CASE 1

Short case number: 3_2_1
Category: Cardiovascular
Discipline: Medicine
Setting: General Practice
Topic: Cardiomyopathy – Hypertrophic, valvular heart disease and transplantation

Case

Heidi Tanner, aged 59 years presents with shortness of breath. She collapsed at work yesterday after exerting herself but did not go to hospital as she hates doctors and hospitals. However, today she presents because she feels quite odd. On examination she appears short of breathe and clinically is in heart failure with a loud ejection systolic murmur.

Questions

1. What further history and examination is required?
2. What initial investigations would you order?
3. What are the main causes of aortic value disease?
4. Using a table, distinguish the key features of the following valvular diseases on clinical examination: mitral stenosis, aortic stenosis, aortic incompetence, mitral incompetence.
5. How might your management be altered if Heidi was indigenous and lived in a remote community with a population of 90 people whose only medical care was delivered by a health worker once a fortnight?
6. What elements in assessment suggest that valvular surgery or cardiac transplantation is indicated?

Suggested reading:

- Colledge NR, Walker BR, Ralston SH, Penman ID, editors. Davidson's Principles and Practice of Medicine. 22nd edition. Edinburgh: Churchill Livingstone; 2014. Chapter 18.
- Talley NJ, O'Connor Clinical Examination: A systematic guide to physical diagnosis. 9th Edition. Sydney: Elsevier; 2017

Advanced reading:

- Ramaraj R. Sorrell VL. Degenerative aortic stenosis. BMJ. 336(7643):550-5, 2008 Mar 8. http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2265359/

ANSWERS

1. **Further history and examination**

The history needs to focus on:

- the episode of collapse yesterday,
- the perception of feeling "odd",
- the associated breathless.

The most important first step is to better define the "collapse" with the first questions considering whether this was an episode of pre-syncope or syncope. An accurate description needs to be obtained from the patient and if possible a witness.

Further questioning should focus on the precipitating factors which must include a history of exercise, medications, last oral intake and alcohol. The duration of the episode and the duration of recovery must allow be elicited.

A directed history of the collapse allows the clinician to begin to categorise the event into the differential diagnoses with features to argue for and against the differentials.

Physical examination should then be targeted at further differentiating the four most likely causes. In this case, the physical examination has revealed a loud ejection systolic murmur. This physical finding further defines the most likely cause of collapse as cardiac in nature. The combination of history and physical examination makes the most likely diagnosis aortic stenosis (AS).

Other clues to the presence of AS on physical examination include:

- rasping quality to the ejection systolic murmur
- radiation of the murmur to the supra-sternal notch and the carotids,
- an ejection click,
- a single second heart sound,
- left ventricular hypertrophy to palpation
- a slow rising pulse on palpation.

The differential diagnosis includes hypertrophic cardiomyopathy. On physical examination the following would be found in hypertrophic cardiomyopathy:

-jerky pulse

-double impulse at the apex (due to forceful left atrial contraction against a highly non-compliant left ventricle)

- late systolic murmur is audible at the left sternal border (which can be heard better when patient is standing or with Valsalva Maneuver).

- there is an associated pansystolic murmur audible at the apex due to mitral regurgitation.

2. **What are the initial investigations?**

Initial investigations based on the history of syncope and a murmur of AS include:

-ECG (left ventricular hypertrophy, left bundle branch block)

-Chest x-ray (may be normal or show an enlarged ventricle on PA view)

-Echocardiogram with Doppler (a valuable diagnostic tool)

Eventually, cardiac catheterisation will be needed to measure the gradient across the aortic valve and assess the coronary vessels in preparation for surgery.

3. **What are the main causes of aortic valve disease?**

- Infants, children, adolescents: congenital aortic valve disease, congenital supravalvular disease, congenital subvalvular disease
- Young adults to middle-aged: rheumatic valve disease, calcification and fibrosis of a bicuspid valve
- Middle-aged to elderly: senile degeneration of the aortic valve, calcification of a bicuspid valve, rheumatic valve disease.

