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Independent learning program for GPs

Unit 575
September 2020

Chronic conditions

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We acknowledge the Traditional Custodians of the lands and seas on which we work and live, and pay our respects to Elders, past, present and future.

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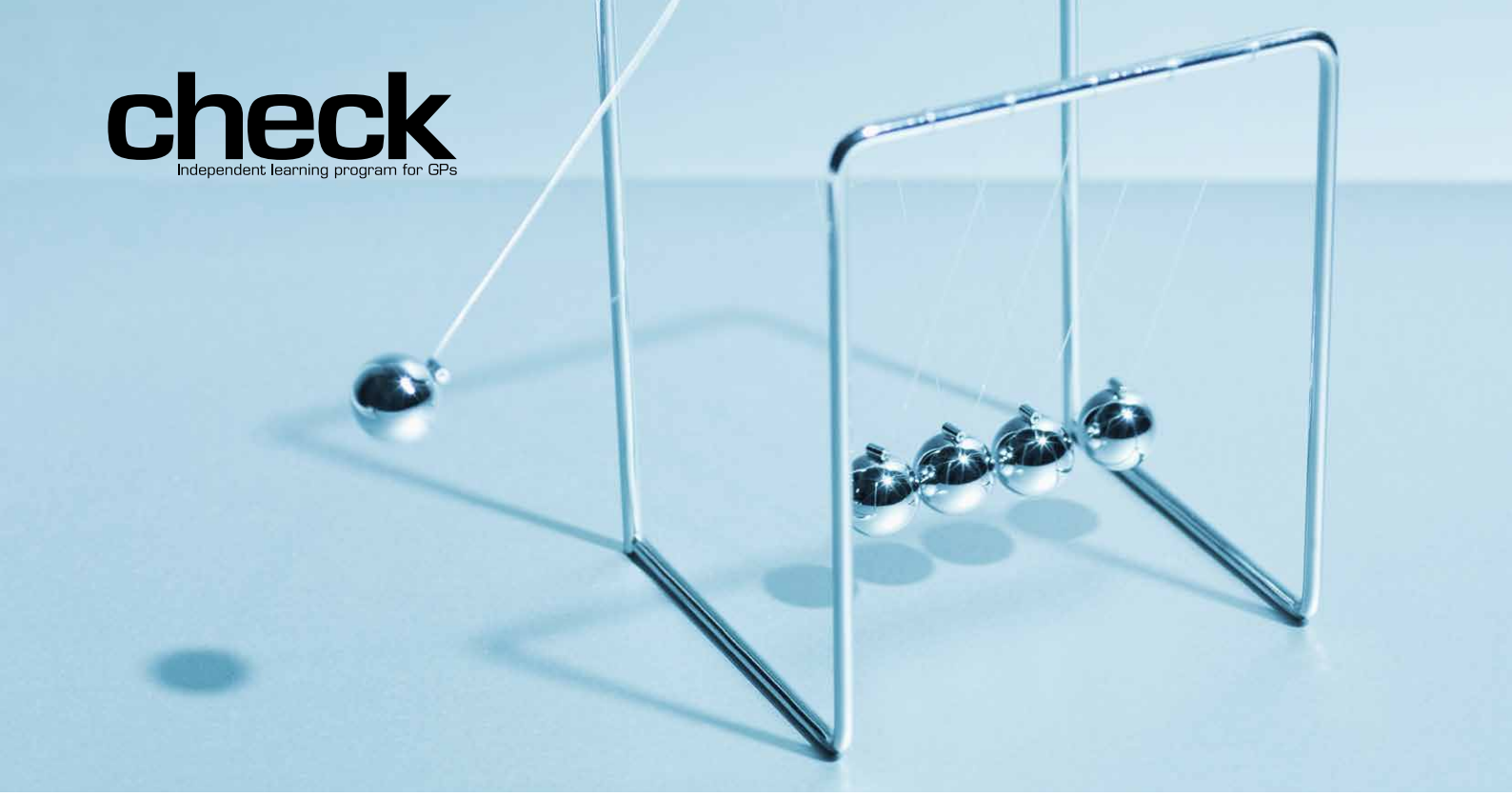

Chronic conditions

Unit 575 September 2020

The five domains of general practice

- Communication skills and the patient–doctor relationship
- Applied professional knowledge and skills
- Population health and the context of general practice
- Professional and ethical role
- Organisational and legal dimensions

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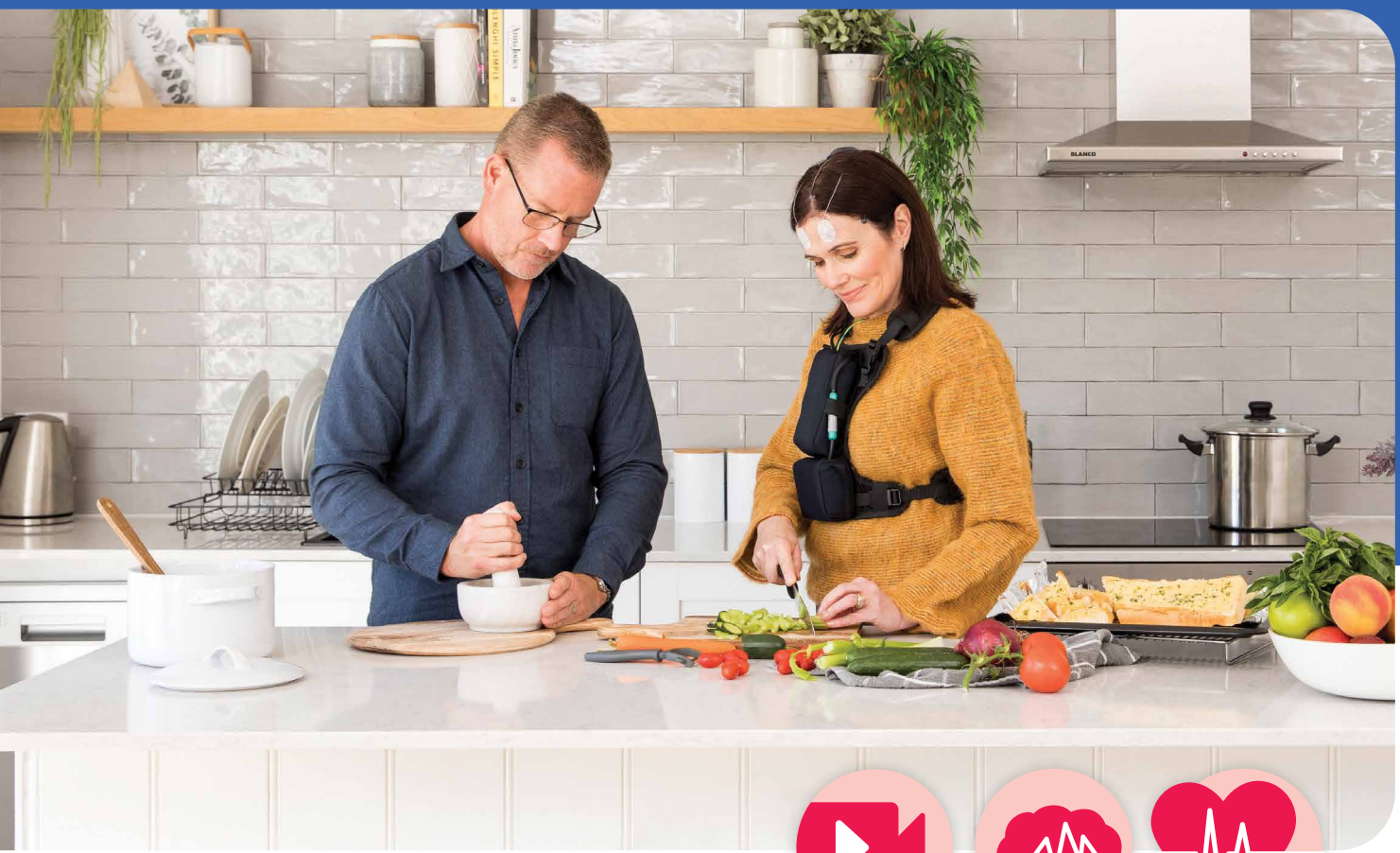

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About this activity

Presentations relating to chronic conditions are common in the general practice setting, with data showing that 40% of Australian general practice encounters involve the management of at least one chronic problem.[1]

The prevalence of cerebral palsy in Australia is 1.4 per 1000 live births.[2] General practitioners (GPs) have an important role in the care of patients with cerebral palsy as they transition to adult services, because the GP may be the only health professional who has known the patient since childhood.[3]

It is estimated that more than 142,000 people in Australia are currently living with active epilepsy.[4] Approximately 3–4% of Australians develop epilepsy throughout their lives, and the condition affects people of all ages and genders.[4]

Approximately 100 Australians are diagnosed with Addison's disease each year, and the condition is thought to affect an estimated 2500 people.[5] GPs can assist with the management of ongoing therapy and educating the patient on what to do in case of an adrenal crisis.

The incidence of bronchiectasis in Australia is unknown; however, it is more prevalent in patients living in low socioeconomic areas and in remote Aboriginal and Torres Strait Islander communities.[6] It is thought that improved access to healthcare, medication adherence and medical follow-up would contribute to decreased rates of bronchiectasis.[6]

This edition of *check* considers the investigation and management of chronic conditions in general practice.

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Learning outcomes

At the end of this activity, participants will be able to:

- outline the investigations and corresponding results that would lead to a diagnosis of bronchiectasis
- identify common causes of pain in patients with cerebral palsy
- discuss the management of Addison's disease
- describe the process of diagnosing epilepsy in a patient presenting after their first seizure.

