Short Cases in Postgraduate Clinical Exams of INTERNAL MEDICINE

Wanis Hamad Ibrahim

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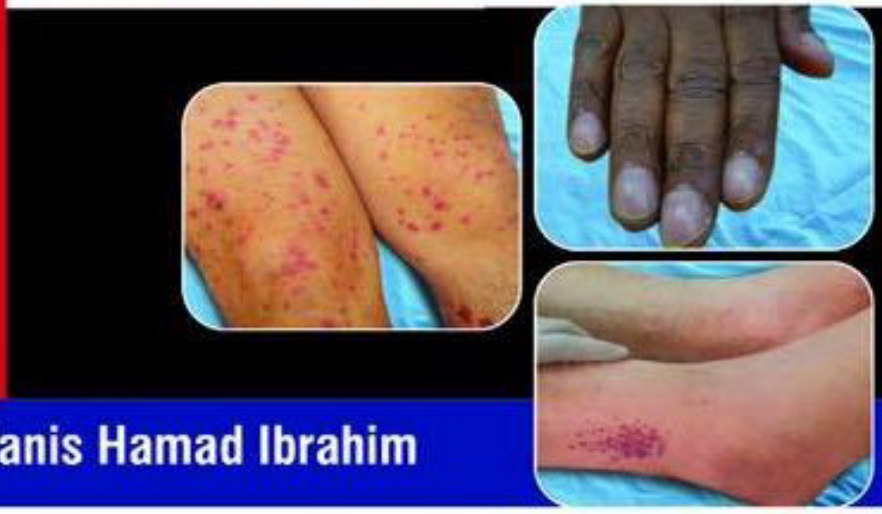
for PACES, ARAB BOARD, FRACP, FCPS, MD and Other National Board Exams



Wanis Hamad Ibrahim

Foreword
RA Malik

JAYPEE

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How to Present your Findings to the Examiners?

In addition to the proper technique of physical examination and identification of the correct findings, the way that candidates present their findings to the examiners will affect the overall examiner's impression about the performance of the candidates and the final short case mark. Many candidates of high standard can fail the examinations simply because they cannot convey the correct findings and diagnosis to the examiners. Presenting your ideas to listeners is an art in itself; and, therefore, candidates should practice repeatedly presenting their clinical findings to their peers or senior colleagues. The common two scenarios after a candidate completes physical examination of a patient are that either he/she is confident about the diagnosis or he/she identified the findings but is not confident about the exact diagnosis (in the latter scenario, the candidates has 2 or 3 possible differential diagnoses). If the former scenario is applicable and the candidate is confident about the diagnosis (for example, the candidate found a pansystolic murmur of maximal intensity at the mitral area radiating to the axilla suggestive of mitral regurgitation), then the candidate should tell the diagnosis first and then refer to the findings. For example, the examiner asks what your diagnosis is. The typical answer should be: 'Well, this pleasant gentleman has features to suggest mitral regurgitation as evidenced by muffled first heart sound, a pansystolic murmur of grade 3 of 5 heard best at the mitral area radiating to the axilla'. There are no signs of heart failure or infective endocarditis and I would like to request echocardiography to confirm my findings and assess the severity of the valve lesion. A candidate who is confident about combined aortic valve disease (stenosis and regurgitation) can provide the following answer: 'Well, this pleasant lady has features to suggest combined aortic stenosis and regurgitation as evidenced by an ejection systolic murmur heard best in the aortic area grade 3 of 5 radiating to the neck as well as an early diastolic murmur at the aortic area. The patient seems to be in heart failure as I could hear bilateral crackles over the lung bases. The predominant valvular lesion seems to be aortic regurgitation as the pulse is collapsing and, I could find peripheral signs of aortic regurgitation.

Alternatively, the candidate may have established some findings but he/she is not confident regarding the diagnosis. In other words, he/she is confused as the findings could fit more than one diagnosis. In that scenario, I suggest that the candidate should present his/her findings first, and then suggest a diagnosis and justify or defend his/her thinking. Example of this, a candidate found a harsh systolic murmur over the base (aortic area), but could also hear a loud murmur over the mitral area, and is not confident whether it is aortic stenosis or mitral regurgitation. The typical candidate answer will be: 'Well, I examined this pleasant lady who has a holosystolic murmur that is best heard over the aortic area; however, I could also hear the same murmur with the same intensity over the tricuspid and mitral area. Although the murmur is heard loudly over the mitral area it does not radiate to the

axilla and I could hear radiation of the murmur in the neck. This makes aortic stenosis the most likely diagnosis in my mind; however, coexistent mitral regurgitation needs to be ruled out by echocardiography. The patient is not in heart failure and there are no signs of infective endocarditis'. In the second scenario, the examiners usually ask questions that can lead the candidate to the correct diagnosis. Now think what will be the candidate mark, if he/she stated that the diagnosis was aortic stenosis and stopped, and it turned to be mitral regurgitation or vice versa. The second important point candidates need to consider when presenting their findings is to show extreme respect to the patient. A male patient should always be referred to as *pleasant gentleman* and a female patient as *pleasant lady*. Although each candidate is given a mark before the next candidate is examined, the examination is a competition between candidates and examiners, will usually compare your performance to other candidates. A candidate who starts his answer by: 'Well, I examined this pleasant gentleman/lady….' is definitely considered more courteous to the one who starts by: 'This patient or this old woman, etc.' Candidates in clinical examinations are usually under tremendous anxiety and stress, and a simple question by the examiner might be interpreted by the anxious candidate as a trick or trap. Always think simple and in case, you have a doubt as to what the examiner means by the question, do not just give any answer, simply request the examiner politely to repeat or rephrase the question.

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Cardiovascular Cases

HOW TO EXAMINE A PATIENT WITH HEART DISEASE IN THE CLINICAL EXAMINATION?

- Wash your hands.
- Shake hands with the patient, introduce yourself and take permission
- Position the patient at a 45° angle and request the patient to remove their upper clothes.
- Inform the patient to alert you in case you cause any discomfort or pain to him/her during the examination.
- Start by inspecting the patient and the surrounding. Allow some time for a quick surveillance of the patient and their surroundings. This may give you important clues about the patient condition and diagnosis. A tall marfanoid habitus may suggest the diagnosis of aortic regurgitation due to Marfan's syndrome. Inability of the patient to move his or her neck (particularly while examining the Jugular venous pressure (JVP) should raise the suspicion of ankylosing spondylitis with associated aortic regurgitation. External appearance of Down's syndrome may suggest an atrioventricular septal defect. Similarly, a woman who looks short with a webbed neck may indicate the reason for cardiovascular system examination is coarctation of the aorta as a complication of Turner' syndrome. A patient on an intravenous heparin infusion may suggest atrial fibrillation or presence of a metallic valve. A patient who is using oxygen may suggest the diagnosis of heart failure. A patient in a semi-sitting position from the start who appears in respiratory distress may suggest significant heart failure, while one lying comfortably in a flat position at the start of the examination suggests that significant pulmonary edema is unlikely. Look for cyanosis, malar flush due to mitral stenosis, obvious pedal edema, etc.

- Candidates may hear different instructions from different examiners such as examine the cardiovascular system, examine the heart or examine the precordium. Many candidates get confused whether to examine only the precordium or look for peripheral signs of cardiac diseases. I suggest that even if the instruction is to examine the heart, candidates should start by looking for peripheral signs of cardiovascular disease unless they are redirected by the examiners to examine only on the precordium. A common cause of failure is when candidates miss important peripheral signs such as clubbing or signs of infective endocarditis in a patient with valvular heart disease.
- Hold the right hand of the patient and feel the pulse. Pay attention to the rhythm as an irregular pulse can be easily missed by the anxious candidate. Record the rate, rhythm, volume, any special character, the presence of a synchronous pulse on the other arm and feel for radiofemoral delay. Make sure that the patient does not have shoulder pain before you check for a collapsing pulse.
- Examine the hands for clubbing, cyanosis, pallor, splinter hemorrhage, Janeway lesions and Osler's nodes **(Figure 1.1)**.
- Move to the face and mouth. Examine the eyes for pallor, cheeks for malar flush of mitral stenosis, cyanosis and dental caries **(Figure 1.2)**.
- Examine the neck for JVP, dancing carotid pulsations "Corrigan pulse" of aortic regurgitation and thyroid gland (particularly in a patient with atrial fibrillation).
- Follow the steps of inspection, palpation and auscultation to examine the precordium.
- Inspect for visible pulsations, shape of the chest, pectus excavatum **(Figure 1.3)** or carinatum **(Figure 1.4)**.
- Before you start palpating the chest ask the patient if he/she has any pain.

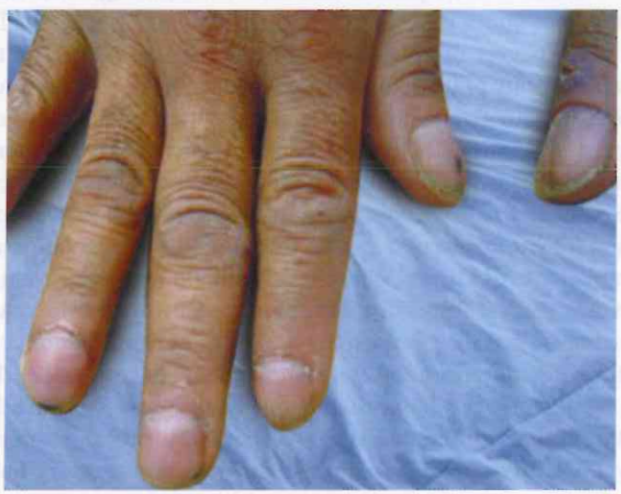


FIGURE 1.1 Finger clubbing and splinter hemorrhage in a patient with infective

FIGURE 1.2 Facial appearance in mitral stenosis

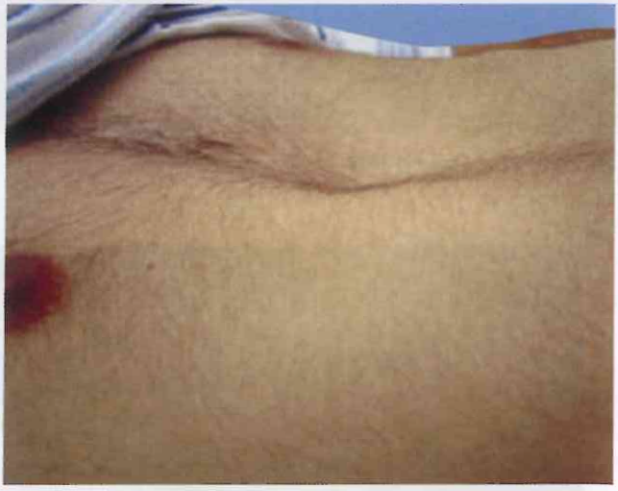


FIGURE 1.3 Pectus excavatum

- Feel for the apex beat; determine its location and character. Feel for a parasternal heave, thrill and palpable second heart sound.
- Start listening at the apex and simultaneously place your free hand over the carotid to enable you to time the heart sounds, this is essential. First concentrate on the first and second heart sounds. Determine whether they are normal in intensity, muffled or loud. Check whether the splitting of second heart sound is normal wide, or fixed. Once you hear a murmur, determine the place in which you hear the murmur at its maximal intensity. This is usually the site of origin of the murmur. For example, if you hear a pansystolic murmur loudest at the mitral area, that murmur is most likely to be due to mitral valve disease. Determine also the character, radiation and effect of respiration on the murmur. Once you finish listening to the four areas, ask the patient to tilt to the left lateral position to listen for the mid-diastolic murmur of mitral stenosis then ask the patient to sit forward, breath out and hold breath to examine for the murmur of aortic regurgitation. Listen carefully over the left axilla for radiation of the mitral regurgitation murmur

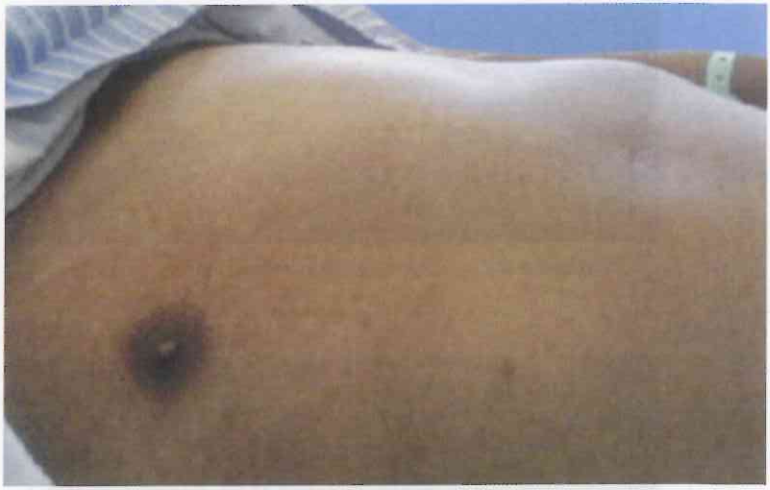


FIGURE 1.4 Pectus carinatum

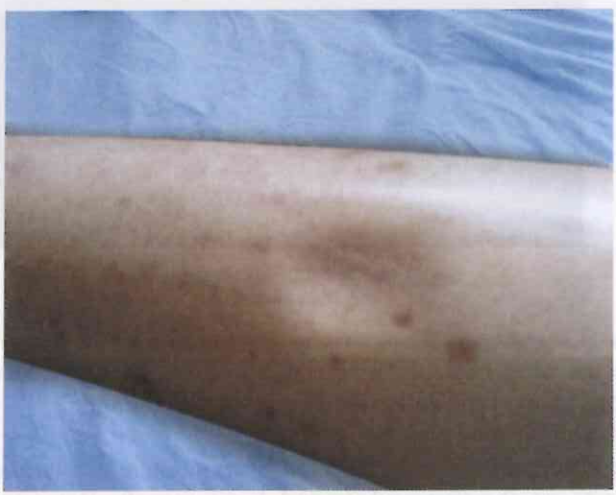


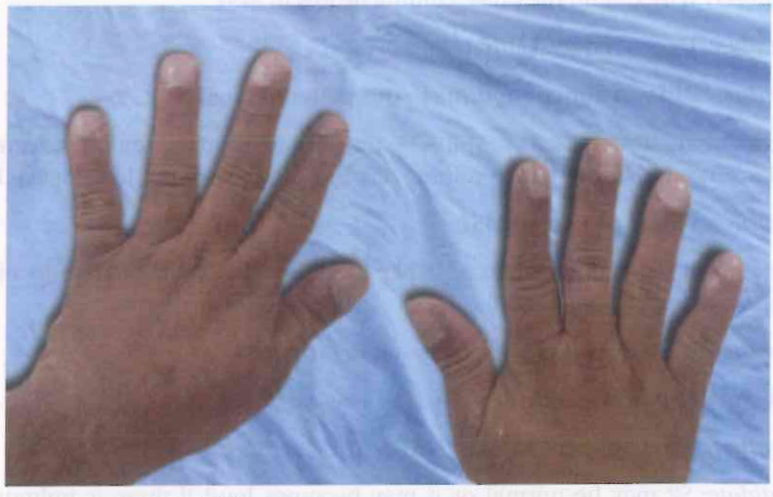
FIGURE 1.5 Pitting edema of the legs (observe also the presence of diabetic dermopathy)

and over the carotids in the neck for the radiation of the murmur of aortic stenosis.

