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CASE REPORT

Hearing impairment: an unexpected diagnosis

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Introduction

Most patients complaining of hearing impairment have problems with the peripheral auditory apparatus, either conductive or sensorineural, traditionally distinguished with tuning fork tests. Defects of central auditory processing such as pure word deafness, also called auditory agnosia (an inability to appreciate the meaning of sounds despite normal perception), are much less common. Nevertheless, clinicians need to be aware of this broad differential when assessing patients with hearing complaints.

Case report

A previously healthy 55-year-old woman presented with an approximately 1-month history of hearing difficulty, subjectively greater on the right, without tinnitus, vertigo or aural fullness. A few days after onset she developed difficulties with word finding and pronunciation, and made spelling errors when texting. On direct questioning she had subjective unsteadiness when walking but no history of dizziness. Past medical history was unremarkable, aside from occasional migraine with visual aura but without headache. There was no family history of neurological or otological disease. There were no occupational risk factors for cochlear trauma.

On examination, communication was mildly affected because of the hypoacusis. There was no eye movement disorder, no bulbar abnormality, reflexes were normal, but heel-toe walking appeared ataxic. Initial magnetic resonance imaging of the brain was normal.

Thereafter, the patient deteriorated rapidly. She repeatedly reported that she could not hear and was unable to communicate either by lip reading or writing. Speech output was paraphasic with impaired repetition; it was difficult to assess comprehension. Examination now showed nystagmus, and unequivocal gait and truncal ataxia.

Repeat magnetic resonance brain imaging now showed abnormal signal intensity on T2-weighted imaging in the basal ganglia, particularly the caudate nuclei. On diffusion-weighted imaging, cortical hyperintensity was evident, more marked in the left hemisphere. In this clinical context, the imaging changes were suggestive of Creutzfeldt–Jakob disease. Electroencephalography showed a generalised encephalopathic picture (no clear alpha rhythm, background dominated by mixed frequency slow wave activity), but neither epileptiform changes nor periodic slow wave complexes were seen. CSF analysis showed normal cell count and protein.

The patient's relentless deterioration continued, developing into an akinetic rigid syndrome. She died within 3 months of symptom onset. CSF subsequently proved positive for 14–3-3 protein and S100b was elevated (0.56 ng/ml, normal <0.41 ng/ml). Permission for postmortem examination was not granted.

The combination of clinical and investigation findings (magnetic resonance imaging, CSF) was consistent with a diagnosis of probable sporadic Creutzfeldt–Jakob disease, according to World Health Organization clinical diagnostic criteria (Zerr et al, 2009).

Discussion

Prion diseases have heterogeneous pathogenesis (sporadic, inherited and iatrogenic forms) and phenomenology (cerebellar, visual cortical, extrapyramidal, pyramidal and psychiatric symptoms). Many unusual phenotypes are also recognised, but are confined to case descriptions or small case series.

Hearing loss or deafness has been reported as the presenting feature of Creutzfeldt–Jakob disease in sporadic (Tobias et al, 1994; Bigelow et al, 1998; Krishna and Bauer, 2004; Salazar et al, 2014), iatrogenic (Fernández Pérez et al, 2014), and familial (Cataldi et al, 2000; Reñé et al, 2009; Miyagawa et al, 2018) forms, the latter all carrying the common prion protein (PrP) gene E200K point mutation. Clinical and audiometric studies in these

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Learning points

- Sporadic Creutzfeldt–Jakob disease may occasionally present with symptoms of hearing impairment.
- This may be a result of sensorineural hearing loss, retrocochlear pathology (auditory agnosia, cortical deafness, pure word deafness), or a combination of both.
- Because of the frequency of unusual variants of Creutzfeldt–Jakob disease, early diagnosis is often difficult, with implications for early intervention.
- The diagnosis of Creutzfeldt–Jakob disease may need to be considered, even if a remote possibility, in patients with hearing impairment, particularly if there are additional neurological symptoms and signs and when initial neuroimaging fails to disclose a diagnosis.

cases have sometimes indicated sensorineural hearing impairment, although these are sometimes normal. However, word recognition difficulties greater than predicted by the degree of hearing loss have sometimes been noted, suggesting more central pathology (such as 'cortical deafness', reported by Tobias et al, 1994). Auditory agnosia has been described in patients with Creutzfeldt–Jakob disease (Gold et al, 1997; Orimo et al, 2000), sometimes as pure word deafness (Hillis and Selnes, 1999; Fernández Pérez et al, 2014), and this may have been a contributory factor in other cases (Krishna and Bauer, 2004; Salazar et al, 2014). Linguistic and balance problems have been noted as early symptoms in some patients with hearing impairment (Tobias et al, 1994; Hillis and Selnes, 1999; Krishna and Bauer, 2004).

Auditory agnosia is associated with bilateral temporal lobe lesions, including primary auditory cortex (transverse gyri of Heschl), auditory radiations and insular cortex. In patients with Creutzfeldt–Jakob disease and hearing impairment, who have undergone magnetic resonance imaging of the brain, high signal intensity change has sometimes been observed not only in the basal ganglia (as typically seen in cases of sporadic Creutzfeldt–Jakob disease), but also in superior temporal cortex (Cataldi et al, 2000; Fernández Pérez et al, 2014). Electroencephalography has often been non-specific, as in this case.

Neuropathological studies are few. Tobias et al (1994) found confluent vacuolation and positive PrP immunocytochemistry in both transverse temporal gyri. Reñé et al (2009) found neuronal loss, gliosis and deposition of PrP in the vestibular and cochlear nuclei of their patient.

The current case confirms and extends the existing literature on hearing impairment as a presenting feature of sporadic Creutzfeldt–Jakob disease. More generally, the case highlights hearing impairment as a feature of dementia disorders. While Creutzfeldt–Jakob disease presenting with visual symptoms is well recognised (Heidenhain variant), hearing impairment is less common in Creutzfeldt–Jakob disease and may go unrecognised (Creutzfeldt–Jakob disease was not considered in the differential diagnosis when this patient was first seen, by a clinician with a specialist interest in neurodegenerative disorders). The authors suggest that neurodegenerative disorders, including Creutzfeldt–Jakob disease, merit consideration in patients presenting with hearing impairment, especially if, as in the present case, other neurological features are present, making an isolated otological diagnosis unlikely.

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