CASE REPORT

Superficial siderosis misdiagnosed as idiopathic bilateral neurosensorial deafness

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Accepted 27 April 2018

SUMMARY

Superficial siderosis (SS) is a rare condition resulting from different sources of bleeding into the subpial space. The most common symptoms are: hypoacusia, ataxia, incontinence, dementia and parkinsonism. Since several neurodegenerative disorders may present with same clinical features, SS is often misdiagnosed. Here we present a case of SS misdiagnosed as idiopathic bilateral neurosensorial deafness.

BACKGROUND

This case is important since it shows diagnostic pitfalls when you do not order specific gold standard tests. In some cases, diagnostic delay may last years.

CASE PRESENTATION

Superficial siderosis (SS) of the central nervous system (CNS) is a rare acquired condition resulting from bleeding into the subarachnoid space, leading to deposition of hemosiderin into the subpial layers of the brain, cranial nerves or spinal cord. The first report in the literature was described as 'melanosis of the brain' by Ralph C Hamil in 1908 in one patient autopsy.^{1 2} Since then, many causes of SS have been found as a source of chronic hemosiderin accumulation in the CNS.^{3–11} A prior history of trauma or brain surgery can be found in the history of some patients.⁹ The time that elapses between the bleeding episode and the onset of symptoms varies widely and recall bias may delay the diagnosis.¹ In spite of many technological advances in diagnosis, some cases still remain idiopathic.¹²

Because a great variety of unspecific symptoms may be present in some patients, it is the adult onset of progressive neurosensory hearing loss, gait ataxia and pyramidal dysfunction that ultimately suggest the diagnosis.^{1 13} In some patients, gradual bilateral hearing loss is the first complaint and it may lead to a wrong diagnosis if other symptoms are absent or without an appropriate approach. Less common clinical features are headache, bladder disturbances, urinary incontinence, dementia, spastic paraplegia, anosmia and parkinsonian symptoms which may develop many years later and could be considered as distinct entities, leading to misdiagnosis.^{14–22}

Here we present the case of a 65-year-old right-handed man referred to our movement disorder clinic with a previous history of gradual hearing loss associated with unsteadiness, intermittent falls

and speech changes; he was diagnosed for 8 years with idiopathic bilateral neurosensorial deafness by an Ear-Nose-Throat specialist. No further investigation was done before he arrived at our clinic. Over time he developed urinary incontinence, which was attributed to prostate hypertrophy. Finally, he developed dysarthria and unsteadiness, which progressed to the point of inability to walk without aid, so he was referred to our clinic for workup.

His medical history was unremarkable, except for a significant head injury 20 years ago, which required observation in the ER for 8 hours. No brain imaging was done at that time since patient progression was favourable in ER and no warning signs were detected.

Physical examination revealed dysarthria, bilateral neurosensorial deafness and Babinski signs, without hyperreflexia or spasticity. However, symmetric rigid-akinetic parkinsonism of four limbs and truncal ataxia was found (video 1). No sensitivity symptoms were found. A CBC, glu, urea, crea, Na, K, Cl, Ca, Mg CT scan were normal.

INVESTIGATIONS

Because we initially suspected a neurodegenerative disorder associated to parkinsonism, a levodopa trial was done without response and a brain MRI, which showed low-signal-intensity in axial T2-weighted and gradient echo image in midbrain cisterns and mesial temporal sulci bilaterally, compatible with hemosiderin deposition at the subpial surface. Cerebellar atrophy was also observed (figure 1).

DIFFERENTIAL DIAGNOSIS

Differential diagnosis of SS is broad, including hereditary disorders such as spinocerebellar ataxias (SCAs) or multiple system atrophy. SCA type 36 (SCA36) is characterised by a late-onset, slowly progressive cerebellar syndrome associated with sensorineural hearing loss. However, this is an autosomal-dominant rare disorder with less than 5% of de novo presentation, which makes it unlikely in sporadic cases, such as our patient. More frequent SCAs, such as SCA 1, 2 and 3, associated with parkinsonism, may be possible, since hearing loss is a frequent finding in general population with ageing; however, in our patient, hearing loss was the initial symptom and cerebellar signs were detected later. Again, lack of family history and age of onset make the diagnosis unlikely. In the absence of history of alcohol

To cite: González-Usigli HA, Perez-Torres T, Saenz-Farret M, et al. *BMJ Case Rep* Published Online First: [please include Day Month Year]. doi:10.1136/bcr-2017-222639

