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Review Article

Case review: hearing and balance loss in Creutzfeldt-Jakob disease

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ABSTRACT

Creutzfeldt-Jakob disease (CJD) is a rare transmissible neurodegenerative disease caused by misfolded prion proteins leading to rapid mental deterioration and death. Misfolded prion proteins form insoluble aggregates that cause irreversible neurological damage. While typically presenting with symptoms such as cognitive decline and behavioral changes, atypical presentations of CJD include symptoms such as sensorineural hearing loss or balance loss. The goal of this study is to systematically characterize atypical presentations of CJD with hearing and balance loss following Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) guidelines. The PubMed database was utilized to analyze all case reports from the years 1990 to 2024 using the keywords "Creutzfeldt-Jakob disease", "hearing loss", and "balance". A total of 16 cases were retrieved, and 11 were included in the study. Among cases retrieved, we identified eight patients with hearing loss and three with balance loss. Of hearing loss patients, four (50%) were male and four (50%) were female. The average age was 63.75 years (SD=11.17 years). The most common symptoms that accompanied hearing loss were analyzed (N, %), and include gait disturbances (8, 100%), myoclonus (6, 75%), cognitive impairment (4, 50%), akinetic mutism (4, 50%), and vision disturbances (4, 50%). The tests utilized to diagnose CJD in patients with hearing loss included MRI (5, 63%), EEG (5, 63%), and 14-3-3 protein (4, 50%). These findings highlight the importance of recognizing hearing and balance loss as potential early symptoms of CJD to aid in earlier diagnosis and a better understanding of disease progression.

Keywords: Creutzfeldt-Jakob disease, Neurodegenerative disorders, Sensorineural hearing loss

INTRODUCTION

Creutzfeldt-Jakob disease (CJD) is a rare and invariably fatal neurodegenerative disorder classified under the spectrum of transmissible spongiform encephalopathies (TSEs).[1] CJD was first described in the 1920's by neurologists Hans Gerhard Creutzfeldt and Alfons Maria Jakob and is the most commonly reported prion disease.[2] The prion hypothesis, proposed by Stanley B. Prusiner in 1982, posits that misfolded prion protease-resistant proteins (PrPSc) propagate by inducing the misfolding of normal cellular prion proteins (PrPc), leading to protein accumulation throughout the brain and subsequent neurodegeneration.[1,3] Global CJD incidence is estimated at 1-2 cases per million people per year though most PrD surveillance programs are operational only in high- and upper-middle-income countries.[2,4] CJD can be further classified into different subtypes, with sporadic CJD (sCJD) constituting 85-90% of all cases, while genetic CJD (fCJD) makes up another 10-15% of cases. Acquired CJD, including iatrogenic CJD, Kuru, and variant of CJD (vCJD) forms of the disease are far less common.[2,5] Despite its low incidence, CJD's rapid progression and devastating clinical manifestations contribute to a 90% percent mortality rate within one year of onset with sCJD.[1]

CJD is typically detected late into disease progression with rapidly progressive dementia, myoclonus, pyramidal or extrapyramidal signs, visual or cerebellar disturbances, and akinetic mutism followed by death.[6] Diagnostic modalities for CJD involve a combination of these clinical features with MRI findings of hyperintensity in the caudate nucleus and putamen on diffusion-weighted imaging or fluid-attenuated inversion recovery (FLAIR), periodic sharp wave complexes on EEG, cerebral spinal fluid analysis for 14-3-3 and elevated tau proteins, prion protein (PRNP) genetic mutation testing, and spongiform changes with prion protein accumulation found in brain biopsy for a definitive diagnosis. Recent advancements in prion-specific protein aggregate assays like real-time quaking-induced conversion (RTQuIC) or other biomarkers such as Neurofilament light chain or Alpha-synuclein have shown promise in improving pre-mortem diagnosis of CJD.[7,8] Despite an expanding understanding of the disease and improved diagnostic strategies, current treatment modalities remain palliative.[9]

