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DIAGNOSTIC INSIGHTS IN RAPIDLY PROGRESSIVE CREUTZFELDT-JAKOB DISEASE: A CASE SERIES EMPHASIZING THE ROLE OF MRI

Radio-Diagnosis

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ABSTRACT

Creutzfeldt - Jakob disease (CJD) is a rare, rapidly progressive neurodegenerative disorder caused by prion proteins. Its clinical presentation often includes rapidly worsening cognitive decline, motor dysfunction, and visual disturbances, which can easily be mistaken for other neurological conditions. Early and accurate diagnosis is essential for management and prognosis. In this context, radiological imaging, particularly Magnetic Resonance Imaging (MRI), plays a crucial role in the diagnostic process. MRI findings in CJD typically include hyper intensities in the basal ganglia, thalamus, and cortical regions with diffusion-weighted imaging (DWI) features as pivotal in the diagnostic workup of sCJD. These findings, when interpreted alongside clinical symptoms and supportive testes (EEG, and CSF Analysis) help in diagnosing CJD. This abstract reviews the importance of radiology in the early diagnosis of CJD, highlighting the distinctive MRI patterns that can aid in clinical decision-making, contributing to better patient outcomes and more effective management.

KEYWORDS

CASE 1

73year old female patient came with history of forgetfulness for 15 days. MRI brain was done which shows – T2W/FLAIR hyper intensities involving bilateral caudate nucleus, putamen and thalami and corresponding areas shows diffusion restriction. Gyriform diffusion restriction involving right high parietal cortex noted.

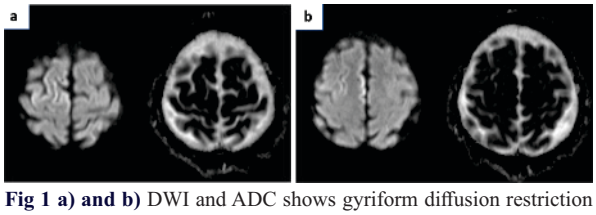

Fig 1 a) and b) DWI and ADC shows gyriform diffusion restriction involving right high parietal lobe (Cortical Ribbon sign) and right superior frontal gyrus.

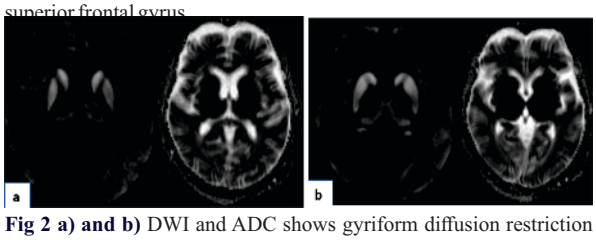

Fig 2 a) and b) DWI and ADC shows gyriform diffusion restriction involving bilateral basal ganglia and bilateral thalamus.

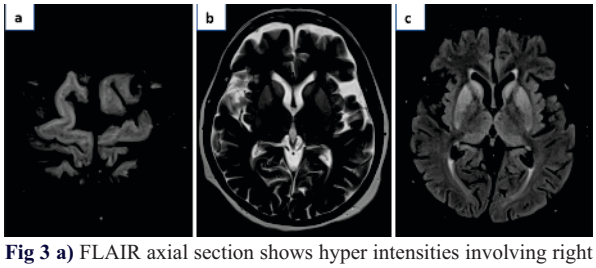

Fig 3 a) FLAIR axial section shows hyper intensities involving right superior frontal gyrus.
Fig 3 b) and c) T2W/FLAIR axial section shows hyper intensities involving bilateral basal ganglia giving typical Double Hockey Stick Sign.

Following Up The Patient,

Patient soon developed left lower limb weakness with left upper limb weakness. Numbness on the feet on and off. Involuntary jerking movements in left upper limb.

CSF analysis for 14.3.3 – 52,800; JE antibody – Negative
Follow up MRI was taken after 1 year.

