Unit 534 December 2016

Cardiology
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About this activity

Acronyms

Case 1  Ken has trouble walking when he goes to buy the paper  
Case 2  Doug presents for his cholesterol results  
Case 3  Anthea comes for a check-up  
Case 4  John has a murmur  
Case 5  Bill presents with fatigue and shortness of breath on exertion

Multiple choice questions

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
ABOUT THIS ACTIVITY

Cardiovascular disease (CVD) affects one in six Australians and is one of the major causes of death in Australia; in 2013, about 30% of deaths were attributed to CVD. Risk factors for CVD include diabetes, smoking, obesity, diet and physical activity.

Peripheral arterial disease is estimated to affect 10–15% of the general population and approximately half of those with the disease are asymptomatic. Familial hypercholesterolaemia is the most common inherited cholesterol defect and occurs in at least 1 in 500 in the general population.

More than six million Australians were considered to have hypertension or were on antihypertensive medication in 2014–16. Congenital heart disease has been estimated to affect 7.8 cases per 1000 births in Australia in the general population. Chronic heart failure affects more than 300,000 Australians and more than 30,000 new cases are diagnosed each year.

General practitioners have a key role in the early identification of CVD risk factors and in providing preventive healthcare, where possible, as well as diagnosing and managing various aspects of CVD. This edition considers assessment and management of CVD in general practice.

LEARNING OUTCOMES

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

• describe the assessment of peripheral artery disease
• discuss the approach to diagnosis and management of familial hypercholesterolaemia
• outline the management of elevated blood pressure
• list the recommendations for management of congestive heart failure
• discuss the assessment and management of congenital heart disease.

AUTHORS

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Jonathan Golledge (Case 1) BA, MBChir, MChir, FRCS, FRACS, is director of the Queensland Research Centre for Peripheral Vascular Disease (ORCPVD) at the School of Medicine and Dentistry, James Cook University. Professor Golledge joined James Cook University in 2002 and established the Vascular Biology Unit with the aim of carrying out research intended to be translated into improved management of aortic aneurysm and other vascular conditions. Trained as a vascular specialist, Professor Golledge took two years out of specialist training to obtain experience in research techniques as part of a Cambridge MChir (Doctoral equivalent), UK. His research commitment is illustrated by a large number of presentations at international and national meetings and publications in peer-reviewed journals, including a large number in top specialised journals.

Matthew Henry (Case 4) MBBS, is a second year cardiology fellow at the Royal Children’s Hospital, Melbourne. He obtained his medical degree and paediatric training in Adelaide, South Australia and has been involved in a number of teaching programs including those for medical students and paediatric trainees.

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Andrew Maiorana (Case 5) PhD, is a research academic at Curtin University and a practising clinical exercise physiologist with the Advanced Heart Failure and Cardiac Transplant Service at Fiona Stanley Hospital, Perth, Western Australia. Associate Professor Maiorana’s research interests focus on improving the health of patients with heart failure through lifestyle interventions and multidisciplinary care. He has published internationally on the effects of exercise training in heart failure and has coordinated several trials on the management of heart failure in primary care, including the use of telemonitoring as an adjunct to heart failure management.

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Joel Rhee (Case 3) BSc (Med), MBBS (Hons), GCULT, PhD, FRACGP, is an associate professor and the head of Assessment at the School of Medicine, University of Notre Dame Australia; and conjoint academic at UNSW Medicine. He is a general practitioner with special interest in chronic disease management, aged care and primary palliative care.

PEER REVIEWERS

Mark Beebay MBBS, FRACGP, DipPalMed, has been a general practitioner for 34 years in Lalor Plaza Medical Centre, Lalor, Vic. Dr Beebay is also an examiner for the RACGP and is a clinical supervisor for general practice registrars with Eastern Victoria GP Training (EVGPT).

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REFERENCES


ACRONYMS

<table>
<thead>
<tr>
<th>AAA</th>
<th>abdominal aortic aneurysm</th>
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<tbody>
<tr>
<td>ABI</td>
<td>ankle:brachial index</td>
</tr>
<tr>
<td>ASD</td>
<td>atrial septal defect</td>
</tr>
<tr>
<td>AVSD</td>
<td>atrioventricular septal defect</td>
</tr>
<tr>
<td>BMI</td>
<td>body mass index</td>
</tr>
<tr>
<td>BNP</td>
<td>brain natriuretic peptide</td>
</tr>
<tr>
<td>CHD</td>
<td>coronary heart disease</td>
</tr>
<tr>
<td>CHF</td>
<td>congestive heart failure</td>
</tr>
<tr>
<td>CT</td>
<td>computed tomography</td>
</tr>
<tr>
<td>CVD</td>
<td>cardiovascular disease</td>
</tr>
<tr>
<td>DLCNS</td>
<td>Dutch Lipid Clinic Network Score</td>
</tr>
<tr>
<td>ECG</td>
<td>electrocardiogram</td>
</tr>
<tr>
<td>eGFR</td>
<td>estimated glomerular filtration rate</td>
</tr>
<tr>
<td>FH</td>
<td>familial hypercholesterolaemia</td>
</tr>
<tr>
<td>HF</td>
<td>heart failure</td>
</tr>
<tr>
<td>LDL-C</td>
<td>low-density lipoprotein-cholesterol</td>
</tr>
<tr>
<td>LV</td>
<td>left ventricular</td>
</tr>
<tr>
<td>MBS</td>
<td>Medicare Benefits Schedule</td>
</tr>
<tr>
<td>NYHA</td>
<td>New York Heart Association</td>
</tr>
<tr>
<td>PAD</td>
<td>peripheral artery disease</td>
</tr>
<tr>
<td>PBS</td>
<td>Pharmaceutical Benefits Scheme</td>
</tr>
<tr>
<td>RACGP</td>
<td>The Royal Australian College of General Practitioners</td>
</tr>
<tr>
<td>RV</td>
<td>right ventricular</td>
</tr>
<tr>
<td>SNAP</td>
<td>smoking, nutrition, alcohol and physical activity</td>
</tr>
<tr>
<td>UACR</td>
<td>urinary albumin-to-creatinine ratio</td>
</tr>
<tr>
<td>URTI</td>
<td>upper respiratory tract infection</td>
</tr>
<tr>
<td>VSD</td>
<td>ventricular septal defect</td>
</tr>
</tbody>
</table>
CASE 1

KEN HAS TROUBLE WALKING WHEN HE GOES TO BUY THE PAPER

Ken, 74 years of age, presents for his annual flu vaccination. After receiving his vaccination, Ken mentions that he has been having pain in his right calf muscle during his daily 1 km walk to his local newsagency.

QUESTION 1

How would you manage this consultation? What would you ask Ken to help make a diagnosis?

FURTHER INFORMATION

Ken has pain in his right calf when walking to the newsagency. It settles within minutes if he rests at the bus stop but returns on reaching his destination. He has no history of diabetes, heart disease, stroke, hypertension (last clinic measure 138/67 mmHg) or dyslipidaemia. Ken had been a smoker but quit recently after being diagnosed with emphysema. He is on regular tiotropium and salbutamol prn inhalers for his lung condition but on no other medications.

QUESTION 2

What is your provisional diagnosis? What are the differential diagnoses?

QUESTION 3

What tests would you order to confirm the diagnosis?

QUESTION 4

How would you manage this condition?
CASE 1 Answers

ANSWER 1
You should ask Ken about classical symptoms of intermittent claudication, which include that the pain is located in a lower limb muscle group, brought on by exercise and relieved by rest. Classical symptoms of rest pain should also be sought, including severe pain located in the foot, present for at least four weeks, worse on leg elevation (eg in bed) and relieved by lowering the foot to the ground. A history of gangrene and lower limb ulceration should also be sought. Although Ken does not have a history of ischaemic heart disease or cerebrovascular disease, he should also be asked specifically about symptoms related to those vascular beds (eg exertional chest pain/tightness, past focal neurological symptoms typical of transient ischemic attack or stroke).