While previous literature has largely concentrated on classical neurological symptoms, neuropathological characteristics, and genetic underpinnings of the disease, several studies and cases have highlighted CJD presentation and spread within the setting of otolaryngology. Specifically, the role that the ENT surgeon might play as a vector for transmission of CJD.[10,11] Studies of early disease presentation have also established that dizziness, vertigo, and other cerebellar disturbances are some of the most common early presentations of CJD while documentation of auditory disturbances is extremely rare.[6,12,13] One study has posed that certain focal neurological deficits, including dizziness and vertigo, may be associated with a younger age of onset of disease and also with the subtype of disease.[14] Addressing this gap in the literature surrounding auditory disturbances is crucial for the use of otolaryngologists as early identification of these symptoms could lead to more timely and accurate diagnoses as well as avoidance of iatrogenic spread.

The primary aim of this study was to conduct a comprehensive review of case series to identify and characterize otolaryngological presentations of CJD, with a particular focus on hearing loss and balance disturbances. Secondary aim included evaluating the temporal relationship between these otolaryngological symptoms and the more classically recognized neurological signs. Our investigation posits that hearing loss and balance disturbances may precede the classical neurological symptoms of CJD, providing a critical window for earlier detection and diagnosis.

METHODS

Data sources and search strategy

This case review was performed in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) 2020 guidelines.[15] Between July 10, 2024 and July 12, 2024, C.S. searched the PubMed database for articles published between January 1990 and June 2024 to identify case reports on patients diagnosed with CJD who initially presented with a loss of hearing or balance. Studies were found using the keywords “Creutzfeldt-Jakob Disease” AND “hearing loss” OR “balance”. We included case reports published in a non-English language. References were searched and cross-matched and full-text articles were then reviewed.

Eligibility criteria and study selection

During the initial screening process, C.S. identified abstracts which met the inclusion criteria. Inclusion criteria consisted of all English and non-English language articles discussing CJD cases with either loss of hearing or balance as one of the signs and symptoms. Articles published in a non-English language were translated using an online tool prior to eligibility assessment. Studies that did not explicitly mention the loss of balance or hearing as a presenting symptom were excluded. Descriptive studies, editorials, meta-analyses and systematic and narrative reviews were excluded. Full-text articles were reviewed independently by three authors. Disagreements were resolved by consensus between all authors (C.S., D.H., B.H., K.B., B.G., A.O., S.S.B., and K.M.).

Data abstraction and analysis

Three independent reviewers (C.S., D.H., and B.H.) screened results, extracted and tabulated data. Any dispute was settled by consensus involving all authors (C.S., D.H., B.H., K.B., B.G., A.O., S.S.B., and K.M.). A data extraction sheet was created to document patient age, gender, presenting signs and symptoms, pertinent imaging and examinations, treatments, and outcome measures. Descriptive data analysis was performed using Microsoft Excel.

The initial search retrieved 16 case reports discussing either a loss of hearing or balance as a symptom in patients ultimately diagnosed with CJD. After screening full-text articles and applying the inclusion and exclusion criteria, we included 11 articles for analysis as shown in Figure 1. Of the articles excluded, one was a retrospective chart review, two were out of scope, and two did not state our symptoms of interest in the patient presentation.

Quality assessment and risk of bias

Studies were assessed for quality against the Joanna Briggs Institute (JBI) case reports critical appraisal tool.16 Further, all articles were reviewed and screened by three independent authors to eliminate potential bias (C.S., D.H., and B.H.).

Table 1: Key findings from case analysis.