Authors

Case 1

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Case 4

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Abbreviations

AAC	augmentative and alternative communication
ACTH	adrenocorticotrophic hormone
BMI	body mass index
bpm	beats per minute
CAP	community-acquired pneumonia
CT	computed tomography
DHEAS	dehydroepiandrosterone sulfate
EEG	electroencephalography
GMFCS	Gross Motor Function Classification System
GORD	gastro-oesophageal reflux disease
GP	general practitioner
HRCT	high-resolution computed tomography
Ig	immunoglobulin
IV	intravenous
LCQ	Leicester Cough Questionnaire
LFT	liver function test
MR	Medium Rigid
MRI	magnetic resonance imaging
NDIS	National Disability Insurance Scheme
PEG	percutaneous endoscopic gastrostomy
SUDEP	sudden unexpected death in epilepsy
TFT	thyroid function test

CASE

1 Eniola has a cough

Eniola, aged 68 years, is a retired accountant who presents with a cough. She reports being frustrated and embarrassed by the cough, which seems to have come and gone since she was hospitalised with pneumonia three years ago. Eniola says that the cough has been getting worse over the past month. She has a past medical history of asthma but is generally well. She is a non-smoker and currently uses a fluticasone propionate/salmeterol 250/25 μg metered dose inhaler twice per day and salbutamol 100 μg as needed.

Question 1

In regard to Eniola's cough, what further history would you like to know?

Further information

Reviewing her notes, you see that Eniola was seen by one of your colleagues three weeks ago. Physical examination at the time recorded a temperature of 38.1°C, bilateral wheeze in the lower zones, an oxygen saturation level of 98% and no increased work of breathing. Eniola was treated for an infective exacerbation of asthma and given a course of amoxicillin 500 mg three times per day and prednisolone 50 mg daily for a week in addition to her usual asthma management. The previous year, she underwent a spirometry test that demonstrated a reversible obstructive pattern with an increase in forced expiratory volume in one second of 13% post-bronchodilator.

Eniola reports that the initial treatment with amoxicillin and prednisolone resulted in mild improvement of symptoms; however, she continues to have a cough with shortness of breath and daily sputum production. She has been using her inhaler on multiple occasions daily and has been unable to complete her daily walk without requiring her inhaler. She has had persistent rhinorrhoea but does not report any haemoptysis, weight loss, chest pain or other symptoms. On examination today, Eniola's temperature is 37.8°C with an oxygen saturation of 98% on room air, a respiratory rate of 12 breaths per minute and a right lower zone wheeze. Nasal examination shows no abnormalities and equal airflow from both nostrils.

Question 2

What further investigations would you order?

Further information

The investigation results are reported in Table 1. The sputum result is still pending.

Table 1. Full blood examination results

	Result	Reference range
Haemoglobin	152 g/L	110–160 g/L
White cell count	**11.2 × 10^9/L**	3.5–10 × 10^9/L
Neutrophils	**7.62 × 10^9/L**	1.5–6.5 × 10^9/L
Lymphocytes	2.67 × 10^9/L	0.8–4.0 × 10^9/L
Monocytes	0.82 × 10^9/L	0–0.9 × 10^9/L
Eosinophils	0.11 × 10^9/L	0–0.6 × 10^9/L
Basophils	0.01 × 10^9/L	0–0.15 × 10^9/L
Platelets	236 × 10^9/L	150–400 × 10^9/L
Sodium	141 mmol/L	135–145 mmol/L
Potassium	4.2 mmol/L	3.5–5.5 mmol/L
Chloride	107 mmol/L	95–110 mmol/L
Bicarbonate	25 mmol/L	20–32 mmol/L
Urea	3.4 mmol/L	3.0–8.5 mmol/L
Creatinine	59 mmol/L	45–85 mmol/L
Estimated glomerular filtration rate	>90 mL/min/1.73 m^2	>59 mL/min/1.73 m^2
Random glucose	4.7 mmol/L	3.6–7.7 mmol/L
Total protein	**82 g/L**	63–80 g/L
Albumin	39 g/L	32–44 g/L
T-bilirubin	7 μmol/L	<16 μmol/L
Alkaline phosphatase	78 U/L	30–115 U/L
Aspartate aminotransferase	22 U/L	10–35 U/L
Alanine aminotransferase	14 U/L	5–30 U/L
Gamma-glutamyl transferase	26 U/L	5–35 U/L

Eniola's chest X-rays show focal consolidation and collapse in the right lower lobe (Figure 1).

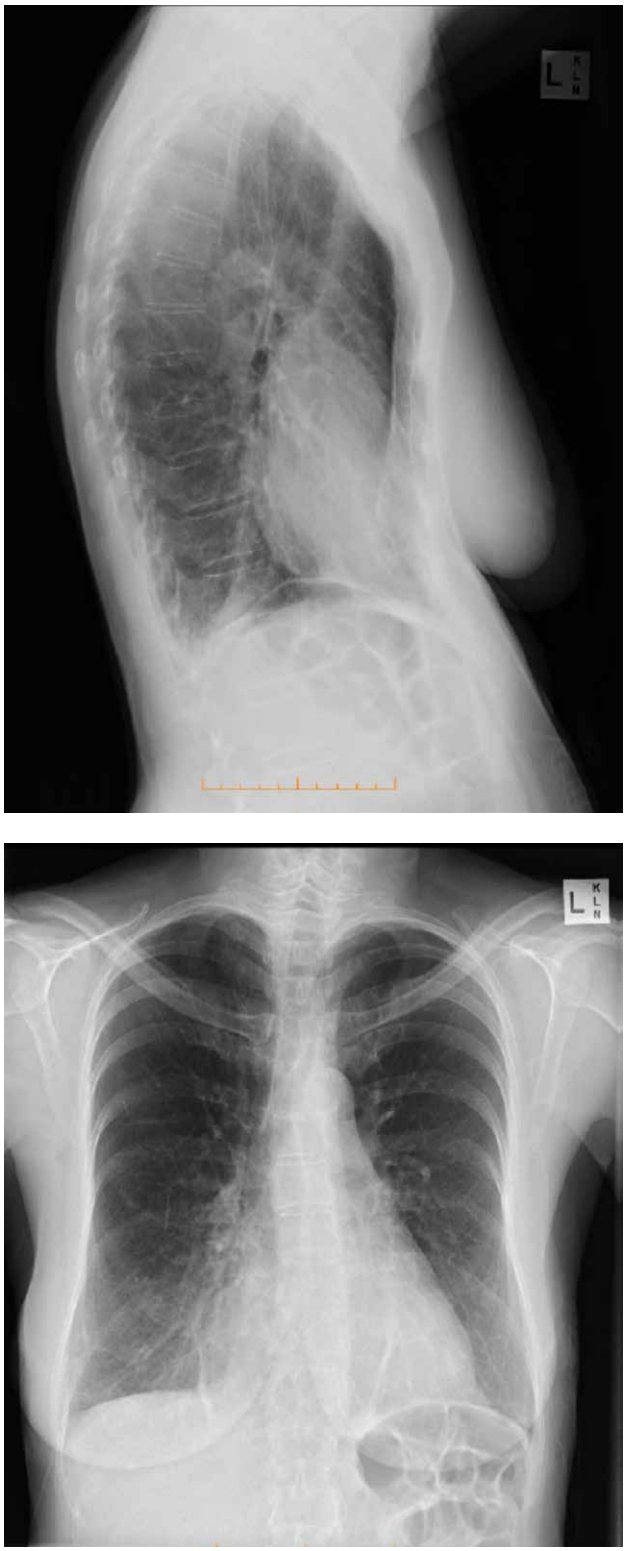

Figure 1. Initial chest X-ray

Question 3

How would you manage Eniola's condition?

Question 4

When would you follow up with Eniola to assess her response to treatment? If Eniola fails to improve following management, what alternative diagnosis should be considered?

Further information

Eniola fails to attend her review in 48 hours but presents for review one month later. Her sputum was positive for leucocytes and gram-negative bacilli. She reports ongoing symptoms of cough, progressive fatigue and production of more viscous sputum, which is darker in appearance than previously**.** Yesterday she coughed up enough 'muck' to fill a small glass. She has a temperature of 37.4°C. Respiratory examination reveals a bilateral wheeze. You repeat her chest X-ray (Figure 2), which shows wedge-based opacification in the right lower lobe that is similar in appearance to the initial chest X-ray (Figure 1). The lungs and pleural recesses are otherwise clear. The cardiac contour is normal. The radiologist also comments that there may be a degree of bronchiectasis as suggested by dilated bronchi with wall thickening (tram track sign).

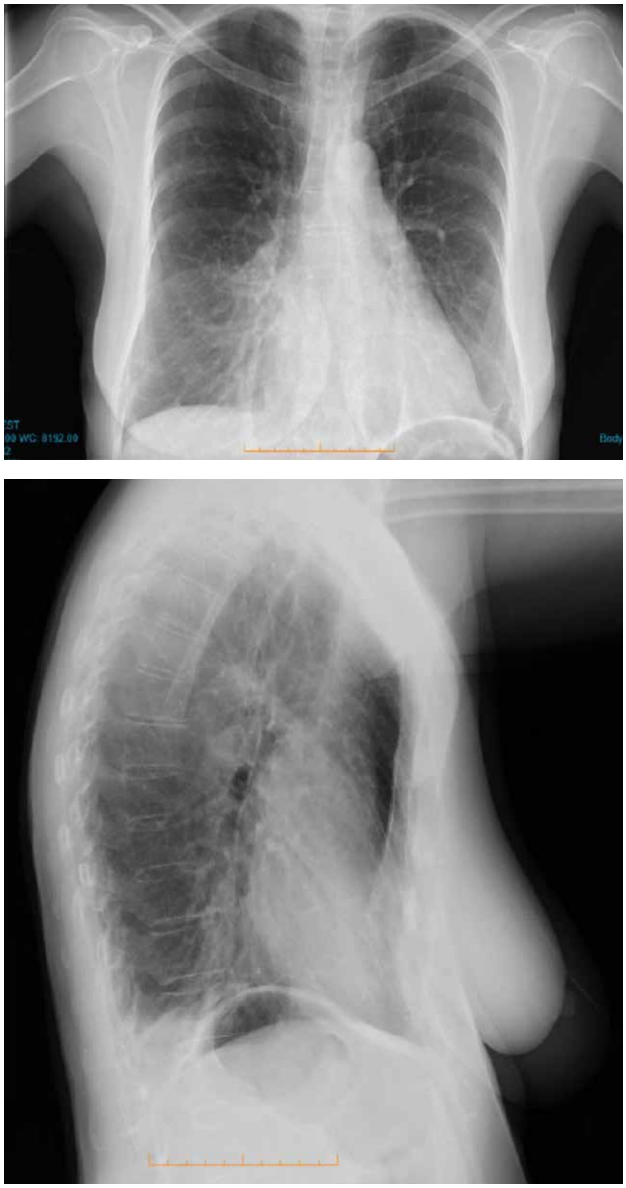

Figure 2. Follow-up chest X-ray

Question 5

To further evaluate the possibility of bronchiectasis, what imaging would you arrange for Eniola?

