

Case Report: A difficult diagnosis to swallow

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A 23 year old woman was referred to Queen Elizabeth Central Hospital (QECH) from Thyolo District Hospital with a 15 year history of persistent post-prandial vomiting. She had associated dysphagia to solids and liquids, heartburn and weight loss, without nausea or change in appetite. Two weeks prior to referral she gave birth to a premature live infant. Her symptoms worsened significantly during the pregnancy. Previous presentations to her local health centre had resulted in advice to improve her nutritional status only. This was a single mother with no psychiatric history.

On admission she was cachectic with ketotic breath. She was afebrile and hypotensive with a blood pressure of 90/50 mmHg. Her weight was 34kg and height 1.55m (BMI 14.2). On inspection she had dry mucous membranes, pink conjunctivae and clear oral cavity. There was no oedema or jaundice. Cardiovascular, respiratory, neurological and abdominal examinations were normal. First impressions were severe malnutrition and hypotension, for which immediate fluid resuscitation and antiemetics were commenced.

Initial Investigations

Abdominal ultrasound scan, urea and electrolytes, creatinine and HIV testing were ordered. The abdominal ultrasound scan showed no abnormalities, HIV negative status was confirmed and other lab results were unobtainable due to technical issues.

What is the diagnosis? How would you treat this? What management challenges may arise?

Diagnosis

Differential diagnoses included:

Mechanical	Motility	Other
<p>Benign:</p> <ul style="list-style-type: none"> Oesophageal web Unlikely as tends to affect White females, dysphagia to solids+liquids and bullous skin lesions sometimes associated. Peptic stricture Possible as heartburn is common but there is no progressive dysphagia to solids then liquids or increased risk of aspiration. Pharyngeal pouch Unlikely as there is no halitosis, regurgitation of undigested food or aspiration. <p>Malignant:</p> <ul style="list-style-type: none"> Oesophageal/stomach Cancer Unlikely with such a chronic history 	<ul style="list-style-type: none"> Achalasia Possible as there is dysphagia to solids and liquids, vomiting, heartburn, weight loss. Systemic sclerosis possible but no evidence of any other connective tissue disease in this case. Myasthenia Gravis Unlikely given no preceding ocular muscle weakness. 	<ul style="list-style-type: none"> Oesophagitis <p>Candida/HSV/CMV Unlikely as no odynophagia/evidence of any oral lesions</p> <p>Reflux Possible with symptoms of heartburn, vomiting and dysphagia.</p>

The patient deteriorated with increased frequency of vomiting so it was decided that as upper GI endoscopy was readily available at QECH, this would be performed first instead of after Barium swallow to confirm a diagnosis. As the procedure began undigested food emptied out onto the floor. A dilated oesophagus with significant food residue and narrowing of the lower oesophageal sphincter was found. The stomach and duodenum were empty and severe oesophageal candidiasis was noted. The endoscopic diagnoses were:

1. Achalasia
2. Oesophageal candidiasis

Treatment

Now that the diagnosis was made, establishing nutritional support to preserve the patient's life was critical. Despite attempted dilatation of the lower oesophageal sphincter, vomiting continued therefore a nasogastric tube (NGT) was inserted with the plan to use F-75 and F-100 formulae (therapeutic milk products used to treat severe malnutrition, measuring 75 and 100 kcal/100ml respectively) to re-establish feeds. Fluconazole was started to control the oesophageal candidiasis. The patient pulled out her first NGT so re-insertion and feeding were only possible after her parents were counselled. She then developed a sixth nerve palsy, confusion and psychosis, which led to a diagnosis of Wernicke's encephalopathy. She was too weak to determine ataxia. Feeds were stopped and high dose Thiamine given (in the form of compound vitamin B tablets) for three days, after which the symptoms settled. Feeds were restarted 5 days later with supplementary multivitamins and this was tolerated well.

Plans are being made for definitive management of her achalasia once her nutritional status is adequate.

Discussion

Achalasia is a rare condition where there is failure of relaxation of the lower oesophageal sphincter due to degeneration of ganglion cells in the myenteric plexus of the oesophagus and the lower oesophageal sphincter. Its underlying cause is not clear with viral, genetic and autoimmune mechanisms postulated in its aetiology.

This case raised a number of interesting management challenges for the team. Initially the possibility of a psychosomatic cause had been suspected but continued investigation revealed achalasia. Once diagnosed, the challenges of re-feeding – both practically and physiologically – had to be overcome.

Practical and ethical management challenges

The outcome of this lady's health hinged on her refusal of NGT feeding. There is a locally held belief that the insertion of an NGT heralds the end of life, such that families may refuse to accept the placement of a tube, or as in this case, the patient took her own action to remove it. Ethically it is her right to refuse treatment, but only when she can have been considered to have full knowledge of the benefits and disadvantages of that decision. At the time of initial withdrawal of the tube she was too weak to converse so it

was not possible to gain informed consent. Thus the parents (as next of kin) were called in, and it was explained to them that the nasogastric feeding, rather than signalling death was in fact the only possible route back to life. Initially the mother wanted the patient discharged due to prolonged hospital stay. She also had a young baby of her own (as well as that of the patient) to care for. After discussion, her parents came to provide consent to NGT reinsertion.

Physiological management challenges

Wernicke's encephalopathy is a well-recognised complication of thiamine deficiency and has been described in re-feeding syndrome. It is a neurological disorder characterised by a triad of confusion, ataxia and ophthalmoplegia, which tends to be the result of prolonged starvation or, as is more common in the Western world, chronic alcoholism. Treatment is urgent Thiamine replenishment to prevent Korsakoff's syndrome, where memory loss and confabulation feature.

Learning points

- Organic causes must be excluded where psychogenic/psychiatric diagnoses are being queried
- Achalasia should be considered in anyone who presents with recurrent vomiting
- Communication can greatly influence the outcomes of care

in hospital

- Re-feeding in severe malnutrition, a syndrome may occur which can manifest with thiamine deficiency.

References

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