Author, Year	Age (years)	Gender	Patient presentation	ENT specific symptoms	Other CJD symptoms	Other unrelated symptoms	Symptomatic treatment?	Outcome	Imaging/labs ordered+significance	Takeaway they made (look at discussion/conclusion)?	Family history?
Na[4]	76	Male	Patient presented with bilateral sudden hearing impairment and dizziness that had been going on for 10 days. 2 weeks later he had gait impairment, was disoriented, and had cognitive impairment. 2 months later was couldn't move anything in body (akinetic mutism).	Hearing loss and dizziness.	Gait impair-ment, disorientation, cognitive impair-ment, akinetic mutism.	Hypertension and diabetes.	Patient received steroid treatment for hearing loss but it was ineffective.	2 months after initial presentation total akinetic mutism.	Pure tone audiometry showed bilateral severe hearing impairment. Initial MRI at time of hearing loss was normal. Follow up MRI 2 weeks later showed cortical ribboning. RT-QuIC was positive and 14-3-3 detected.	Consider CJD when working up older patients with hearing loss.	Not reported.
Brito-Marques[2]	61	Male	Patient presented with progressive bilateral hearing loss and gait disturbance over 2 months. Upon examination he had deafness, cerebellar syndrome, myoclonus, and frontal signs. All tests came back normal except hearing. Patients condition worsened to akinetic mutism and cortical blindness. Patient was hospitalized and died 5 weeks from aspiration pneumonia and death. Pathology was positive for PrPSc protein.	Hearing loss.	Myoclonus, cerebellar syndrome, akinetic mutism.	Not reported.	Not reported.	Patient died after being hospitalized for 5 weeks from aspiration pneumonia and sepsis.	MRI showed bilateral striatum hyperintensity. EEG showed three phase periodic activity. CSF analysis of 14-3-3 was negative, tau was slightly elevated. Auditory evoked potential revealed severe bilateral dysfunction of the CN8. Upon autopsy, PrPSc protein was positive.	Keep CJD on diagnosis when working up patients with progressive encephalopathies and auditory symptoms.	Not reported.
Miyagawa[20]	43	Female	Patient presented with severe bilateral hearing loss and unusual behavior including loss of balance and frequent falling. Ataxia and deep tendon reflexes were diminished bilaterally in the lower extremities. Symptoms continued to progress and on day 14 of hospitalization following initial presentation she developed respiratory failure.	Hearing loss and loss of balance.	Not reported.	Not reported.	Not reported.	Respiratory failure.	Upon blood testing, C-reactive protein was slightly elevated. CSF analysis showed negative neuron specific enolase. Brain MRI showed no abnormalities. EEG showed a small amount of alpha waves in the posterior region. Audiometry could not be performed due to the patient's lack of cooperation. Abnormal prion was detected in CSF using RT-QuIC. Genetic testing was done and a diagnosis of familial CJD due to E200K mutation.	Cases of CJD can present with negative MRI- there were no signs of cortical or basal ganglia abnormalities in this case.	Family healthy.
Salazar[11]	67	Male	Patient presented with behavioral decline, unsteady gait, and bilateral hearing loss. 3 months prior he abruptly developed hearing loss with no tinnitus or vertigo. EEG showed generalized slowing without periodic complexes. Patient developed myoclonus and gait problems. MRI showed cortical ribboning and FLAIR hyperintensities in	Bilateral hearing loss.	Myoclonus, unsteady gait, behavioral decline (prosopagnosia that led to delusions and psychomotor agitation). Encephalopathy, mutism, and	Not reported.	Not reported.	Mutism and death 8 months after initial presentation.	No audiometry was ordered. Immunological, infectious, metabolic, and toxic tests all came back negative. Initial MRI was negative. EEG showed slowing without short wave complexes. Audiometry was not ordered due to patient state. No retinal vasculitis was found. A complete paraneoplastic panel was conducted and all came back negative. A	This case illustrated the variability of presentation of CJD. Consider CJD when patients present with unclear origin of bilateral hearing loss and loss of cognitive function.	Not reported.

Continued.

