radiology, TUTH Hospital, Kathmandu, Nepal.
[5] Department of Laboratory Medicine, Manmohan Institute of Health sciences, Kathmandu, Nepal

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ABSTRACT

Transmissible Spongiform Encephalopathy is sub-acute transmissible neurodegenerative disease which affects both human and animals. Among those that affect humans, Creutzfeldt-Jakob disease is the most common. This disease has a genetic basis and characteristic of an infectious disease. Magnetic Resonance Imaging is an important tool in diagnosis of Creutzfeldt-Jakob disease and can assist especially in the early stages of the disease, when the clinical features might not be typical and Electroencephalogram shows normal or nonspecific abnormalities. Newer Magnetic Resonance Imaging techniques such as Diffusion Weighted Imaging have shown to be more sensitive in detecting this disease at its early stage. We report a single case of probable Creutzfeldt-Jakob disease and its peculiar findings on brain magnetic resonance imaging.

Key words: *Creutzfeldt-Jakob disease; Transmissible Spongiform Encephalopathy*

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Correspondence: Dr. Hari Om Joshi, Department of Radiology, Bhaktaur Hospital, Bhaktapur, Nepal, E mail: dr.hariomjoshi@gmail.com

INTRODUCTION

Transmissible Spongiform Encephalopathy (TSE) is sub-acute transmissible neurodegenerative disease which affects both human and animals. Among those that affect humans, Creutzfeldt-Jakob disease (CJD) is the most common. The CJD manifests itself in the presence of an abnormal isoform (PrPSc) of a protein present naturally (PrPC) in our body. This isoform is the precursor of a protein of lower weight protein called prion or Prp, found in infected brains. In familial cases the genetic factors are thought to be responsible where as in sporadic cases no mutation has been described and evidence shows the coexistence of a prion environment favoring the change in confirmation of the normal protein PrPC to its isoform.[1]

Here we report a rare case of probable CJD and its peculiar findings on brain magnetic resonance imaging.

CASE REPORT

A 74 years' gentleman, a local resident of northern part of China was admitted with history of intermittent confusion, irrelevant talks and obsessive compulsive disorder for six months. After admission, he developed left sided partial seizure with dysphasia progressing to aphasia. Myoclonic jerking movements and choreoathetoic movements were also noted. The general physical examination was normal. With periods of attack he became confused and started talking irrelevantly.

The laboratory tests, carotid doppler ultrasound, echocardiogram, cerebral arteriography, CSF examination and CT head were normal. He underwent electroencephalogram (EEG) which showed seizure like activity in the occipital area. Magnetic resonance imaging (MRI) showed hyperintense signal in left occipital and frontoparietal cortex in Diffusion weighted image (DWI) and Fluid Attenuated Inversion Recovery (FLAIR) (Fig 1). However, T1WI and T2WI images in our case were without any signal abnormality. He also underwent lumbar puncture with CSF protein analysis of 14-3-3 protein and neuron-specific enolase. The concentration of the enzyme neuron-specific enolase was high, 14-3-3 protein in CSF was also positive. During his stay in hospital, two EEG showed triphasic sharp waves, periodic, sometimes predominant in posterior areas were consistent with CJD. With these findings, it was diagnosed as a case of probable sporadic CJD according to MRI- CJD consortium criteria (Table 1). However, brain biopsy was not performed and the patient is alive till date.

Table 1. MRI-CJD Consortium Criteria of sporadic CJD [2]

MRI-CJD Consortium criteria for diagnosis of sporadic CJD
1. Clinical signs
Dementia
Cerebellar or visual signs
Pyramidal or extrapyramidal signs

Akinetic mutism

2. Tests

Periodic sharp wave complexes (PSWC) in EEG

14-3-3 detection in CSF (in patients with a disease duration less than 2 years)

High signal abnormalities in caudate nucleus and putamen or at least two cortical regions

(temporal-parietal-occipital) either in DWI or FLAIR.