Question 6

What further investigations should be considered at this stage?

Further information

Eniola undergoes further investigations. Her full blood examination, serum immunoglobulin (Ig) levels and *Aspergillus* serology are unremarkable. Sputum culture grows gram-negative bacilli but does not grow mycobacteria. No clear underlying cause for Eniola's bronchiectasis can be identified.

Question 7

How would you manage Eniola's acute exacerbation of bronchiectasis?

Question 8

What further long-term management strategies should be initiated?

CASE 1 Answers

Answer 1

Cough is one of the most common presenting conditions in primary healthcare, and it is vital to obtain a detailed history.[1] Chronic cough is defined as lasting more than eight weeks, and determining its cause and management can be clinically challenging because of the broad range of possible causes including infections, autoimmune reactions, gastrointestinal causes and as a result of medication.[1,2] Consideration should be given to the four most common aetiologies (upper airway cough syndrome, asthma, gastro-oesophageal reflux disease and non-asthmatic eosinophilic bronchitis).[3]

To guide investigations accurately, Eniola should be asked about the following:

- detailed history of her cough including duration; frequency; aggravating, relieving or precipitating factors
- any red-flag symptoms that suggest a sinister cause of cough including night sweats, unexplained weight loss, haemoptysis or a new hoarseness of voice[2]
- details about sputum, if productive cough (ie volume, appearance/colour, additions to the sputum)
- previously tried measures and their effect
- associated symptoms noted such as sinus problems, fever, chest pain, dyspnoea or wheezing, gastro-oesophageal reflux disease symptoms
- past history of medical conditions, particularly lung problems experienced by Eniola or any family members
- occupational or environmental factors
- alcohol, smoking and drug use.

Answer 2

A typical work-up for investigation for a suspected lower respiratory tract infection would include:

- full blood examination
- urea and electrolytes
- creatinine and estimated glomerular filtration rate
- liver function tests
- sputum – microscopy, culture and sensitivity, and acid-fast bacteria culture
- chest X-ray.

Although a chest X-ray is not always diagnostic, it is a useful tool to refine a differential diagnosis.[2] A chest X-ray is indicated when a clear cause of the cough cannot be determined using history and examination alone.[3] It is also recommended that a validated tool be used to assess the impact of the cough on the patient's life as well as assessing treatment effects.[4]

Answer 3

A diagnosis of partially treated community-acquired pneumonia (CAP) should be considered. [5] The clinical syndrome of productive cough and dyspnoea combined with a chest X-ray showing focal consolidation is a typical finding in CAP. As initial monotherapy with amoxicillin was only partially effective, Therapeutic Guidelines recommend amoxicillin 1 g eight-hourly plus doxycycline 100 mg 12-hourly for 5–7 days.[6]

Answer 4

Timely follow-up and review 48 hours after commencing treatment should be arranged. Failure to improve should prompt consideration of conditions that mimic CAP, including:[7]

- heart failure
- lung cancer
- pulmonary embolism
- bronchiectasis
- vasculitis
- collagen vascular disease
- interstitial lung diseases
- aspiration or chemical pneumonitis
- medication reactions.

Answer 5

High-resolution computed tomography (HRCT) of the chest is considered the gold standard for the radiological diagnosis of bronchiectasis.[8] Plain chest X-rays are relatively insensitive when compared with HRCT. Chest X-rays have a reported sensitivity of 88% and specificity of 74% and are often normal or show non-specific findings.[9] In comparison, HRCT has a sensitivity of 95% and a specificity approaching 100%.[10,11]

Answer 6

Bronchiectasis is a disease characterised morphologically by the permanent dilatation of bronchi and bronchioles, and clinically by recurrent or persistent bronchial infection, cough and often sputum.[12]

Investigations to determine the cause of bronchiectasis are considered best practice as identification of aetiology can significantly change management. For up to 50% of patients, however, no clear cause can be found.[13]

For all patients, the Thoracic Society states it is necessary to obtain:[8,14]

- full blood examination
- *Aspergillus* serology
- serum IgE, IgA, IgM, IgG
- sputum with routine and mycobacterial culture
- spirometry.

Other additional testing to determine an underlying cause may be undertaken in specific patients in conjunction with respiratory specialist input. These include bronchoscopy; cystic fibrosis screening; testing for human immunodeficiency virus, human T-cell leukaemia virus type 1 and alpha-1-antitrypsin deficiency; and additional immunological testing.[8,14]

Answer 7

An acute exacerbation of bronchiectasis is defined as '>72 hours of increased cough, change in sputum, breathlessness, haemoptysis and/or constitutional upset'.[15]

The goal of antibiotic therapy is to reduce the bacterial load and decrease inflammation.[16] According to the Therapeutic Guidelines, antibiotics are not indicated unless all three of the following clinical features are present:[12]

- increased sputum production
- increased sputum purulence
- increased cough, which may be associated with wheeze, haemoptysis or breathlessness.

A sputum sample should be taken for all patients during an acute exacerbation to guide therapy if the patient fails to improve with empiric antibiotics. For non-severe exacerbations, treatment with amoxicillin 1 g three times daily or doxycycline 100 mg twice daily should continue for 14 days. Treatment duration can be shortened to 10 days if the response to antibiotics is rapid and the exacerbation is not caused by a resistant isolate.[12]

Answer 8

Bronchiectasis is a complex disease entity that requires multidisciplinary management. The evidence supporting the management of bronchiectasis is limited, and guidelines are informed mostly by expert opinion;[17] however, early and effective management reduces morbidity in both the short and long term.[18]

The general aim of treatment is to increase quality of life, preserve lung function, reduce frequency of exacerbations and achieve improved symptom control.[15]

This is achieved through the following strategies:[14,19,20]

- Airway clearance through the use of strategies such as the active cycle of breathing technique and the forced expiration technique (huff). Physiotherapists can further guide and individualise treatment.
- Reduction of the risk of further infective exacerbations by offering influenza and pneumonia immunisations.
- Prompt treatment of exacerbations with sputum surveillance. Patients should be offered pulmonary rehabilitation following an acute episode.
- Treatment of the underlying cause when known.
- Smoking cessation counselling, when appropriate.

Patients with bronchiectasis should undergo annual review including:[18]

- oximetry and spirometry
- sputum culture
- assessment and management of comorbidities
- assessment of treatment effects
- assessment of the impact of the disease on the patient
- development and review of a Bronchiectasis Action Plan.

Validated tools to help assess the severity and impact of the illness on the patient include the Bronchiectasis Severity Index [21] and the Leicester Cough Questionnaire (LCQ).[22]

Conclusion

Eniola returns, after two months, experiencing her third exacerbation this year. You refer her to a respiratory physician to consider further treatment. The respiratory physician repeats the sputum investigation to confirm there is no nontuberculous mycobacterium infection. After discussing the increased risk of acquiring antibiotic-resistant organisms, Eniola agrees to commence azithromycin 250 mg daily.[23] Eniola returns to your care, and the respiratory physician requests regular sputum collection. Specifically, they request that you re-refer Eniola for review if she has a positive sputum result for *Pseudomonas* spp. for consideration of eradication therapy.[24]

After commencing daily antibiotics, Eniola has less frequent exacerbations but does experience some gastrointestinal side effects. You monitor her LCQ score, which suggests that the antibiotics have not resulted in an improvement in her quality of life.

Summary

Bronchiectasis is characterised by cough with/without sputum, recurrent or persistent bronchial infection and the presence of bronchial wall thickening and bronchial dilatation on HRCT. The diagnosis should be considered in patients with a chronic productive cough.[11] While there is somewhat limited evidence for treatment, patients with bronchiectasis require individualised care and multidisciplinary management including respiratory specialist care when required.[17]

Resources for doctors

- National Institute for Health and Care Excellence (NICE) – Bronchiectasis (acute exacerbation): Antimicrobial prescribing, www.nice.org.uk/guidance/ng117/resources/visual-summary-pdf-6606081325
- The Thoracic Society of Australia – Bronchiectasis Toolbox, https://bronchiectasis.com.au

Resources for patients

- Lung Foundation Australia – Bronchiectasis, https://lungfoundation.com.au/patients-carers/conditions/bronchiectasis/overview/

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CASE

2 Madeline isn't happy

Madeline, aged 15 years, comes to your practice with her mother, Katie, who reports that Madeline's behaviour has changed over the past few weeks. She seems 'grumpy' and often appears to be in pain.

Madeline has cerebral palsy and uses a motorised wheelchair for mobility; she is not able to weight-bear, stand or walk. Madeline does not use speech to communicate. You find it difficult to judge her level of understanding and cognition.

You have seen Madeline intermittently, mostly for coughs, colds and immunisations. Much of her care in childhood was coordinated by her paediatrician, whom she has not seen for more than four years. Katie tells you she stopped attending follow-up appointments with the paediatrician because she did not find the appointments useful; the focus seemed to be only on weight and hips.