- Finish your examination by listening to the lung bases and feeling for pitting pedal edema (**Figure 1.5**). Remember during the examination for pitting edema to enquire from the patient about leg pain before pressing over the legs and to direct your face towards the patient for any tenderness rather than towards the examiner.

IMPORTANT CLUES REGARDING CARDIOVASCULAR CASES IN THE CLINICAL EXAMINATION

- Many candidates feel the time given for cardiovascular cases in the clinical examination is not enough. Frequent timed practice of the cardiovascular system examination under supervision of a registrar or a consultant will be very helpful.
- A systolic murmur that radiates to the neck is due to aortic stenosis.
- A systolic murmur that radiates to the axilla is due to mitral regurgitation.
- If you find a displaced apex beat in a patient with mitral stenosis, search carefully for other coexistent valvular lesions.
- The early diastolic murmur of aortic regurgitation may mimic breath sounds. Make sure that you ask the patient to hold breath when you listen for this murmur.
- When you find pure aortic regurgitation in the examination make sure you identify ankylosing spondylitis or Marfan's syndrome, if present.
- Do not forget to examine for peripheral signs of aortic regurgitation when you find an early diastolic murmur in the aortic area.
- Aortic stenosis murmur may be harsh and heard also in the mitral area and might be confused with a systolic murmur of mitral regurgitation (Gallavardin phenomenon). The clue is in the radiation to the neck and an absence of radiation to the axilla.
- In Eisenmenger syndrome, the murmur of the ventricular septal defect (VSD) may disappear. If you are asked to examine the cardiovascular system of a patient with cyanosis, clubbing and elevated JVP, think of Eisenmenger syndrome even if you cannot hear a murmur (other differential diagnosis include chronic lung diseases) **(Figure 1.6)**.



- If you do not feel the apex beat and you do not hear heart sounds keep in mind dextrocardia. Some candidates developed a good habit of feeling both sides of the chest for the apex beat as they begin their examination.
- Remember that the indications for infective endocarditis prophylaxis in valvular heart disease have been modified recently.

MITRAL STENOSIS

Common Pitfalls

- Candidates fail to recognize that the patient has stroke due to systemic embolization.
- Candidates fail to recognize signs of infective endocarditis.
- Candidates miss the presence of atrial fibrillation (AF).
- Candidates find a displaced apex beat and do not think of other coexistent valvular lesion aortic regurgitation (AR)/mitral regurgitation (MR).

Examiner Instructions

- Examine this patient's cardiovascular system
- Examine this patient's precordium
- Listen to this patient's heart.

Candidate

Well, this gentleman has features to suggest mitral stenosis as evidenced by a low pulse volume, tapping apex, loud S1, opening snap and mid-diastolic rumbling murmur. There were no signs to suggest infective endocarditis or pulmonary hypertension.

Examiner: **Why should the pulse volume be low in MS?**

Candidate: Due to reduced stroke volume.

Examiner: **Did you notice anything in this patient's face?**

Candidate: He has malar flush (pinkish-purple cheeks) resulting from decrease cardiac output (reduced stroke volume) leading to vasoconstriction **(Figure 1.2)**.

Examiner: **Why is S1 loud in MS?**

Candidate: It is due to the increase in left atrial pressure leading to closure of the mitral valve from a wide distance.

Examiner: **What does it mean if S1 is soft in pure MS?**

Candidate: It indicates that the valve is heavily calcified.

Examiner: **What happens to S2 in MS?**

Candidate: S2 may be normal or it may becomes loud if there is pulmonary

Examiner: **What is the mechanism of the opening snap in MS and what are its clinical implications?**

Candidate: It is heard after S2 at the apex and is best heard at the left sternal border. It is caused by sudden and rapid opening of the mitral valve in early diastole due to the high pressure in the left atrium. The opening snap helps in determining the severity of mitral stenosis; when the valve becomes severely stenotic or heavily calcified, the interval between S2 and opening snap becomes shorter and the opening snap may disappear.

Examiner: **Which sound is usually confused for the opening snap and how would you differentiate them?**

Candidate: Splitting of S2 (For example, due to pulmonary hypertension). Variation with respiration may help in differentiation.

Examiner: **Is opening snap pathognomonic for MS?**

Candidate: No. It can be heard in tricuspid stenosis and left atrial myxoma. (*Remember:* In the examination opening snap is always due to mitral stenosis).

Examiner: **In which condition will you not be able to hear presystolic accentuation of the MS murmur?**

Candidate: If the patient has atrial fibrillation.

Examiner: **What are the factors that indicate the severity of MS?**

Candidate:

- A short S2-OS interval
- Signs of pulmonary arterial hypertension
- Long mid-diastolic murmur
- Mitral valve area less than 1.5 cm^2.

Examiner: **What are the causes of MS?**

Candidate:

- Rheumatic heart disease
- Congenital MS/Lutembacher's syndrome atrial septal defect (ASD) with mitral stenosis (MS)
- Mitral annular calcification [For example, in patients with end-stage renal disease (ESRD)]
- Systemic lupus erythematosus (SLE).

Examiner: **What are the complications of MS?**

Candidate:

- Systemic embolization
- PA hypertension
- Infective endocarditis
- Hemoptysis.

Examiner: **What if this patient comes with hoarseness of his voice?**

Candidate: Compression of the left recurrent laryngeal nerve against the pulmonary artery by an enlarged left atrium may rarely cause hoarseness.

Examiner: **How would you manage this patient?**

Candidate:

- Perform transthoracic echocardiogram (TTE) to confirm the diagnosis, quantify hemodynamic severity, PA hypertension and assess concomitant valvular lesions.
- Surgical intervention is indicated in
  - Very severe MS with a mitral valve area < 1.0 cm^2
  - Severe MS with a mitral valve area <1.5 cm^2 with:
    - *Severe symptoms:* NYHA III or IV
    - Presence of pulmonary hypertension (PA systolic pressure ≥ 25 mm Hg)
    - Possibly in new onset AF or multiple systemic embolization despite adequate anticoagulation
- Anticoagulation for prevention of thromboembolism (high risk groups include prior embolic event, AF, small mitral valve area, presence of AR and left atrial thrombus).
- Management of AF.
- Prevention of rheumatic fever recurrence.

Examiner: **Would you recommend routine antibiotic prophylaxis for infective endocarditis (IE) in this patient before procedures?**

Candidate: The prophylaxis against IE has recently been updated and not all valvular lesions or procedures require prophylaxis. (See infective endocarditis section).

MITRAL REGURGITATION

Common Pitfalls

- Candidates hear radiation of a systolic murmur to the axilla and fail to diagnose MR.
- Candidates confuse harsh AS with MR.

Examiner Instructions

- Examine this patient's cardiovascular system.
- Examine this patient's precordium.
- Listen to this patient's heart.

Candidate

This patient has brisk pulse, displaced apex beat, systolic thrill in the mitral area, soft S1 and a loud pansystolic murmur heard best in the mitral area which

Examiner: **How would you differentiate a high volume pulse in AR from that of MR?**

Candidate: In MR the pulse pressure will be normal while in AR it will be wide.

Examiner: **What are the causes of MR?**

Candidate:

- Mitral valve prolapse (most common cause in developed countries)
- Rheumatic heart disease
- Infective endocarditis
- Acute MR in ischemic heart disease (rupture papillary muscle)
- Left ventricular failure (dilatation of valve)
- Hypertrophic cardiomyopathy.

Examiner: **What are the main complications of MR?**

Candidate:

- Development of left ventricular dysfunction
- Atrial fibrillation
- Infective endocarditis.

Examiner: **Which factors indicate severity of MR?**

Candidate:

- Acute MR (from acute coronary syndrome)
- Development of symptoms and signs of left ventricular dysfunction
- Regurgitant fraction >50%
- Regurgitant volume > 60 mL
- Left ventricular ejection fraction (LVEF) <60%
- Left ventricular end systolic dimension(LVESD) >40 mm (LV dilatation).

Examiner: **In the presence of coexisting MS, how would you determine the predominant valvular lesion?**

Candidate: Presence of displaced apex beat; high volume pulse and muffled first heart sound suggest that MR is the predominant lesion.

Examiner: **What are the indications for surgical intervention in MR?**

Candidate:

- Patients with acute MR who are symptomatic
- Symptomatic severe MR (regurgitant fraction >50% or volume > 60 mL) with LVEF> 30% [ejection fraction (EF) should be reasonable to go for surgery]
- Asymptomatic severe MR with one of the following:
  - LVEF between 30% and 60%
  - LVESD < 40 mm
  - Development of AF
  - Development of pulmonary hypertension

Examiner: **How would you manage this patient?**

Candidate:

- Serial echocardiography to follow LVEF
- Afterload reduction by diuretics and nitrates
- Infective endocarditis prophylaxis is not routinely recommended in mitral and aortic rheumatic heart disease (RHD) except in high-risk groups (see MS)
- Treat AF.

AORTIC REGURGITATION

Common Pitfalls

- Failure to recognize clinical features of Marfan's syndrome or ankylosing spondylitis in a patient with AR
- Failure to diagnose AR in a patient with dancing carotid pulsation
- Failure to examine for peripheral signs of AR
- Missing AR murmur as breath sound particularly when there is another valvular lesion.

Examiner Instructions

- Examine this patient's cardiovascular system
- Examine this patient's precordium
- Listen to this patient's heart

Candidate: This patient has a collapsing pulse, there is a dancing carotid (Corrigan's sign) and a blowing early diastolic murmur heard best at the left second intercostal space, which suggests AR.

Examiner: **What are the causes of AR?**

Candidate:

- *Valve disease:* Rheumatic heart disease—Infective endocarditis—Congenital bicuspid valve
- *Aortic root disease:* Marfan's syndrome—Long standing hypertension—Syphilitic aortitis—Ankylosing spondylitis—Ehlers-Danlos syndrome—Aortic dissection

Examiner: **How could you identify the cause of AR from the site of the murmur?**

Candidate: AR due to valvular disease is usually heard at the third and fourth intercostal space of the left sternal border, while that due to aortic root disease is heard best at the right sternal border.

Examiner: **If you hear an ejection systolic murmur at the aortic area, what could it be?**

Candidate: It could be due to coexisting AS or a functional stenosis resulting from the large volume of blood passing through the aortic valve because of AR.

Examiner: **If you hear an associated mid-diastolic murmur at the apex, what could it be?**

Candidate: It can be an associated MS or an Austin-Flint murmur.

Examiner: **How would you differentiate MS from an Austin-Flint murmur?**

Candidate: An Austin-Flint murmur occurs because of the turbulence of blood at the mitral valve due to regurgitation of blood from the aorta into the left ventricle. In Austin-Flint, S1 will be normal and there is no opening snap.

Examiner: **What are the peripheral signs of AR?**

Candidate:

- *Becker sign:* Visible systolic pulsations of the retinal arterioles
- Corrigan pulse (dancing carotid pulsation)
- *de Musset sign:* Bobbing of the patient's head with each heartbeat
- *Hill sign:* Popliteal cuff systolic blood pressure 40 mm Hg higher than brachial cuff systolic blood pressure
- *Duroziez sign:* Systolic and diastolic murmur over the femoral artery with mild compression of the artery
- *Müller sign:* Visible systolic pulsations of the uvula
- *Quincke sign:* Visible capillary pulsations of the fingernail bed
- *Pistol-shot sign:* Systolic and diastolic sounds heard over the femoral artery.

Examiner: **Why do these signs occur in AR?**

Candidate: All these signs result from widened pulse pressure (exaggerated difference between systolic and diastolic blood pressure) because of elevated stroke volume during systole that falls significantly during diastole due to the incompetent aortic valve.

Examiner: **What are the main complications of chronic AR and what is the single-most important prognostic factor?**

Candidate: Progressive LV dysfunction and failure, angina, arrhythmia, and sudden death. Patients with AR and New York Heart Association (NYHA) class III or IV heart failure have an annual mortality of about 25%. The single-most important prognostic factor is LV function.

Examiner: **What are the factors indicating severity of AR?**

Candidate:

- *Clinical factors:* Displaced apex beat, longer AR murmur, presence of peripheral signs that suggest wider pulse pressure (see above)
- *Echocardiographic factors:* LVEF < 50% and LVESD > 50 mm (*Remember the number 50. They all represent dilatation or poor function of LV*).

Examiner: **How would you manage this patient?**

Candidate:

- Serial echocardiography to assess LVEF and LVESD

- Cardiac catheterization to assess the presence of coronary artery disease (CAD) and severity of AR
- Vasodilator drugs like angiotensin-converting enzyme (ACE) inhibitors and calcium channel blockers to decrease afterload
- Infective endocarditis prophylaxis is not routinely recommended in mitral and aortic RHD except in the high risk groups (see MS).