A history of risk factors for atherosclerosis, including hypertension, diabetes, dyslipidaemia and smoking, should be sought. Current medications should be recorded, as should allergies and family history of cardiovascular disease. The functional effect of the pain on Ken’s life should be estimated. For example, is it stopping him from enjoying a significant range of activities?

A general examination of Ken’s cardiovascular system should be performed. His lower limbs should be examined, looking for evidence of poor perfusion, such as skin changes (reduced capillary refill, cold periphery and pallor) and reduced or absent pulses. Ankle:brachial index (ABI) should also be measured as noted in Answer 3.

ANSWER 2
Ken’s symptoms of pain on exertion that subsides with rest is consistent with intermittent claudication, which is a classical manifestation of peripheral arterial disease (PAD). Claudication, from the Latin ‘to limp’ has a classical origin, being named after the Roman Emperor Claudius, who was said to have walked with a limp.

The differential diagnosis would be spinal claudication. It is important to note that patients with PAD may present with atypical symptoms, or indeed no symptoms, as the exercise tolerance may be limited by other disease processes. A significant number of patients with PAD also have abdominal aortic aneurysms (AAAs) so screening them for an AAA is suggested. Pain location can vary with location of the disease, from the buttock to the foot.

ANSWER 3
ABI (<0.9) is a reliable indicator of the PAD, providing the appropriate equipment is available. ABI is usually measured with a hand-held Doppler and an appropriate blood pressure cuff, although there are more expensive devices available for this.

Arterial duplex ultrasonography (and computed tomography angiography) can identify sites of occlusion or stenosis but requires a highly experienced sonographer to provide reliable information. ABI alone is adequate prior to referral to a specialist, who can decide whether further imaging is appropriate.

ANSWER 4
Medical management is the priority. PAD is a sign of generalised vascular disease. Ken’s condition, therefore, warrants prescriptions for antihypertensive and cholesterol-lowering medication despite not crossing individual risk factor thresholds. An angiotensin converting enzyme inhibitor (eg ramipril) or calcium channel blocker (eg amlodipine) and a statin (eg atorvastatin or rosuvastatin) are recommended. Beta-blockers are best avoided unless required for cardioprotection. The aim is to improve circulation and walking distance, but maximally tolerated medication is also required because of this high risk for cardiovascular events. Antiplatelet medication is also indicated (eg aspirin 100 mg).

Supervised exercise programs are effective at improving symptoms but not currently funded by the healthcare system in Australia. Ken should be encouraged to continue his regular walks and not to fall back into his previous smoking habit.

Revascularisation procedures are usually only considered if symptoms are severe or if tissue destruction is evident. Referral to a vascular specialist is recommended for patients with lifestyle-limiting intermittent claudication, rest pain and gangrene. Patients with rest pain, arterial ulcers or gangrene should be seen urgently. Revascularisation is considered for lifestyle-limiting intermittent claudication, but has associated risks and is frequently not durable on its own. This highlights the importance of lifestyle changes and medical management as first-line treatment.

References
CASE 2

DOUG PRESENTS FOR HIS CHOLESTEROL RESULTS

Doug, 36 years of age, presents to discuss the results of a recent cholesterol test. This was requested as part of your work-up for newly diagnosed hypertension. Doug has no history of chest pain or any other symptoms of acute coronary syndrome. He is on perindopril 5 mg daily for hypertension but no other medications. His fasting lipid levels are shown in Table 1.

Table 1. Doug’s lipid levels

<table>
<thead>
<tr>
<th>Lipids</th>
<th>Doug’s levels</th>
<th>Reference range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total cholesterol</td>
<td>11.6 mmol/L</td>
<td>&lt;5.5 mmol/L</td>
</tr>
<tr>
<td>Triglycerides</td>
<td>1.7 mmol/L</td>
<td>&lt;1.5 mmol/L</td>
</tr>
<tr>
<td>High-density lipoprotein-cholesterol</td>
<td>1.0 mmol/L</td>
<td>&gt;1.0 mmol/L</td>
</tr>
<tr>
<td>Low-density lipoprotein-cholesterol</td>
<td>9.8 mmol/L</td>
<td>&lt;3.5 mmol/L</td>
</tr>
</tbody>
</table>

QUESTION 1
What other history do you want to know from Doug? Are there any particular examination findings you would look for?

FURTHER INFORMATION
Apart from newly started medication for hypertension, Doug is on no other medications and has no other significant medical history. He does not smoke and classifies himself as an occasional drinker of alcohol.

Doug tells you that his father had a heart attack at 46 years of age, requiring coronary artery stenting and ongoing medication for high cholesterol. He is still alive and is currently aged 59 years. His paternal grandfather had a heart attack at the age of 45 years and died shortly after. In addition, Doug has an uncle, his father’s brother, who is also on cholesterol-lowering treatment. Doug has one sister aged 32 years.

As far as he knows, she is well and on no medications. He has three children aged 7, 9 and 11 years.

On examination in your office, Doug’s cardiovascular examination findings are normal. His blood pressure is 132/84 mmHg by sphygmomanometer on a peripheral cuff on the left arm. You note he has prominent xanthelasma, but no arcus cornealis and no tendon xanthomata.

QUESTION 2
What is the most probable diagnosis?

QUESTION 3
How do you calculate Doug’s likelihood of having an inherited cholesterol disorder?

QUESTION 4
What are the most important confounding factors to consider before making this diagnosis?
### Table 2. Calculation of Dutch Lipid Clinic Network Score (DLCNS) for Doug

<table>
<thead>
<tr>
<th>Criteria</th>
<th>Score</th>
<th>Patient score (Doug)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Family history</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>First-degree relative with premature coronary and/or vascular disease (male &lt;55 years, female &lt;60 years) OR First-degree relative with known low-density lipoprotein-cholesterol (LDL-C) above 95th percentile for age and gender</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>First-degree relative with tendinous xanthomata and/or arcus cornealis OR Children (&lt;18 years) with LDL-C above 95th percentile for age and gender</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td><strong>Clinical history</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient with premature coronary artery disease (male &lt;55 years, female &lt;60 years)</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>Patients with premature cerebral or peripheral vascular disease (male &lt;55 years, female &lt;60 years)</td>
<td>1</td>
<td>0</td>
</tr>
<tr>
<td><strong>Physical examination</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tendinous xanthomata</td>
<td>6</td>
<td>0</td>
</tr>
<tr>
<td>Arcus cornealis prior to 45 years of age</td>
<td>4</td>
<td>0</td>
</tr>
<tr>
<td><strong>Investigation</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>LDL-C (mmol/L) &gt;8.5</td>
<td>8</td>
<td>8</td>
</tr>
<tr>
<td>LDL-C 6.5–8.4</td>
<td>5</td>
<td></td>
</tr>
<tr>
<td>LDL-C 5.0–6.4</td>
<td>3</td>
<td></td>
</tr>
<tr>
<td>LDL-C 4.0–4.9</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>Patient total</td>
<td></td>
<td>9</td>
</tr>
<tr>
<td><strong>Diagnosis</strong></td>
<td><strong>Total</strong></td>
<td></td>
</tr>
<tr>
<td>Definite familial hypercholesterolaemia (FH)</td>
<td>&gt;8</td>
<td></td>
</tr>
<tr>
<td>Probable FH</td>
<td>6–8</td>
<td></td>
</tr>
<tr>
<td>Possible FH</td>
<td>3–5</td>
<td></td>
</tr>
<tr>
<td>Unlikely FH</td>
<td>&lt;3</td>
<td></td>
</tr>
</tbody>
</table>

A diagnosis of FH is most confidently made for a score >5 and especially >8

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**FURTHER INFORMATION**

You calculate Doug’s Dutch Lipid Clinic Network Score (DLCNS; Table 2). Doug has a DLCNS of 9, which means he has a definite clinical diagnosis of familial hypercholesterolaemia (FH).

**QUESTION 5**

What are your clinical and management priorities?