Question 1

What is cerebral palsy?

Question 2

From whom would you obtain further history?

Question 3

What further information would you seek?

Further information

Madeline uses an iPad at school but has no communication device or aid of her own. Katie reports that Madeline grimaces and vocalises when moved. Madeline is not able to indicate where she is experiencing pain, and she appears unsettled and uncomfortable in her wheelchair. Currently her only medication is lamotrigine 200 mg twice daily prescribed for epilepsy. Madeline has not gained weight for five years. Mealtimes are slow and appear to be associated with discomfort.

Question 4

What would you focus on in your examination?

Further information

Examination reveals Madeline is an alert teenager who responds appropriately to questions by nodding and shaking her head.

Madeline looks uncomfortable. Her posture is slumped and she sits asymmetrically, leaning to her right side in her wheelchair. She has multiple contractures of her upper and lower limbs.

Madeline's weight is 25 kg and her length is 152 cm. Her body mass index (BMI) is 10.8 kg/m^2. Her previous weight measurement four years ago was 25 kg.

Her chest is clear. Abdominal examination identifies palpable faecal loading in the descending colon. There are no signs of pubertal development.

Question 5

What would be your working diagnosis and problem list?

Further information

Katie says she spends several hours each day feeding Madeline. If Madeline eats or drinks too quickly, she chokes and coughs. Katie's friend's child, who also has cerebral palsy, had a gastrostomy several years ago. Katie does not want Madeline to have a gastrostomy because she wants Madeline to eat and drink like everyone else.

Katie tells you that Madeline has not seen an orthopaedic surgeon for five years. The surgeon wanted to operate and said Madeline's hip was 'on the way out of its socket'. Katie did not want Madeline to undergo major surgery and could not see the point when Madeline would remain unable to walk.

Madeline is now aged 15 years and has not had her first period.

Question 6

What further investigations are recommended at this point?

Further information

Madeline's test results are as follows:

- mild iron deficiency: haemoglobin 105 g/L (reference range: 120–160 g/L); ferritin 25 µg/L (reference range: 20–220 µg/L)
- vitamin D deficiency: 35 nmol/L (reference range: 50–200 nmol/L)
- liver function tests (LFTs), thyroid function tests (TFTs), and urine, creatinine and electrolytes are within normal ranges.

Her pelvic X-ray confirms a dislocated hip.

Question 7

What is your management plan for Madeline? How can allied health professionals assist in her care?

Further information

After several visits, Katie and Madeline agree to referrals to an orthopaedic surgeon, speech pathologist, dietitian and gastroenterologist.

Madeline undergoes a gastrostomy several months later that results in an immediate gain in weight and increase in her energy levels. She now has more time to participate in social activities. With the increase in weight, Madeline's periods have started.

The orthopaedic surgeon felt that surgery for the dislocated hip was needed but wanted to await improvement in Madeline's nutritional status.

Madeline now has an iPad with a communication app and loves sharing her news and photos of her artwork.

Katie reports Madeline is much happier, although both are apprehensive about the upcoming orthopaedic surgery.

Question 8

What would be your management plan for Madeline's ongoing care?

Further information

One year later, Madeline returns for a routine visit. She is now 16 years of age and considering post-school options. She is not sleeping well and often feels sad, particularly in the morning. She is struggling to complete some of her school assignments and feels her career opportunities are limited. She is not seeing her friends very often.

Question 9

What advice would you provide in relation to her mood?

Question 10

What is the long-term prognosis for Madeline, and how could you help to promote optimal quality of life?

CASE 2 Answers

Answer 1

Cerebral palsy has been defined as 'a group of permanent disorders of the development of movement and posture, causing activity limitation, that are attributed to nonprogressive disturbances that occurred in the developing fetal or infant brain. The motor disorders of cerebral palsy are often accompanied by disturbances of sensation, perception, cognition, communication, and behaviour, by epilepsy, and by secondary musculoskeletal problems.'[1] Aetiology is best defined in terms of causal pathways, the most common involving hypoxia and/or ischaemia as a result of preterm birth and neonatal immaturity or birth asphyxia; perinatal stroke; maldevelopment of the brain from genetic, infective or vascular causes; and genetic/chromosomal syndromes. Seventy-five per cent of cases of cerebral palsy occur as the result of events in the antenatal period. In a substantial number of cases, the pathogenesis and timing of brain injury remains unclear.[2] Cerebral palsy is the most common motor disorder in childhood.

Cerebral palsy can be classified by the:[1]

- severity of the motor impairment (Gross Motor Function Classification System [GMFCS] Levels I–V)
- distribution of the motor signs (hemiplegia, diplegia, quadriplegia)
- movement disorder (spasticity, dyskinesias, ataxia, mixed).

The motor disorder commonly affects mobility, coordination, balance, communication and self-care. Associated learning and other cognitive or sensory impairments may or may not be present.

General practitioners (GPs) play a central part in monitoring and optimising health and wellbeing, and coordinating the multidisciplinary team, which may include medical specialists, dentists, allied health professionals, psychologists, nurses, disability support workers and teachers.

Answer 2

It is recommended to take a history directly from your patient, in this case Madeline, as much as possible. Including her in the conversation is essential. Some people with severe physical disability also have intellectual disability; others have normal or high intelligence.[3] It is important not to make assumptions about someone's cognitive ability on the basis of their physical function.

To obtain a history, it is essential to determine how Madeline communicates. She may use sign language or an augmentative and alternative communication (AAC) aid, such as an electronic device (eg iPad with a communication app), communication board or other aid. If she has a communication aid with her, this should be used throughout the consultation. If she does not, yes/no questions may need to be asked.

If use of a communication aid is not possible, questions can be directed towards Katie, while still paying particular attention to Madeline's verbal/non-verbal cues. It is important to respectfully ask Madeline for consent, explaining that the goal is to understand how best to help her.

Communication aids enable people to speak for themselves and thereby promote agency and enable independence.

Answer 3

Pain

While self-reporting pain is the preferred method of information gathering, observing changes in behaviour is an informative alternative indication of symptoms (Table 1).[4,5]

Table 1. Common causes of pain in people with cerebral palsy[5]

Cause of pain	Signs	Management
Oral/dental pain	Pain on eating, brushing teeth; gum or facial swelling	Dental review and treatment
Gastro-oesophageal reflux disease	Pain after meals and/or when supine (eg in bed); pain on eating; anaemia	Postural management, endoscopy, diet, proton-pump inhibitors
Constipation	Hard, infrequent bowel motions (+/– overflow diarrhoea); pain on defecation; abdominal pain and/or bloating, increased flatus	Diet, fluid intake, regular exercise, stool softeners
Muscle spasm	Localised muscle spasm	Physiotherapy, stretching and exercise, posture support, muscle relaxants
Musculoskeletal injury: strains, sprains, subluxations, dislocations, fractures	Pain on movement; joint swelling or deformity; swelling/bruising; tenderness in limbs	Imaging, rest/splinting, surgical repair *Note: There is an increased risk of osteoporosis for people who do not weight-bear and/or are taking anti-epileptic medications*
Pressure injuries	Skin redness; breakdown in pressure areas from wheelchair or orthotics	Immediately relieve pressure to avoid skin breakdown; correct pressure through modification of equipment

Dental care

Dental disease may go unrecognised because of communication difficulties and is a frequent cause of pain.[6] Preventive dental care may be challenging because of the motor control required for brushing and flossing. Regular dental review is essential.

Cerebral palsy can be associated with other factors affecting oral health including inadequate food clearance, reduced salivation associated with medication, and gastro-oesophageal reflux disease (GORD).[7]

Mealtimes

It is important to ascertain the patient's dietary intake details. Has the patient gained or lost weight? Have there been any changes in behaviour during or after mealtimes?

GORD is common and often undetected in people with cerebral palsy. Late presentation may occur with anaemia, weight loss or Barrett's oesophagus.[8] However, in Madeline's case, Barrett's oesophagus would be extremely unlikely at her age.

If the patient is coughing or choking with food or fluids, or has frequent chest infections, it may indicate poor oromotor coordination with increased risk of aspiration. Aspiration pneumonia is an important cause of morbidity, hospitalisation and death in people with cerebral palsy.[9]

Current medications

Anti-epileptic medication can have adverse effects on mood and behaviour.[10] Nonsteroidal anti-inflammatory drugs can result in heartburn and nausea.[11]

Answer 4

Key aspects of the physical examination include:

- vital signs
- weight/height/length/BMI
- oral/dental disease
- spine – scoliosis
- ear, nose and throat
- eyes (strabismus, visual acuity)
- chest – air entry, signs of infection
- abdomen – tenderness and masses
- signs of pubertal development (Tanner staging)
- musculoskeletal – range of movement, joint deformity, contractures, swelling, bruising
- neurological – muscle tone, strength, coordination, symmetry
- skin – pressure areas, chilblains, peripheral perfusion.

It is also important to observe Madeline's communication (the ways she communicates with you and her mother) and her alertness and interest in participating in the discussion and examination.

Answer 5

Madeline has cerebral palsy; she is unable to sit or stand independently (GMFCS Level V).[12] She is severely underweight and has no independent form of communication.

She also has:

- possible left hip dislocation – the incidence of hip displacement in those with GMFCS Level V is 90% and is usually associated with severe pain[13,14]
- constipation
- delayed puberty.

Answer 6

As Madeline is severely underweight, further investigations recommended include a full blood examination, including iron studies, vitamin B12 and folate. It may also be beneficial to assess the risk to bone health by measuring vitamin D and performing a comprehensive metabolic panel, including urea, creatinine and electrolytes, LFTs and TFTs.

A pelvic X-ray is required to confirm the dislocated hip. X-ray films will also help to explain the situation to Madeline and Katie.