Examiner: **What are the indications for surgery in AR?**

Candidate:

- Acute AR from IE or aortic dissection
- Symptomatic patient
- Asymptomatic patient, with a resting EF <50%
- Asymptomatic patient, with LV dilation LVESD >50 mm
- Asymptomatic patient with AR who is undergoing coronary bypass or other cardiac surgery.

AORTIC STENOSIS

Common Pitfalls

- Failure to differentiate aortic sclerosis from aortic stenosis
- Misdiagnosing aortic stenosis (AS) MR in a patient whose murmur radiates to the neck (misinterpretation of the Gallavardin phenomenon)
- Failure to look for AR as it is commonly associated with AS (in 80% of cases)

Examiner Instructions

- Examine this patient's cardiovascular system
- Examine this patient's precordium
- Listen to this patient's heart

Candidate

This patient has a displaced heaving apex beat, with a slow rising pulse (pulsus parvus tardus), S4 and a harsh ejection systolic murmur which is heard best in the right second intercostal space and radiates to the neck. I could hear the same murmur which was a bit softer at the apex in the mitral area but there was no radiation to the axilla, I would suggest pure AS (without coexistent MR) - Gallavardin phenomenon. There are no signs of heart failure.

Examiner: **What do you mean by Gallavardin phenomenon?**

Candidate: An aortic stenosis murmur sometimes has two components, a noisy harsh component which is heard in the right second or third intercostal space and radiates to the neck and a musical component that radiates to the apex (mitral area). This musical component may be mistaken for MR.

Examiner: **Then how do you differentiate between the Gallavardin phenomenon and the presence of true MR?**

Candidate: The murmur of MR usually radiates to the axilla, whereas that due to Gallavardin does not.

Examiner: **Which cause of AS usually produces the Gallavardin phenomenon?**

Candidate: Degenerative aortic stenosis.

Examiner: **What are the common causes of AS?**

Candidate:

- *Congenital aortic stenosis:* Unicuspid, bicuspid and tricuspid valve.
  In adults, bicuspid aortic valve is the most common cause of AS
  In children, a unicuspid valve predominates
- *Acquired aortic stenosis:*
- *Degenerative calcific AS:* The most common cause in older patients (risk factors include: DM, hypertension, hypercholesterolemia and smoking)
- Rheumatic heart disease.

Examiner: **How would you differentiate between aortic stenosis and aortic sclerosis?**

Candidate: In aortic sclerosis, the murmur usually does not radiate to the neck and the volume of the pulse is usually high rather than slow rising. Also, in aortic stenosis there is usually a reduced pulse pressure (difference between systolic and diastolic < 40 mm Hg).

Examiner: **What do you mean by pulsus parvus tardus and how would you check for its presence?**

Candidate: Parvus means low volume and tardus means rising slowly. You need to place one hand over the apex beat and the other over the carotid pulse. In addition to being low in volume, there will be delay in the upstroke of the carotid pulse compared to the apex beat. Tardus is more specific than parvus in AS.

Examiner: **What are the classic triad of symptoms in AS?**

Candidate: Angina, dyspnea and syncope or dizziness

Examiner: **What is the incidence of sudden cardiac death in AS?**

Candidate: About 30% in symptomatic AS.

Examiner: **If this patient presents with bleeding per rectum, what do you suspect as a cause?**

Candidate: Colonic angiodysplasia. Aortic valve replacement may cure angiodysplasia and recurrent bleeding.

Examiner: **What factors indicate the severity of AS?**

Candidate:

Clinical:

- Presence of symptoms
- Prolonged louder murmur
- Paradoxical splitting of S2 (due to delayed closure of A2)
- Presence of S4
- Pulsus parvus tardus

Echocardiographic:

- Valve area < 1 cm^2
- Pressure gradient across the valve > 40 mm Hg

Examiner: **How would you manage this patient?**

Candidate:

- Request echocardiography to assess valve area and pressure gradient
- Caution when prescribing blood pressure lowering medications to avoid excessive lowering of preload (diuretics, beta blockers, vasodilators such as nitrates, hydralazine and nifedipine should be used with caution and in smaller doses)
- Instruct patient to avoid standing suddenly from a sitting or recumbent position
- No routine endocarditis prophylaxis is indicated in valvular AS except in the high-risk groups mentioned in the infective endocarditis section
- Cardiac catheterization to look for coexistent CAD.

Examiner: **What are the indications for surgery in AS?**

Candidate:

- Surgery is the mainstay treatment of symptomatic AS
- Asymptomatic severe AS with EF <50% or abnormal exercise treadmill test
- Asymptomatic severe AS in a patient undergoing other cardiac surgery.

PATIENT WITH A PROSTHETIC HEART VALVE

Common Pitfalls

- Failure to see scar particularly sub-mammary scars in females.
- Candidate can not differentiate mitral from aortic valve prosthesis.
- Failure of the candidate to recognize signs of IE.

Examiner Instructions

- Examine this patient's cardiovascular system
- Examine this patient's precordium
- Listen to this patient's heart

Candidate

This patient has a sternotomy scar and an audible metallic click heard loudest in the mitral area coinciding with S1. There are no signs of heart failure or IE. He has a metallic mitral valve, which seems to be functioning well.

Examiner: **What are the different types of prosthetic heart valves?**

Candidate:

- Metallic:
  - *Caged ball valve:* For example, Starr-Edwards valve
  - *Tilting disc valve:* For example, Medtronic Hall valve
  - *Bileaflet valves:* For example, St. Jude valve
  - On-X mechanical valve (lower thrombosis rate)
- Biological valves (Bioprosthetic).

Examiner: **What are the advantages of the On-X mechanical valves over other mechanical valves?**

Candidate: On-X mechanical valves are made purely of carbon which makes them having smooth surface and lower thrombosis rate. The INR target in these types of valves is therefore lower than other mechanical valves (1.5–2.0). This in turn lowers the bleeding from anticoagulation.

Examiner: **What are the clinical features indicating a malfunctioning metallic valve?**

Candidate:

- Signs of heart failure
- Absence of normal valve closure sound
- Development of abnormal regurgitant murmur (normal metallic valve may be associated with systolic murmurs)
- Signs of infective endocarditis.

Examiner: **What are the complications of metallic valves?**

Candidate:

- Metallic valve malfunction leading to heart failure or sudden death
- Infective endocarditis
- Systemic embolization
- Microangiopathic hemolysis
- Anticoagulation-related bleeding.

Examiner: **If this patient presents with anemia, name 2 possible causes?**

Candidate: Warfarin-related bleeding or microangiopathic hemolysis due to destruction of red blood cells (RBCS) on the metallic valve.

Examiner: **What are the advantages and disadvantages of the metallic and bioprosthetic valves?**

Candidate:

- Metallic valves have a lower rate of valve dysfunction such as paravalvular leak and therefore are more durable
- Bioprosthetic valves do not require anticoagulation
- Survival is equal

Examiner: **How long do artificial valves usually last?**

Candidate:

- *Metallic valve:* Up to 30 years
- *Bioprosthetic valve:* Up to 15 years.

Examiner: **In which group of patients is a bioprosthetic valve recommended?**

Candidate: A bioprosthetic valve although less durable does not require anticoagulation. Therefore, it is recommended for patients aged 65-year-or above, patients at risk of bleeding from warfarin and patients who may be poorly compliant with warfarin therapy.

Examiner: **How would you manage this patient?**

- Patient counseling and education
- Complete blood count (CBC), bilirubin and urine microscopy
- Echocardiography to assess valve function
- Anticoagulation (warfarin is recommended)
- *IE prophylaxis:* Patients with prosthetic heart valves are high risk group and should receive IE prophylaxis (see infective endocarditis section)
- Counseling regarding pregnancy (risk of valve dysfunction, heart failure, thromboembolism and warfarin use).

Examiner: **What is the target INR level in mechanical valves?**

Candidate: Aortic valve INR is 2–3; mitral valve INR is 2.5–3.5.

Examiner: **When would you consider the addition of Aspirin to Warfarin?**

Candidate: Those patients with high risk factors such as atrial fibrillation, venous thromboembolism, left ventricular dysfunction, and a hypercoagulable state.

Examiner: **How would you manage anticoagulation for major surgical procedures?**

Candidate: Warfarin is stopped 5 days prior to surgery to achieve INR < 1.5 at the time of operation and bridging anticoagulation using unfractionated heparin or low molecular weight heparin is given until the day of surgery. Warfarin is started 24 hrs after surgery following confirmation of hemostasis.

VENTRICULAR SEPTAL DEFECT

Common Pitfalls

- Failure to recognize signs of Down's syndrome

Examiner Instructions

- Examine this patient's cardiovascular system
- Examine this patient's precordium
- Listen to this patient's heart and tell me the diagnosis.

Candidate

This patient has a loud harsh holosystolic murmur associated with a systolic thrill heard best in the left 4th intercostal space. There are no signs of pulmonary hypertension and no cyanosis. The patient has VSD.

***Examiner:* What are the types of VSD?**

Candidate:

- *Perimembranous:* Most common type
- *Supracristal:* May be associated with AR
- Muscular
- Posterior.

***Examiner:* How does the size of a VSD correlate with the murmur?**

Candidate: The smaller the size of VSD, the louder the murmur.

***Examiner:* What are the complications of VSD?**

Candidate:

- Pulmonary hypertension
- Polycythemia
- Eisenmenger complex.

***Examiner:* How would you manage this patient?**

- Serial echocardiography
- Cardiac catheterization to quantify the net shunt
- IE prophylaxis is not indicated if no cyanosis and no previous IE
- Diuretic therapy and ACEI
- Surgery.

***Examiner:* What are the indications of surgical repair in VSD?**

Candidate:

- The ratio of total pulmonary blood flow to total systemic blood flow QP/QS > 2
- Prior history of infective endocarditis
- Left ventricular volume overload
- QP/QS >1.5 with PAP < two-third of systemic pressure
- Presence of VSD and AR

***Examiner:* What does it mean if the murmur disappears in a patient with VSD?**

Candidate: It means either the VSD closed (very rare after the age of 4 years) or the patient has developed Eisenmenger complex.

EISENMENGER COMPLEX

Examiner: **What are the clinical manifestations of Eisenmenger complex?**

- The holosystolic murmur disappears
- Signs of pulmonary hypertensions and right heart failure
- Cyanosis
- Finger clubbing **(Figure 1.6)**
- Polycythemia

Examiner: What is the treatment of choice of Eisenmenger complex develops?

Candidate: Heart-Lung transplantation

ATRIAL SEPTAL DEFECT

Common Pitfalls

- Candidates miss the wide fixed splitting of S2
- Candidates think that the cause of the ejection systolic murmur in ASD is the flow across the ASD shunt.

Examiner Instructions

- This patient complains of exertional dyspnea, please listen to his heart
- Examine this patient's heart.

Examiner: **What are the types of ASD?**

Candidate:

- *Ostium secondum:* The most common type of ASD
- *Ostium primum:* The second most common type of ASD. Usually associated with mitral valve abnormalities.
- *Sinus venosus:* The least common.

Examiner: **What are the auscultatory findings in ASD?**

Candidate: An ejection systolic murmur in the pulmonary area and fixed wide splitting of S2.

Examiner: **What causes the ejection systolic murmur in ASD?**

Candidate: The systolic murmur heard in ASD is due to the increased flow across the pulmonary valve (functional stenosis) and not as a result of blood flow across the ASD shunt itself.

Examiner: **Which murmur mimics that heard in ASD and how would you differentiate the two?**

Candidate: Pulmonary stenosis also gives an ESM at the pulmonary area. However, fixed wide splitting of S2 occurs in ASD but not in PS.

Examiner: **If in addition to the ASD murmur, you hear a mid-diastolic murmur in this patient, what is the explanation?**

Candidate: If a mid-diastolic murmur is heard in the tricuspid area, it is due to increased blood flow across the tricuspid valve because of a large ASD. If a mid-diastolic murmur is heard in the mitral area (mitral stenosis murmur) this is called "Lutembacher syndrome" which is a combination of ASD and mitral stenosis.

Examiner: **What are the complications of ASD?**

Candidate:

- Pulmonary hypertension
- Eisenmenger syndrome

Examiner: **How would you manage this patient?**

Candidate:

- Echocardiography to assess the size of the shunt and the presence of pulmonary hypertension
- Spontaneous closure in adults is unlikely (commonly happen in childhood)
- Surgical closure is indicated in patients with significant shunts and patients who develop pulmonary hypertension and right ventricular overload.

DEXTROCARDIA

Common Pitfalls

- Failure to auscultate over the right chest when candidates cannot hear heart sounds on the left
- Failure to recognize associated features of Kartagener's syndrome such as clubbing due to bronchiectasis
- Examiner instructions
- Examine this patient's cardiovascular system
- Examine this patient's precordium
- Listen to this patient's heart.

Candidate

This patient has dextrocardia evidenced by an absent apex beat and heart sounds over the left side of the chest, which can be heard clearly over the right side. He also has finger clubbing, so I would like to examine the chest and ask him a few questions to confirm the diagnosis of Kartagener's syndrome.

Examiner: **What conditions are associated with dextrocardia in adults?**

Candidate:

- *Kartagener's syndrome:* Characterized by the triad of situs inversus, paranasal sinusitis, and bronchiectasis
- Congenitally corrected transposition of the great arteries (TGA)

Examiner: **How would you manage this patient?**

Candidate:

- Take a history of cardiac symptoms, chronic cough or cardiac surgery in early life.
- Chest X-ray
- Echocardiography.

ATRIAL FIBRILLATION

Common Pitfalls

- Missing the presence of AF
- Missing the features of hyperthyroidism as the cause of AF
- Confusion regarding differentiating AF from multiple ventricular ectopic beats.

Examiner Instructions

- Perform a general examination
- Examine the heart
- Examine this patient's pulse.

Examiner: **What are the causes of AF?**

Candidate:

- **M:** Mitral stenosis
- **A :** Alcohol
- **T :** Thyrotoxicosis
- **C :** CAD
- **H :** Hypertension

Examiner: **What simple bedside test differentiates AF from ventricular premature beats?**

Candidate: Ask the patient to perform some exercise. Ventricular premature beats will reduce in frequency with exercise but AF will not.