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**QUESTION 6**

What are your next steps in management, including further investigations, treatment and referral?
ANSWER 1
A full medical history with particular attention to cardiovascular history is required. In Doug’s case, a detailed family history of cardiovascular disease (CVD) and lipid disorder is also indicated. A routine cardiovascular examination should be done. Examination specifically looking for peripheral signs of lipid disorders should be done. These signs are tendon xanthomata, fatty deposits in tendon sheaths and premature arcus cornealis, the presence of a white ring of lipid deposit around the margin of the cornea in a patient under the age of 45 years. Xanthelasma palpebrarum, flat, lipid-rich growths on the eyelids, are commonly associated with elevated cholesterol and increased risk of coronary heart disease (CHD), but are not particularly predictive of inherited hypercholesterolaemia.

ANSWER 2
While lifestyle factors, diet, exercise and metabolic disease are the most likely causes for cholesterol abnormalities in the general population, Doug’s low-density lipoprotein-cholesterol (LDL-C) levels and family history are strongly suggestive of an inherited cholesterol defect. The most common of these is FH. It is estimated to occur with a frequency of at least 1 in 500 in the general population, and at higher levels in selected populations such as people from Mediterranean countries, Christian Lebanese, French Canadians and Afrikaner South Africans. It is autosomally dominant, so that in a family with an index case of FH, the likelihood of other first-degree relatives having FH is 50%. If both parents are carriers, the likelihood of FH increases to 75% with a 25% chance that an offspring will be homozygous for FH, a rare but much more severe form of the disease. Critically, FH is underdiagnosed; only 10–15% of cases are formally identified. GPs underestimate the prevalence of FH in their own practice population. The risk of premature atherosclerotic cardiovascular disease (CVD) is raised many times in patients with FH, in some by 25 times the population risk.

ANSWER 3
Diagnosis can be made on clinical findings, history, examination and fasted, untreated LDL-C levels. An LDL-C of >5 mmol/L raises the suspicion of FH. The likelihood of FH can be quantified using a tool such as the DLCNS, a diagnostic algorithm that incorporates the patient’s cardiovascular history, family history, clinical findings, such as arcus cornealis and tendon xanthomata, and their LDL-C level to give a score and probability level of FH. Online calculators for DLCNS are readily available.

ANSWER 4
Important confounding factors, for which the patient must be screened before making the diagnosis of FH include nephrotic syndrome, diabetes, hypothyroid disease and corticosteroid use, as these will contribute to hypercholesterolaemia.

ANSWER 5
FH can be subdivided into low, intermediate and high complexity. Low-complexity cases can be managed in primary care by the patient’s general practitioner. Intermediate complexity includes patients with CVD risk factors and/or minor issues around achieving ideal LDL-C levels and should involve shared care between the GP and an FH specialist centre. High-complexity cases will involve patients with multiple cardiovascular risk factors and difficult-to-control LDL-C levels, and should be managed by an FH specialist centre.

Clinical priorities include three important dimensions. First, Doug needs treatment. He has an inherited metabolic disorder of cholesterol, which has been present and active from birth. The likelihood of vascular disease in Doug at 36 years of age is very high and will increase the longer he is untreated. He should be screened for existing coronary artery disease. Second, he may need specialist involvement and genetic testing. While management of low-complexity cases of FH is well within the scope of GPs, specialist involvement can assist with difficulties with treatment options, rare cases of homozygous FH, provision of genetic testing and cascade screening of affected families. Genetic testing can confirm the diagnosis and streamline further case discovery within families. The third priority is discussion with Doug about cascade screening of his relatives. Cascade screening is the term used for screening family members of an index case of FH. It is the most cost-efficient way of finding new cases of FH. Cascade screening involves important discussions around disclosure of confidential health information and legal and ethical risks around discovering genetic disease in clinically unaffected family members.

Cascade screening services may be offered by some specialist clinics. Doug has a sibling and three children who are at risk of FH, and may be diagnosed and treated before significant atherosclerosis occurs. Screening of children remains controversial, but should definitely be considered in families affected by FH or with a strong family history of premature atherosclerotic CVD. In the Australian setting a collaborative model of care is advocated involving primary care and specialist services.

ANSWER 6
Further investigations include fasting glucose testing to screen for diabetes, thyroid function tests for thyroid disease, and proteinuria for renal disease, if not already done. In addition, if starting a statin it is worth checking liver function enzymes in the first 4–12 months.

First-line management of Doug will involve advice about lifestyle risks for CVD, including diet, exercise, smoking, and treatment with a cholesterol-lowering agent, which is a high-potency statin in most patients. Other agents may be required if statins cannot be tolerated because of side effects, or if there is incomplete treatment of LDL-C levels. Other agents include ezetimibe and bile acid sequestrants. Target levels for treatment are an LDL-C <2.5 mmol/L with no CVD, and 1.8 mmol/L with evidence of CVD. Referral to specialist lipid and/or cardiology services should be considered for several reasons. First, Doug’s current coronary artery status should be assessed. Computed tomography (CT) coronary angiography is a useful tool in excluding pre-existing atherosclerotic CVD in patients at
low-to-intermediate risk. Under the current Medical Benefits Schedule (MBS), this is restricted to specialist or consultant physician referral. Currently in Australia, use of CT coronary angiography in the asymptomatic patient remains controversial. If FH has been formally diagnosed there may be a stronger case to proceed. Genetic testing should be considered in Doug’s case. Identification of the genetic mutation allows better prognostic evaluation of the dyslipidaemia. It may facilitate novel treatments in more complex cases of FH. It also enables the accurate identification of family members at risk.

CONCLUSION

FH is the most common inherited cholesterol disorder and remains significantly underdiagnosed. All patients will present to general practice at some stage, often after having screening cholesterol tests. A high LDL-C value should trigger further investigations for FH. Identification of a patient with FH should always involve a discussion about the possibility of cascade screening of other family members. A collaborative model of care involving both GPs and specialist service is advocated.

RESOURCES FOR DOCTORS

- FH Australasia Network is an excellent online resource for Australian GPs that provides information on FH, inheritance, diagnosis referral, treatment and an online diagnostic tool can be found here. There is also an updated list of specialists for each state should referral be required, www.athero.org.au/fh

REFERENCES

CASE 3

ANTHEA COMES FOR A CHECK-UP

Anthea, 53 years of age, presents for a routine check-up. She was initially seen by the practice nurse. Her body mass index (BMI) was 25 kg/m². Blood pressure was 155/95 mmHg. Anthea has been otherwise well and has an unremarkable past medical history. She is a smoker, having smoked since the age of 22 years. Her father has type 2 diabetes, hypertension, high cholesterol and ischaemic stroke.

QUESTION 1

What are the key aspects in the assessment of Anthea?

FURTHER INFORMATION

Anthea exercises once or twice per week. This consists of relaxed walking for 15 minutes at a time. She has been a smoker since the age of 22 years, smoking half a pack of cigarettes per day. Three years ago, she tried to quit (‘cold turkey’), but this only lasted for one week. She drinks one to two standard drinks of alcohol per day.

Anthea is a high school librarian. She has been stressed in the past two to three months, as her new boss at work is demanding. Anthea was born in Australia to parents of Anglo-Celtic background.

You measure Anthea’s blood pressure from both arms with an interval of 10 minutes between the two measurements. The blood pressure from the left arm is 155/95 mmHg and right is 150/90 mmHg. Anthea recently purchased a home blood pressure machine.

QUESTION 2

Are Anthea’s clinic blood pressure measurements acceptable? Should you advise her to check her blood pressure at home? What else might you advise? List the pros and cons of each option.

FURTHER INFORMATION

Anthea is referred for 24-hour ambulatory blood pressure monitoring. The results show:
- average blood pressure = 153/93 mmHg
- day time blood pressure average = 158/96 mmHg
- night time blood pressure average = 145/88 mmHg
(<10% difference).

Cardiovascular examination shows:
- pulse 88 beats per minute, regular
- apex beat normal position, heart sounds dual nil murmur
- jugular vein pressure not raised, nil evidence of congestive cardiac failure.