Video 1 This video shows subtle wide based gait with unsteadiness.

abuse, toxic cerebellar drugs or a family history of ataxia, the diagnosis of multiple system atrophy is likely in a patient with idiopathic late onset cerebellar ataxia associated with parkinsonism, dysautonomic features and subtle cognitive decline with no adequate response to levodopa. Nevertheless, our patient's MRI did not show the typical degeneration of the middle cerebellar peduncles pons or a 'hot cross bun' sign. On the contrary, it showed intensity changes at the subpial space of the hindbrain with diffuse cerebellar atrophy, compatible with chronic subpial hemosiderin accumulation.²³

Figure 1 T2 sequence and gradient Echo MRI with subpial hemosiderin deposition. T2 sequence may be not as clear than gradient Echo or SWIFT sequences as it is shown in the figure.

OUTCOME AND FOLLOW-UP

He remains stable, improving hearing with hearing aids and balance with physical therapy.

DISCUSSION

The clinical features previously mentioned, in conjunction with the lack of adequate imaging studies, may lead the clinician to consider a neurodegenerative disorder when evaluating cases of SS; the history of head trauma is also missing in some patients or at least is not taken into account in the evaluation of these patients.²⁴

In our patient, there was considerable head trauma 20 years ago, a possible source of subarachnoid haemorrhage with subsequent hemosiderin accumulation in his brain. We recognise that a spinal imaging would be desirable in order to rule out another possible source of the haemorrhage, such as recurrent spinal bleeding. However, our principal objective in this case is not to establish the precise source, but to point out that SS can be mistaken for other conditions when it is not a suspected diagnosis or when the correct auxiliary diagnostic studies, such as specialised MRI sequences, are not carried out. Furthermore, unfortunately we were unable to follow up the case of our patient and it is not possible to get a spinal MRI in order to rule out other possible sources of recurrent bleeding, which can be especially important, since in that case surgery may arrest the progression of symptoms associated with the accumulation of subpial iron.

Initially, we did not consider a spinal MRI necessary, since there were no clinical symptoms of spinal cord disease, spine surgery or trauma, but there was evidence of brain SS combined with the knowledge of an important head injury in the past. Furthermore, as mentioned above, head trauma has been related to SS as a possible cause in previous reports.^{21 25 26}

Patients with ataxia in association with neurosensorial hearing loss, pyramidal, dysautonomic and parkinsonian features must raise the clinical suspicion of SS and brain MRIs must be done in those cases with adequate sequences as T2-weighted and echo gradient imaging, focusing particularly on the subpial area. A source of old or intermittent bleeding can be sought.²⁷ Nevertheless, if the radiologist does not recognise the initial signs of hemosiderin deposition in these specialised sequences, the MRI is often reported as normal.¹

Many reports do not include SS as a potential cause of cerebellar ataxia. We believe, however, that in the appropriate clinical setting, this condition must be included as a differential, to avoid misdiagnosis when adequate MRI sequences are missing. In our patient, we think that the history of head injury many years ago explains subpial hemosiderin. Even in our patient who had a solid history of head injury which may be the source of the subpial hemosiderin, the evaluation with a MRI of the spine, to rule out other sources of chronic bleeding is desirable. Nevertheless, the lack of spinal disease history and symptoms solidly orient us to the head trauma as the cause of his SS.

SS is a rare condition which must be included in the differential diagnosis of sensorineural hearing loss and ataxia, since the full picture with parkinsonian, pyramidal and urinary symptoms may manifest many years later. Adequate imaging studies are essential to its diagnosis.

Learning points

- Siderosis superficial is another uncommon cause of ataxia associated with neurosensory deafness and dysautonomia.
- When an adequate medical history is missing, siderosis superficial may be misdiagnosed as a number of neurodegenerative diseases, such as multiple systems atrophy or spinocerebellar ataxias, due to its clinical characteristics.
- Specialised MRI sequences, due to their high specificity, may help differentiate superficial siderosis from other, more common, diagnoses.
- If a brain MRI does not show a definitive source of subarachnoid bleeding, a spinal MRI must be included to rule out other sources of it.
- Often follow-up of patients with ataxia and neurosensory deafness may clarify the diagnosis.

Contributors HAG-U is the intellectual author and final reviewer of the case. MS-F, TP-T and PR-M edited the manuscript and recollected information from the patient and his medical records.

Funding The authors have not declared a specific grant for this research from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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