Examiner: **What do you mean by paroxysmal AF, persistent AF, permanent AF and lone AF?**

Candidate:

- *Paroxysmal AF:* Self-terminating
- *Persistent AF:* AF that fails to terminate within 7 days
- *Permanent AF:* AF that lasts for more than one year and failed cardioversion or not attempted
- *Lone AF:* AF occurring in the absence of structural heart disease

Examiner: **How would you manage this patient?**

Candidate:

- Treat the underlying cause, e.g. thyrotoxicosis, mitral stenosis, alcohol
- *Rate control:* Beta-blockers, diltiazem, digoxin, amiodarone
- *Rhythm control:* Amiodarone, flecainide
- *Anticoagulation:* Warfarin or the new oral anticoagulants: Dabigatran, rivaroxaban, apixaban.
- *AF ablation:* Antrum pulmonary vein ablation, pulmonary vein antrum isolation, circumferential ablation.
- *Cardioversion:* If AF < 48 hours and low risk for stroke

***Examiner:* What is the risk of stroke in nonvalvular AF?**

Candidate: Around 5% per year.

***Examiner:* How do you assess (predict) the risk of stroke in nonvalvular AF?**

Candidate: Using the **CHA2 DS2-Vasc** score

C (CHF points 1), **H** (hypertension points 1), **A2** (age > 75 points 2), **D** (Diabetes points 1), **S** (stroke/TIA points 2), **V** (vascular disease "CAD, PVD" points 1), **A** (age 65–74 points 1), **Sc** (sex category female point 1)

Patients with AF and a score of 2 or more should be anticoagulated. Many patients with a score of 1 should be considered for oral anticoagulation. Aspirin can be used if anticoagulation is declined.

INFECTIVE ENDOCARDITIS

Common Pitfalls

- Missing the signs of IE in patients with valvular lesions.
- Failure to recognize the presence of stroke in a patient with IE.

Examiner Instructions

- Examine the cardiovascular system.
- Have a look at this patient's hands and then examine the heart.

***Examiner:* What are the signs of IE? And what are the mechanisms behind them?**

Candidate:

- Splinter hemorrhages (vascular phenomenon "septic emboli")
- Janeway lesions (vascular phenomenon "septic emboli")
- Roth spots (immune complex phenomenon)
- Osler's nodes (immune complex phenomenon)
- Finger clubbing **(Figure 1.1)**

***Examiner:* What organisms cause IE?**

Candidate:

- *Streptococcus viridans*

- *Staphylococcus epidermidis*
- *Enterococcus*
- HACEK group (*Hemophilus parainfluenzae* and *aphrophilus, Actinobacillus, Cardiobacterium, Eikenella* and *Kingella*)

Examiner: **What are the causes of culture negative endocarditis?**

Candidate:

- Certain organisms:
  - HACEK group
  - *Bartonella*
  - Coxiella
  - *Chlamydia*
  - *Legionella*
  - *Brucella*
- Prior antibiotic use
- Fungal endocarditis

Examiner: **What is the significance of growing *Streptococcus bovis* in a blood culture?**

Candidate: There is a significant association between the presence of *Streptococcus bovis* endocarditis and colonic neoplasia.

Examiner: **How do you collect blood cultures in suspected endocarditis?**

Candidate:

- They should be collected prior to antibiotic use unless the patient is severely ill
- Use strict sterile technique
- Minimum number of cultures 3
- Minimum amount of blood for each culture is 10 mL
- Should be from separate veins
- Should be at different times
- Not necessarily during the fever.

Examiner: **What criteria are used to diagnose IE?**

Candidate: The modified Duke criteria: Two major and five minor criteria. The major criteria depend on the blood culture findings and evidence of endocardial involvement. The minor criteria involve the presence of a predisposing heart condition, fever, vascular phenomenon, immunological phenomena or a positive culture that does not meet major criteria.

- *Definite IE:* 2 major, 1 major and 3 minor or 5 minor.
- *Possible IE:* 1 major and 1 minor or 3 minor criteria.

Examiner: **What are the complications of IE?**

Candidate:

- Local complications:

  - Heart failure
  - Valve dysfunction and destruction
- Systemic complications:
  - Systemic embolization leading to stroke, cerebral abscesses, septic pulmonary emboli, disseminated abscesses in other organs and mycotic aneurysms
  - *Immune complex phenomena:* Glomerulonephritis, Osler's nodes and Roth spots
  - Severe sepsis or septic shock.

Examiner: **What do you mean by mycotic aneurysms?**

Candidate: Mycotic aneurysms result from septic embolization into the wall of the blood vessels. It can occur anywhere in the body but intracranial vessels are the most frequently involved with a poor prognosis.

Examiner: **What is the sensitivity of transthoracic and transesophageal echocardiography in diagnosing IE?**

Candidate:

- Sensitivity of TTE is about 60%
- Sensitivity of TEE > 90%

Examiner: **What are the poor prognostic factors in IE?**

Candidate:

- Old age
- Diabetes
- Prosthetic valve IE
- Presence of complications from IE
- Staphylococcal IE
- Fungal IE
- Large vegetation's on echo

Examiner: **What is the usual duration of therapy in IE?**

Candidate: 4–6 weeks.

Examiner: **What are the indications for surgery in IE?**

Candidate:

- Refractory heart failure or cardiogenic shock
- Uncontrolled infection such as abscess formation, fistula or enlarging vegetation despite treatment
- Prosthetic valve IE
- Fungal IE
- Multidrug resistant organism
- Persistent fever and positive blood culture after 7–10 days
- Large vegetation > 10 mm with systemic embolization
- Very large vegetation > 15 mm

Examiner: **Can endocarditis occur due to noninfectious causes?**

Candidate: Yes, vegetations on valves can occur due to other noninfectious conditions. This is called *Nonbacterial thrombotic endocarditis or Marantic endocarditis*. The causes are:

- Malignancies
- Libman Sack's endocarditis of SLE
- Hypercoagulable states

Examiner: **What features differentiate Marantic endocarditis from IE?**

Candidate: Absence of fever and leukocytosis, absence of heart murmur, small size of the vegetation's on echocardiography

Examiner: **How would you manage this patient?**

Candidate:

- CBC, C-reactive protein (CRP) erythrocyte sedimentation rate (ESR), rational functional tester (RFT)
- Blood cultures (minimum 3)
- Echocardiography (TEE better than TTE)
- IV antibiotics (initially empiric combination, e.g. ampicillin and gentamicin or vancomycin, gentamicin and ciprofloxacin and then guided by culture result)
- Surgery as mentioned above
- Colonoscopy if blood culture grows *Streptococcus bovis*.

Examiner: **What are the indications for antibiotic prophylaxis against infective endocarditis in patients with valvular heart disease?**

Candidate: Indications of antibiotic prophylaxis depend on the type of valvular heart disease and the type of surgical procedure.

High-risk patients that need prophylaxis are:

- Patients with prosthetic heart valves
- Patients with previous IE
- Cardiac transplant recipients with abnormal valve
- *Certain patients with congenital heart disease:* Who have unrepaired cyanotic congenital heart disease or repaired congenital heart disease with residual defects adjacent to the prosthetic device or repaired congenital heart defect with prosthetic material placed during surgery (only for the first 6 months after surgery).

Procedures that require prophylaxis in the high-risk patients are:

- Dental procedures requiring manipulation of the gingival, periapical region or perforation of the mucosa
- *Respiratory procedures that involve incision and biopsy of mucosa:* Such as tonsillectomy, adenoidectomy, empyema drainage and bronchoscopy with biopsy (American but not European guidelines).
- Skin surgery (American but not European guidelines).

Examiner: **Is prophylaxis needed before gastrointestinal tract (GIT) or genitourinary (GU) procedures?**

Candidate: No.

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2

Respiratory Cases

HOW TO EXAMINE THE RESPIRATORY SYSTEM?

- Wash your hands.
- Shake hands with the patient, introduce yourself and take permission.
- Position the patient at a 45° angle and request him or her to remove the upper clothes.
- Inform the patient to alert you in case, you cause any discomfort or pain to him/her during the examination.
- Start by inspecting the patient and the surrounding. Pay particular attention to the use of oxygen, non-invasive ventilation machines, ongoing intravenous infusion and sputum pot. Look at the patient for any morphological appearance that may give a clue to the chest findings. Features of Cushing's syndrome due to chronic steroid use for chronic lung diseases such as interstitial lung diseases and severe asthma are frequently missed by candidates in clinical exams. Observe for the presence of cachexia [malignancy, chronic obstructive pulmonary disease (COPD) or tuberculosis (TB)], facial features of scleroderma, systemic lupus erythematosus (SLE) or sarcoid lesions. A patient who cannot move his neck in the exam might have ankylosing spondylitis with apical lung fibrosis.
- Hold the patients hand, put your fingers on the radial pulse and pretend to count the pulse whilst counting the respiratory rate to distract the patient's attention.
- Examine the hands for nicotine stain, finger clubbing, palmar erythema and flapping tremor that might suggest CO_2 retention. Pay particular attention to the presence of rheumatoid hand deformities, tight skin or fingertip ulceration of scleroderma. Asymmetric wasting of the small muscles of one hand may suggest a pancoast tumor compressing the brachial plexus (**Figures 2.1** and **2.2**).

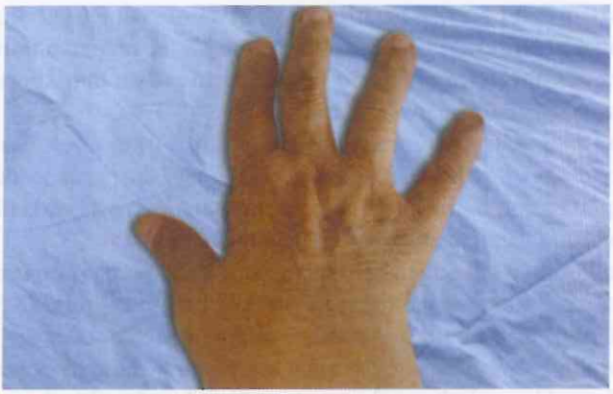

FIGURE 2.1 Small muscle wasting in the hand

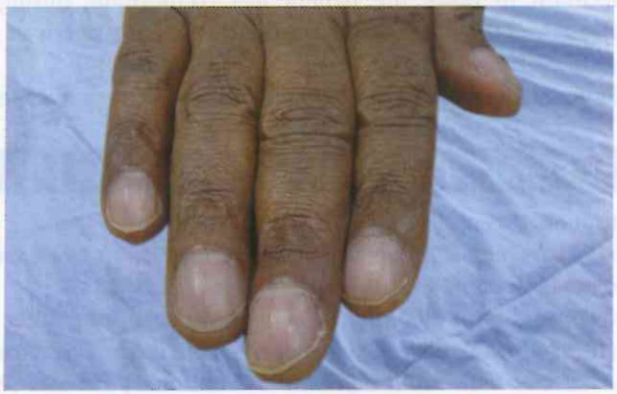

FIGURE 2.2 Finger clubbing and cyanosis in a patient with severe idiopathic pulmonary fibrosis (IPF)

- In the eyes look for features of Horner's syndrome such as ptosis, meiosis and enophthalmos. Look also for conjunctival pallor and the presence of episcleritis (may suggest rheumatoid) or uveitis (ankylosing spondylitis).
- Examine the mouth for cyanosis
- Examine the neck for lymphadenopathy
- Examine the jugular venous pressure (JVP) (cor pulmonale)
- Examine the legs for pitting edema (may be left till the end, but should not be forgotten). Remember when examining for pitting edema to enquire from the patient about leg pain before pressing over the legs and to direct your face towards the patient for tenderness rather than towards the examiner
- Follow the four steps of chest examination—Inspection, palpation, percussion and auscultation.
- *Inspection:* Look for chest deformities such as pectus excavatum and carinatum or kyphoscoliosis (see **Figures 1.3 and 1.4**), asymmetry of the chest,

patients with cystic fibrosis have long lasting venous catheters (Hickman's line or peripherally inserted central catheter (PICC) for repeated administration of antibiotics. Presence of dilated veins over the chest may suggest superior vena cava obstruction.

- There are four things you need to palpate for during chest examination—chest expansion, tactile vocal fremitus, position of the trachea and apex beat
- During percussion of the chest, always compare the right and left side at the same level and listen to any difference in the note.
- If you find dullness at one lung base, perform tidal percussion immediately.
- During auscultation, concentrate first on the breath sounds. They should be of equal intensity on both sides and should be vesicular over the lungs. Next, listen for any adventitial sounds such as crackles, rhonchi or pleural rub. If you find crackles, ask the patient to cough and notice the change in quality of crackles. Do not forget to listen for vocal resonance, it is more reliable than vocal fremitus.
- It is important to make the patient feel comfortable during the chest examination from the back. You may give the patient a pillow to put his/her arms on while sitting on the bed.
- Do not forget to percuss and auscultate the lateral sides of the chest.

BILATERAL BASAL CRACKLES

The most common 4 scenarios:

1. *Bilateral crackles with clubbing in a young patient:* Bronchiectasis most likely secondary to cystic fibrosis.
2. *Bilateral crackles with clubbing in a older patient:* Idiopathic pulmonary fibrosis (IPF) (consider also chronic hypersensitivity pneumonitis).
3. *Bilateral crackles with hand deformities:* Pulmonary fibrosis secondary to rheumatoid arthritis.
4. *Bilateral crackles with signs of scleroderma:* Scleroderma.

Common Pitfalls

- Failure to see Hickman line for long-term antibiotics in cystic fibrosis (CF).
- Diagnosing idiopathic pulmonary fibrosis (IPF) in a young patient.

Examiner Instructions

- Examine this patient's respiratory system
- Examine this patient's chest
- Listen to the chest.