QUESTION 3

What other physical examinations and/or office tests would you perform?

FURTHER INFORMATION

Anthea is referred for 24-hour ambulatory blood pressure monitoring. The results show:
- average blood pressure = 153/93 mmHg
- day time blood pressure average = 158/96 mmHg
- night time blood pressure average = 145/88 mmHg
(<10% difference).

Cardiovascular examination shows:
- pulse 88 beats per minute, regular
- apex beat normal position, heart sounds dual nil murmur
- jugular vein pressure not raised, nil evidence of congestive cardiac failure.

QUESTION 4

What (if any) investigations would you order?

FURTHER INFORMATION

The rest of Anthea’s physical examination is normal. The urinalysis shows no proteins, glucose or blood.

The results of Anthea’s blood test and urine examination, which you receive a few days later, are shown in Table 1.
Table 1. Anthea’s blood test and urine examination

<table>
<thead>
<tr>
<th></th>
<th>Anthea’s results</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Urine</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Creatinine</td>
<td>14.7 mmol/L</td>
<td>12.0–18.0 mmol/L</td>
</tr>
<tr>
<td>Albumin</td>
<td>5.2 mg/L</td>
<td>0.3–1.5 mg/L</td>
</tr>
<tr>
<td>Albumin:creatinine</td>
<td>0.4 mg/mmol</td>
<td>0.0–3.0 mg/mmol</td>
</tr>
<tr>
<td><strong>Blood</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sodium</td>
<td>139 mmol/L</td>
<td>135–145 mmol/L</td>
</tr>
<tr>
<td>Potassium</td>
<td>3.5 mmol/L</td>
<td>3.5–5.5 mmol/L</td>
</tr>
<tr>
<td>Chloride</td>
<td>103 mmol/L</td>
<td>95–110 mmol/L</td>
</tr>
<tr>
<td>Urea</td>
<td>3.6 mmol/L</td>
<td>1.5–6.5 mmol/L</td>
</tr>
<tr>
<td>Creatinine</td>
<td>65 µmol/L</td>
<td>45–95 µmol/L</td>
</tr>
<tr>
<td>Estimated glomerular filtration rate</td>
<td>&gt;90 mL/min/1.73 m$^2$</td>
<td>&gt;59 mL/min/1.73 m$^2$</td>
</tr>
<tr>
<td>Cholesterol</td>
<td>5.6 mmol/L</td>
<td>2.9–5.5 mmol/L</td>
</tr>
<tr>
<td>Triglycerides</td>
<td>3.3 mmol/L</td>
<td>0.5–1.7 mmol/L</td>
</tr>
<tr>
<td>High-density lipoprotein-cholesterol</td>
<td>0.9 mmol/L</td>
<td>0.9–2.1 mmol/L</td>
</tr>
<tr>
<td>Low-density lipoprotein-cholesterol</td>
<td>4.5 mmol/L</td>
<td>1.7–3.5 mmol/L</td>
</tr>
<tr>
<td>Glucose fasting</td>
<td>5.0 mmol/L</td>
<td>3.6–6.0 mmol/L</td>
</tr>
</tbody>
</table>

**QUESTION 7**
How will you manage Anthea? Consider non-pharmacological and pharmacological options.

**FURTHER INFORMATION**

After two months of trying non-pharmacological options, Anthea’s blood pressure has remained unchanged. She is started on ramipril 10 mg daily and atorvastatin 20 mg daily.

Anthea returns to you after four weeks. Her blood pressure is 147/93 mmHg.

**QUESTION 8**
What is your blood pressure target for Anthea?

**FURTHER INFORMATION**

You discuss with Anthea and agree to increase her medications. Two options are available – increase the ramipril dose or start a second medication.

**QUESTION 9**
What will you do? If you opt for the latter, what class of medications will you start?
ANSWER 1

The key aspects in further assessment of Anthea include:

- Establishing a diagnosis of hypertension
- Determining the severity of hypertension
- Ruling out secondary causes of hypertension, including screening for obstructive sleep apnoea (e.g., using the Pittsburgh Sleep Quality Index)
- Assessing contributing lifestyle and other modifiable risk factors for CVD
- Assessing for complications/target organ damage
- Assessing the absolute CVD risk.

Other preventive activities include:

- Comprehensive smoking assessment and smoking, nutrition, alcohol and physical (SNAP) risk factor assessment
- History of screening activities – relevant ones including cervical cancer screening, breast cancer screening, colorectal cancer screening, osteoporosis screening (coupled with determination of menopause status).

ANSWER 2

The 2016 edition of the National Heart Foundation of Australia’s Guide to management of hypertension in adults recommends that patients with a new diagnosis (or potential diagnosis) of hypertension be evaluated initially with either home blood pressure monitoring or with 24-hour ambulatory blood pressure monitoring (Table 2). Studies have shown that these measurements correlate better with complications and morbidity, compared with in-clinic measurements. Some patients experience the ‘white coat’ effect, which results in a higher blood pressure when taken in clinical environments.

ANSWER 3

Given Anthea’s presentation, it is prudent to:

- Do a urinalysis (protein and blood in the urine may indicate end-organ renal disease)
- Listen for carotid artery bruits
- Examine the fundi for hypertensive changes
- Examine the peripheral vascular system for evidence of arterial disease
- Record an electrocardiogram (ECG) to look for left ventricular hypertrophy and ischaemic changes
- Examine the abdomen, looking for evidence of arterial disease (e.g., abdominal aortic aneurysm and renal bruits that may signify renal artery stenosis), and ballot the kidneys for polycystic kidney disease.

ANSWER 4

It is appropriate to check:

- Fasting cholesterol and lipids
- Blood glucose levels
- Electrolytes and renal function
- Urine examination, especially urinary albumin-to-creatinine ratio (UACR) to exclude end-organ damage from hypertension and exclude renal parenchymal disease that may contribute to high blood pressure.

ANSWER 5

Anthea’s clinic blood pressure and 24-hour ambulatory blood pressure monitoring confirm that she has hypertension. The next step after confirming the diagnosis is to determine the severity. Table 3 provides good guidance. According to the current National Heart Foundation of Australia’s guidelines, Anthea has Grade 1 (mild) hypertension.

In some cases, the application of 24-hour blood pressure monitoring or home measurements may result in a lower grade of hypertension, compared with clinic measurements. In this case, the clinic measurement should be used when estimating absolute CVD risk using available risk assessment calculators.

ANSWER 6

It is recommended that the Australian absolute cardiovascular disease risk calculator (an initiative of the National Vascular Disease Prevention Alliance and endorsed by The Royal Australian College of General Practitioners [RACGP]) be used to determine a patient’s CVD risk. A convenient online calculator is available for those with internet access (www.cvdcheck.org.au). Different charts are available for women and men, smokers and non-smokers, and those with or without diabetes. The chart suitable for Anthea is shown in Figure 1. Anthea has a 14% risk of having a cardiovascular event (e.g., acute myocardial infarction, stroke, cardiac death) over the next five years. The Australian absolute cardiovascular disease risk chart and

<table>
<thead>
<tr>
<th>Table 2. Home blood pressure monitoring compared with 24-hour ambulatory blood pressure monitoring</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
</tr>
<tr>
<td><strong>Patient monitoring at home</strong></td>
</tr>
<tr>
<td>Advantages</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>Disadvantages</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
</tbody>
</table>

(1) According to the current National Heart Foundation of Australia’s guidelines, Anthea has Grade 1 (mild) hypertension.
check Cardiology

CASE 3

The Framingham heart study – a long-term cohort study of more than 5000 people living in the town of Framingham in Massachusetts, US. It is useful for estimating the cardiovascular risk but has several limitations. For instance, it does not take into account a person’s family history, ethnicity, body mass or waist circumference, or other associated conditions except for diabetes. The use of the chart in Aboriginal and Torres Strait Islander patients may result in underestimation of their risk, so it should be used carefully in this patient group. However, available evidence suggests that this approach will provide an estimate of minimum CVD risk (refer to https://heartfoundation.org.au/images/uploads/publications/aust-cardiovascular-risk-charts.pdf).

