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Failure to diagnose: Addison disease

On 11 May 2007, the State Coroner of Western Australia, made a number of recommendations regarding the recognition of serious illnesses in a general practice setting, and the supervision of medical practitioners undertaking prevocational training. The recommendations arose out of a Coronial Inquest into the death of a 16 year old patient from Addison disease.¹ This article discusses the case and the recommendations made by the coroner.

Case history

The patient, 16 years of age, saw her general practitioner on 18 November 2003. She was complaining of a sore throat. The GP noted 'pus +++ tonsils +++ dry lips ++'. The GP thought the patient may be suffering from glandular fever but did not order any blood tests at this time. He prescribed antibiotics and the patient's condition improved.

On 3 April 2004, the patient attended her year 12 school ball. At this time she was reasonably well. Her weight was about 49 kg. Not long after the ball however, the patient's health deteriorated. The patient's mother took her to the GP on 24 May 2004. The mother reported that her daughter was picking up 'more bugs' on a regular basis and that she had lost a significant amount of weight. The GP felt that the patient's symptoms were related to glandular fever, which he believed she had suffered from in November 2003. He ordered blood tests, which were normal. The GP advised the patient's mother of the test results by phone on 27 May 2004.

On 10 June 2004, the patient's mother took her to see another GP at the practice. The GP noted that the patient had stopped attending school at the end of May because she was so tired. She noted in the records:

'Typical day – wakes up at 11.30, still feels tired, then will have some breakfast and usually fall asleep on the couch. The most energy req. activity in last 1 month is – cooking herself a pasta meal. Then, totally exhausted will sleep more in pm, then eat some dinner. Goes to bed at 11 pm – latest. Not able to concentrate...Used to weigh 45 kg. Now weighs 42 kg'.

The patient re-attended the GP on 18 June 2004 at which time the GP recorded:

'Feels better now after taking some herbal remedies...Has been trying to do some walks with mum. Going to school is still hard – has organised with teachers to take the rest of the term off – 2 weeks. Will go back next term'.

At this stage, the GP felt the patient might be suffering from depression and wrote a letter of referral to the mental health team asking for her to be assessed. The GP told that patient that she thought her problems were related to glandular fever and not chronic fatigue syndrome.

On 7 July 2004, the patient was seen at the mental health service who advised that although the patient presented with some symptoms of mild depression, these could just as easily be attributed to her physical health issues.

The patient's father became increasingly concerned about his daughter's health and made another appointment at the practice on 20 September 2004. On that occasion they saw Dr W who was a postgraduate year two trainee who had been seconded from his position as a resident medical officer at a teaching hospital to the general practice. Dr W was supervised by the other GPs within the practice. There was always another GP on duty while Dr W was consulting and there was a rostered doctor as a 'second on call' when Dr W was covering after hours from whom he could seek advice. Dr W doubted the diagnosis of glandular fever because of the length of time that the patient had been unwell. He performed a physical examination, which he

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noted was unremarkable. He did not weigh the patient. The patient reported occasional vomiting which occurred 3–5 times per week, general weakness and lethargy. These symptoms were thought to have commenced around November 2003. According to the patient's father, there was some discussion about changes to the patient's skin colour. Dr W apparently did not appear to be concerned about the patient's skin colour and stated that if she had liver problems this would cause yellowing of the eyes. Dr W thought that the patient may have been suffering from chronic fatigue syndrome, a depressive illness or an eating disorder such as anorexia nervosa. He ordered screening blood tests and a chest X-ray. The patient fainted while having her blood taken. She was unable to sit up without feeling dizzy for about 30 minutes after her collapse. Dr W asked one of the senior GPs for advice about how to manage the situation. He was told to give the patient something to drink and to send her home. The patient remained too dizzy to walk to her father's car and arrangements were made for an ambulance to transport her to the local hospital where Dr W arranged for the emergency department nurse to provide intravenous (IV) rehydration. The nurse was unable to obtain IV access and Dr W attended the hospital to site the IV cannula. Dr W subsequently received a phone call from the ED nurse who said that IV fluids had been completed. The patient had eaten something and was feeling better. She had passed urine and was able to walk without fainting. Dr W advised that the patient could go home and should see him later that week to obtain the results of the blood tests. The patient remained unwell but refused her parents' requests to go to hospital for review. On 29 September, as the patient remained unwell her parents decided to make an appointment for her to see a doctor at the practice. Her mother took two of her children to school and her husband to work while one other child remained at home with the patient. On her return home the patient's mother was on the phone to the practice when she went into the patient's bedroom and found that she had died. A postmortem examination concluded that the cause of death was adrenal insufficiency. The patient's weight at death was 38 kg.