Answer 7

Pain relief

In view of her hip pathology, Madeline needs regular pain relief such as sustained-release paracetamol.

Her position and sling for hoisting need review to ensure that she is as comfortable as possible during transfers.

Referrals

It is important to work closely with Madeline and her mother to gain their trust and to enable appropriate referrals.

Speech pathology

A swallowing assessment can determine the risk of choking and aspiration.

Communication assessment

The selection of communication aids is a specialised area of speech pathology practice. Giving Madeline her own voice will enable her to have an active role in her life and healthcare. Madeline's National Disability Insurance Scheme (NDIS) Support Coordinator can advise about appropriate local speech pathologists. Madeline has been using an iPad at school for learning and communication. The school and speech pathologist can advise about appropriate equipment, which can then be purchased through Madeline's NDIS plan.

Dietitian

A dietitian can design a diet with appropriate consistency to minimise risk of aspiration. They can advise about oral supplements to improve nutritional and caloric intake,[15] and reduce the time taken for meals.

If Madeline is unable to eat safely or efficiently enough to enable adequate intake in a reasonable mealtime, she may need a gastrostomy. Gastrostomy placement has proven benefits for weight gain and health.[16] This is a difficult decision for Madeline and her family, and clear information and support will be needed. Connecting Katie and Madeline with someone who has had a gastrostomy may be helpful.

Gastroenterologist

A gastroenterologist can advise on gastrostomy – usually percutaneous endoscopic gastrostomy (PEG). They can also offer advice for managing constipation and GORD.

Occupational therapy

An occupational therapist can provide immediate modifications to Madeline's wheelchair to improve support and comfort. A full wheelchair assessment for a new chair should not be undertaken until after any orthopaedic surgery.

Orthopaedic surgeon

Madeline's hip is causing considerable pain, and surgery may be necessary. Sensitive discussion with the family is required as this recommendation previously led to a withdrawal from medical care. Hip surveillance in childhood and early surgical treatment may prevent progression.[14]

Rehabilitation physician

A rehabilitation physician can provide management of spasticity including contractures and painful spasms, assess function and determine and advise on ways to maximise independence and improve and maintain quality of life.

Constipation

Constipation in people with cerebral palsy is so common that they and their families may come to accept it as normal. Constipation is, however, a frequent and preventable cause of discomfort and pain and is associated with life-threatening complications including volvulus and bowel infarction.[17] A continence clinic can be helpful when constipation is resistant to treatment.

Endocrinology

An endocrinologist can assist with management of delayed puberty and bone health.

Social/mental health

Madeline needs friends, as all young people do. She may need support to connect with peers at her school or through interest groups. She may also like to connect with other young people with cerebral palsy through social groups. Her NDIS Support Coordinator can help her find these opportunities and arrange funding for one-on-one support.

Answer 8

Madeline presents with many issues that must be prioritised and addressed over time. Regular appointments, Annual Health Assessments and working as part of a multidisciplinary team all contribute to good management. Chronic Disease Management Medicare items including the GP Management Plan, Team Care Arrangement and Adult Health Assessment are helpful for these appointments.

It is important to ensure that Madeline is linked with appropriate healthcare professionals (Table 2).

Answer 9

Mental health problems are more common in people with cerebral palsy than in the general population.[18] GPs can review the biopsychosocial aspects of Madeline's life that contribute to her wellbeing by:

- addressing any physical ill-health, pain and discomfort
- monitoring medication side effects that may affect function and wellbeing (eg nausea, dizziness)

- exploring goals (friends, activities, interests, education, employment) and how they can be supported through Madeline's NDIS plan
- engaging an occupational therapist to optimise Madeline's function and enable her participation in recreation, education or employment
- providing access to counselling – Madeline's NDIS Support Coordinator could be a useful source of local providers. The Australian Psychological Association may be able to suggest someone experienced in working with people using AAC.

Table 2. Healthcare professionals and their management role for people with cerebral palsy

Healthcare professional	Role in patient management
Dentist	Monitor oral health and treat disease Advise on strategies to maintain oral health and prevent disease Provide appropriate orthodontic interventions as indicated
Dietitian	Enteral formula – monitor weight, nausea, vomiting, reflux, constipation with changes in formula
Occupational therapist	Address seating needs including wheelchair modification and prescription of new wheelchair Advise on home management equipment to promote independence in self-care and ease of personal care assistance including hoists, bathroom and other home modifications
Speech pathologist	Assess swallowing and provide advice on safe options for any oral intake Communication assessment and training (specialist area of knowledge)
Physiotherapist	Monitor and address muscle function, tone, contractures Create exercise program to improve and maintain fitness, muscle range and strength, bone health
Nurse	Continence nurse – assist in monitoring bowel function and continence Stomal therapist – provide percutaneous endoscopic gastrostomy stoma care Practice nurse – assist with medical reviews, annual health assessment
Orthotist	Assess for, provide and modify orthotics
Optometrist/ ophthalmologist	Monitor vision
Audiologist/ear, nose and throat surgeon	Monitor hearing
Psychologist	Educational psychologist – identify learning strengths, weaknesses and strategies to optimise learning Clinical psychologist – provide counselling

As for all adolescents, Madeline needs opportunities to:

- find and maintain friendships and engage in activities with her friends
- explore interests and hobbies and try new activities
- experience increasing agency, autonomy and independence
- investigate education and employment options.

Answer 10

Preventive healthcare is important to ensure Madeline remains as well as is possible to optimise her function and quality of life. Mortality rates are twice as high as in the general population at 35 years of age for people with severe cerebral palsy.[19] Respiratory disease is the main cause of death.[9]

Not all Madeline's health issues will stem from her cerebral palsy; she is at risk of the same conditions as any woman of her age. Your role as a GP is to provide and arrange preventive health interventions (sexual health, cancer screening, immunisation, health promotion) and coordinate the multidisciplinary health team that will be required throughout her life.

The pneumococcal vaccine and a yearly influenza vaccine are particularly important considering her respiratory risk factors.

The NDIS enables access to therapy, equipment and personal support Madeline requires to participate in education, employment, recreational and social activities.

Resources for doctors

- Therapeutic Guidelines: Management Guidelines: Developmental Disability version 3, https://tgldcdp.tg.org.au/fulltext/quicklinks/management_guideline.pdf
- Cerebral palsy for general practitioners – Fact sheets, www.ausacpdm.org.au/resources/cerebral-palsy-for-general-practitioners-fact-sheets
- GMFCS: Gross Motor Classification System for Cerebral Palsy, https://cerebralpalsy.org.au/our-research/about-cerebral-palsy/what-is-cerebral-palsy/severity-of-cerebral-palsy/gross-motor-function-classification-system/
- National Disability Insurance Scheme, www.ndis.gov.au/applying-access-ndis/how-apply/information-gps-and-health-professionals
- The Royal Australian College of General Practitioners – National Disability Insurance Scheme: Information for general practitioners, www.racgp.org.au/running-a-practice/practice-resources/general-practice-guides/ndis-information-for-general-practitioners
- The Royal Australian College of General Practitioners – Guidelines for preventive activities in general practice, www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/red-book
- Cerebral Palsy Alliance, https://cerebralpalsy.org.au

- Oral Health for people with disability
  - www.nidcr.nih.gov/sites/default/files/2017-09/practical-oral-care-cerebral-palsy.pdf
  - https://inclusionmelbourne.org.au/projects/your-dental-health/

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CASE

3 Albert feels fatigued

Albert, aged 45 years, has been experiencing worsening fatigue, nausea, abdominal pain and low mood for the past few months. He has had bouts of similar symptoms in the past few years but put it down to 'hitting the grog too hard'. This led to his occasionally seeing a general practitioner (GP) for some related abdominal pain and nausea for which he has trialled esomeprazole as needed, with some effect.

Question 1

What further history would you want to explore with Albert?

Further information

Albert advises he has also experienced unintentional weight loss, loss of appetite, dizzy spells, poor concentration and confusion for the past few months. He has not noticed any change in bowel habit or urinary symptoms, nor has he experienced any shortness of breath or chest pain. There is no other past medical history of note, and he is not taking any regular medications.

Albert does not smoke but drinks 2–3 cans of full strength beer four nights per week. He does not report any illicit drug use. He works the day shift in the building supplies yard of his local hardware store, is married with three children and tries to exercise regularly. He enjoys being involved in his children's soccer club and being a team manager.

He has an older sister with thyroid problems and a younger brother with coeliac disease but there is no family history of malignancy. As a result of his low mood, his wife and work colleagues think he might be depressed.

Question 2

What diagnoses might you consider at this point?

Further information

You decide to further explore Albert's abdominal pain and nausea. Albert discloses that he was reviewed a few years ago by two different gastroenterologists, which resulted in endoscopic investigations that identified gastritis but no evidence of coeliac disease. He does not report any other constitutional symptoms.

Albert is worried about cancer and coeliac disease as his brother has had a terrible time managing this illness. On further review you establish that his work–life balance is good, and there are no issues at work or home that may be contributing to his low mood or confusion. He does not have thoughts of harm to himself or others.

Question 3

What examination(s) will you undertake?

Further information

The results of Albert's physical examination include:

- alert and orientated, but looks fatigued, reactive mood
- euthymic mood with congruent affect
- mildly dehydrated
- temperature – 36.8 °C
- weight – 71 kg (11 months ago Albert's weight was 79 kg)

- body mass index (BMI) – 21.7 kg/m^2
- blood pressure sitting – 98/65 mmHg, heart rate 89 beats per minute (bpm)
- blood pressure standing – 72/55 mmHg, heart rate 101 bpm, with symptomatic dizziness
- cardiovascular and respiratory examinations – normal
- abdominal exam – essentially normal, with the exception of a darkening pigmentation of the oral mucosa and some slight epigastric tenderness
- other noted features – no lymphadenopathy, slight hyperpigmentation of the palmer creases and knuckles.

Question 4

Are there any concerning features of Albert's presentation?