Examiner: **How would you differentiate crackles in bronchiectasis, lung fibrosis and pulmonary edema?**

Candidate

- *In bronchiectasis:* Course and change with cough

- *In pulmonary edema:* Mostly fine or coarse, mid to late inspiratory and do not change with cough
- *In lung fibrosis:* Fine, late inspiratory and do not change with cough (usually Velcro crackles)

Examiner: **Which crackles are heard during early inspiration?**

Candidate:

- Crackles heard during early inspiration are indicative of airways disease, such as chronic bronchitis and emphysema.
- Crackles heard during late inspiration are indicative of parenchymal disorders, such as pulmonary fibrosis, interstitial pneumonitis, and pneumonia

Examiner: **Where do you examine for a right middle lobe abnormality?**

Candidate: The right middle lobe is represented in the right mid chest anteriorly and laterally (between 4th and 6th ribs) but not posteriorly. When you examine the chest posteriorly, you examine for abnormalities in upper and lower lobes only.

IDIOPATHIC PULMONARY FIBROSIS AND INTERSTITIAL LUNG DISEASE

Examiner: **What is the most common cause of interstitial lung disease (ILD)?**

Candidate: Idiopathic pulmonary fibrosis is the most common cause.

Examiner: **What are the diagnostic criteria for idiopathic pulmonary fibrosis (IPF)?**

Candidate: The three main diagnostic criteria are:

1. Exclusion of a known cause of ILD.
2. High resolution computed tomography (CT) scan criteria.
3. Pathologic criteria (lung biopsy).

Examiner: **What do you know about the high-resolution computed tomography (HRCT) findings in IPF?**

Candidate:

- Honeycombing
- Bilateral, predominantly subpleural basal fibrosis
- Absence on HRCT of features that point to other etiology.

Examiner: **What is the name given to the histopathologic appearance of a lung biopsy in IPF?**

Candidate: Usual interstitial pneumonia (UIP)

Examiner: **What are the causes of pulmonary fibrosis with honeycombing (UIP pattern) on CT scan of the chest? Or what diseases may mimic IPF?**

Candidate:

- Idiopathic pulmonary fibrosis
- Chronic hypersensitivity pneumonitis (extrinsic allergic alveolitis)
- Connective tissue diseases such as RA and scleroderma
- *Drug induced:* Methotrexate used in treatment of RA
- Sarcoidosis
- Asbestosis

Examiner: **In a patient with RA treated with methotrexate; how would you differentiate pulmonary fibrosis due to methotrexate from that due to RA?**

Candidate: This may be difficult to differentiate, but the presence of lymphocytes in the bronchoalveolar lavage (BAL) fluid, granuloma in the lung biopsy specimens and peripheral eosinophilia can suggest it is methotrexate rather than RA-related ILD. Presence of active RA and neutrophilic BAL suggest RA-related ILD.

Examiner: **Can you name some new drugs for the treatment of IPF?**

Candidate:

- Pirfenidone
- Nintedanib

Examiner: **What are the complications or comorbidities associated with IPF?**

Candidate:

- Pulmonary hypertension
- Lung cancer
- Coronary artery disease
- Pulmonary embolism
- Respiratory failure

Examiner: **How would you manage this patient?**

Candidate:

Investigations:

- High resolution CT chest
- Pulmonary function tests, arterial blood gas (ABG)
- 6 minute walk test
- Bronchoscopy with BAL
- Autoimmune work-up [rheumatoid factor (RF), anti-cyclic citrullinated peptide (ACCP), ASCL-70, ENA]
- Hypersensitivity pneumonitis panel (serum precipitins)
- Surgical lung biopsy.

Treatment:

- Stop smoking

- Home oxygen therapy
- Vaccination
- *Drugs:* Pirfenidone and nintedanib
- Referral for lung transplantation
- Pulmonary rehabilitation

Examiner: **What is the prognosis for IPF?**

Candidate: The prognosis of IPF is poor with a median survival of 2.5–3.5 years.

BRONCHIECTASIS

Examiner: **What are the causes of bronchiectasis?**

Candidate: "OK CHAPS"

- **O:** Obstruction of the bronchus (tumor and foreign body)
- **K:** Kartagener's syndrome
- **C:** Cystic fibrosis
- **H:** Hypogammaglobulinemia
- **A:** Allergic bronchopulmonary aspergillosis
- **P:** Pneumonia and previous chest infections (whooping cough and measles)
- **S:** Sjögren's syndrome and CTD.

Examiner: **What is Kartagener's syndrome?**

Candidate: This is an autosomal recessive disorder, which results in dextrocardia, sinusitis and bronchiectasis. It is caused by defective ciliary function in the airways "ciliary dyskinesia" leading to defective mucus clearing and repeated infections and bronchiectasis. The main diagnostic method is nasal and bronchial brushings or biopsy.

Examiner: **What are the complications of bronchiectasis?**

Candidate:

- Massive hemoptysis
- Repeated chest infections
- Lung abscess
- Cor pulmonale
- Systemic amyloidosis

Examiner: **How do you investigate this patient?**

Candidate:

- Complete blood count (CBC)
- Sputum gram stain and culture
- Chest X-ray
- HRCT lungs
- Serum immunoglobulin levels
- Workup for cystic fibrosis
- Skin prick test for aspergillus and aspergillus specific IgE level

- Bronchoscopy and BAL
- Pulmonary function text (PFT)

Examiner: **How would you treat this patient?**

Candidate:

- Antibiotics
- Bronchodilator therapy
- Mucolytic therapy
- Saline nebulization
- Treat the underlying cause
- Vaccination
- Chest physiotherapy
- Surgery for localized bronchiectasis or massive hemoptysis.

CYSTIC FIBROSIS

Examiner: **How would you diagnose cystic fibrosis?**

Candidate:

- Clinical features, PFT, sputum culture and HRCT chest
- Two samples of sweat chloride > 60 mmol/L
- *Genetic testing:* Indications.
  - Intermediate results of sweat chloride test (40–60)
  - Family history of CF
  - Pre-pregnancy

Examiner: **What is the basic abnormality in CF?**

Candidate: Defect in the cystic fibrosis transmembrane conductance regulator (CFTR) gene results in abnormal cAMP-regulated chloride transport across epithelial cells. This results in decreased secretion of chloride and increased reabsorption of sodium and water across epithelial cells resulting in decreased hydration of the mucus, which becomes stickier to bacteria leading to recurrent infections and inflammation.

Examiner: **Can you name some conditions that give false positive high sweat chloride?**

Candidate:

- *Endocrine diseases:* Adrenal insufficiency—hypothyroidism—pan hypopituitarism
- Glycogen storage disease
- Mucopolysaccharidosis
- Atopic dermatitis

Examiner: **How would you manage a cystic fibrosis patient?**

Candidate:

- Chest physiotherapy and chest clearance
- Mucolytic nebulization
- Dornase alfa (Pulmozyme)
- Nebulized hypertonic saline
- Bronchodilators
- Antibiotics
- Pancreatic enzyme supplements
- Multivitamins (including fat-soluble vitamins to treat associated conditions or complications (e.g. insulin, bisphosphonates)
- *New drug:* Ivacaftor alone or in combination with lumacaftor
- Patient and family counseling

Examiner: **If this patient presents with acute right iliac fossa pain what should come to mind?**

Candidate: Meconium ileus equivalent (MIE). Usually manifests with right lower quadrant abdominal pain. One may feel a mass in the RIF. Management is with oral laxatives and enemas, rehydration and correction of associated electrolyte abnormalities. In severe cases, surgery may be indicated.

Examiner: **Which bacteria commonly cause infections in CF patients?**

Candidate:

- *Pseudomonas aeruginosa,*
- *Burkholderia cepacia,*
- *Stenotrophomonas maltophilia,*
- *Staphylococcus aureus,*
- *Haemophilus influenzae*

Examiner: **What is the mechanism of action of ivacaftor in cystic fibrosis?**

Candidate: It is a CFTR protein potentiator.

Examiner: **If this man gets married what is the chance that he is infertile and why?**

Candidate: More than 95% of men with CF are infertile. This is because of absence of vas deferens and not due to a defect in spermatogenesis. Female infertility occurs in 20% of CF patients and is related to amenorrhea of malnutrition.

DULLNESS AT THE LUNG BASE

Common Pitfalls

- Failure to mention pleural thickening in the differential diagnosis
- Failure to observe pleural aspiration or biopsy scar
- Failure to observe thoracotomy scar suggestive of lobectomy or

Examiner Instructions

- Examine this patient's respiratory system
- Examine this patient's chest.

The differential diagnosis of dullness at the lung base includes:

- **Pleural effusion**
  Signs: Site of needle aspiration, stony dullness to percussion, decreased breath sounds and decreased vocal resonance.
- **Collapse**
  Signs: Dullness to percussion, tracheal deviation to the same side and decreased breath sounds.
- **Lobectomy/pneumonectomy**
  Signs: Thoracotomy scar, asymmetrical chest (depressed on the affected side), dullness to percussion, tracheal deviation to the same side and decreased breath sounds (signs are similar to a collapse in addition to the presence of a scar).
- **Elevated hemidiaphragm**
  Signs: Dullness, decreased breath sounds, decreased vocal resonance and fremitus.
- **Consolidation**
  Signs: Dullness to percussion, decreased breath sounds, bronchial breathing, increase vocal resonance, egophony and whispering pectoriloquy.
- **Pleural thickening**
  Signs: Similar to pleural effusion.

Examiner: **How would you differentiate an elevated hemidiaphragm from other causes of dullness at lung base?**

Candidate: By tidal percussion—percuss down the back until you reach the area of dullness. Keep your finger over that level and ask the patient to breathe in deeply and hold the breath. Now percuss again at that level. If the note becomes resonant then the cause is supradiaphragmatic. In diaphragmatic paralysis or infradiaphragmatic pathology, the note will remain dull.

PLEURAL EFFUSION

Examiner: **What are the causes of an exudative pleural effusion?**

Candidate:

- *Common causes*
  - Malignancy
  - Parapneumonic
  - Tuberculosis
- *Less common causes*
  - Rheumatoid and autoimmune disease
  - Asbestosis
  - Pancreatitis

  - Acute MI and post CABG
- *Rare causes*
  - Drugs
  - Yellow nail syndrome.

Examiner: **What are the causes of a transudative pleural effusion?**

Candidate:

- Congestive heart failure (CHF)
- Liver cirrhosis
- Nephrotic syndrome
- Constrictive pericarditis
- Hypothyroidism
- Hypoalbuminemia
- Urinothorax
- Meigs' syndrome

Examiner: **What are the causes of a lymphocytic pleural effusion?**

Candidate:

- Malignancy
- Lymphoma
- TB
- Sarcoidosis
- Rheumatoid arthritis

Examiner: **What are Light's criteria?**

Candidate: These are criteria for differentiating exudative from transudative effusion. Presence of any of the following suggests an exudative effusion:

- Pleural protein/serum protein > 0.5
- Pleural LDH/serum LDH > 0.6
- Pleural LDH > 2/3 upper limit of laboratory normal value for serum LDH.

Examiner: **What is the main disadvantage of using the light criteria to differentiate an exudative from transudative effusion?**

Candidate: The main disadvantage is in patients with CHF who are receiving diuretic therapy. In these patients, pleural fluid may turn exudative.

Examiner: **How would you differentiate a pleural effusion of CHF with diuretic use from other causes of an exudative effusion?**

Candidate: By measuring the serum-pleural albumin gradient or measuring the NT-pro BNP in pleural fluid. A serum-pleural albumin gradient > 1.2 g/dL suggests that the patient has a transudative pleural effusion

Examiner: **How would you investigate a patient with suspected pleural effusion?**

Candidate:

- Chest X-ray to confirm the presence of an effusion

- If the patient has a cause of a transudative effusion such as heart failure or liver cirrhosis, we correct that condition.
- *US guided pleural aspiration:* If no obvious cause of transudative effusion
- We send pleural fluid for—protein, LDH, pH, GS and CS, AFP smear and culture, cell count and cytology.
- *CT scan chest:* If pleural aspiration did not give a diagnosis.
- VAT pleural biopsy.

Examiner: **How much fluid should be in the pleural space for it to be seen on a Chest X-ray?**

Candidate: Around 200 mL.

Examiner: **What are the advantages of US in guiding pleural aspiration?**

Candidate: It increases the likelihood of successful aspiration particularly if the effusion is small, reduces the risk of organ puncture and differentiates pleural thickening from effusion.

Examiner: **What pleural fluid test should you send for if you suspect mesothelioma?**

Candidate: Pleural mesothelin (tumor marker for mesothelioma)

Examiner: **What are the characteristic findings in rheumatoid pleural effusion?**

Candidate: Glucose < 1.6 mmol/L, pH < 7.30, high LDH and presence of rheumatoid factor.

Examiner: **What are the causes of pleural thickening?**

Candidate:
- Empyema
- Mesothelioma
- Tuberculosis (TB)

PNEUMONECTOMY/LOBECTOMY

Common Pitfall

Missing the scar (particularly lateral thoracotomy scar) **(Figure 2.3)**

Signs: Thoracotomy scar, asymmetrical chest (depressed on the affected side) dullness to percussion, tracheal deviation to the same side and decreased breath sounds (signs are similar to collapse in addition to presence of surgical scar)

Note: If you do not see the scar you may miss the diagnosis.

Most common causes in the exam:
- Pneumonectomy or lobectomy for lung cancer (in young patient surgery for carcinoid)
- Old surgical treatment for TB

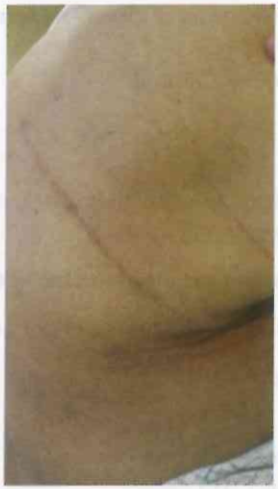

FIGURE 2.3 Right thoracotomy scar for resection of lung lobe

- Surgery for localized bronchiectasis
- Decortication for empyema.

UNILATERAL LUNG FIBROSIS (POST-TUBERCULOUS)

Signs: Asymmetry of the chest (affected side is depressed), decreased chest expansion on the affected side, trachea shifted to the affected side, dull percussion note on the affected side, decreased breath sounds on the affected side. Bronchial breathing, increased vocal fremitus and vocal resonance as in consolidation may be found. (There might be a scar if the patient was treated surgically before the anti-TB drugs era).