It is recommended that absolute CVD risk assessment be performed regularly in all adults aged 45 and older (or 35 years and older for Aboriginal and Torres Strait Islander peoples) without existing CVD or not already known to be at increased risk of CVD. The calculator should not be used in those that are already at increased (high) risk of cardiovascular disease. These patient groups include those with:

- diabetes and aged >60 years
- diabetes with microalbuminuria (>20 µg/min or UACR >2.5 mg/mmol for males and >3.5 mg/mmol for females)
- moderate or severe chronic kidney disease (persistent proteinuria or estimated glomerular filtration rate (eGFR) <45 mL/min/1.73 m²)
- patients with familial hypercholesterolaemia
- systolic blood pressure ≥180 mmHg or diastolic blood pressure ≥110 mmHg
- serum total cholesterol >7.5 mmol/L.

A number of other calculators have been developed overseas and some of these take into account factors such as family history. An example is the Joint British Societies recommendations on the prevention of cardiovascular disease (JBS3; www.jbs3risk.com), which is recommended for use in the UK.

### Table 3. Classification of clinic blood pressure levels in adults

<table>
<thead>
<tr>
<th>Diagnostic category*</th>
<th>Systolic (mmHg)</th>
<th>Diastolic (mmHg)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Optimal</td>
<td>&lt;120</td>
<td>and</td>
</tr>
<tr>
<td>Normal</td>
<td>120–129</td>
<td>or</td>
</tr>
<tr>
<td>Normal-to-high</td>
<td>130–139</td>
<td>and/or</td>
</tr>
<tr>
<td>Mild hypertension (Grade 1)</td>
<td>140–159</td>
<td>and/or</td>
</tr>
<tr>
<td>Moderate hypertension (Grade 2)</td>
<td>160–179</td>
<td>and/or</td>
</tr>
<tr>
<td>Severe hypertension (Grade 3)</td>
<td>≥180</td>
<td>and/or</td>
</tr>
<tr>
<td>Isolated systolic hypertension</td>
<td>&gt;140</td>
<td>and</td>
</tr>
</tbody>
</table>

*If systolic and diastolic blood pressure levels fall into different categories, the higher diagnostic category and recommend actions apply.

---

**Figure 1. CVD chart for women without diabetes**

**Women**

<table>
<thead>
<tr>
<th>Systolic blood pressure (mm Hg)</th>
<th>Non-smoker</th>
<th>Smoker</th>
</tr>
</thead>
<tbody>
<tr>
<td>179*</td>
<td></td>
<td></td>
</tr>
<tr>
<td>160</td>
<td></td>
<td></td>
</tr>
<tr>
<td>150</td>
<td></td>
<td></td>
</tr>
<tr>
<td>140</td>
<td></td>
<td></td>
</tr>
<tr>
<td>120</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Total cholesterol: HDL ratio**

*In accordance with Australian guidelines, patients with systolic blood pressure ≥180 mm Hg, or a total cholesterol of >7.5 mmol/L, should be considered at clinically determined high absolute risk of CVD.

### Risk level for 5-year cardiovascular (CVD) risk

<table>
<thead>
<tr>
<th>High risk</th>
<th>Moderate risk</th>
<th>Low risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>≤30%</td>
<td>10–15%</td>
<td>5–9%</td>
</tr>
<tr>
<td>25–29%</td>
<td>16–19%</td>
<td>&lt; 5%</td>
</tr>
<tr>
<td>20–24%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>16–19%</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Charts in this age bracket are for use in Aboriginal and Torres Strait Islander Populations only.
ANSWER 7
Advice on lifestyle changes is paramount in managing Anthea’s CVD risk and her blood pressure. This includes SNAP risk factors – smoking cessation, nutrition, alcohol and physical activity. Dietary advice should include lowering salt intake, reducing saturated fat intake, avoiding eating liquorice, and increasing the intake of oily fish and dietary fibre. Anthea should be encouraged to be active – most guidelines recommend at least 30 minutes a day, five days a week of moderate intensity aerobic exercise. Referral to a dietitian or exercise physiologist may be useful. Motivational interviewing and the use of the 5As framework (www.racgp.org.au/your-practice/guidelines/smoking-cessation/the-5-structure-for-smoking-cessation), including pharmacotherapy could be used to help Anthea quit smoking.

Anthea’s stress may also be contributing to her high blood pressure. Counselling, advice on relaxation and stress reduction techniques may prove to be helpful. Referral to a psychologist or counselling service may be required if Anthea does not respond to interventions initiated by the general practitioner.

Initially, Anthea should be trialled on non-pharmacological management for a few weeks. Given she has intermediate CVD risk (10–15% over five years), if Anthea’s blood pressure remains above 140/90 mmHg despite lifestyle changes, then pharmacological treatment is indicated. On the other hand, if Anthea’s CVD risk were <10% (ie low risk), then antihypertensive therapy would be recommended only for persistently high blood pressure ≥160/100.

Once a decision has been made to start antihypertensives, consideration should be given to starting a statin to further reduce her CVD risk. The benefits of statins in reducing CVD risk appears to go beyond what may be expected from the reduction in cholesterol levels. Because she has hypertension, if Anthea’s total cholesterol remains >5.5 mmol/L and HDL <0.9 mmol/L after six weeks of lifestyle changes, then she would qualify for Pharmaceutical Benefits Scheme (PBS) subsidy (Table 4).

ANSWER 8
Until recently, it was recommended that patients should aim for a blood pressure target of 140/90 mmHg when titrating medications (higher in elderly or those with side effects). However, the results of a large randomised controlled trial (the SPRINT study) showed that patients whose medications were titrated to a more intensive lower blood pressure target had significantly better outcomes compared with those with a higher target.

Therefore, the revised 2016 National Heart Foundation of Australia’s guidelines recommend that in selected high CVD-risk populations (ie in those with an estimated absolute CVD 10-year risk of at least 20% who were recruited into the SPRINT trial), a more intense treatment can be considered, aiming for a target of <120 mmHg systolic blood pressure to improve CVD outcomes (grade of recommendation = strong, level of evidence = II). This would be more important in those with end-organ damage or existing CVD, or those with significant risk factors. These patients will need to be monitored carefully for symptoms of postural hypotension and other adverse effects (e.g hyperkalaemia in patients taking angiotensin converting enzyme inhibitor or angiotension receptor antagonist).

ANSWER 9
It is preferable to start Anthea on a second medication. Maximising the dose of one medication may result in more side effects and may not result in satisfactory control of the blood pressure.

Anthea is currently taking an angiotensin converting enzyme inhibitor, which is an appropriate first-line agent for hypertension. An angiotension receptor II antagonist is an acceptable alternative, but the two should not be prescribed together because of the risk of side effects (hyperkalaemia...