Author, Year	Age (years)	Gender	Patient presentation	ENT specific symptoms	Other CJD symptoms	Other unrelated symptoms	Symptomatic treatment?	Outcome	Imaging/labs ordered+significance	Takeaway they made (look at discussion/conclusion)?	Family history?
			basal ganglia. 14-3-3 was positive supporting diagnosis of CJD. Patient died 8 mos later. No autopsy was obtained.		death 8 months after initial test.				second MRI demonstrated cortical ribboning. 14-3-3 came back positive.		
Rene[7]	53	Male	Patient presented with bilateral hearing loss and tinnitus in the left ear. Over the next months the hearing loss worsened and he developed stocking paresthesia and gait instability. Audiometry showed bilateral sensorineural hearing loss and nerve conduction studies showed mixed polyneuropathy. CT and MRI were normal. EEG showed non-specific changes. Patient died of respiratory infection 10 mos after onset of symptoms. Abnormal prions were found during autopsy consistent with CJD.	Bilateral sensorineural hearing loss.	Gait ataxia, myoclonus, mental status deteriorated.	Not reported.	Not reported.	Patient died 10 months after initial presentation from respiratory infection.	All cranial nerves were normal, audiometry showed bilateral senorienueral hearing loss. CT and MRI were both normal and EEG showed non-specific changes. 14-3-3 protein test was positive. EEG showed nonspecific changes. Autopsy found deposits of PrPSc in the cerebral and cerebellar cortices, as well as in the cerebellum. Genetic sequencing showed a heterozygous E200K mutation.	CJD can present with deafness due to involvement of brains tem auditory nuclei (found from autopsy). Sporadic CJD presenting with deafness follows a clinical course similar to other presentations of sporadic CJD.	Mother died from pathologically confirmed CJD.
Krishna et al[1]	74	Female	Patient presented with a sudden-onset, right-sided hearing loss of 2 weeks duration. She then experienced sudden memory loss, ataxia and diplopia. Patient developed progressive imbalance and had fallen 3 times in the 2 weeks prior to symptoms. family noted scanning speech. Local urgent care diagnosed her with Meniere's disease. 2 weeks following admission, developed involuntary jerking of UE and increased tremor and startle myoclonus, aphasia and apraxia.	Sudden-onset right-sided hearing loss.	Memory loss, ataxia and diplopia, poor eye tracking, shuffling gate, truncal ataxia with intention tremor.	Scanning speech.	Admitted to a rehabilitation unit to start physiotherapy.	Dementia and level of consciousness deteriorated rapidly to where she passed away less than 2 months following admission.	Audiometric testing revealed moderate-to-severe bilateral sensorineual hearing loss with poorer word recognition than would be predicted by the degree of hearing loss. EEG showed excessive diffuse slowing of theta and delta waves and occasional triphasic wave, suggestive of a mild diffuse encepholapathy. Western blot analysis of a right frontal brain biopsy detected the presence of PRPSc, confirming CJD.	Patient had progressive increase in baseline hearing loss with dysequilibrium but no vertigo. Reports with these symptoms may be initially diagnosed with a peripheral vestibulocochlear disorder, as was the case with this patient. Ultimate diagnosis of CJD depends on neuropathologic findings together with clinical history.	Not reported.
Bigelow et al[3]	71	Female	Patient presented with a sudden and consistent change in hearing associated with aural fullness and described the sensation of lightheadedness and vague feelings of imbalance, with no associated tinnitus or true vertigo. Initially treated by her family physician for otitis media without improvement. Patient noted worsening of hearing loss, more noticeable on left side and rapid progression of her gait instability. 7 weeks after onset, patient	Sudden, consistent hearing loss with aural fullness, mostly left-sided; lightheadedness and imbalance.	Gait, paresthesias, blurry vision.	Not reported.	At 4 weeks, a course of oral prednisone provided no symptomatic relief. Symptoms continued to progress, and she began to have episodes of intermittent mild mental	During the course of days, neurologic status deteriorated, and the patient became aphasic and unresponsive. She died 3 months after her initial presentation.	Audiometric testing showed bilateral sensorineural hearing loss. A neurologic evaluation revealed a mild nystagmus, a questionable left visual field deficit, and a moderately unsteady gait. MRI revealed only a small area of white matter infarct in the left corona radiata while MR angiography showed no abnormalities. EEG revealed periodic sharp wave complexes consistent with CJD.	Rapidly consistent, rather than episodic signs and symptoms, suggest that the hearing loss was attributable to the patient's CJD pathology and not from a separate peripheral cause.	Not reported.

Continued.