Further information

You undertake full fasting pathological tests. Box 1 shows Albert's results.

Box 1. Albert's serum electrolytes results

Sodium 131 mmol/L (reference range 137–145 mmol/L)
Potassium 5.6 mmol/L (reference range 3.5–5.2 mmol/L)
Chloride 103 mmol/L (reference range 100–109 mmol/L)
Bicarbonate 22 mmol/L (reference range 22–32 mmol/L)
Urea 6.1 mmol/L (reference range 2.7–8.0 mmol/L)
Creatinine 0.02 mmol/L (reference range 0.05–0.12 mmol/L)
Glucose 3.1 mmol/L (reference range 3.8–5.5 mmol/L)

Adapted with permission from Phillips PJ, Great tan but I feel awful, Aust Fam Physician, 2008;37(8):648–49.

Other results include:

- full blood examination – slight eosinophilia
- liver function tests – normal
- iron studies – normal
- thyroid tests – mildly decreased thyroid stimulating hormone but normal T3 and T4
- lipids – normal
- urine albumin to creatinine ratio – normal
- coeliac serology – negative.

Question 5

What are the concerning features of these pathology results?

Question 6

What further tests might you request?

Question 7

What is primary adrenal insufficiency and what results would you expect from the additional pathology tests?

Further information

Albert's fasting/morning cortisol result is 147 nmol/mL (reference range: 200–700 nmol/mL), and his adrenocorticotrophic hormone (ACTH) result is 220 pmol/mL (reference range: 2–11 pmol/mL).

Albert returns to the clinic and you explain the results and your concern. You recommend referral to an endocrinologist. You explore further Albert's presenting history that may be relevant for the endocrinologist and Albert advises he has noticed a salt craving over the past few months.

Question 8

How would a diagnosis of primary adrenal insufficiency be confirmed?

Further information

Albert is referred to an endocrinologist who undertakes a short synacthen test, and investigations for plasma renin, aldosterone and 21-hydroxylase antibody, which confirm primary adrenal insufficiency (hypocortisolism or autoimmune adrenalitis).

The short synacthen test is positive and confirmatory.

Question 9

How should Albert's condition be treated and managed?

Further information

Albert returns as planned and advises he has been feeling much better since starting the medication. His fatigue, dizziness and nausea are resolving, he is not as confused, his abdominal pain is almost gone and he is noticing the darkening skin is getting lighter.

His electrolytes have now normalised, his BMI is normal for his age and sex, his blood pressure is in the normal range and he is gaining weight. As winter is approaching, you suggest he has an influenza vaccination, to which he agrees.

Two months later, Albert presents and advises he is feeling 'stuffed' again. He has significant nausea, abdominal pain and confusion. A few days ago he developed flu-like symptoms and a fever. He has been compliant with his medication but has not employed the crisis action plan or filled the necessary medication scripts.

Question 10

What would you do now?

CASE 3 Answers

Answer 1

This constellation of non-specific symptoms poses an often-typical dilemma experienced by GPs when managing patients with undifferentiated problems. Murtagh's masquerades are often a useful guide.[1] The history-taking may include, but not be limited to, a diagnostic approach that considers a probability diagnosis, serious disorders that should not be missed[1] and the primary seven masquerades (depression, diabetes, medications, anaemia, thyroid and endocrine disorders, spinal dysfunction and urinary tract infection). Additionally, the inclusion of social and collateral history can provide a valuable insight into the presentation.

Answer 2

Albert's symptoms suggest several conditions that may be considered as part of the differential diagnosis including autoimmune thyroid disease, coeliac disease, depression, irritable bowel syndrome, diabetes mellitus and colorectal cancer.

Answer 3

A full physical examination is recommended, as a general mood and body habitus assessment and inspection may help to guide any further focused physical examinations. In this setting, a focus on vital observations in addition to assessing for postural

blood pressure changes and a cardiovascular and gastrointestinal review would be a reasonable initial approach.[2]

Answer 4

Albert's symptoms of profound weight loss, hypotension (including postural) and a family history of thyroid disease raise concern regarding an autoimmune illness. Additionally, his skin hyperpigmentation indicates the possibility of haemochromatosis[1,3] among other illness states including, but not limited to, Hashimoto's disease, pernicious anaemia, vitiligo and Addison's disease.

Answer 5

These pathology results are suggestive of an endocrine problem, as indicated by hyponatraemia, hyperkalaemia, hypoglycaemia and slight eosinophilia.[5]

Answer 6

Urgent additional tests may include fasting/morning cortisol and ACTH. Tests for dehydroepiandrosterone sulfate (DHEAS), renin, aldosterone and 21-hydroxylase antibody could also be considered.[4,6]

Owing to the combination of Albert's history, signs, symptoms and primary electrolyte disturbances, an endocrine cause needs consideration. In this context, further investigation for primary adrenal insufficiency confirmatory testing would not be unreasonable.

Answer 7

Primary adrenal insufficiency, also known as Addison's disease, is a rare disorder that is complicated by a range of non-specific symptoms. It has a prevalence of 100–150 per million people and an incidence of 4.4–6.0 new cases/million/year.[4,7–9] The most frequent cases of primary adrenal insufficiency occur as a result of autoimmune destruction of the adrenal cortex leading to the failure of the gland to produce cortisol, aldosterone and DHEAS.[10,11] The resulting autoimmune adrenalitis produces the signs, symptoms and pathological findings listed in Table 1.[2,12,13]

For patients with Addison's disease, results for fasting cortisol would be expected to be low, while results for ACTH are likely to be elevated. The resulting adrenal gland cortex destruction in primary adrenal insufficiency also affects the sodium/potassium balance, leading to hypoaldosteronism and androgen dysfunction with low DHEAS.[13]

Answer 8

The classic Murtagh triad of fatigue + anorexia/nausea/vomiting + abdominal pain (+/– skin discolouration) is useful in the setting of chronic primary disease. Of interest is the relationship between primary adrenal insufficiency and hyperpigmentation of the skin, which is a cardinal sign.[13,14] This is a direct result of elevated ACTH, as this pituitary-secreted hormone has a common precursor protein to melanocyte-stimulating hormone. Increased production of ACTH via a negative feedback loop leads to concomitant increased production of melanocyte-stimulating hormone.[3,10,11,15]

The diagnosis of primary adrenal insufficiency is confirmed by a positive short synacthen test, elevated ACTH and elevated plasma renin.[14]

Answer 9

Albert has most likely had symptoms of chronic adrenal insufficiency[8,16] for an extended and protracted period of time.

The usual treatment is a combination of hydroxycortisone with a dosing pattern that follows a circadian rhythm to mimic cortisol production for glucocorticoid replacement and fludrocortisone for mineral corticoid substitution.[4,7,13,14] A suitable starting dose for an adult with adrenal insufficiency is hydrocortisone 12 mg orally, in the morning immediately after waking, and 8 mg in the mid-afternoon. Alternatively, patients can commence cortisone acetate 15 mg orally in the morning immediately after waking and 10 mg in the mid-afternoon.[14] For mineralocorticoid replacement for adrenal insufficiency in an adult, fludrocortisone 100 μg orally, daily, is recommended.[14] The dose should be adjusted according to postural blood pressure and serum potassium and plasma renin concentrations.[14] The usual dose is 50–300 μg daily.[14]

Table 1. Symptoms and signs of primary adrenal insufficiency[2,12,13,19]

Symptoms (prevalence %)*	Signs (prevalence %)*	Routine laboratory finding
• Weakness, malaise (100%) • Weight loss and anorexia (100%) • Pigmentation or 'persistent tan' • Abdominal pain, diarrhoea, vomiting (92%) • Dizziness and syncope (12%) • Myalgia, joint pain (6–13%) • Depression or confusion • Impotence/amenorrhoea • Acute back pain (if patient is taking anticoagulants)	• Generalised pigmentation – especially extensors and palmar creases (94%) • Buccal and tongue pigmentation • Scar pigmentation • Hypotension (systolic blood pressure usually <110 mmHg; 88–94%) • Postural hypotension (88–94%) • Weight loss and general wasting • Signs of autoimmune disease (eg vitiligo) • Signs of other diseases (eg congenital adrenal hyperplasia, amyloidosis, adrenoleukodystrophy, haemochromatosis)	• Hyponatremia • Hyperkalaemia • Hypoglycaemia • Hypercalcaemia

**Prevalence is included where available.*

Patient education, such as information from the Endocrine Society and the Australian Addison's Disease Association,[7,13,14] is a critical component of Addison's disease management along with a lifelong annual endocrinology review and screening for other autoimmune diseases.[6,8] Education relating to adrenal crisis and how to manage when unwell, prior to surgery and (for female patients) during pregnancy is significant, as complacency can have serious consequences.[13,14]

Other management options to consider may include:

- medi-alert bracelet[13,14,16]
- action plan for crisis management (Box 2)[7,13,14,17]
- baseline bone mineral density assessment[7]
- consideration of dehydroepiandrosterone treatment[16]
- consideration of GP Management Plan and Team Care Arrangements for Chronic Disease Management.

Box 2. Self-care action plan

Key aspects of a self-care plan for a patient with adrenal insufficiency:[14]

- increase glucocorticoid dose during intercurrent illness
- recognise early features of adrenal crisis (eg nausea, vomiting, dehydration, hypotension)
- carry injectable hydrocortisone when away from medical care
- wear an alert bracelet or necklace
- carry a wallet card with details about their condition and treatment.

Answer 10

Albert's symptoms indicate a possible adrenal crisis, and it is recommended to assess his vital signs and undertake a general examination. Crisis situations will ultimately require hospitalisation and stabilisation with a short admission and can be managed with a combination of intravenous (IV) fluids and hydrocortisone replacement.

Conclusion

Albert is afebrile, and his abdominal examination is unremarkable. His sitting blood pressure is 130/70 mmHg, heart rate 81 bpm, and his standing blood pressure is 95/56 mmHg, heart rate 105 bpm.