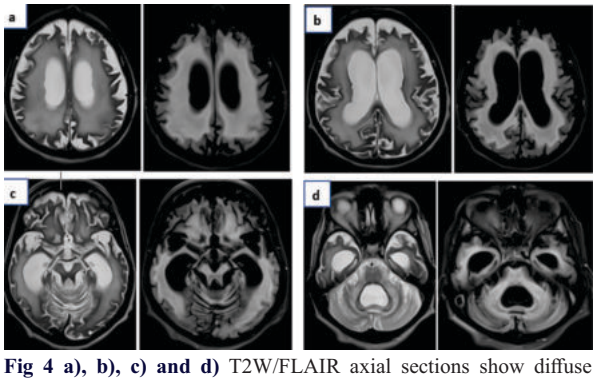

Fig 4 a), b), c) and d) T2W/FLAIR axial sections show diffuse cerebral and cerebellar atrophy with ex-vaco dilatation of ventricles. Mid-brain atrophy with prominent cerebral and cerebellar peduncles.

CASE 2

70-year-old female came with C/O increasing difficulty following commands, forgetfulness, recognizing familiar faces, and caring for herself.

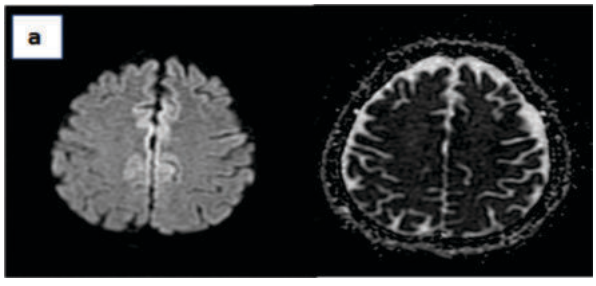

Fig 5 a) Faint diffusion restriction in the parasagittal frontal lobes.

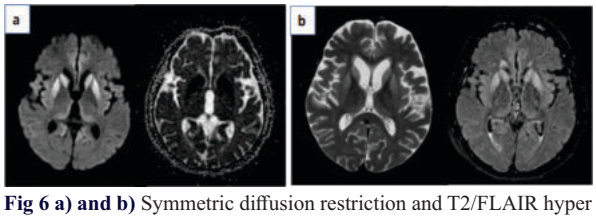

Fig 6 a) and b) Symmetric diffusion restriction and T2/FLAIR hyper

intensity in both caudate heads and bilateral putamen. Faint diffusion restriction and T2 hyper intensity in the dorsomedial thalami (hockey stick sign).

Laboratory results from CSF fluid showed a positive CSF RT-QuIC, elevated T-tau protein, and elevated 14-3-3 protein, consistent with CJD.

CASE 3

65year old male patient with history of forgetfulness and unsteady gait.

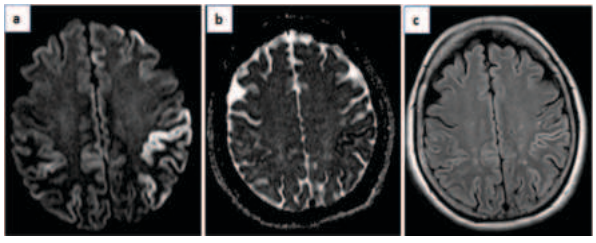

Fig 7 a), b) and c) Gyriform diffusion restriction involving left parietal lobe with corresponding areas shows FLAIR hyper intensities.

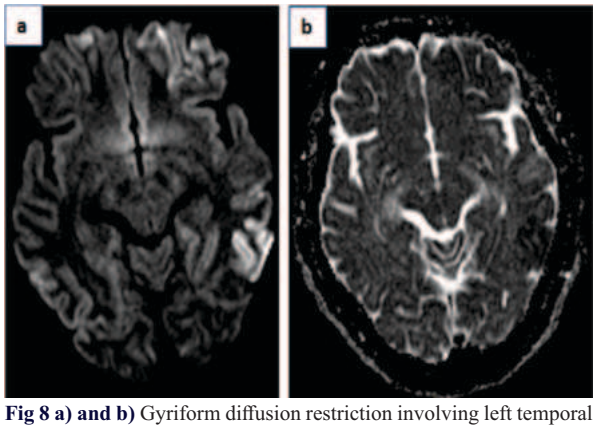

Fig 8 a) and b) Gyriform diffusion restriction involving left temporal lobe. Laboratory results shows elevated 14-3-3 protein consistent with CJD.

