Acute Dysphagia as the Presenting Feature of Syringobulbia in a Sixteen-Year-Old Patient

Hong Gao[1], Anita Cheah[1], Neli Avramova[1], Martin Whyte[1], Bassel Zebian[2], Kholoud Al-Majali[1]

1. Department of Acute Medicine, King's College Hospital, Denmark Hill;
2. Department of Neurosurgery, King's College Hospital, Denmark Hill

Introduction

Doctors often use heuristics (pattern recognition) to aid in the diagnostic process[1-2].

This case emphasises the importance of keeping an open mind about the pathology behind a presenting complaint and the relevance of multi-specialty cooperation.

Case History

16-year-old female, with no significant past medical or drug history, presented with "worsening sore throat, cough, unable to swallow + drooling of saliva, feeling increasingly unwell" [A&E notes].

She reported:

- 1-week history of increasing dizziness, nausea and dry wretching episodes
- 2-week history of bowels not opening
- 3-week history of fevers and increased amount of phlegm
- Reduced oral intake due to inability to swallow (painless) - "it doesn't go any further"

She lives with her family (both teachers) and recently had a GCSE Maths exam.

Timeline

6th Jan 2015
05:39
Admitted under the medical team.

Neurological examination: Normal eye movements, normal power and sensation throughout lower limbs.

11:42
Post-Take Ward Round: History and bloods reviewed (Fig. 1).

Neurological examination: Downgoing plantars.

Impression: Need to rule out meningitis.

Initial management: Intravenous Aciclovir & Ceftriaxone, CT Head, lumbar puncture after CT Head.

White Cell Count	12.1
Neutrophil count	10.1
CRP	133
Liver function tests	Normal
Bone and renal profile	Normal

12:04
CT Head: Medulla and cervical cord are expanded by a centrally-located longitudinal cerebrospinal fluid-density lesion.

Timeline

6th Jan 2015
16:36
CT Head results explained.

Neurological examination: Left-beating nystagmus. Normal cranial and peripheral nerve examination.

18:25
Difficult fundoscopy examination: patient was unable to focus her eyes on a single point.

19:52
Bladder scan: Residual volume > 999 mLs.

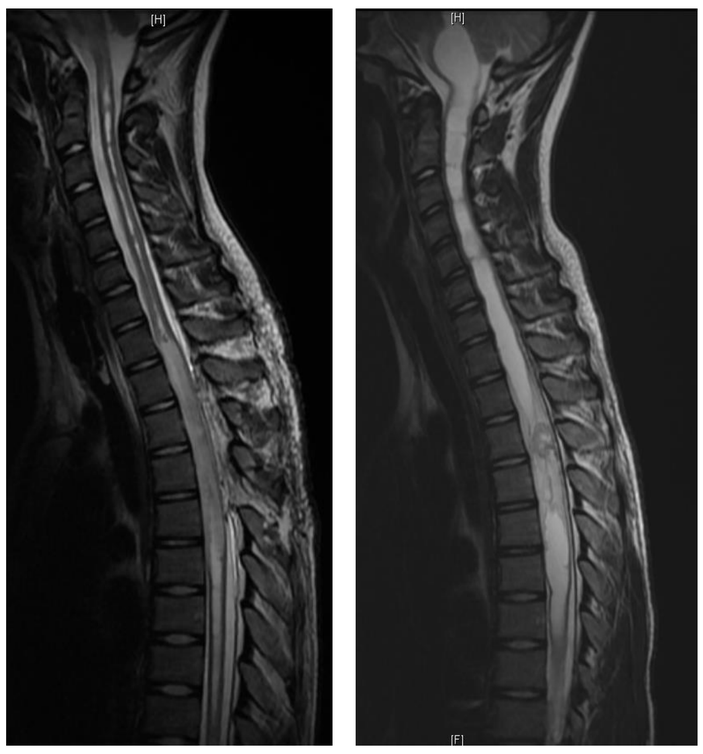
Urinary catheter inserted.

7th Jan 2015
00:28
Neurosurgical review.

Neurological examimation: Bilateral IX, X, XI, XII cranial nerve weakness, bilateral nystagmus, vertical nystagmus, glove and stocking paraesthesiae.

06:55
Revised management: NBM, Dexamethasone 12mg stat and 8mg BD with PPI cover.

9th Jan 2015
"Highly-vascular" tumour resected in theatre; syrinx decompressed.



Before (7th Jan 2015)

MRI Spine (Fig. 1): Extensive, complex hydro-syringomyelic cavity extending from obex to T10 level…heterogenous components at T3-4 level suggestive of a neoplastic lesion at this level

After (9th Jan 2015)

MRI Spine (Fig. 2): Significant reduction in the size of the cervical thoracic syrinx with marked reduction in cord and brainstem expansion…distal syrinx seen

Timeline

13th Jan 2015
Histology: WHO Grade I Haemangioblastoma.

Neurological examination: Normal eye movements and speech formation. Normal tone, power and reflexes in all limbs and downgoing plantars. Normal pinprick, light touch and pain in all limbs. Discharged from hospital.

13th April 2015
Reviewed in clinic. Patient well and neurologically normal.

Currently being investigated for Von Hippel Lindau Disease, which involves retinal screening, abdominal ultrasonography, genetic analysis and follow-up MRI imaging of the spine.

Learning Points

Think of Bulbar/ Pseudobulbar palsies in any patients with dysphagia or has neurological deficit – including the young.

Neurological signs can change quickly – regular full neurological examination is key!

Discussion

- Haemangioblastomata are vascular tumours in the spinal cord which can cause Syringomyelia[3]. They are uncommon tumours of the Central Nervous System (CNS), accounting for less than 3% of all CNS tumours[4]. They are usually well-circumscribed, highly-vascular and benign[4].
- Syringomyelia can present with a protean range of neurological deficits. Case reports in literature show that dysaesthesiae[5], quadriparesis[6], hiccups[7], headaches, ataxia and cerebellar signs[3] could all be presenting signs of Syringomyelia or Haemangioblastomata. They can also exist in asymptomatic patients[8].
- If Syringomyelia is still suspected after CT imaging of the CNS, regular neurological examination should be carried out to detect evolving neurological signs and prompt referral to Neurosurgery is warranted.
- Von Hippel Lindau Disease (VHL) is an autosomal dominant condition characterised by haemangioblastomata in the CNS, retina, kidneys, adrenal glands, endolymphatic sac and pancreas[9]. A 2014 analysis of 92 cases of patients who underwent resection of intraspinal haemangioblastomata showed that 35% had VHL.

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Correspondence to hong.gao@nhs.net or anita.cheah1@nhs.net.
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