You arrange urgent hospital transfer and discuss Albert's situation with the accident and emergency consultant. You commence IV fluids and 100 mg hydrocortisone IV, initially, then 50 mg IV, every six hours until stable.[14]

Albert is reviewed in hospital by his endocrinologist. He recovers well and is subsequently reviewed by the specialist a few weeks later and receives further education, an updated crisis management plan and appropriate prescriptions for intramuscular hydrocortisone and prednisolone. His wife is also provided with education.

You see Albert a year later after his annual endocrine review, in which he has was diagnosed with thyroid disease. As Addison's disease is related to other autoimmune states and is categorised as a polyglandular syndrome (type 2),[3,6] it is not unusual for subsequent diagnoses of thyroid disease, coeliac disease or pernicious anaemia to present. As a result, all first-degree relatives should be screened.[6]

Summary

Addison's disease can have serious consequences if not recognised early, particularly when in crisis. Despite Addison's disease being a rare illness with a slow and often chronic path, when patients present with nonspecific signs and symptoms, it may be worthwhile to consider this condition.[18]

Resources for doctors

- Therapeutic Guidelines – Adrenal insufficiency, https://tgldcdp.tg.org.au/viewTopic?topicfile=adrenal-insufficiency&guidelineName=Bone%20and%20Metabolism&topicNavigation=navigateTopic

Resources for patients

- healthdirect – Addison's disease, www.healthdirect.gov.au/addisons-disease
- BetterHealth Channel – Addison's disease, www.betterhealth.vic.gov.au/health/conditionsandtreatments/addisons-disease?viewAsPdf=true
- Hormones Australia – Adrenal insufficiency, www.hormones-australia.org.au/endocrine-diseases/adrenal-insufficiency
- Australian Addison's Disease Association Inc., https://addisons.org.au

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CASE

4 Sosuke thinks he had a 'fit'

Sosuke, aged 51 years, has been a regular at your practice every few months for a check-up after having an ST-elevation myocardial infarction two years ago. He has successfully changed his lifestyle since that event; he has stopped smoking, and he exercises regularly and has reduced his weight. As a result, his coronary artery disease has been stable. He takes aspirin 100 mg, atorvastatin 40 mg, ramipril 5 mg and metoprolol 50 mg every day.

Today, he comes in to see you earlier than his scheduled visit and says he thinks he had a 'fit' a few days ago.

Question 1

What would you ask Sosuke?

Further information

Sosuke remembers that while sitting at the table at home, he fell down without warning. His wife told him afterwards that she heard the noise and entered the room to see his arms and legs jerking and his mouth frothing. He recovered after a few minutes and discovered that he had been incontinent. He felt dazed for approximately half an hour afterwards. He has never experienced anything like this before.

Question 2

What further information would you seek?

Question 3

What examination(s) would you perform?

Question 4

What are the other possible explanations for Sosuke's symptoms besides a seizure?

Further information

Further inquiry into Sosuke's history is unremarkable. Other than a laceration on the lateral border of his tongue, his physical examination, including a full neurological examination, is essentially normal. There is no information that suggests a seizure mimic. You determine that Sosuke had an unprovoked tonic-clonic seizure without aura followed by postictal confusion.

Question 5

What investigations would you perform?

Further information

Sosuke's blood, metabolite and cardiovascular examinations show no abnormalities. His EEG and CT scan and MRI of the brain are normal. These clinical and investigative findings reinforce your initial working diagnosis of a generalised tonic-clonic seizure. The next step is to determine if Sosuke has epilepsy.

Question 6

How would you classify epilepsy?

Question 7

What would you do next?

Question 8

Is there anything else you should discuss with Sosuke?

Further information

Sosuke has been a courier driver all his life and is worried he will not be able to continue his work. Sosuke is a holder of Medium Rigid (MR) and C class licences.

Question 9

What would you tell Sosuke about his ability to continue driving?

Further information

Sosuke returns to see you after his appointment with a neurologist, who elicited a history of absences, myoclonic jerks and photosensitivity suggestive of an underlying epileptic syndrome. A diagnosis of generalised tonic-clonic epilepsy was made. The neurologist recommended anti-epileptic medication, and Sosuke has commenced carbamazepine 200 mg/day with a scheduled increase in the daily dose by 200 mg every three days to a maximum of 400–600 mg/day.

Question 10

What does Sosuke need to know about this medication?

Further information

As Sosuke's employment involves driving, he will have to consider alternative employment and possibly retraining.

Question 11

As his general practitioner (GP), how could you support him in this life-changing moment?

CASE 4 Answers

Answer 1

A careful history is the single most important component of the initial diagnosis and classification of seizures as well as differentiation from seizure mimics. History from a witness, if present, or access to CCTV recording is invaluable.[1] It is important to inquire about symptoms present before the seizure (ie aura, which can be autonomic [rising epigastric sensation], psychic [déjà vu or jamais vu], sensory [metallic taste, coloured spots, tinnitus, tingling] or lightheadedness).

It is recommended that the patient is asked to describe the seizure.

Did he lose awareness? Was he aware of sudden irregular jerks involving the trunk or limbs (myoclonus), regular shaking (clonic), stiffening (tonic) or spasms (dystonic)? A witness may describe impaired awareness such as absences (motionless stare, eyelid fluttering or disruption of awareness and activity) and automatisms (lip-smacking, fiddling, rubbing), pseudo-purposeful movements (dressing or undressing, hitting out), vocalisations and impaired communication.[2]

Any other physiological events that occurred during the seizure should be noted; for example, tongue biting, cyanosis, lateral gaze, torticollis, urinary incontinence.

It is also essential to ask the patient how they felt after the seizure. Postictal confusion or drowsiness of more than 10 minutes is characterised by disorientation, poor concentration, short-term memory loss and decreased verbal and interactive skills.

Answer 2

Further questioning could include:

- Was there any unusual activity, illness or fever in the 24 hours preceding the event?
- Was there anything that could have provoked the seizure, such as alcohol, drugs or trauma?
- Was the patient sleep deprived?
- Is the patient taking any medications that can lower the seizure threshold, such as antipsychotics, antidepressants, antihistamines, analgesics, bowel preparations and drug withdrawals?

In addition, it is important to ask about the patient's past medical history, family history and social history.

Answer 3

A full physical examination is recommended for any patient presenting with an episode of seizure, including an assessment of cardiac, neurological and mental status.[3] The purpose of the examination is to look for seizure mimics in the systemic examination.

The cardiovascular examination would look for an irregular pulse, heart murmurs, cardiac enlargement and carotid bruits. Electrocardiography could be requested. The neurological examination would focus on finding a neurological deficit and investigating abnormal motor movements. A fundoscopic examination should also be performed to look for papilloedema. A mental state examination is essential to record the patient's current mental health and pre-existing psychological symptoms.

Answer 4

Conditions included in the differential diagnosis for seizures are:[4]

- cardiac syncope – arrhythmias, tachycardia, aortic stenosis, cardiomyopathy
- noncardiac syncope – vasovagal syncope, postural hypotension
- migraine
- narcolepsy–cataplexy
- tremors
- tics
- movement disorders
- psychological and psychiatric – pseudo seizures, hyperventilation, panic attacks, dissociative reactions
- metabolic derangements – hypoglycaemia, hypoxia, severe acid–base disturbances, febrile illness, hyperthermia, respiratory, liver and renal failure, intoxications and poisonings, drug and alcohol withdrawal
- head injury – acute concussive convulsions: extradural haematoma, subdural haematoma, raised intracranial pressure
- cerebral infections – bacterial meningitis, viral encephalitis
- stroke – embolism or thrombosis, subarachnoid haemorrhage, intracerebral haemorrhage.

Answer 5

It is recommended that Sosuke undergoes basic blood and metabolite examinations including fasting blood sugar, renal

function tests, liver function tests, calcium and magnesium, vitamin B12 and folate. Serum prolactin, when measured 10–20 minutes after a suspected seizure, can provide insight into the type of seizure experienced; this could be considered if Sosuke presented soon after another seizure.[5] A blood alcohol and urine drug screen for opiates and amphetamines can be useful in selected cases. Cardiovascular screening is also recommended, including an electrocardiogram, 24-hour Holter monitoring and two-dimensional echocardiography.[6]

Electroencephalography (EEG) is an indispensable investigation in the diagnosis of epilepsy. If generalised 3–4 cycle per second discharges are present, the diagnosis is generalised epilepsy. The definition of epilepsy is any of the following:[7]

- two or more unprovoked or reflex seizures occurring >24 hours apart
- diagnosis of an epilepsy syndrome
- single unprovoked (or reflex) seizure and high risk of recurrence over the next 10 years.

If the initial EEG is non-diagnostic, a sleep-deprived EEG should be obtained.[8] A computed tomography (CT) or magnetic resonance imaging (MRI) scan of the brain is important for patients who have had their first seizure episode and is mandatory for all patients with a history of trauma, focal seizure onset, suspected malignancy, fever, meningism or neurological signs. As a rule, all adult patients who have had their first seizure should be considered for MRI.[3]

Answer 6

The classification of epilepsies comprises a multilevel diagnosis.[2]

The first diagnostic level is seizure type, which can be divided into three major groups on the basis of the onset – focal, generalised or unknown.

The second diagnostic level is epilepsy type, and it assumes the patient has a diagnosis of epilepsy based on the 2014 definition.[7] Epilepsy types are focal, generalised, combined focal and generalised, and unknown.

The third diagnostic level is an epilepsy syndrome diagnosis. An epilepsy syndrome refers to a cluster of features incorporating seizure types, EEG and imaging features that tend to occur together (ie juvenile myoclonic epilepsy).

An aetiological diagnosis should be considered from the first presentation and at every stage along the diagnostic pathway. A diagnosis of epilepsy may have more than one aetiological category and more than one or two seizure types.