DISCUSSION

Creutzfeldt-Jakob Disease (CJD) is a rare, rapidly progressive neurodegenerative disorder characterized by a swift decline in cognitive function, motor skills, and, eventually, death, typically within months of diagnosis. CJD falls under the umbrella of prion diseases, which are distinct from other neurodegenerative diseases due to their unique pathology driven by prions—infectious agents composed of misfolded proteins rather than conventional pathogens like bacteria or viruses.

At the molecular level, CJD is caused by the alteration of a naturally occurring prion protein (PrPc) found on Chromosome 20, which transforms into an abnormally folded, misbehaving version known as the scrapie prion protein (PrPSc). This misfolded protein has the ability to induce conformational changes in other normal prion proteins, leading to a cascade of pathological events in the brain. Over time, these changes result in the characteristic spongiform encephalopathy, with the formation of vacuoles in the brain tissue, ultimately leading to the rapid deterioration of the affected areas.

CJD is most commonly seen in its sporadic form, which accounts for approximately 85% of all cases. Sporadic CJD typically affects individuals between the ages of 65 and 75, although it can occur at any age. The exact cause of the sporadic form remains unclear, but environmental factors or yet-to-be-identified mutations may play a role in triggering the disease.

In addition to sporadic cases, CJD can also occur in inherited and infectious forms. The inherited types, including Gerstmann-Straussler-Scheinker syndrome and familial CJD, make up about 10-15% of cases and are caused by genetic mutations that affect prion protein folding. The infectious form of CJD, though rare (<1% of cases), is most commonly associated with variant CJD (vCJD), which is linked to exposure to bovine spongiform encephalopathy (BSE), a prion disease in cattle. The discovery of vCJD highlighted the potential zoonotic transmission of prions, emphasizing the importance of public health measures to prevent transmission of prion diseases from animals to humans.

Criteria for diagnosing CJD –

1) Definitive diagnosis - Neuropathological confirmation through brain tissue examination.
2) Probable diagnosis – Includes at least two of the clinical features with a positive supportive test.

a) Clinical features - Myoclonus.
Visual or cerebellar signs.
Pyramidal/extrapyramidal signs
Akinetic mutism

b) Supportive test - EEG: Periodic sharp wave complexes.
CSF Analysis: Presence of 14-3-3 protein
MRI: Characteristic imaging findings brain.

Radiologically, CJD presents unique challenges due to its rapid progression and varied symptomatology. MRI plays an essential role in diagnosing CJD, especially in its early stages. The hallmark imaging findings, particularly on diffusion-weighted imaging (DWI), include bilateral basal ganglia hyperintensities, which are highly suggestive of the disease. Additionally, involvement of the thalamus and cortical regions may also be observed, further aiding in the differentiation of CJD from other neurodegenerative disorders such as Alzheimer's disease or frontotemporal dementia. These MRI features, combined with clinical symptoms and other diagnostic tests like cerebrospinal fluid (CSF) analysis for 14-3-3 protein and real-time quaking-induced conversion (RT-QuIC), are crucial for timely and accurate diagnosis, ultimately improving patient care and management.

In conclusion, CJD remains a complex and devastating disease with a challenging diagnostic process. Advances in radiology, particularly MRI, have significantly improved our ability to identify this disease at an earlier stage, thereby aiding in the differential diagnosis of rapidly progressive dementia. Despite ongoing research, however, much remains to be understood about the underlying mechanisms and potential therapeutic interventions for CJD and other prion diseases.

CONCLUSION

Creutzfeldt-Jakob disease should be suspected in any case in which areas of abnormal signal hyperintensity are depicted on diffusion-weighted images in the cerebral cortex and deep gray matter, especially in the caudate nucleus. In early stages of Creutzfeldt-Jakob disease, the areas of abnormal high signal intensity on diffusion-weighted images may be restricted to the cerebral cortex. It is crucial to consider these findings, in conjunction with the patient's clinical history, to suspect Creutzfeldt-Jakob disease as early as possible.

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