An executive with poor vision, weight loss and stress

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A former business executive who is smoking and drinking excessively presents to the emergency department with visual loss and poor nutritional status. Despite the entreaties of family and friends, he repeatedly refused support and medical management.

PHOTOLIBRARY

As a GP also working shifts in the local hospital's emergency department, you sometimes find people you know socially ask to be looked after by you when they have to attend the department.

The unexpected state of the patient

While on duty one day in the emergency department, you get a phone call from an ophthalmologist regarding a patient who asked him to call you rather than the patient's own GP.

The patient is a 51-year-old former business executive, whom you know and until a few years ago saw regularly on a social basis. He had been referred to the ophthalmologist by the optometrist he had consulted for new glasses. The ophthalmologist expresses concern about the vision and general health of the man, who he is worried may have toxic optic neuropathy as well as possibly something more serious, such as a cancer.

You agree to see the patient, emphasising that he is likely to be a low priority patient and all that probably will be achieved is to start him on the diagnostic management pathway, which will have to include his own GP.

When the patient attends, he is allocated by the triage sister to the triage category of 3 (urgent: to be seen within 30 minutes) because of his poor vision and feeble state. At his request, the sister also contacts you.

You are somewhat surprised by the patient's poor state when you see him. He tells you the details of his downward spiral – total loss of self-esteem, disastrous business venture, inability to get a job, a hostile family situation and, between the lines, little food and copious alcohol and cigarettes. When questioned about regular medical care, he admits to making occasional single visits to different doctors for chest infections and viral illnesses. He is happy to be assessed and seen in the queue as long as you keep an eye on him.

Toxic and nutritional optic neuropathies[1]

Toxic and nutritional optic neuropathies are rare progressive and painless reductions in visual acuity. The vision loss is usually bilateral.

Toxic optic neuropathy has been associated with lead, ethylene glycol, tobacco use, arsenic and various therapeutic agents, including cisplatin. It is sometimes misnamed toxic amblyopia.

Nutritional optic neuropathy is caused by nutritional deficiencies, especially of vitamin B_{12}, and is sometimes misnamed nutritional amblyopia. Alcoholics are particularly susceptible, although the cause is more likely to be malnutrition resulting from excessive alcohol use rather than the alcohol itself. Restoration of at least some vision is usually achievable by treatment with dietary supplements, although if the cause is vitamin B_{12} deficiency, injections of supplemental vitamin B_{12} are required.

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continued

Table. The patient's blood test results

Blood test	The patient's result	Normal range
Full blood count		
WBC	7.4 x 10^9/L	4.0 - 11.0 x 10^9/L
RBC	3.4 x 10^{12}/L*	4.5 - 6.5 x 10^{12}/L
Haemoglobin	121 g/L*	130 - 180 g/L
Haematocrit	0.35*	0.40 - 0.54
Mean corpuscular volume	104 fL*	76 - 96 fL
Platelets	268 x 10^9/L	150 - 400 x 10^9/L
ESR	45 mm/h*	0 - 10 mm/h
C-reactive protein	52.1 mg/L*	<10.0 mg/L
Anaemia tests		
Serum vitamin B_{12}	481 pmol/L	>126 pmol/L
Serum folate	20.7 nmol/L	6.8 - 38.5 nmol/L
Ferritin	1665 µg/L*	30 - 400 µg/L
Serum iron	8.6 µmol/L*	10.0 - 30.0 µmol/L
Transferrin	1.5 g/L*	2.0 - 3.5 g/L
Transferrin saturation	23%	15 - 50%
Blood chemistry		
Sodium	130 mmol/L*	137 - 146 mmol/L
Potassium	3.5 mmol/L	3.5 - 5.0 mmol/L
Chloride	90 mmol/L*	95 - 105 mmol/L
Bicarbonate	27 mmol/L	24 - 31 mmol/L
Urea	1.1 mmol/L*	3.0 - 8.5 mmol/L
Creatinine	0.07 mmol/L	0.06 - 0.12 mmol/L
Glucose (random)	6.3 mmol/L	3.0 - 7.8 mmol/L
Osmolality	271 mmol/kg*	275 - 295 mmol/kg
Phosphate	0.79 mmol/L	0.70 - 1.40 mmol/L
Magnesium	0.65 mmol/L*	0.70 - 1.05 mmol/L
Calcium, total	2.46 mmol/L	2.10 - 2.60 mmol/L
Albumin	41 g/L	36 - 47 g/L
Protein, total	77 g/L	66 - 82 g/L
Bilirubin, total	15 µmol/L	<18 µmol/L
ALT	46 U/L*	<30 U/L
ALP	175 U/L*	30 - 100 U/L
GGT	674 U/L*	<35 U/L
Creatine kinase	<30 U/L	<130 U/L
Lipase	426 U/L*	<60 U/L
Amylase	147 U/L*	<100 U/L
TSH	2.00 mIU/L	0.30 - 4.00 mIU/L
Alpha-fetoprotein	4.7 kIU/L	<15.0 kIU/L