<table>
<thead>
<tr>
<th>Patient category</th>
<th>Lipid levels for PBS subsidy</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patients with diabetes mellitus not otherwise included</td>
<td>total cholesterol &gt;5.5 mmol/L</td>
</tr>
<tr>
<td>Aboriginal or Torres Strait Islander patients</td>
<td>total cholesterol &gt;6.5 mmol/L</td>
</tr>
<tr>
<td>Patients with hypertension</td>
<td>OR total cholesterol &gt;5.5 mmol/L and HDL cholesterol &lt;1 mmol/L</td>
</tr>
<tr>
<td>Patients with HDL cholesterol &lt;1 mmol/L</td>
<td>total cholesterol &gt;6.5 mmol/L</td>
</tr>
<tr>
<td>Patients with familial hypercholesterolaemia identified by:</td>
<td>If aged more than 18 years at treatment initiation:</td>
</tr>
<tr>
<td>• DNA mutation; or</td>
<td>LDL cholesterol &gt;5 mmol/L</td>
</tr>
<tr>
<td>• tendon xanthomas in the patient or their first or second degree relative</td>
<td>OR total cholesterol &gt;6.5 mmol/L</td>
</tr>
<tr>
<td>Patients with:</td>
<td>OR total cholesterol &gt;5.5 mmol/L and HDL cholesterol &lt;1 mmol/L</td>
</tr>
<tr>
<td>• family history of coronary heart disease which has become symptomatic before the age of 60 years in one or more first degree relatives; or</td>
<td></td>
</tr>
<tr>
<td>• family history of coronary heart disease which has become symptomatic before the age of 50 years in one or more second degree relatives</td>
<td></td>
</tr>
<tr>
<td>Patients not eligible under the above:</td>
<td>total cholesterol &gt;7.5 mmol/L OR triglyceride &gt;4 mmol/L</td>
</tr>
<tr>
<td>• men aged 35 to 75 years</td>
<td></td>
</tr>
<tr>
<td>• post-menopausal women aged up to 75 years</td>
<td>total cholesterol &gt;9 mmol/L OR triglyceride &gt;8 mmol/L</td>
</tr>
<tr>
<td>Patients not otherwise included</td>
<td></td>
</tr>
</tbody>
</table>


Table 4. PBS-subsidy for lipid-lowering drugs
and increased risk of renal dysfunction). The second agent could be a calcium channel antagonist. Thiazide diuretics could also be considered. Beta-blockers, while not contraindicated, are not recommended at this stage because of adverse effects profile, unless Anthea has pre-existing ischaemic heart disease.

RESOURCES FOR DOCTORS

- [www.cvdcheck.org.au](http://www.cvdcheck.org.au)

REFERENCES

CASE 4

JOHN HAS A MURMUR

John, four months of age, was seen by a colleague at your practice two months ago. At the time, John was unwell with a viral upper respiratory tract infection (URTI) and a heart murmur was heard. His mother brings him to see you for a review.

QUESTION 1

What are the important points from the history?

FURTHER INFORMATION

There is no family history of congenital heart disease in John’s extended family. His mother had poorly controlled gestational diabetes mellitus and denies taking any illicit drugs. John used to feed well and was thriving; however, this has changed over the past two months and he feeds more frequently but only takes small volumes at each feed. His mother says, he ‘looks exhausted’ after each feed.

QUESTION 2

What are the important clinical findings?

FURTHER INFORMATION

On examination, John has subtle features of trisomy 21 (Down syndrome) and has mildly increased work of breathing. John’s heart rate is 130 beats per minute, respiratory rate 45 breaths per minute, and saturations of 98% (SpO2). On palpation, he has an obvious heave, strong femoral pulses and mild hepatomegaly. On auscultation, he has a 3/6 pan-systolic murmur at the left sternal edge with no obvious radiation. There are no clicks or additional heart sounds.

QUESTION 3

What are your differential diagnoses?

FURTHER INFORMATION

You referred John for cardiology review. When seen by the cardiology team, John was diagnosed with a complete atrioventricular septal defect (AVSD). John underwent surgery to repair the AVSD. The postoperative period was uneventful and he was discharged on day seven with a planned review by the cardiologist after two weeks and you in one week.
CASE 4

At the time of follow-up, John’s mother reports that his feeding has improved and he no longer appears out of breath. She is coping with giving the medication and he continues to be more and more active. There has been no history of fevers and John’s sternal wound and drain sites are healing well with no evidence of infection. On examination, he is well hydrated and active. On palpation, John has a normal precordium, pulses and no hepatomegaly; on auscultation, he has dual heart sounds with no murmurs.

When John is seen for suture removal in the postoperative period, his mother reports that John’s sister Jody, 13 years of age, has been complaining of chest pain and occasional dizziness. She asks whether this could be related to John’s heart problem.

QUESTION 5

How would you respond to John’s mother?

ANSWER 1

Symptoms arising from congenital heart disease vary widely. Infants may initially present with failing to thrive and a history of short, frequent feeds. These may be associated with diaphoresis, tachypnoea and signs of respiratory distress such as subcostal recession and nasal flaring. By contrast, older children may have a history of unexplained lethargy and reduced exercise tolerance when compared with their peers for age-specific activities (e.g., walking up the stairs, playing organised sports). Of course, it is also possible that children with even quite significant congenital heart lesions may be completely asymptomatic and have normal exercise tolerance.

A comprehensive history starts from the antenatal period, looking for potential risk factors of congenital heart disease. In up to 20% of cases, the condition is directly related to either genetic abnormalities or teratogens, but these factors may not be identified as part of the history. A family history should be sought of structural cardiac abnormalities and of maternal complications of pregnancy (especially gestational diabetes mellitus), illness during pregnancy (e.g., infection, although less common these days), prescribed or non-prescribed medications.

ANSWER 2

A basic set of observations is an important first step in assessing the child with a murmur. This includes respiratory rate, heart rate, peripheral saturations and blood pressure. Particular attention should be made to respiratory effort as a sign of cardiac failure. It is important to examine for any dysmorphic features that may be associated with congenital heart disease (Table 1). As children can be difficult to examine, it is important to be flexible and opportunistic in approach.

Characterisation of the murmur is important; however, this can be challenging as heart rate is much faster in infants. Innocent murmurs are those arising from increased velocity of blood flow through a structurally normal heart and, typically, are soft, with an intensity ≤ 2/6*, heard in systole and ejection in character.

They may be continuous in the setting of a venous hum, although these murmurs disappear on lying flat. Conversely, harsh murmurs, those that are pan-systolic, diastolic, or continuous (except venous hums) are pathological. Similarly, murmurs that increase in intensity with either positional changes (e.g., squatting to standing) or Valsalva manoeuvres are pathological. While of interest, most young children are not cooperative enough to perform these physiological manoeuvres. Furthermore, additional sounds, such as clicks and fourth heart sounds, are, by definition, pathological. With experience, clinicians may evaluate with high sensitivity whether murmurs are innocent or pathological.

Particular attention must be taken to feel for femoral pulses, looking for a significant coarctation, and hepatomegaly. The internal jugular vein is difficult to see in infants and young children, so clinicians tend to rely on liver size as a marker of elevated central venous pressure. Oedema is rarely seen in heart failure in children, but if present in the infant, is typically seen in the peri-orbital/scrotal/labial regions; in older children, it is more likely to be pedal in location. Basal lung crepitations are rarely heard in young children with heart failure.

* A 1/6 murmur is barely audible; 2/6 audible but soft; 3/6 is easily audible but without a thrill; 4/6 murmur has a thrill; 5/6 is with a thrill and heard with the stethoscope hardly touching the chest; 6/6 murmur has a thrill and is audible without the stethoscope on the chest.

Table 1. Selected syndromes and associated cardiac lesions

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Cardiac lesion</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fetal alcohol syndrome</td>
<td>Ventricular septal defect (VSD), atrial septal defect (ASD)</td>
</tr>
<tr>
<td>Trisomy 21</td>
<td>Atrioventricular septal defect (AVSD), ASD, VSD</td>
</tr>
<tr>
<td>Turner syndrome</td>
<td>Coarctation of the aorta, bicuspid aortic valve</td>
</tr>
<tr>
<td>Noonan syndrome</td>
<td>Pulmonary stenosis, ASD, hypertrophic cardiomyopathy</td>
</tr>
<tr>
<td>Williams syndrome</td>
<td>Supravalvular aortic stenosis, peripheral pulmonary stenosis</td>
</tr>
<tr>
<td>Di George syndrome</td>
<td>Aortic arch and conotruncal abnormalities</td>
</tr>
<tr>
<td>Marfan syndrome</td>
<td>Mitral valve prolapse</td>
</tr>
</tbody>
</table>
ANSWER 3
The prevalence of congenital heart disease has been reported as up to 1 in 100,\(^6\) with ventricular septal defects being the most common, making up to one-third of these, followed by atrial septal defects, patent ductus arteriosus and pulmonary stenosis. However, the incidence of severe congenital heart disease requiring expert management early in life is roughly 3 in 1000.\(^7\)

Congenital heart disease can be broadly divided into cyanotic and acyanotic lesions. Acyanotic lesions can be further subdivided into left-to-right shunts and obstructive lesions. Septal defects and patent ductus arteriosus are examples of left-to-right shunts, whereas pulmonary/aortic valve stenosis and coarctation are typical obstructive lesions.