4. On clinical examination, differentiate mitral stenosis, aortic stenosis, aortic incompetence and mitral incompetence

Murmur	Symptoms	Peripheral signs	Examination of the precordium	Auscultation of the heart	Signs of severity
Mitral stenosis	Breathless Fatigue Oedema Palpations Haemoptysis Cough Chest pain Symptoms of thrombo-embolic disease	Irregular pulse (AF) Mitral facies (rosy checks with a bluish tinge due to dilation of the malar capillaries) JVP may be normal; Loss of "*a*" wave in AF; Prominent "a" wave if pulmonary hypertension is present.	Taping quality of the apex beat (palpable S1) RV heave and palpable P2 if pulmonary hypertension is present	Loud S1 Opening snap Mid-diastolic rumbling murmur best heard in the left lateral position	Small pulse pressure Soft first heart sound Early opening snap Long diastolic murmur Diastolic thrill Pulmonary hypertension
Aortic stenosis	May be asymptomatic early in disease; Later may develop exertional angina, increasing shortness of breath and syncopal episodes	Low volume, plateau pulse.	Hyperdynamic apex beat that may be displaced laterally Systolic thrill at the base of the heart	Narrow split or reversed S2 Harsh mid-systolic ejection murmur that is maximal over the aortic area and radiates to the carotids; Murmur is attenuated with person sitting up and in full expiration.	Plateau pulse Aortic area thrill Length of the murmur S4 Paradoxical splitting of S2 Left ventricular failure

Murmur	Symptoms	Peripheral signs	Examination of the precordium	Auscultation of the heart	Signs of severity
Aortic regurgitation	Symptoms occur late in the disease and include: Exertional dyspnoea Fatigue Palpitations Exertional angina	Collapsing pulse. Prominent carotid pulsations	The apex beat is displaced and hyperkinetic A diastolic thrill may be palpated with the person sitting upright	A decrescendo high pitched diastolic murmur is audible beginning immediately after the second heart sound and extending for a variable duration into diastole. An Austin-flint murmur should be sought which is low pitched mid-diastolic rumbling at the apex.	Collapsing pulse Wide pulse pressure Long diastolic murmur S3 Soft A2 Austin-Flint murmur Signs of LVF
Mitral regurgitation (Chronic)	Dyspnoea Fatigue	Tachypnoea Atrial fibrillation is common	The apex is displaced, diffuse and hyperkinetic. A pansystolic thrill maybe present at the apex. A parasternal impulse may be palpable.	Soft or absent S1. S3 Pansystolic murmur that is loudest at the apex and radiates to the axilla.	Small flume pulse Loud S3 Soft S1 Signs of pulmonary hypertension and LVF
Mitral regurgitation (acute)	Unwell breathless patients due to acute pulmonary oedema		Systolic thrill	Loud apical systolic murmur	

5. **How might your management be altered if Heidi was indigenous and lived in a remote community with a population of 90 people whose only medical care was delivered by a health worker once a fortnight?**

   The only definitive treatment for aortic stenosis is aortic valve replacement. The development of symptoms due to aortic stenosis provides a clear indication for replacement. For patients who are not candidates for aortic replacement, percutaneous aortic balloon valvuloplasty may provide some symptom relief.

   The medical treatment options are limited in symptomatic patients with aortic stenosis who are not candidates for surgery. In patients with pulmonary congestion, cautious use of digitalis, diuretics, and angiotensin-converting enzyme (ACE) inhibitors might attempted, whereas beta-blockers might be used if the predominant symptom is angina. In any case, excessive decrease in preload or systemic arterial blood pressure should be avoided.
   Antibiotic prophylaxis for the prevention of bacterial endocarditis is no longer recommended in patients with isolated valvular aortic stenosis.

   The management of AS depends upon the severity of the stenosis. The development of symptoms such as angina, syncope or increasing breathlessness are indications for objective evaluation of the severity of the stenosis and prompt consideration of aortic valve replacement.

6. **What elements in assessment suggest that valvular surgery or cardiac transplantation is indicated?**

   All patients with AS should be kept under observation. However the development of any further problems such as angina, syncope, symptoms of low cardiac output or cardiac failure indicate that surgery is promptly indicated.

   Unfortunately, the woman in this case has clinical signs of heart failure and a history consistent with syncope. An aortic valve replacement needs to be considered.