* indicates abnormal result. Abbreviations: ALT = alanine aminotransferase; ALP = alkaline phosphatase; GGT = gamma-glutamyltransferase; TSH = thyroid stimulating hormone.

The patient's problems

The file records the patient's presenting complaint as 'possible legal blindness, weight loss, referred by ophthalmologist'. Surprisingly, the history reveals very little specifically, only 'decreasing vision, nearly blind at night, lethargy and weight loss'. The systems review does not elicit other CNS symptoms such as headaches or blackouts. A background history of 40 to 50 cigarettes a day over 30 years is given. He is now smoking more, and drinks wine continuously, on a daily basis. He is on no medication, has no allergies and, except for a fractured sternum from a yachting accident two years ago, has no history of medical or surgical illnesses.

Detailed physical examination reveals no specific signs apart from low weight (42 kg; height, 164 cm; BMI, 15.6 kg/m^2). There are no focal CNS signs, no stigmata of chronic liver disease and fundoscopy is unremarkable. No pathology is shown on ECG and chest x-ray.

Abnormal blood results

The patient's blood tests are quite abnormal (Table), especially when compared with those recorded two years ago when he had fractured his sternum . The resident looking after the patient asks you to help interpret these results. Apart from the abnormalities in the levels of the liver and pancreas enzymes (increased alkaline phosphatase, alanine aminotransferase and gamma-glutamyltransferase, and increased amylase and lipase) consistent with excessive alcohol intake, neither you nor the resident can put a simple label on the laboratory picture.

Provisional diagnosis

Given the patient's history of excessive alcohol intake and smoking, it is thought likely that he may indeed have toxic optic neuropathy, with the underlying cause being vitamin B deficiency – that is, nutritional optic neuropathy (see the box on page 53).

Vitamin deficiency, particularly of folate (vitamin B_9) or vitamins B_6 or B_{12}, is one of the common harms related to alcohol misuse and excessive smoking. Although night blindness is suggestive of vitamin A deficiency, the history of alcohol abuse made it more likely that the patient had a vitamin B deficiency.

Gastroenterology review

The gastroenterology registrar elicits no additional history or signs but agrees that nutritional deficiency and/or underlying malignancy may be causing the optic neuropathy. An abdominal CT is organised, as well as review by the gastroenterology consultant on his round later that afternoon. The imaging reveals only a fatty liver.

The gastroenterology consultant agrees that the optic neuropathy may be due to nutritional deficiencies. He orders discharge of the patient, who is given multivitamin (folate and other B group vitamins) tablets to take with him and a vitamin B_{12} injection before discharge. Follow up and endoscopy are planned.

The sad outcome

Over the next months, the patient attended his GP only once. Follow up by the ophthalmologist revealed no improvement in his vision.

You hear much later from a mutual friend that the patient's work and family situation became too much for him. Seriously depressed and with a legal blindness certificate, he left his family and continued to drink and smoke until his death in a coastal country town.

In spite of all entreaties and manipulation by his friends, he had repeatedly and staunchly refused support and antidepressive management. MT

Reference

1. Optic neuropathy. Merck Manual Home Edition. www.merck.com/mmhe/sec20/ch235/ch235d.html (accessed December 2006).

DECLARATION OF INTEREST: None.