Cyanotic lesions include tetralogy of Fallot, truncus arteriosus, transposition of the great arteries, total anomalous pulmonary venous drainage and more complex single ventricle hearts (eg hypoplastic left heart syndrome). Trisomy 21 is typically associated with an AVSD (Table 1).

ANSWER 4
John’s murmur may have changed over the two months since your colleague saw him because of the natural ongoing fall in pulmonary vascular resistance in the first three months of life and also because John had a viral URTI at the time of his last assessment. His tachypnoea, increase in work of breathing and hepatomegaly are indicators of an important left-to-right shunt now and he should have a fairly prompt cardiology review. An electrocardiogram (ECG) may be helpful in making the diagnosis of an AVSD with demonstration of a superior axis.

An AVSD can be repaired surgically. This is a semi-elective procedure usually performed in children with Down syndrome at around the age of three to four months to avoid the complications of pulmonary hypertension. While waiting for surgery, medical treatment with diuretics, with or without an angiotensin converting enzyme inhibitor, and a high-calorie diet can be commenced.\(^8\)

ANSWER 5
Chest pain is a very uncommon manifestation of heart disease in children and is much more likely to be musculoskeletal, gastrointestinal or pulmonary in nature. A thorough history usually confirms the non-cardiac origin of the chest pain. The nature, location and timing of chest pain often provide the aetiology; however, associated features should also be discussed. In a teenager, it should be possible to elicit whether there are symptomatic palpitations. A history of palpitations should lead to questioning over how these start and stop (eg abruptly in re-entrant tachycardia), duration, frequency and associated symptoms. A family history of arrhythmia, sudden death or epilepsy is useful to stratify risk. Timing and precipitating factors for presyncopal/syncopal episodes are very important to elicit because those with symptoms during exertion or sudden fright may be at higher risk.\(^9\)

CONCLUSION
John’s mother brings Jody in to see you. She describes a sharp, right-sided chest pain during which it is difficult to take a deep breath and which is not associated with palpitations or pre/syncope. The dizzy episodes occur only on hot days when she has been standing for long periods.

REFERENCES
CASE 5
BILL PRESENTS WITH FATIGUE AND SHORTNESS OF BREATH ON EXERTION

Bill, 55 years of age, has been feeling tired and lethargic since ‘coming down with the flu’ four weeks ago. He is breathless on exertion and describes an irritating cough, which is worse at night. Bill visited another general practitioner (GP) two weeks ago and was commenced on antibiotics, but is feeling worse, so he is seeking a second opinion. He is not on any other medication.

QUESTION 1
What assessments would you conduct to establish the diagnosis?

FURTHER INFORMATION
Bill reports that he first noticed being breathless prior to visiting the previous GP. Despite the antibiotics, his symptoms have been getting worse. He now notices that he gets breathless when he has to walk quickly or up a slope. Bill reiterates that he feels very tired, but that this may be due to not sleeping well because of frequently coughing at night. He gets relief by propping himself up on three pillows.

Chest auscultation reveals inspiratory crepitation, which does not clear with coughing. On further examination, Bill looks pale and has a blood pressure of 100/60 mmHg, heart rate of 95 beats per minute (regular) and body temperature of 36.5°C. He mentions that his weight has increased by 3 kg in the past month. He also has bilateral pitting oedema in the lower legs.

QUESTION 2
What are your initial diagnosis and differential diagnoses? What further assessments would you undertake to investigate the cause of Bill’s dyspnoea?

FURTHER INFORMATION
The electrocardiogram (ECG) reveals a left bundle branch block and suggested left ventricular hypertrophy. A third heart sound is noted on chest auscultation. Further examination reveals elevated jugular venous pressure. These observations support the diagnosis of congestive heart failure (CHF).

QUESTION 3
What investigations should you request to confirm the diagnosis?

FURTHER INFORMATION
Echocardiography confirms a diagnosis of impaired left ventricular function, with an ejection fraction of 32%.

QUESTION 4
How will you manage Bill’s condition?

FURTHER INFORMATION
Echocardiography confirms a diagnosis of impaired left ventricular function, with an ejection fraction of 32%.

QUESTION 5
How will you monitor Bill’s cardiac function? How will you manage any changes in cardiac function?
QUESTION 6
What is the prognosis for CHF? What secondary prevention measures can be implemented to improve the prognosis?

ANSWER 1
You should seek further information about the primary symptom of dyspnoea. Questions to ask include:
• What is the time-course of its onset?
• How severe are the symptoms?
• Are there other associated symptoms (ie chest pain/discomfort)?
You should also examine Bill to check for signs of oedema and, if present, whether this is affected by body position. Undertake a respiratory examination and check his vital signs.

ANSWER 2
Bill’s elevated heart rate, low blood pressure and peripheral oedema indicate the need for a cardiovascular investigation for a possible diagnosis of CHF. Differential diagnoses on initial assessment may include respiratory conditions, including pulmonary emboli, respiratory tract infection, pneumonia or asthma. Swollen ankles may indicate nephrotic syndrome. Further assessments would be to perform cardiac auscultation and a resting ECG.

ANSWER 3
Chest X-ray
A chest X-ray is useful in making a diagnosis of heart failure, but a normal result does not exclude the diagnosis. An increase in cardiothoracic ratio of more than 0.50 indicates cardiomegaly.

Trans-thoracic echocardiography
All patients with suspected heart failure should have an echocardiogram. This is the most effective investigation for determining suspected heart failure because of impaired left ventricular (LV) systolic function or other causes. The echocardiogram gives information about:

- LV and right ventricular (RV) size, volumes and wall thickness
- the presence of regional scarring
- LV and RV systolic function – ejection fraction <40% is highly indicative of heart failure. An ejection fraction of 41–49% may be considered borderline heart failure, but does not always indicate that a person has heart failure
- LV thrombus
- LV diastolic function and filling pressures
- left and right atrial size – enlargement is an important manifestation of chronically elevated filling pressure
- valvular structure and function – assessment of the severity of valvular stenosis or incompetence and whether CHF can be explained by the valve lesion
- pulmonary systolic pressure – in most patients this can be estimated by Doppler echo.

Blood tests
- Brain natriuretic peptide (BNP) – an elevated BNP level is helpful in making a diagnosis of heart failure in patients with unexplained dyspnoea, especially when an echocardiogram is not immediately available.
- Full blood evaluation – anaemia may occur in patients with CHF.
- Urea, creatinine and electrolytes – usually normal in mild-to-moderate heart failure, but may become deranged with more severe heart failure, use of diuretics and angiotensin converting enzyme inhibitors and renal dysfunction secondary to worsening heart failure. It may help to rule out nephrotic syndrome if the values are normal.
- Liver function tests – congestive hepatomegaly results in abnormal liver function tests.
- Thyroid function tests – hyperthyroidism and hypothyroidism are less common causes of heart failure.
- D-dimer testing – to exclude pulmonary emboli.

Bill’s symptoms and examination findings all indicate a diagnosis of CHF consistent with cardiomyopathy, with probable viral aetiology (myocarditis). A definitive diagnosis should be established urgently. Bill should be advised to return once the diagnosis is confirmed for review of the results, symptoms and medication tolerance, and measurement of blood pressure and heart rate. If his condition deteriorates further, he should be referred for urgent cardiology review or present to an emergency department.

ANSWER 4
Provide symptomatic relief initially while awaiting a confirmed diagnosis:
- diuretic regime: 40 mg frusemide for three days
- fluid restriction of <1.5 litres/day and salt avoidance.

Advise Bill to avoid alcohol consumption.