Author, Year	Age (years)	Gender	Patient presentation	ENT specific symptoms	Other CJD symptoms	Other unrelated symptoms	Symptomatic treatment?	Outcome	Imaging/labs ordered+significance	Takeaway they made (look at discussion/conclusion)?	Family history?
			complained of increasing gait difficulty, numbness in her left hand and foot, and blurry vision. Patient exhibited rapid neurologic deterioration and worsening gait such that she could no longer live independently at home. She developed considerable cognitive impairment and blunted affect. Decreased vibratory and pinprick sensation, brisk reflexes, and positive Babinski responses were also noted.				status changes.				
Tobias et al[9]	65	Male	Patient presented with bilateral hearing loss which progressively developed over 2 months. Patient reported "gurgling" noises in ears and that hearing loss could initially be overcome by shouting or increasing TV volume. He had difficulty using telephone and could only communicate by writing. Perception of speech was more impaired than his hearing. One month before admission, pt's wife noted that he became hesitant and nervous, unsteady (falling to the left) and developed speech difficulties.	Bilateral hearing loss with aural fullness, imbalance.	Speech difficulties, paratonic rigidity of all limbs and recurred primitive reflexes, spontaneous myoclonus and prominent startle response, akinetic and mute; possibly blind (no response to visual menace), nystagmus.	Not reported.	Not reported.	Died 2 weeks following admission from bilateral acute bronchopneumonia.	Audiometry confirmed bilateral hearing loss. EEG showed prolonged repetitive sharp wave complexes. Following death, histological exam revealed subacute spongiform encephalopathy with vacuolation in the frontal occipital and both temporal lobes, BG, thalamus and cerebellum, with extensive neuronal loss and reactive astrocytosis in areas of severe spongiform change in the temporal lobes. Staining for PrP was positive in areas of vacuolation. Gene sequencing of PrP revealed patient was methionine-valine heterozygous at codon 129.	Spongiform changes in temporal lobes support the diagnosis of cortical deafness as cause of patients hearing loss.	Healthy.
Kroeze et al[8]	68	Female	Patient presented to the neurology clinic with progressive balance disorder and spasticity of the left side of the body and right arm as well as increasing forgetfulness, less talkativeness and change in character.	Progressive balance disorder.	Severe dysarthria, spasticity of the arms, a pyramidal syndrome of both legs, and significant ataxia. Patient also had altered mental status.	Not Reported.	Not reported.	Not reported.	Electroencephalogram (EEG) showed diffusely slowed background pattern with paroxysmal waves with a triphasic aspect. Magnetic Resonance Imaging (MRI): T2-weighted images, the fluid-attenuated inversion recovery images, and the diffusion-weighted images in both hemispheres a striking hyperintensity of the putamen and the caudate nucleus. Additionally, some hyperintensity of the right occipital cortex was visible. These same areas showed a hypointense signal on the apparent diffusion coefficient images. Cerebrospinal Fluid showed elevated 14-3-3 protein.	A hyperintense signal of the neostriatum, which includes the putamen and the caudate nucleus, and a high concentration of the 14-3-3 protein in the CSF are commonly seen in Creutzfeldt-Jakob disease.	Not reported.
Zarei et al[6]	55	Male	Patient presented with progressive difficulty with maintaining balance	Progressive balance	Hypomimia, Gait	Not Reported.	Not reported.	Died 6 months following	Hematological, biochemical, and immunological tests—including	Quantitative Oculometric and neuropsychological	Not reported.

Continued.