An inquest was held to determine how the patient died from a treatable condition and why that condition was not diagnosed and treated. An endocrinologist gave evidence at the inquest. He stated that Addison disease is a rare disorder. While all medical graduates would have some awareness of Addison disease, only a small minority of GPs would have had any direct experience of the diagnosis or management of the disorder. The endocrinologist noted that the patient's blood tests did not show a marked reduction in sodium concentration and elevated potassium levels, which are often present in severe adrenocortical insufficiency, but he commented that these changes are not always seen. Otherwise the endocrinologist felt that the patient's presentation was a typical case of Addison disease with the history of chronic fatigue, loss of weight and ultimately changes in skin pigmentation. The endocrinologist noted that it appeared that the doctors who saw the patient had not fully appreciated the severity of her illness and, in particular, the severity of her weakness, the full extent of her weight loss and the history of repeated vomiting. By 20 September 2004, the endocrinologist stated that the patient was critically ill and in urgent

need of investigation and treatment. He concluded that the patient had a serious unexplained illness that required more intensive and more expert assessment than it received, which should have led to the correct diagnosis and commencement of effective treatment. That said, the endocrinologist recognised that Dr W, the last doctor who saw the patient, was a very inexperienced medical practitioner of only 2 years postqualification practice who was not well placed to identify the patient's rare condition.

Discussion and risk management strategies

Clinical features of Addison disease include:

- lethargy/excessive fatigue
- anorexia and nausea
- diarrhoea/abdominal pain
- weight loss
- dizziness/funny turns
 - hypoglycaemia (rare)
 - postural hypotension (common)
- hyperpigmentation, especially mucous membranes of mouth and hard palate.

Diagnostic features include:

- fatigue + anorexia/nausea/vomiting + abdominal pain (+/- skin discolouration) = Addison disease.

The synacthen stimulation test is the definitive test for the diagnosis of Addison disease. Elevated serum potassium, low serum sodium and low plasma cortisol levels may also be present.²

The coroner concluded that this case highlighted the tragic consequences that can result from failure to diagnose Addison disease, a rare but potentially fatal illness. The coroner made the following recommendations:

- the arrangements in place to give prevocational doctors experience in general practice should include specified criteria for referral of very unwell patients seen by those doctors to those responsible for supervising their work. Such referrals should occur on a regular basis and should include, for example, cases where a patient is suffering from an illness that has significantly altered the patient's quality of life or has impacted on the patient's ability to conduct important functions involved in the patient's work, schooling or other activities
- before patients are seen in a general practice by a trainee doctor undertaking prevocational training, patients should be informed of the doctor's medical status and informed consent obtained
- the issues raised by this case should be discussed with relevant GP bodies and the health department, with particular

emphasis on the management of patients with serious undiagnosed illness. The outcomes of these discussions should be brought to the attention of all GPs and relevant specialist groups

- steps should be taken to ensure that GPs seek and are able to obtain assistance from relevant specialists when managing patients with serious undiagnosed illness. This can be done by patient referral, telephone contact, or patient transfer to an appropriately staffed hospital. All practices should have a mechanism that is well understood by all medical and nursing staff for obtaining urgent specialist assistance when necessary. Barriers to specialist access for metropolitan and rural practitioners should be identified and addressed
- particular attention should be given to developing mechanisms to ensure that serious illness is recognised by GPs and nursing staff. These should include a range of continuing educational activities.

Conflict of interest: none.

References

1. Inquest into the death of Emma Louise Robson; Ref No:14/07. State Coroner, Western Australia, 11 May 2007.
2. Murtagh J. General practice. 3rd edn. Sydney: McGraw Hill, 2003.

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