Answer 7

All patients who have had an initial seizure should be referred to a neurologist for a semi-urgent neurology assessment in 4–6 weeks and advice regarding commencement of anti-epileptic medication.[9]

Answer 8

It is recommended to have an open discussion with patients regarding the implications of a potential diagnosis of epilepsy. Approximately 60% of single seizures recur within two years, and the recurrence rate is highest within the first few months.[10] Patients should be advised of first aid measures, which they can discuss with their families. It is also important to discuss activities that may affect the patient's safety such as using heavy machinery, working at heights, being around water, sports, recreation and general safety issues. Some lifestyle measures will help reduce seizures and give patients a sense of control.

It is recommended that a patient who experiences seizures follows a ketogenic (high fat, low carbohydrate, adequate protein) diet[11] and avoids illicit drugs of abuse.[12]

Sosuke should be advised to get adequate sleep, as lack of sleep is a common trigger for seizures.[13] He should be encouraged to maintain a healthy lifestyle by practising muscle relaxation exercises, breathing techniques, meditation and obtaining regular exercise.

It is important to discuss any psychological symptoms such as mood and emotions, as even patients with well-controlled seizures have higher rates of depression than the general population.[14] Suicide rates are triple that of the general population, especially in the first six months after diagnosis.[15] Sosuke should be made aware of the availability of professional support and therapy.

Sudden unexpected death in epilepsy (SUDEP) is a rare complication of epilepsy. It occurs in benign circumstances, usually at night, and is unrelated to seizure duration. Although rare, a discussion about SUDEP, its causes and how to reduce its risk is recommended. If appropriate, this discussion can include the family.[16]

An epilepsy management plan should also be discussed, including emergencies, safety, risks, medications and monitoring.[17] Well-controlled epilepsy has a good prognosis. It is considered resolved if the patient is seizure-free for 10 years, including five years off anti-seizure medications.[7]

Answer 9

Austroads is the peak organisation of Australasian road transport and traffic agencies. It is the final authority with regard to all matters relating to fitness to drive.

As Sosuke's symptoms are of a potentially serious nature, he should be advised not to drive until his condition can be adequately assessed. For further information, refer to the Austroads website (https://austroads.com.au/__data/assets/pdf_file/0022/104197/AP-G56-17_Assessing_fitness_to_drive_2016_amended_Aug2017.pdf). Further advice can be given after the neurologist's evaluation and recommendations are received.

Answer 10

The choice of initial therapy is complex, with many new anti-epileptic medications available. They are classified as either

broad- or narrow-spectrum medications with regard to efficacy against different seizure types and epilepsy syndromes.[1] Carbamazepine is a narrow-spectrum hepatic enzyme–inducing medication.

Common side effects of carbamazepine are dizziness, diplopia, blurred vision, ataxia, sedation, rash, weight gain, nausea, hyponatraemia and benign leukopenia. Rare side effects are agranulocytosis, aplastic anaemia, Stevens–Johnson syndrome and heart block.[1]

It is recommended that patients taking carbamazepine undergo regular monitoring of sodium, liver function tests, full blood examination and electrocardiography.[1,18] However, in general, routine monitoring of anti-epileptic medication levels is not required.[9]

It is recommended that anti-epileptic medication is initiated by a specialist with epilepsy experience; likewise, cessation of anti-epileptic medication should be guided by specialist evaluation.[9]

Long-term enzyme-inducing anti-epileptic medications can result in loss of bone density.[19] Sosuke should receive supplemental vitamin D and calcium and undergo periodic bone-density measurements.

Answer 11

For patients recently diagnosed with epilepsy, it is important to offer support and reassurance as they work through the changes that will occur in their lives in the following few months to years. A consultation with Sosuke's close family could include discussing transport, living arrangements, safety and help with paperwork.

As Sosuke's GP, you can consider providing a letter of support to access government-funded financial support. Sosuke can also be referred to a disability service provider for retraining and suitable employment.

Resources for doctors and patients

- Epilepsy Action Australia, www.epilepsy.org.au
- Epilepsy Foundation, https://epilepsyfoundation.org.au
- International League Against Epilepsy, www.ilae.org/patient-care

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ACTIVITY ID **213556**

Chronic conditions

This unit of *check* is approved for six CPD Activity points in the RACGP CPD Program. The expected time to complete this activity is three hours and consists of:

- reading and completing the questions for each case study
  - you can do this on hard copy or by logging on to the RACGP website (www.racgp.org.au), clicking on the My Account button and selecting the *gplearning 2020* link from the drop-down
- answering the following multiple choice questions (MCQs) by logging on to the RACGP website (www.racgp.org.au), clicking on the My Account button and selecting the *gplearning 2020* link from the drop-down
  - you must score ≥80% before you can mark the activity as 'Complete'
- completing the online evaluation form.

You can only qualify for CPD points by completing the MCQs online; we cannot process hard copy answers.

If you have any technical issues accessing this activity online, please contact the *gplearning* helpdesk on 1800 284 789.

If you are not an RACGP member and would like to access the *check* program, please contact the *gplearning* helpdesk on 1800 284 789 to purchase access to the program.

Case 1 – Timmy

Timmy, aged 34 years, was recently diagnosed with adrenal insufficiency (Addison's disease). He has been reviewed by an endocrinologist, and today you are checking for the resolution of his initial presenting symptoms.

Question 1

Which one of the following best describes the key featured symptoms of chronic adrenal insufficiency?

A. Fatigue + anorexia/nausea/vomiting + abdominal bloating (+/– skin hypopigmentation)

B. Headache + anorexia/nausea/vomiting + abdominal pain (+/– skin discolouration)

C. Fatigue + anorexia/nausea/vomiting + abdominal pain (+/– skin hyperpigmentation)

D. Low mood + anorexia/nausea/vomiting + reflux (+/– skin discolouration)

Further information

Timmy advises he is feeling much better since commencing hydrocortisone and fludrocortisone. He tells you the endocrinologist suggested he have a self-care action plan. You decide to check his understanding of his condition, the need for lifelong therapy and the importance of an action plan.

Question 2

Which one of the following lists includes the key crisis symptoms to note when rescue treatment is needed?

A. Abdominal pain, confusion, hypertension

B. Abdominal pain, delirium, hypertension

C. Abdominal pain, confusion, hypotension

D. Abdominal pain, acute psychosis, hypotension

Case 2 – Anthea

Anthea, aged 63 years, is a smoker with osteoarthritis and hypertension who presents to you with a persistent cough with copious tenacious sputum. She is currently taking ramipril 2.5 mg at night, meloxicam 15 mg in the morning and paracetamol (slow release) 1330 mg three times per day. She has no known allergies. You suspect bronchiectasis.

Question 3

Which one of the following is the probable cause of Anthea's bronchiectasis?

A. Idiopathic bronchiectasis

B. Long-term nonsteroidal anti-inflammatory drug use

C. Osteoarthritis

D. Hypertension

Question 4

Which one of the following investigations is the gold standard for diagnosis of bronchiectasis?

A. Chest X-ray

B. Bronchoscopy

C. Chest high-resolution computed tomography scan

D. Full lung function tests

Further information

Anthea is diagnosed with bronchiectasis and chronic obstructive pulmonary disease. She is seen by a respiratory physician who prescribes a bronchodilator daily for ongoing management. She presents to you two months later with increased cough, mucopurulent sputum and wheeze. You diagnosis an exacerbation of bronchiectasis.

Question 5

Which one of the following treatments is recommended?

A. Amoxycillin and clavulanic acid 875 mg/125 mg twice per day for 10 days

B. Cefuroxime 250 mg twice per day for seven days

C. Amoxycillin 1000 mg three times per day for 14 days

D. Doxycycline 100 mg twice per day for five days

Case 3 – Sriyan

Sriyan, aged 15 years, is brought in to see you by his brother, who witnessed Sriyan having a seizure. He has never had seizures before. After your initial history-taking and examination, you formulate your differential diagnosis, which includes epilepsy.

Question 6

Which one of the following statements is most accurate regarding the diagnosis of epilepsy?

A. An electroencephalogram is diagnostic.

B. Diagnosis relies on magnetic resonance imaging of the brain.

C. History and examination are vital.

D. Serum prolactin level has a high positive predictive value.

Further information

Sriyan undergoes further investigations and is diagnosed with epilepsy. You consider your knowledge of anti-epileptic medication and the possible complications of epilepsy for Sriyan.

Question 7

Which one of the following statements is true regarding anti-epileptic medication for Sriyan?

A. Therapy should be initiated only by a specialist .

B. Therapy should be lifelong.

C. Routine monitoring of therapeutic drug levels is required.

D. Valproate can cause Stevens–Johnson syndrome.

Question 8

Which one of the following is a possible complication of epilepsy?

A. There is an increased association with depression .

B. There is a 10% incidence rate of Sudden Unexpected Death in Epilepsy (SUDEP).

C. There is an increased risk of SUDEP if a seizure is prolonged for >10 minutes.

D. There is an increase in suicidal risk with prolonged duration of illness.

Case 4 – Ilaria

Ilaria, aged 24 years, has cerebral palsy and presents to see you with her Disability Support Worker, Nabil. You see from her records that she is non-verbal.

Question 9

Which one of the following **best** describes how you would proceed with the consultation?

A. Ask Ilaria to return with an augmentative and alternative communication device.

B. Direct your questions to Nabil.

C. Modify your questions to allow for only 'yes' or 'no' responses.

D. Ask Ilaria how she would prefer to communicate.

Further information

As part of your examination of Ilaria, you assess how she is managing her oral hygiene.

Question 10

Which one of the following factors is **most likely** to contribute to poor dentition in a person with cerebral palsy?

A. Behavioural difficulties

B. Reduced salivation due to medication

C. Gastro-oesophageal reflux disease

D. Limited motor control

check

Independent learning program for GPs