Differential Diagnosis of Unilateral Pulmonary Fibrosis

- Post-TB sequelae (most common in examination)
- Radiation induced pulmonary fibrosis (for lung or breast cancer)
- Post-bronchiectasis/splenomegaly (CVID)
- Ankylosis spondylitis (uncommon in examination)

CHRONIC OBSTRUCTIVE PULMONARY DISEASE

Common Pitfalls

- Candidates do not suspect chronic obstructive pulmonary disease (COPD) (emphysema) when there is diffuse reduction in breath sounds over the chest
- Attributing clubbing to COPD
- Failure to recognize the presence of Cor pulmonale

- Failure to observe biphasic positive airway pressure (BIPAP) machine beside patient
- Failure to recognize nicotine staining

Examiner Instructions

- This patient complains from exertional dyspnea, examine his chest
- Examine the respiratory system.

Examiner: **What is the most important predictor of COPD exacerbation?**

Candidate: The most important predictor is a history of previous exacerbation.

Examiner: **How would stage COPD?**

Candidate:
The new COPD staging depends on three factors:
1. Severity of symptoms (by CAT)
2. Degree of air flow limitation (using spirometry)
3. Frequency of exacerbations

Stage	*Characteristics*	*Spirometric classification*	*Exacerbation per year*	*CAT*
A	Low-risk Less symptoms	GOLD 1–2	≤ 1	<10
B	Low-risk More symptoms	GOLD 1–2	≤ 1	≥10
C	High-risk Less symptoms	GOLD 3–4	≥ 2	<10
D	High-risk More symptoms	GOLD 3–4	≥ 2	≥10

Examiner: **What are the comorbidities and extrapulmonary manifestations of COPD?**

Candidate:
- Cor pulmonale
- Cardiovascular disease
- Metabolic syndrome
- Polycythemia
- Muscle wasting
- Osteoporosis
- Depression
- Lung cancer

Examiner: **How would you manage this patient?**

Candidate:
- CBC, hematocrit and ABG
- Chest X-ray

- Stop smoking
- Bronchodilators with or without inhaled steroids (depending on the stage)
- Treat exacerbations
- Vaccination (influenza and pneumococcal)
- Long-term oxygen therapy (LTOT)
- Non-invasive ventilation
- Pulmonary rehabilitation

Examiner: **What are the indications of LTOT in COPD?**

Candidate:

- PaO_2 < 55 mm Hg or $SpaO_2$ < 88% on two occasions when patient stable
- PaO_2 between 55–60 mm Hg or $SpaO_2$ 88% with evidence of pulmonary hypertension, cor pulmonale, or polycythemia (HCT > 55%)

Examiner: **What are the spirometric findings in COPD?**

Candidate:

- FEV1/FVC < 70%
- Irreversibility with bronchodilator use (increase in FEV1 < 12%)
- Decreased diffusing capacity of the lungs for carbon monoxide (DLCO)
- Evidence of air trapping (increase RV)

Examiner: **How do you differentiate asthma from COPD on spirometry?**

Candidate:

- Irreversibility of air way obstruction
- Decreased diffusing capacity of the lungs for carbon monoxide (DLCO)

Examiner: **Is COPD a cause for finger clubbing?**

Candidate: COPD per se is not a cause of finger clubbing. Presence of clubbing in COPD patient should raise the suspicion of lung cancer or bronchiectasis.

Examiner: **What is the best way to deliver O_2 in acute COPD exacerbation and why?**

Candidate: Venturi masks are preferred because they permit a precise delivered fraction of O_2 (24–60%)

Examiner: **What value of pulse oxymetry corresponds to PaO_2 of 60 mm Hg?**

Candidate: $SpaO_2$ of 90% corresponds to PaO_2 of 60 mm Hg

FURTHER READING

1. Boyle MP, Bell SC, Konstan MW, et al. A CFTR corrector (lumacaftor) and a CFTR potentiator (ivacaftor) for treatment of patients with cystic fibrosis who have a phe508del CFTR mutation: a phase 2 randomised controlled trial. Lancet Respir Med. 2014;2(7):527-38.
2. Global strategy for the diagnosis, management and prevention of chronic obstructive pulmonary disease. Updated 2013. Available from: *www.goldcopd.org/uploads/*

3. Light RW, MacGregor MI, Luchsinger PC, et al. Pleural effusions: the diagnostic separation of transudates and exudates. Ann Intern Med. 1972;77:507-13.
4. Light RW. Diagnostic principles in pleural disease. Eur Respir J. 1997;10(2):476-81.
5. Raghu G, Collard HR, Egan JJ, et al. An official ATS/ERS/JRS/ALAT statement: idiopathic pulmonary fibrosis: evidence-based guidelines for diagnosis and management. Am J Respir Crit Care Med. 2011;183(6):788-824.
6. van der Doef HP, Kokke FT, van der Ent CK, et al. Intestinal obstruction syndromes in cystic fibrosis: meconium ileus, distal intestinal obstruction syndrome, and constipation. Curr Gastroenterol Rep. 2011;13(3):265-70.

3

Abdominal Cases

HOW TO EXAMINE THE ABDOMEN?

- Wash your hands.
- Shake hands with the patient, introduce yourself and take permission to examine them.
- Ask the patient to lie flat with one pillow behind the head
- I suggest that for abdominal examination the candidate asks the patient to remove the upper clothes completely and not only to the level of the nipple. The reason is that a spider angiomata over the shoulder may be easily missed if the shoulders are not exposed **(Figure 3.1)**.
- A quick surveillance of the patient and the surrounding may give important clues to the diagnosis of the case. A patient with thalassemia major may have a typical thalassemic face. A patient who appears hyperpigmented may have hemochromatosis. A patient with Cushing's syndrome may have had a renal transplant. A patient using a hearing aid may have Alport syndrome. Inspect also the surrounding for a blood transfusion set (hematological diseases, hemolytic anemia such as thalassemia or bleeding varices), intravenous antibiotics (spontaneous bacterial peritonitis), dialysis machine, etc.
- Examine the hands for clubbing, leukonychia **(Figure 3.2)**, koilonychia, Beau's lines of chemotherapy, palmar erythema, Dupuytren's contracture **(Figure 3.3)** and flapping tremor.
- Examine the arms for the presence of an arteriovenous (AV) fistula.
- Examine the eyes for jaundice, pallor and xanthelasma (may suggest primary biliary cirrhosis) **(Figures 3.4 and 3.5)**
- Examine the face, ears and mouth for parotid enlargement, hyperpigmentation, use of hearing aids, pallor and jaundice.
- Feel the neck for lymphadenopathy

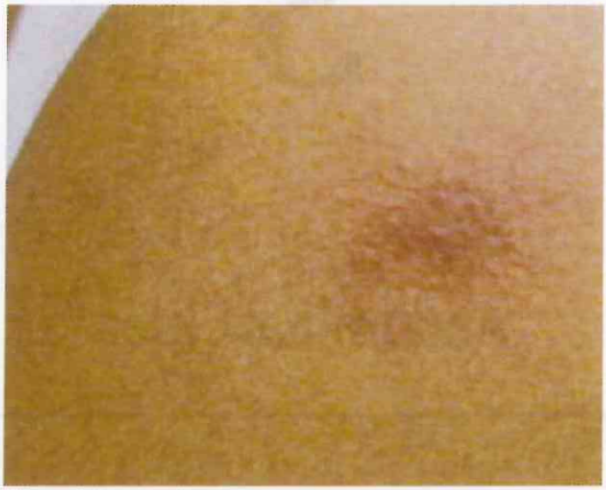

FIGURE 3.1 Spider angiomata over the shoulder may be missed if not properly exposed

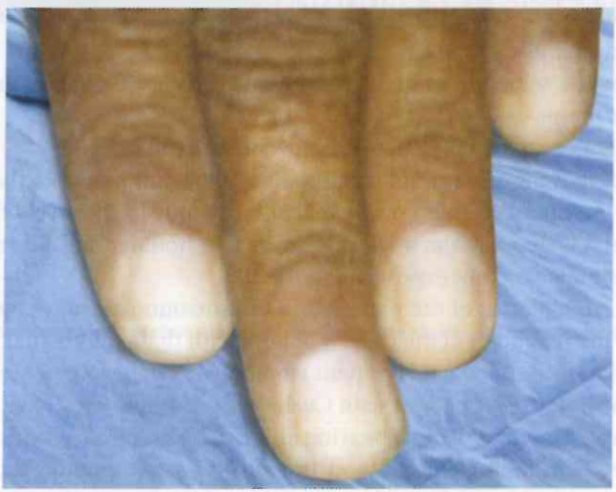

FIGURE 3.2 Leukonychia

- Inspect and feel the chest for spider angiomata, hair distribution, gynecomastia and paper money skin.
- Examine the legs for pitting edema (may be left till the end but should not be forgotten). Remember when examining for pitting edema to enquire from the patient about leg pain before pressing over the legs and to direct your face towards the patient to elicit tenderness rather than towards the examiner.
- Inspect the abdomen for distension, full flanks, scars (particularly Mercedes-Benz scar of liver transplant (**Figure 3.6**) and hidden scars in the right iliac

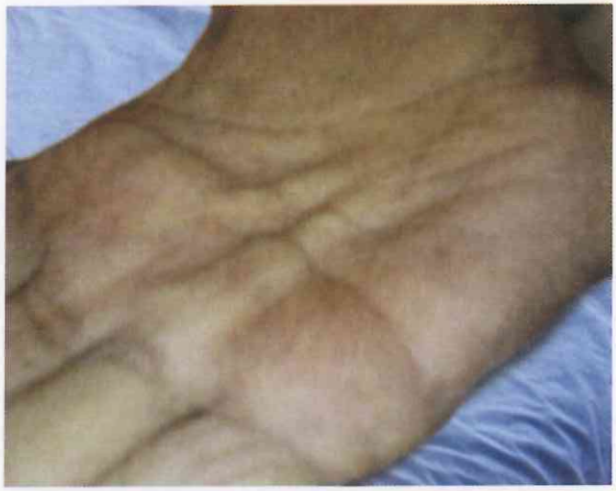

FIGURE 3.3 Dupuytren's contracture

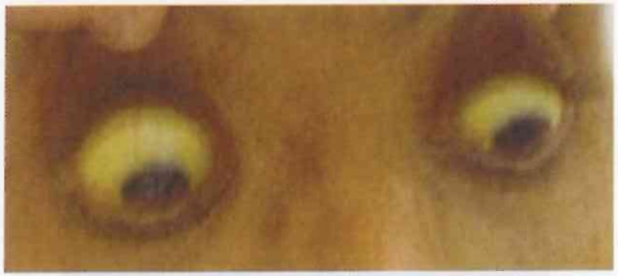

FIGURE 3.4 Jaundice

FIGURE 3.5 Xanthelasma in a patient with jaundice may suggest primary biliary cirrhosis

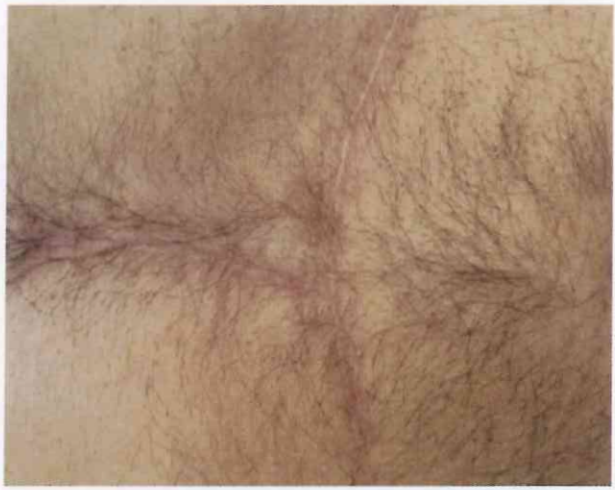

FIGURE 3.6 Mercedes-Benz scar in a patient with a liver transplant

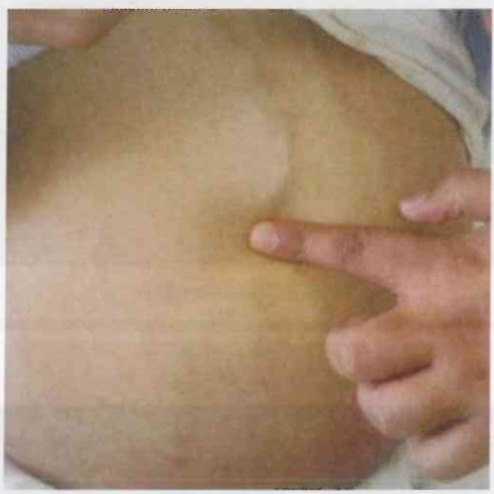

FIGURE 3.7 Dilated veins over the abdomen (check for direction of blood flow)

fossa of a renal transplant), dilated veins and caput medusa. If dilated veins seen, check the direction of blood flow (**Figure 3.7**). Look for the shape of the umbilicus (everted or inverted) and hernial orifices

- Before palpating the abdomen ask the patient if he/she has any abdominal pain. Palpate the abdomen superficially and then for organomegaly and minor splenomegaly
- Percuss the abdomen for organomegaly and shifting dullness
- Listen for bowl sounds, renal and hepatic bruit

- Inform the examiner that you would normally perform genital examination and rectal examination to complete the examination.

CHRONIC LIVER DISEASE

Common Pitfalls

- Failure to look for spider nevi above the nipple area
- Failure to observe increased skin pigmentation suggesting hemochromatosis
- Failure to mention primary biliary cirrhosis and autoimmune hepatitis in women with chronic liver disease (CLD) or jaundice.

Examiner Instructions

- Examine this patient's abdomen
- Examine the gastrointestinal system

Examiner: **What is your diagnosis?**

Candidate: This patient has stigmata of CLD in the form of spider nevi, palmar erythema, paper-money skin and jaundice. Abdominal examination revealed ascites and splenomegaly. He has CLD.

Examiner: **What is the most common cause of chronic liver disease?**

Candidate: Hepatitis C virus in both developed and developing countries.