Author, Year	Age (years)	Gender	Patient presentation	ENT specific symptoms	Other CJD symptoms	Other unrelated symptoms	Symptomatic treatment?	Outcome	Imaging/labs ordered+significance	Takeaway they made (look at discussion/conclusion)?	Family history?
			and tracking objects as well as diplopia. No cognitive, sensory, or motor deficits were reported on initial presentation. Towards the end, the patient became verbally unresponsive, and developed myoclonic jerks and fasciculations before passing.	disorder and ataxia.	disturbance, saccades, impairment in comparing objects during assessment of mental status. Psychomotor speed was reduced.			presentation from bronchopneumonia.	paraneoplastic markers, serum caeruloplasmin, autoimmune profile, serology for Whipple's disease, and routine cerebrospinal fluid (CSF) examination all normal. MRI and single photon emission CT normal. Quantitative oculomotor exam, using an infrared oculometer, with the head immobilized on a bite bar and appropriate visual stimuli presented at a distance of 57 cm. Visually evoked saccades and OKN quick phases were dramatically slowed to around 12% of typical normal values. Leftward saccades were significantly faster than rightward, and their latencies were higher but not abnormal. The relation between peak saccadic velocity and amplitude was similar to that of a normal subject, albeit with much lower velocity values. Neither smooth pursuit nor slow phases of optokinetic nystagmus were affected, and there was no obvious abnormality in the accuracy of saccades in either direction. CSF showed very high titers of S-100b, neuron specific enolase, and 14-3-3 proteins. Necropsy examination confirmed the diagnosis of sporadic CJD, with a diffuse pattern of lesions involving many areas of the brain including the brain stem, and no clear focus of degeneration.	studies in sCJD suggest that sCJD should be included in differential of patients presenting with supranuclear palsy.	
Hintz[12]	20	Male	Patient presented to Children's hospital with acute onset of ataxia noticed by his parents after returning from a trip. The patient had been receiving pituitary growth hormone treatments from the age of 3 years old and had been responding well.	Ataxia.	Ataxia and a 'series of other neurological events'.	Insulin-dependent diabetes mellitus and a history of persistent growth failure and secondary hypothyroidism with growth hormone deficiency.	Not Reported.	Pt died 6 months following onset of ataxia.	Autopsy showed 'classic' signs of CJD.	Contamination of purified human growth hormone with CJD infectious agent resulted in this case. 28 cases of CJD have been identified globally associated with human growth hormone treatment. This report claims that these and other risks make the use of human growth hormone purified from pituitary glands unjustifiable and favors the use of synthetic growth hormone.	Not reported.