Diagnostic Criteria for Sporadic CJD

Probable CJD: Two out of 1 and at least one out of 2

Possible CJD: Two out of 1 and duration less than 2 years

Definite: Neuropathological examination or detection of Scrapie prion protein (PRP^{sc}) by

Western Blot analysis of brain biopsy or autopsy specimen

DISCUSSION

Creutzfeldt-Jakob disease most commonly begins between 55 and 70 years of age. The average age in most cases is 60-69 years. In about one third of cases the prodromal signs are gait ataxia, aphasia or visual loss.[1] Disease progression is gradual and dementia and myoclonus is often characteristically present. Visual disturbances and cerebellar signs such as incoordination and difficulty walking are less common. In our case, the patient had prodromal signs mentioned above, most notably with gait ataxia and aphasia. EEG may show typical periodical sharp wave complexes. When the "bursts" are stereotyped and periodic interval is 1 to 2 seconds, the diagnosis of CJD is more probable.[3] This case presented the aforementioned EEG changes during a phase of disease.

Recently, three proteins have been detected by immunoassay or Western blot-CSF in patients with CJD: neuron-specific enolase (NSE), S-100, protein 14-3-3. The presence of CSF markers with typical clinical history from a patient strongly suggests the diagnosis of CJD. The 14-3-3 protein has good sensitivity and specificity.[4] MRI is an important tool in CJD diagnosis and can assist especially in the early stages of the disease, when the clinical features might not be typical and EEG shows normal or nonspecific abnormalities. Although MRI can be normal in 20%, signal changes occur in 80% of patients in the early stage of disease. MRI findings may be bilateral or

unilateral and may be symmetric or asymmetric. MRI may show increase signal in T2WI in the basal ganglia, thalamus, occipital cortex or white matter.[5] Newer MRI technique such as DWI has shown to be more sensitive in detecting this disease at its early stage.[3] The DWI sequence has also been shown superior to FLAIR in the detection of cortical lesions, which was indeed observed in our case.[6] Our patient has left cortical hyper intensities at area of frontal and occipital lobe which can be detected in FLAIR images but were more prominent in DWI images.

CONCLUSION

Diffusion-weighted magnetic resonance imaging provides a highly sensitive method of identifying areas of involvement in CJD. DWI shows positive findings even when other MRI sequences lack the signal abnormalities. This observation may facilitate the earlier diagnosis of this disease.

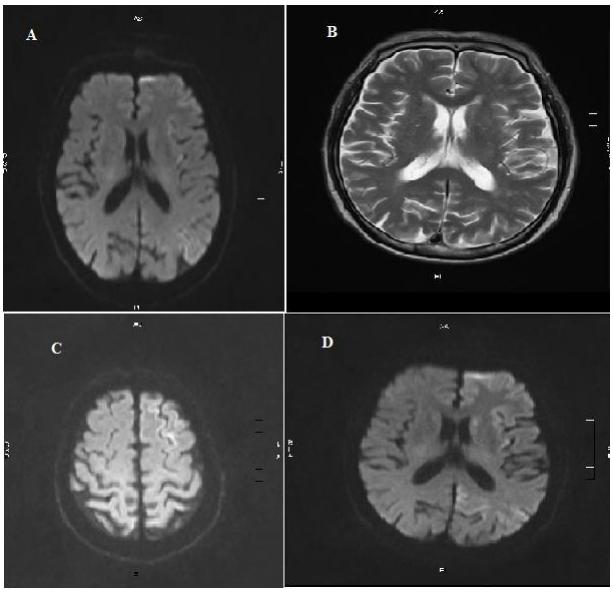

Figure 1. A. FLAIR B.T2WI, C and D. DWI sequences of MRI showing hyperintense signal in left occipital and frontoparietal cortex in DWI and FLAIR sequences.

CONFLICT OF INTEREST

None

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