Examiner: **What are the causes of CLD?**

Candidate:

- Viral hepatitis C and B
- Alcoholic liver disease
- Nonalcoholic fatty liver
- Cryptogenic
- Autoimmune hepatitis
- Primary biliary cirrhosis
- Hemochromatosis
- Wilson disease
- Alpha-1 antitrypsin deficiency
- Schistosomiasis
- Budd-Chiari syndrome
- Drug-induced liver disease, e.g. methotrexate, alpha methyldopa, amiodarone
- Sarcoidosis.

Examiner: **What is portal hypertension and how does it manifest?**

Candidate: Clinically significant portal hypertension is defined as a hepatic venous pressure gradient (HVPG) (pressure gradient between the portal vein

and the inferior vena cava) ≥10 mm Hg. Variceal hemorrhage develops when the gradient exceeds 12 mm Hg.

Manifestations of portal hypertension include: Ascites, splenomegaly and portacaval anastomoses (esophageal varices, gastric varices, anorectal varices).

Examiner: **What are the mechanisms of anemia in CLD?**

Candidate: Anemia in CLD can arise from various causes such as:

- Folate deficiency
- Hypersplenism
- Bleeding

Examiner: **What is the incidence of hepatocellular carcinoma in cirrhotic patients?**

Candidate: 3% per year in patients with viral hepatitis and alcohol-induced cirrhosis.

Examiner: **Which cause of CLD carries the least risk for hepatocellular carcinoma?**

Candidate: Wilson's disease.

Examiner: **Which type of CLD rarely manifests with dermatologic stigmata of CLD?**

Candidate: NAFLD

Examiner: **How would you assess the severity of CLD?**

Candidate:

- The Child-Turcotte-Pugh Scoring System for Cirrhosis which uses the following criteria:
  - Encephalopathy
  - Ascites
  - Bilirubin
  - Albumin
  - Prothrombin time
- *Model for end-stage liver disease (MELD) scoring system:* Which uses serum bilirubin, serum creatinine, and the INR to predict survival. The modified MELD (MELD-Na) incorporates serum sodium also. Patient should be referred for liver transplantation if the MELD score is 15 or more

Examiner: **What are the clinical manifestations that suggest the cause of CLD is alcohol?**

Candidate:

- Parotid enlargement
- Dupuytren's contracture
- Paper-money skin

Examiner: **What are the clinical manifestations that suggest hepatitis C as the cause of CLD?**

Candidate:

- Cryoglobulinemia **(Figure 3.8)**
- Porphyria cutanea tarda

Examiner: **What are the clinical features that suggest primary biliary cirrhosis as the cause of CLD?**

Candidate:

- Presence of xanthelasma
- Scratch marks of pruritus
- Presence of dry eyes and mouth (sicca syndrome)

Examiner: **When do you see clinical jaundice?**

Candidate: Jaundice, is usually recognizable when serum bilirubin levels exceed 2.5 or 3.0 mg/dL

Examiner: **What is the paper-money skin sign and where do you see it?**

Candidate: Paper-money skin (or "dollar-paper" markings) is seen on the upper trunk. The skin is covered by needle-thin superficial capillaries. These thin capillaries resemble silk threads in the American dollar.

Examiner: **Are spider nevi specific for CLD? Why do they occur in CLD?**

Candidate: Spider nevi commonly occur in the upper trunk, face and the neck (above the level of the nipple) possibly because they drain into the superior vena cava. The presumed mechanism is the high estrogen level in CLD due to reduced

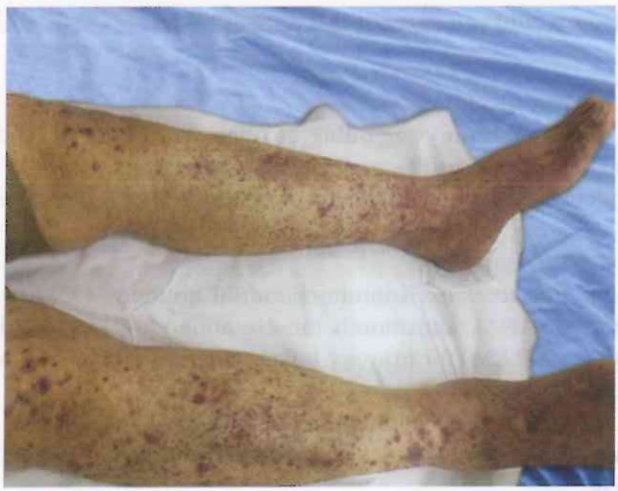

FIGURE 3.8 Cryoglobulinemia in a patient with chronic liver disease from HCV

estrogen metabolism as they can be seen in pregnant women and in patients receiving estrogen therapy. A significant number is more than 3 but any number of spider angiomata in an adult deserves workup.

Examiner: **How does porphyria cutanea tarda manifest?**

Candidate: Porphyria cutanea tarda usually manifests with blisters on sun-exposed skin. In chronic porphyria cutanea tarda skin scarring and ulceration develop. Alopecia, melasma-like hyperpigmentation and hypertrichosis may also be seen.

Examiner: **What cause of CLD other than HCV may present with porphyria cutanea tarda?**

Candidate: Hemochromatosis

Examiner: **If you find hepatomegaly in patients with cirrhosis, what you suspect?**

Candidate:
Causes of hepatomegaly in patients with cirrhosis are:

- Nonalcoholic fatty liver disease
- Alcoholic hepatitis on top of cirrhosis
- Budd-Chiari syndrome
- Hemochromatosis
- Hepatocellular carcinoma

Examiner: **What are the complications of CLD?**

Candidate: Recurrent ascites, spontaneous bacterial peritonitis, hepato-renal syndrome, hepatopulmonary syndrome, hepatic encephalopathy, hepatocellular carcinoma, portal vein thrombosis and muscle cramps.

Examiner: **How would you manage this patient?**

Candidate:

- History and investigations
  - Take a proper history regarding alcohol consumption, IV drug abuse, sexual history, history of surgery or blood transfusion
  - Complete blood count (CBC), liver function test (LFT), renal function test (RFT), clotting profile and ultrasound (US) abdomen
  - Viral hepatitis serology
  - *Autoimmune workup:* Antimitochondrial antibody (AMA), antinuclear antibodies (ANA), antismooth muscle antibodies (ASMA), liver-kidney microsomal (LKM) for primary biliary cirrhosis (PBC) and autoimmune hepatitis
  - Iron, transferrin and ferritin for hemochromatosis
  - Serum ceruloplasmin level, 24 hours urinary copper excretion for Wilson's disease
  - Slit lamp examination for Kayser-Fleischer (KF) rings for Wilson's disease

  - Alpha 1 anti-trypsin level
- Treat the underlying cause (stop alcohol, steroids for autoimmune hepatitis, hepatitis B and C treatment, etc.)
- Avoid hepatotoxic drugs
- Vaccination for hepatitis A, B, influenza and *Pneumococcus*
- Ascites management:
  - *Avoid nonsteroidal anti-inflammatory drug (NSAID):* Cause renal vasoconstriction, renal failure and less response to diuretics
  - *Avoid angiotensin converting enzyme inhibitors (ACEI):* May cause a significant drop in BP and worsening of kidney function
  - Sodium restriction to 80–120 mmol per day
  - Fluid restriction is not recommended unless sodium below 120 mEq/L
  - *Diuretic therapy:* Combination of spironolactone and furosemide
  - Paracentesis: Up to 5 L
  - Transjugular intrahepatic portosystemic shunt (TIPS)
  - Prophylaxis against variceal bleeding with nonselective beta-blockers.
- *Indications for a liver transplant:* MELD score 15 or more, recurrent ascites, variceal bleeding, encephalopathy, hepatorenal syndrome or hepatocellular carcinoma. Contraindications include—Cardiopulmonary disease, malignancy outside the liver, failure to stop alcohol or drug abuse and acquired immune deficiency syndrome (AIDS).
- Treat other complications like variceal bleeding and hepatic encephalopathy.

Examiner: **What is the mechanism of action of lactulose in hepatic encephalopathy?**

Candidate: Due to the absence of the disaccharidase enzyme in the human small intestine, lactulose (synthetic disaccharide) passes to the colon where it is catabolized by the colonic bacteria to lactic acid, which lowers the colonic pH. This in turn converts NH3 to NH4, which leads to reduced colonic bacterial growth and plasma ammonia concentration. Lactulose through its osmotic effect can reduce the transit time for colonic bacteria.

Examiner: **What is the difference between lactitol and lactulose?**

Candidate: Lactitol is as effective as lactulose but is better tolerated and has fewer side effects.

Examiner: **How would you screen this patient for hepatocellular carcinoma?**

Candidate: By US abdomen every 6 months (Alpha-fetoprotein is not recommended because it typically starts to rise only after vascular invasion has occurred. Also it has a high false-positive rate in patients with active hepatitis.

Examiner: **When do you screen this patient for varices?**

Candidate: OGD screening for varices should be performed as soon as cirrhosis is diagnosed.

Examiner: **Are you aware of any recent drugs for the treatment of hepatitis C virus infection with a very high response rate?**

Candidate:

- Combined ledipasvir and sofosbuvir
- Combined ombitasvir and dasabuvir with ribavirin

Examiner: **What are the indications to start prophylactic antibiotics in patients with cirrhotic ascites?**

Candidate:

- Any patient who has recovered from an episode of SBP should receive long-term antibiotic prophylaxis
- If the ascites protein level is low < 15 g/L.

Examiner: **What is the preferred antibiotic used for SBP prophylaxis?**

Candidate: Norfloxacin.

HEMOCHROMATOSIS

Important Clues

Think twice before you diagnose hemochromatosis in women in the menstruating or childbearing age in the exam. Usually menstruation protects against the development of frank signs of hemochromatosis such as liver disease or hyperpigmentation.

Common Pitfalls

Missing hyperpigmentation during the examination of a patient with CLD.

Examiner: **Which organs are affected in hemochromatosis?**

Candidate:

- *Liver:* Hepatomegaly or frank cirrhosis
- *Pancreas:* Diabetes mellitus
- *Thyroid:* Hypothyroidism
- *Gonads:* Hypogonadism and testicular atrophy
- *Skin:* Hyperpigmentation and porphyria cutanea tarda
- *Heart:* Heart failure
- *Skeletal:* Commonly second and third metacarpophalangeal (MCP) joints, chondrocalcinosis and osteoporosis.

Examiner: **What is "bronze diabetes"?**

Candidate: A name given to the classic manifestation triad of hemochromatosis: skin pigmentation, diabetes and cirrhosis.

Examiner: **How would you diagnose a patient with hemochromatosis?**

Candidate:

- Serum transferrin saturation and ferritin level as screening tests
- If the serum transferrin saturation and ferritin levels are high undertake genetic testing (*C282Y* and *H63D*)
- *Liver biopsy:* Not needed in *C282Y* homozygous patients with elevated transferrin saturation and ferritin levels
- *MRI liver:* To quantify hepatic iron content (still not validated).

Examiner: **How would you manage a patient with hemochromatosis?**

Candidate:

- The main treatment is repeated phlebotomy to keep serum ferritin < 50 ug/L
- Iron chelating agents
- Check blood sugar, TFT and gonadal function and treat if abnormal
- Do echocardiography and treat heart failure
- Do dexa scan and give drugs for osteoporosis prevention and treatment
- Screening for hepatocellular carcinoma and management of cirrhosis as in CLD.

Examiner: **What complications of hemochromatosis do you expect to improve with phlebotomy?**

Candidate:

- Liver disease including enzymes, degree of fibrosis, and esophageal varices
- Skin pigmentation

Recovery from diabetes, gonadal function and cardiac complications depends on the degree of damage to these organs.

PRIMARY BILIARY CIRRHOSIS

Important Clues

PBC patients in clinical exams are exclusively women in middle age or older with jaundice and pruritus with or without stigmata of CLD and portal hypertension.

If you are asked by the examiner to examine the abdomen of a patient with pruritus, the three main diagnoses you should consider are primary biliary cirrhosis, polycythemia vera or cholestatic jaundice from another cause.

Common Pitfalls

- Failure to suspect PBC in a middle aged female patient with jaundice, itching and/or stigmata of CLD
- Missing xanthelasmas **(Figure 3.5)**
- Failure to explain the cause of gritty sensations in eyes due to Sicca syndrome

Examiner: **What makes you to suspect PBC in this woman?**

Candidate:

- Being middle aged
- Presence of xanthelasmas
- Presence of scratch marks suggestive of pruritus
- Presence of dry eyes and mouth indicating associated Sicca syndrome

Examiner: **What are the two most common symptoms of PBC?**

Candidate: Fatigue and pruritus

Examiner: **How would you explain this patient's gritty eye sensation?**

Candidate: Sicca syndrome is common in patients with PBC

Examiner: **How would you investigate a patient with PBC?**

Candidate:

- Elevated alkaline phosphatase (ALP) is always present
- Positive AMA in 95% of cases (the serologic hallmark)
- Increased immunoglobulin levels
- Liver biopsy is needed in AMA negative cases
- Work-up for CLD as above.

Examiner: **How would you treat a patient with PBC?**

Candidate:

- Ursodeoxycholic acid in adequate doses is the cornerstone of treatment
- Management of pruritus
  - Cholestyramine
  - Rifampicin
  - Opiate antagonist (naltrexone)
  - *Antidepressants:* Sertraline
- *Management of sicca syndrome:* Artificial tears, moisturizers and saliva substitutes
- Statins for hyperlipidemia
- Screening for hepatocellular carcinoma and management of cirrhosis as per CLD
- Dexa scan, prevention and treatment of osteoporosis
- Avoid oral contraceptive pills (worsen cholestasis)
- Replacement of vitamins A, D, E, K.