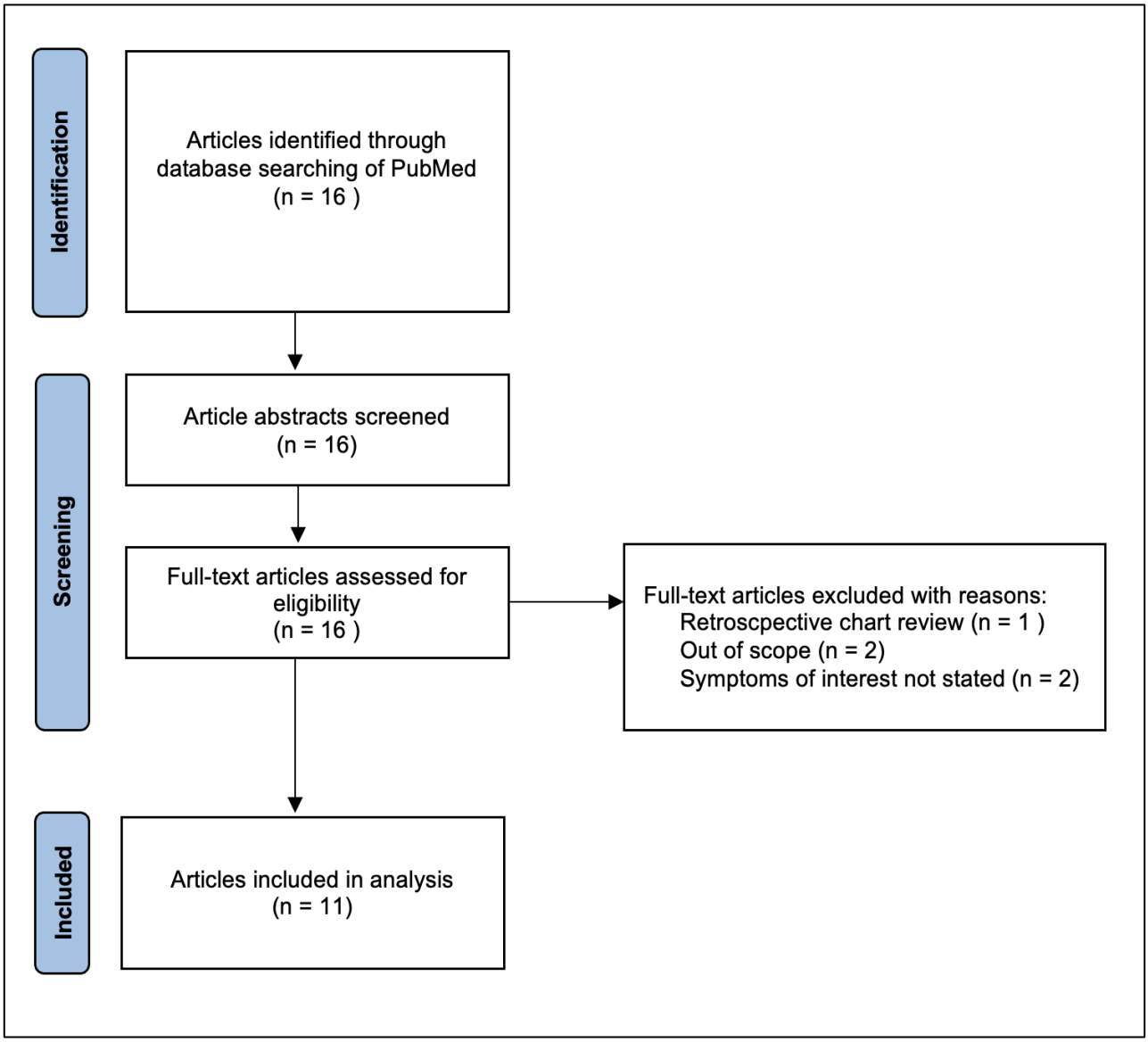

Figure 1: PRISMA flow diagram.

RESULTS

Analysis of CJD cases revealed a total of eight patients with hearing loss (73%) and three patients with balance loss (27%). Table 1 includes a summary of key findings. Further analysis revealed that of patients that presented with hearing loss, 4 (50%) were males, and 4 (50%) were females. The average age of a CJD patient that presented with hearing loss was 63.75 years (SD=11.17 years). Of the three patients that presented with balance loss, two (67%) were males and one (33%) was female. The average age for a patient that presented with balance loss was 46.67 years (SD=24.83 years). The most common CJD symptoms seen in patients that presented with hearing or balance loss were gait impairment (8, 73%), followed by myoclonus (6, 55%), and then cognitive impairment (4, 36%), akinetic museum (4, 36%), and vision disturbances (4, 36%).

As a neurodegenerative disorder, CJD has several common findings. Table 2 lists all CJD associated symptoms in patients with either hearing or balance loss. These are the symptoms that are classically seen in CJD. The most commonly seen symptom was gait impairment (8, 73%), followed by myoclonus (6, 55%), cognitive impairment (4, 36%), akinetic mutism (4, 36%), and vision disturbances (4, 36%). Importantly, gait disturbances are abnormalities in the patterns of walking commonly seen in CJD, whereas balance loss is defined as postural instability rarely reported with CJD.

Next the most commonly ordered tests were analyzed in CJD patients with hearing and balance loss. The most commonly ordered tests were electroencephalogram (5, 45%), magnetic resonance imaging (5, 45%), Real-time quaking-induced conversion (5, 45%), 14-3-3 protein (4, 36%), and audiometry (27%). These findings can be seen in Table 3.

Table 2: Most common CJD symptoms seen in hearing and balance loss patients.

Symptoms	Article number
Gait impairment	1, 4, 5, 6, 7, 9, 10, 11
Myoclonus	2, 4, 5, 6, 8, 9
Cognitive impairment	1, 7, 9, 8
Akinetic mutism	1, 2, 4, 8
Vision disturbances	6, 7, 8, 10
Speech impairment	6, 8
Cerebellar syndrome	2
Prosopagnosia	4
Psychomotor agitation	4
Encephalopathy	4
Mental status deterioration	5
Memory loss	6
Intention tremor	6
Hypomimia	10

Table 3. Most commonly ordered tests in CJD patients.

Tests	Article number
EEG	2, 3, 4, 5, 6
MRI	1, 2, 3, 4, 5
RT-QuIC (PrP^{SC})	1, 2, 3, 5, 6
14-3-3 Protein	1, 2, 4, 5
Audiometry	1, 5, 6
Genetic testing	3, 5
Auditory Evoked Potential	2
CT	5
CRP	3
Neuron Specific Enolase	3

CJD, Creutzfeldt-Jakob Disease; EEG, Electroencephalogram; MRI, Magnetic Resonance Imaging; RT-QuIC, Real-Time Quaking-Induced Conversion; PrPSC, prion scrapie protein; CT, Computed tomography; CRP, C-reactive protein.