Following heart failure diagnosis:
- Initially, prescribe an angiotensin converting enzyme inhibitor, titrating as tolerated to the optimal dose.
- Add a heart failure–specific beta blocker, titrating as tolerated to the optimal dose.
For patients with end-stage heart failure (New York Heart Association Functional Class III–IV) and refractory to conventional medical management (Table 1), advanced therapies include inotropic medication, left ventricular assist devices and cardiac transplantation (through an Advanced Heart Failure/Cardiac Transplant Service).

In patients with end-stage heart failure, advance care planning and palliation should be implemented when advanced therapies are not an option.

**Table 1. Description of patient symptomatic status according to New York Heart Association (NYHA) Functional Class**

<table>
<thead>
<tr>
<th>Classification</th>
<th>Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Class I</td>
<td>No limitations – Ordinary physical activity does not cause undue fatigue, dyspnoea or palpitations</td>
</tr>
<tr>
<td>Class II</td>
<td>Slight limitation of physical activity – Ordinary physical activity results in fatigue, palpitation, dyspnoea or angina pectoris (mild heart failure [HF])</td>
</tr>
<tr>
<td>Class III</td>
<td>Marked limitation of physical activity – Less than ordinary physical activity leads to symptoms (moderate HF)</td>
</tr>
<tr>
<td>Class IV</td>
<td>Unable to carry on any physical activity without discomfort – Symptoms of HF present at rest (severe HF)</td>
</tr>
</tbody>
</table>

**ANSWER 5**

Serial echocardiography is not indicated to monitor heart failure unless signs or symptoms change or to monitor the effect of therapeutic interventions. In the medium term (once diagnosis has been established and evidence-based medication initiated), Bill should be referred to a supervised cardiac rehabilitation program for exercise rehabilitation. Initially, this will involve short bouts of low-to-moderate intensity aerobic exercise, as symptoms allow (the patient should not become so short of breath that they cannot conduct a conversation). Frequency of exercise sessions (days per week) and duration of exercise bouts should be increased initially before increasing intensity.

In the longer term, patients with CHF will benefit from ongoing, regular aerobic exercise, as well as moderate resistance exercise (weight lifting) involving major muscle groups of the upper and lower body. Exercise prescription and supervision can be provided by an accredited exercise physiologist or physiotherapist, or a home-based exercise training regimen can be provided.

**ANSWER 6**

Prognosis, secondary prevention

Survival in patients with heart failure has improved markedly over the past 20 years, with better pharmacological therapy and the widespread use of implantable cardioverter defibrillators in patients at risk of life-threatening arrhythmias. Among patients aged 60 years, five-year survival is approximately 70% in men and 75% in women; however, prognosis remains unpredictable. With good medical management, many people with mild-to-moderate heart failure survive for extended periods, whereas in other patients, the course of the disease is more aggressive and LV function may deteriorate quickly.

To improve long-term health, lifestyle interventions should be encouraged. These include maintaining weight within the healthy range; regular exercise and physical activity; abstinence from smoking and use of illicit drugs; and avoiding excessive alcohol consumption (patients with alcoholic cardiomyopathy should abstain from alcohol). Maintenance of blood pressure at low to normal levels is essential. Monitor blood glucose regularly for diabetes or pre-diabetes.

**CONCLUSION**

Heart failure occurs in 1–2% of Australians. While prevalence increases markedly with age, and is often precipitated by myocardial infarction or hypertension, heart failure may occur in younger people following a viral illness or with no discernible cause (idiopathic cardiomyopathy). These patients frequently present to GPs in the first instance.

The course of heart failure can be unpredictable, with episodes of exacerbation between periods of relatively good health. Periods of clinical deterioration occur more frequently and are more severe as the condition progresses.
REFERENCES


CASE 1 – MICK
Mick, 70 years of age, presents with a history of pain in his right leg. He tells you that he has pain mainly in the calf area when he walks, climbs stairs or does any other exercise. The pain usually subsides when he rests, although sometimes it persists even when he sitting or resting. Your examination of Mick’s leg, including measurement of his ankle:brachial index (ABI), support a diagnosis of peripheral artery disease (PAD).

QUESTION 1
Which of the following ABI results supports a diagnosis of PAD?
A. 0.98  
B. 0.96  
C. 0.94  
D. 0.88

CASE 2 – WILLIAM
William, 35 years of age, comes to see you because he injured his hand in a game of volleyball. While examining William, you notice that he has xanthelasma around his eyelids. You ask William about his medical and family history and he tells you he has no medical problems. His grandfather had a heart attack at the age of 50 years, but William is not aware of any other family history. As far as he knows his parents and siblings are generally well. You refer William for an X-ray of his hand and order blood tests to check his blood lipid levels.

William returns two days later for his test results. His X-ray is normal but his blood tests show that his cholesterol and low-density lipoprotein-cholesterol (LDL-C) levels are elevated (11 and 8.9 mmol/L, respectively).

QUESTION 2
To determine whether William’s elevated cholesterol and LDL-C levels are due to familial hypercholesterolaemia (FH), you would need to:
A. calculate his Dutch Lipid Clinic Network Score (DLCNS)  
B. refer him for genetic testing  
C. exclude confounding factors such as nephrotic syndrome, diabetes, corticosteroid use and hypothyroid disease  
D. confirm a family history of FH.

FURTHER INFORMATION
You order further tests, which exclude kidney disease, hypothyroid disease and diabetes as possible contributors to William’s elevated LDL levels. Using his test results and the information he has given, you calculate his DLCNS score.

QUESTION 3
From the information given, William’s DLCNS is:
A. 8  
B. 9  
C. 10  
D. 12

QUESTION 4
William’s DLCNS indicates a diagnosis of:
A. possible FH  
B. probable FH  
C. definite FH  
D. unlikely FH.

CASE 3 – LAURA
Laura, 45 years of age, has familial hypercholesterolaemia, controlled by treatment with a statin. She is not on any other medications. She saw you recently for a repeat prescription for her cholesterol-lowering medication and a general health check. At that visit, her blood pressure was 150/98 mmHg. Previously, her blood pressure had been around 130/90 mmHg. You referred her for 24-hour ambulatory blood pressure monitoring, which showed an average blood pressure of 139/98 mmHg.
QUESTION 5
According to the 2016 Australian guidelines, how would you classify Laura’s blood pressure level?
A. Normal to high
B. Mild (Grade 1)
C. Moderate (Grade 2)
D. Severe (Grade 3)

QUESTION 6
Ideally, management of Laura should include:
A. assessing absolute cardiovascular disease (CVD) risk
B. maintaining systolic blood pressure at 139 mmHg
C. reducing diastolic blood pressure to <90 mmHg
D. all of the above

CASE 4 – ALICE
Alice, 65 years of age, presents complaining of breathlessness on exertion and general lethargy. She has also noticed some swelling around her ankles. Your examination and tests lead you to a provisional diagnosis of congestive heart failure (CHF). You refer Alice for further investigation, including blood tests, X-ray and echocardiography.

QUESTION 7
What can you recommend/prescribe for relief of Laura’s symptoms while awaiting her test results?
A. Restrict fluid intake to <1.5 L per day
B. Frusemide 40 mg daily for three days
C. Avoid alcohol
D. All of the above

FURTHER INFORMATION
Laura’s echocardiogram confirms the diagnosis of CHF. She commences treatment, which includes pharmacotherapy and recommendations for non-pharmacological management.

QUESTION 8
What advice should you give Alice about non-pharmacological management of her condition?
A. Avoid caffeinated beverages.
B. Contact you if she gains more than 2 kg in 48 hours.
C. Limit salt intake to <1 g/day
D. Limit sugar intake to <2 g/day

CASE 5 – KIRAN
Jess brings her son Kiran, six months of age, to see you because he seems to be hungry all the time and needs to feed frequently, but only has small amounts of milk at each feed. When you examine Kiran, you notice that he has a heart murmur.

QUESTION 9
The murmur might be pathological if it:
A. increases in intensity with positional changes
B. has an intensity of at least 2/6
C. is heard in systole
D. is ejection in character.

QUESTION 10
A common feature of heart failure in children is:
A. hepatomegaly
B. oedema
C. basal lung crepitations
D. jugular venous distension.