eNeurologicalSci 22 (2021) 100321

Contents lists available at ScienceDirect

eNeurologicalSci

journal homepage: www.elsevier.com/locate/ensci

Case report

Basilar invagination: A mimicker of bulbar-onset amyotrophic lateral sclerosis

Jason Seng Hong Tan *, Shirley Lee, Fu Liong Hiew

Department of Neurology, Kuala Lumpur General Hospital, Wilayah Persekutuan Kuala Lumpur, Malaysia

ARTICLE INFO

Keywords:
Basilar invagination
Amyotrophic lateral sclerosis
Bulbar onset
Clinical neurology

ABSTRACT

Amyotrophic lateral sclerosis (ALS) is characterized by progressive onset motor deficits with heterogenous presentations ranging from dysarthria to foot drop. Approximately 20% of the patients present with focal bulbar symptoms, in which some may remain restricted to bulbar region (isolated bulbar palsy), and the remaining eventually spreads to involve other body regions (classical ALS). Without accompanying upper and lower motor neurons signs elsewhere, differential diagnoses for isolated bulbar symptoms are extensive, include ALS variants as well as potentially treatable mimics. Therefore, it is important to take heed on every possible aetiology that may disrupt the hypoglossal nucleus, nerve, or lingual muscle itself. Herein, we illustrated a rare presentation of Group A basilar invagination, which mimicked bulbar-onset ALS.

1. Introduction

Amyotrophic lateral sclerosis (ALS) is characterized by progressive onset motor deficits with heterogenous presentations ranging from dysarthria to foot drop. Approximately 20% of the patients present with focal bulbar symptoms, in which some may remain restricted to bulbar region (isolated bulbar palsy), and the remaining eventually spreads to involve other body regions (classical ALS) [1,2]. Without accompanying upper and lower motor neurons signs elsewhere, differential diagnoses for isolated bulbar symptoms are extensive, includes ALS variants as well as potentially treatable mimics [2]. Therefore, it is important to take heed on every possible aetiology that may disrupt the hypoglossal nucleus, nerve, or lingual muscle itself. Herein, we illustrated a rare presentation of Group A basilar invagination, which mimicked bulbar-onset ALS.

2. Case report

A 55-year-old lady presented with progressive hoarseness of voice and nasal regurgitation over a period of 4 months. She denied any visual abnormality, bowel and bladder symptoms or limbs weakness. Abnormal cranial neurological findings were bulbar speech with bilateral tongue wasting and fasciculation, involving more on the right. Gag reflex and other cranial nerves were preserved. Reflexes were brisk in all limbs, however, without other associated upper motor neuron or cerebellar signs. Initial brain and cervical MRI scans were normal. Nerve conduction study showed no evidence of large fibre neuropathy and electromyography demonstrated fasciculation restricted to both sides of the tongue, sparing other body segments including paraspinal muscles. Therefore, the above clinical and electrophysiological findings did not meet El Escorial criteria for ALS. Clinical assessment by the otolaryngology team revealed no swallowing dysfunction and ruled out infiltrative nasopharyngeal lesion. A subsequent CT neck demonstrated suspicious base of right hemitongue infiltrative lesion, which proved normal on biopsy. A follow-up MRI tongue demonstrated asymmetrically atrophic tongue with fatty infiltration and highlighted basilar invagination and cerebellar tonsillar herniation (Fig. 1a,b). Following this, a dynamic CT cervical spine confirmed the Group A basilar invagination in association with atlanto-axial instability (Fig. 2a,b,c).

Upon further probing, patient recalled a minor car accident two years ago with minor head impact onto the car roof but without sustaining immediate head or neck injuries. The antecedent trauma could have been the precipitating factor of compression at cervico-medullary junction caused by basilar invagination, impinging on the hypoglossal nerves. Likewise, compression of medullary pyramids could explain the brisk limbs reflexes. She was planned for neurosurgical stabilisation of atlanto-axial joint.

* Corresponding author at: Department of Neurology, Kuala Lumpur General Hospital, 23, Jalan Pahang, 50586 Wilayah Persekutuan Kuala Lumpur, Malaysia.
E-mail address: jasontan90@gmail.com (J.S.H. Tan).

https://doi.org/10.1016/j.ensci.2021.100321
Received 11 September 2020; Received in revised form 4 December 2020; Accepted 13 January 2021
Available online 20 January 2021

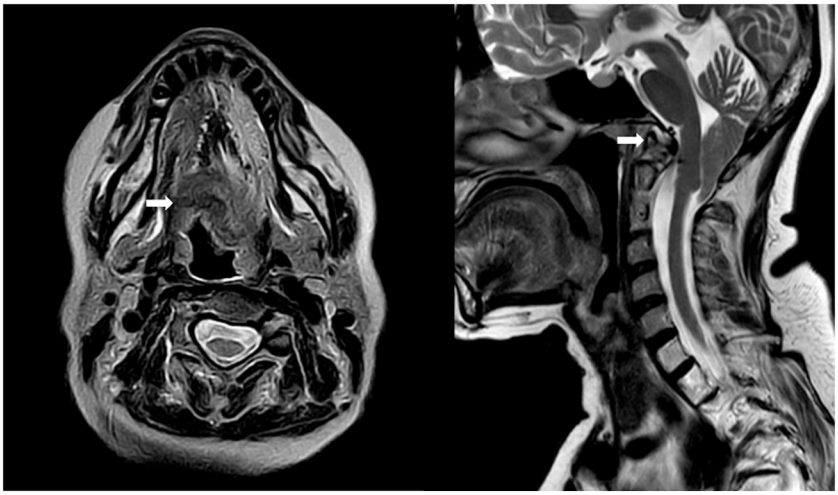

Fig. 1. MRI of tongue and cervical.
a: T2 weighted MRI of tongue showed asymmetrically atrophic tongue with fatty infiltration.
b: T2 weighted MRI of cervical spine highlighted basilar invagination and cerebellar tonsillar herniation.