DISCUSSION

Understanding common symptoms seen in CJD patients is critical for prompt diagnosis. While the loss of hearing or balance are uncommon presentations of CJD, their presence alongside more typical symptoms should raise clinical suspicion. In our reviewed patient cohort, gait impairment was the most frequent symptom accompanying hearing loss. Following gait impairment, other symptoms that accompanied hearing loss included myoclonus, cognitive impairment, akinetic mutism, and vision disturbances, all of which are symptoms typically seen in CJD. Otolaryngologists should maintain a high index of suspicion for CJD when patients present with unexplained auditory or vestibular symptoms, particularly if accompanied by other neurological signs. A thorough history, including past surgeries and potential exposure to infected individuals or animals through hunting or consumption of wild game is essential. Early neuroimaging and appropriate referral can aid in timely diagnosis. Ultimately, recognizing atypical presentations such as hearing or balance loss may play a key role in identifying CJD at an earlier stage.

The gold standard for diagnosing CJD is brain biopsy.[5,13] However brain biopsy is extremely invasive, and typically not done until autopsy. In this review, the most common imaging ordered was MRI, EEG, and RT-QuIC (PRPSC), followed by audiometry. Common MRI findings seen in patients with CJD include cortical ribboning and basal ganglia hypersensitivity on MRI, periodic sharp wave complexes on EEG, and release of 14-3-3 protein into the CSF indicative of rapid neuronal death.[14] Increased levels can help confirm or point to a diagnosis of CJD. Audiometry is not a typical imaging test ordered in CJD, however as these patients presented with hearing loss, it was ordered to determine the underlying cause. Lastly, PRPSC is an abnormally folded protein that should not be found in the CSF and elevated levels are indicative of CJD.[17]

Gait abnormalities and balance impairments may appear similar in presentation but originate from different neurological pathways. Balance disturbances are generally due to the interruption of the vestibular system and gait abnormalities typically suggest disruptions of cerebellar function. Since otolaryngologists may be among the first to evaluate a patient experiencing the onset of CJD-related gait or balance impairments, establishing such a distinction offers a better foundation for recognizing the otolaryngological presentations of CJD. Additionally, differentiation is key as gait impairment is common in CJD, whereas balance loss is extremely rare. This study provides important insight relating to the early identification of Creutzfeldt-Jakob Disease in patients that present with the aforementioned atypical symptoms. Further, the review has identified ancillary tests and imaging that may further aid clinicians in providing a clear diagnosis. Such knowledge can serve as a framework for the management of CJD patients and ultimately lead to better outcomes.

Limitations

Since hearing and balance loss are not well-established symptoms of CJD, they might be overlooked in the differential diagnosis of CJD. The impact of this is two-fold; it hinders the timely diagnosis and management of CJD and limits and availability of published care reports and large-scale studies linking hearing and balance loss to CJD. Additionally, variability in the diagnostic modalities used to assess hearing and balance loss might introduce challenges in the identification of symptoms and reduce their potential use in early detection and differential diagnosis of CJD. Given that the definitive diagnosis of CJD typically requires neuroimaging, hearing and balance loss could serve as additional clinical indicators supporting the diagnosis. A standardized approach to CJD diagnosis would minimize these limitations and help establish a link between these atypical symptoms and

CJD. Moreover, further research is needed to understand the neurodegenerative mechanisms affecting hearing and vestibular function in CJD to aid in its diagnosis and explore potential therapeutic interventions to improve patient outcomes.

CONCLUSION

CJD is a rare degenerative disease that can present with hearing and balance loss. It is important to recognize that there can be atypical presentations of this disease. Characterizing and recognizing atypical presentations can lead to clinicians recognizing and diagnosing these diseases earlier leading to improved outcomes and quality of care. Specifically, otolaryngologists and hearing specialists should keep CJD on their differential when working up atypical cases of hearing and balance loss.

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