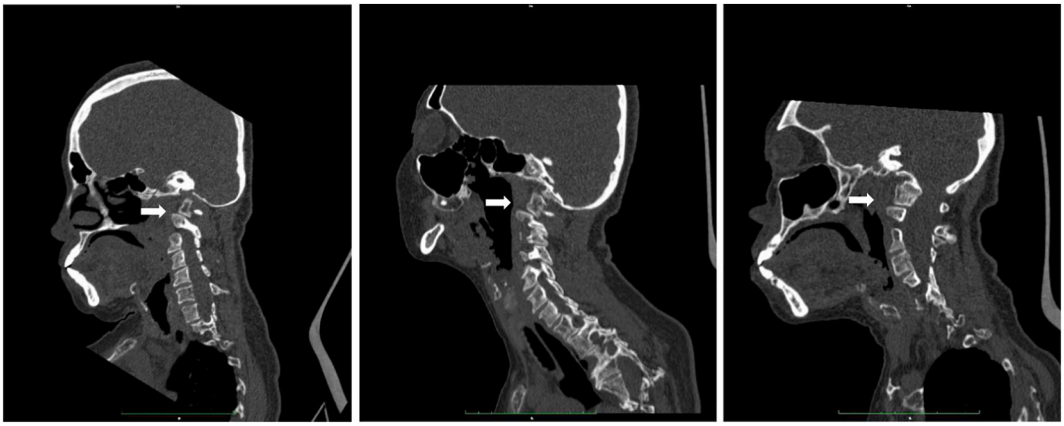

Fig. 2. Dynamic CT cervical spine.
a,b,c: Computed tomography (CT) scan with cut passing through the facets in flexion, extension and neutral showing basilar invagination and atlantoaxial facetal instability.

3. Discussion

Basilar invagination is a pathology at the craniovertebral junction, where the odontoid process of the C2 vertebrae prolapses into the foramen magnum, causing compression. It is broadly classified into 2 groups: Group A with atlantoaxial instability, and Group B without [3]. The aetiologies are often associated with developmental anomaly such as hypoplasia of the basiocciput (clivus), occipital condyle, or atlas, incomplete ring of C1 with spreading of the lateral masses, achondroplasia, and atlanto-occipital assimilation [4]. A third of patients with basilar invagination have associated neural axis abnormalities like Chiari malformation, syringomyelia, syringobulbia and hydrocephalus [1]. If left unrecognized or untreated, basilar invagination can result in profound neurological deficits and potentially be fatal due to complications related to brainstem compression [5].

Patients with basilar invagination commonly presented with limb weakness (97%), with half had paresthesia (54%), a third with neck pain (33%) and one-fifth with bowel and bladder disturbance (22%) [6]. Presence of ataxia was supportive of those with associated Chiari malformation despite only seen in about half (47%) of them. Bulbar symptoms were noted in 9 to 16% of patients, more common among those with associated Chiari malformation.

From a neuroanatomical perspective, the hypoglossal nerve which arises from its nucleus at the medulla, exits between the pyramid and olive in a groove, and subsequently leaves the skull base anterior to the occipital condyle, travelling inferiorly through the hypoglossal foramen, lateral to the jugular foramen to cross the anterior aspect of lateral masses of C1-C2 [7]. Therefore, compression at the cervico-medullary junction by the odontoid process in basilar invagination can cause various neurological presentations in association with lower cranial nerves and upper cervical spinal cord, resembling the core features of a bulbar-onset ALS. More importantly, more than half of the patients are lacking in key 'red flag' features for ALS such as paresthesia, neck pain or bowel and bladder disturbances [8]. Other features against ALS are the presence of dissociated sensory loss, vestibular function impairment and cerebellar signs associated with Chiari malformation, syringomyelia, syringobulbia as seen in up to a third of patients with basilar invagination [1]. Referral to neurosurgical colleagues is warranted, and surgical intervention may alleviate patient's progressive neurological deficits.

4. Conclusion

We illustrated the diagnostic workup of a rare case of focal onset,

bulbar predominant basilar invagination with Chiari malformation in association with atlanto-axial instability mimicking bulbar-onset ALS following antecedent trauma, reversing a diagnosis in favour of a treatable disorder.

Funding source

None.

Declaration of Competing Interest

The authors declare they have no conflict of interest.

Acknowledgement

We would like to thank Dr. Hairuddin bin Achmad Sankala, Radiologist, Department of Radiology, Kuala Lumpur General Hospital for providing the MRI images for this case report. We would also like to thank the Director General of Health Malaysia for his permission to publish this article.

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