JAUNDICE

Common Pitfalls

- Forgetting hemolysis as a cause of jaundice (particularly patients with thalassemia and typical thalassemic facies)
- Missing scratch marks of itching **(Figure 3.9)**

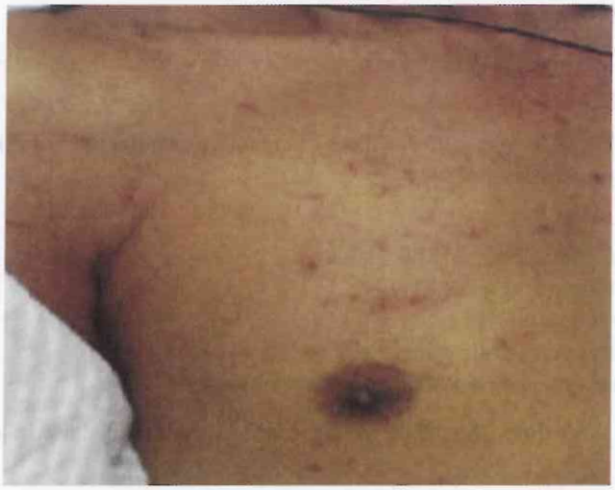

FIGURE 3.9 Scratch marks from severe itching in a patient with cholestatic jaundice. (In females consider primary biliary cirrhosis)

- Forgetting alcohol and drugs (paracetamol) in the differential diagnosis
- Forgetting Wilson's disease in young patients
- Missing stigmata of CLD
- Missing xanthelasma in primary biliary cirrhosis or hyperpigmentation in hemochromatosis.

Common Exam Scenarios

- Young patient with jaundice (Wilson's disease, thalassemia, drugs)
- Middle aged woman (viral hepatitis, PBC, autoimmune hepatitis, viral hemochromatosis)
- Middle aged man (alcohol, viral hepatitis or hemochromatosis).

Examiner Instructions

- Examine this patient's abdomen
- Have a look at this patient and proceed accordingly
- Examine the gastrointestinal system.

Examiner: **Can you name a condition other than jaundice that gives yellow discoloration of the skin?**

Candidate: Carotenemia from excessive consumption of carotene-rich foods such as carrots and sweet potatoes. Typically carotenemia starts in the palms and soles and then spreads to the rest of the skin. Unlike jaundice the sclera are spared.

Examiner: **When do you see clinical jaundice?**

Candidate: Jaundice, is usually recognizable when serum bilirubin levels exceed 2.5 or 3.0 mg/dL.

Examiner: **How do you differentiate obstructive (cholestatic) jaundice from hepatic or hemolytic causes?**

Candidate:

- Pale stool (clay colored) and dark urine
- Severe itching **(Figure 3.9)**
- Dark (green) jaundice
- Conjugated hyperbilirubinemia
- Predominant elevation of alkaline phosphatase.

Examiner: Why does the stool become pale and urine dark in obstructive jaundice?

Candidate: In normal situations, bilirubin is excreted from the bile into the intestine and is converted by intestinal bacteria into urobilinogen and stercobilinogen. The stercobilinogen is responsible for the color of the stool and the urobilinogen for the color of urine. In obstructive jaundice the bilirubin does not reach the intestine and therefore no stercobilinogen is formed in the intestine leading to pale stool and no urobilinogen in the urine. Conjugated bilirubin that does not reach the intestine due to biliary obstruction goes back into the circulation leading to conjugated hyperbilirubinemia and goes to the kidneys leading to dark urine. This is the reason why we find decreased urobilinogen and increased bilirubin in the urine of patients with obstructive jaundice.

Examiner: **What are the causes of jaundice?**

Candidate:

- *Pre-hepatic (unconjugated, hemolytic):* Sickle cell anemia, spherocytosis, thalassemia, malaria, G6PD splenomegaly, Gilbert's syndrome
- *Hepatic:*
  - *Hepatitis:* Viral, alcohol, autoimmune, drugs, infections, toxins, etc.
  - Cirrhosis
  - CHF
- *Post-hepatic (cholestatic):*
  Extra-hepatic cholestasis:
  - Gallbladder and biliary stones
  - *Tumors:* Cholangiocarcinoma—head of pancreas
  - Sclerosing cholangitis
  - Ascariasis
- *Intrahepatic cholestasis:*
  - Primary biliary cirrhosis
  - Alcohol
  - Drugs

- Sepsis
- NASH
- *Infiltrative diseases:* Sarcoidosis, TB, amyloidosis
- Dubin-Johnson syndrome

Examiner: **How would you differentiate Dubin-Johnson syndrome from other causes of conjugated hyperbilirubinemia?**

Candidate: Dubin-Johnson syndrome is an autosomal recessive disorder characterized by conjugated hyperbilirubinemia with normal liver enzymes and liver function tests. The jaundice is usually mild, and not associated with itching.

Examiner: **What clinical features suggest that the cause of jaundice is unlikely due to CLD?**

Candidate:

- Presence of hepatomegaly
- Absence of splenomegaly and ascites (portal hypertension)
- Absence of stigmata of CLD.

Examiner: **How helpful is ultrasound of the abdomen in investigating obstructive jaundice?**

Candidate: It is a simple and non-invasive investigation, which will identify a dilated CBD suggesting extra-hepatic biliary obstruction from biliary stones, cholangiocarcinoma or cancer of the head of the pancreas.

Examiner: **How would you manage this patient?**

Candidate:

- Take a proper history regarding alcohol consumption, drug ingestion particularly paracetamol, IV drug abuse, travel history, sexual history, history of surgery, etc.
- *Liver function tests:* Total, direct and indirect bilirubin, INR, AST, ALT and ALP
- Serum paracetamol level
- US abdomen
- Viral hepatitis serology
- *Autoimmune work up:* AMA, ANA, ASMA, LKM for PBC and autoimmune hepatitis
- Iron, transferrin and ferritin for hemochromatosis
- Serum ceruloplasmin level, 24 hours urinary copper excretion for Wilson's disease
- Slit lamp examination for KF rings for Wilson's disease
- Alpha-1 antitrypsin level
- Magnetic resonance cholangiopancreatography (MRCP), endoscopic retrograde cholangiopancreatography (ERCP), magnetic resonance imaging (MRI)
- Cholestyramine for itching
- Treat the underlying cause.

THALASSEMIA MAJOR

Important Clues

- The typical patient with thalassemia major in clinical exams is a patient with jaundice, hepatosplenomegaly and a classic thalassemic face.
- Hemolytic anemia should always be considered in the differential diagnosis of jaundice with hepatosplenomegaly
- Thalassemic facies are an important clue to the diagnosis in the exam and are easily missed by candidates
- Scars in the abdomen of a patient with thalassemia are either due to splenectomy as a treatment for thalassemia or cholecystectomy due to bilirubin stones (it may be a laparoscopy scar)
- Heart disease secondary to iron overload is the most important cause of mortality in these patients.

Common Pitfalls

- Candidates frequently miss the typical thalassemic face of the patient
- Forget hemolytic anemia as a differential diagnosis of a patient with jaundice and hepatosplenomegaly.

Examiner Instructions

- Examine this patient's abdomen
- Look at this patient's face and proceed accordingly
- What do you think is the cause of this patient's jaundice?

Examiner: **What is the basic mechanism of beta-thalassemia?**

Candidate: Beta-thalassemia is an autosomal recessive disease characterized by defective synthesis of the beta chain in the Hb due to a genetic mutation in chromosome 11.

Examiner: **What are the complications of beta-thalassemia major?**

Candidate:

- Anemia
- Iron overload from repeated blood transfusion
- Bone marrow expansion from ineffective erythropoiesis and erythroid hyperplasia leading to skeletal deformities
- Gallbladder stones (bilirubin stones) from hemolysis
- Heart failure secondary to iron overload and anemia
- Hepatic fibrosis and CLD from iron overload
- Endocrine abnormalities from iron overload—diabetes, osteoporosis, hypothyroidism and hypogonadism
- Growth retardation
- Increased risk of infections due to repeated blood transfusion.

Examiner: **Why does this patient have a midline scar in the abdomen?**

Candidate: Most likely he has undergone splenectomy

Examiner: **What are the indications of splenectomy in thalassemia major?**

Candidate: Nowadays splenectomy is rarely used in the treatment of thalassemia major. This is because of the improvement in the transfusion management of thalassemia and the risk of infection and complications from splenectomy. Splenectomy is reserved for patients in whom transfusion requirement exceeds 250 mL/kg/year and in those with severe hypersplenism.

Examiner: **What is the most common cause of mortality in thalassemia major patients?**

Candidate: Cardiac complication of iron overload leading to heart failure and arrhythmia.

Examiner: **How would you manage this patient?**

Candidate:

- Request CBC, hemoglobin electrophoresis, LFT
- Genetic and family counseling and screening
- US abdomen
- Echocardiography and ECG
- Repeated blood transfusion and use of iron chelating agents are the mainstay therapy for thalassemia major
- Bone marrow transplantation can cure the disease.

Examiner: **When would you start long-term blood transfusion therapy in thalassemia major?**

Candidate:

- Hb level <7 g/dL on two successive occasions
- Patient growth or activity affected by the disease
- Skeletal abnormalities from bone marrow expansion
- Development of organ failure such as cardiac failure, edema
- Hb between 7 g/dL and 10 g/dL with any of the above clinical features.

Examiner: **What is your target Hb when giving a blood transfusion?**

Candidate: Blood transfusion may be given every 2–5 weeks to maintain pre-transfusion Hb >9 g/dL but post-transfusion Hb should not exceed 12 g/dL.

ADULT POLYCYSTIC KIDNEY DISEASE (APKD)

Common Pitfalls

- Misdiagnosing PKD as hepatosplenomegaly
- Failure to observe AV fistula
- Failure to feel a kidney transplant

Examiner Instructions

- Could you examine the abdomen of this patient with loin pain and tell me what caused his pain?
- Examine this patient's abdomen.

Examiner: **How do you differentiate an enlarged kidney from splenomegaly?**

Candidate:

- Unable to get above the swelling in case of splenomegaly (no space between the spleen and the costal margin)
- Kidney is ballotable
- Presence of splenic notch
- Percussion note is dull above splenomegaly and resonant above enlarged kidney due to the presence of the colon
- Both move with respiration.

Examiner: **What are the possible causes of this patient's loin pain?**

Candidate:

- Cyst hemorrhage
- Nephrolithiasis
- Urinary tract infection (UTI).

Examiner: **What is the mode of inheritance and the diagnostic criteria for APKD?**

Candidate:

Mode of inheritance is AD and the diagnostic criteria are:

- At least 2 cysts in 1 kidney or 1 cyst in each kidney in an at-risk patient younger than 30 years
- At least 2 cysts in each kidney in an at-risk patient aged 30–59 years
- At least 4 cysts in each kidney in an at-risk patient aged 60 years or older.

Examiner: **What are the indications for MRA screening for intracranial aneurysms in patients with APKD?**

Candidate:

- Family history of intracranial aneurysms or hemorrhage
- Symptoms suggesting an intracranial aneurysm
- High-risk occupations such as bus drivers and air pilots
- Before major elective surgeries
- Prior to anticoagulation

Examiner: **What are the complications of APKD?**

Candidate:

- Cyst hemorrhage
- Recurrent UTI
- Nephrolithiasis
- Renal cancer

- Renal failure
- Extra-renal manifestations

Examiner: **What are the extra-renal manifestations of APKD?**

Candidate:
- Cysts in the liver (up to 70%)
- Intracranial aneurysm (5–10%)
- Colonic diverticulosis
- Abdominal wall hernias
- Mitral valve prolapse

Examiner: **What is the risk of developing ESRD in APKD?**

Candidate: Risk increases with age and occurs in 50–75%, by 75 years of age

Examiner: **How would you manage this patient?**

Candidate:
- CBC (increased hematocrit), urine analysis, urea and electrolytes and US abdomen
- *Control hypertension:* ACEI and ARB are the drugs of choice because of increase renin-angiotensin system activity in these patients
- Manage renal failure if present
- Treat UTI
- Family screening and counseling

Examiner: **What are the indications for surgery in APKD?**

Candidate:
- Hemorrhage into cyst
- Very large kidneys
- Development of renal cancer.

RENAL TRANSPLANT

Important Clues

Make sure you do not miss features of associated Cushing's syndrome from long-term exogenous steroid use in patients with a renal transplant. This is a common association that is easily missed by candidates.

Common Pitfalls

- Failure to observe AV fistula or PD scar
- Failure to observe steroid and cyclosporine side effects (Cushing's features, hirsutism or gingival hyperplasia)
- Failure to observe hearing aid in Alport syndrome
- Failure to recognize coexistent APKD or SLE signs
- Failure to expose enough to see a scar in the RIF

Examiner Instructions

- Feel this patient's abdomen
- Examine this patient's abdomen.

Examiner: **What are the common reasons for renal transplantation?**

Candidate:

- Diabetes mellitus
- Glomerulonephritis
- Adult polycystic kidney disease (APKD)
- Hypertension

Examiner: **Which diseases tend to recur in transplanted kidney?**

Candidate:

- IgA nephropathy
- Mesangiocapillary glomerulonephritis
- Focal segmental glomerulosclerosis (FSGS)
- Hereditary oxalosis
- Membranous GN

Examiner: **What are the signs indicating that the graft is not functioning?**

Candidate:

- Signs of volume overload
- AV fistula or PD catheter (the presence of functioning AV fistula or PD catheter is important clue to the candidate that the graft may not be functioning. Candidates often miss that in the discussion)
- Proteinuria
- Worsening renal function
- Tenderness over the graft

Examiner: **What are the contraindications of renal transplantation?**

Candidate:

- Metastatic cancer
- Ongoing or recurring infections that are not effectively treated
- Severe cardiac or peripheral vascular disease
- Hepatic insufficiency
- Short life expectancy
- Noncompliance
- *AIDS:* Patient should have CD4 count > 200/μl for > 6 months, undetectable viral RNA, on antiretroviral > 3 months and no ongoing infection.

Examiner: **What are the complications of renal transplantation?**

Candidate:

- Infections
- Renal artery thrombosis or stenosis
- Lymphocele

- Stenosis of ureter and urine leak
- Allograft dysfunction and rejection
- Drug side effects
- *Increased risk of malignancy:* Lymphomas and Kaposi sarcoma

Examiner: **What are the types of transplant rejection?**

Candidate:

- *Hyperacute rejection:* Occurs within hours of the transplant (treatment is nephrectomy)
- *Acute rejection:* Occurs within the first 6 months after transplantation
- *Chronic rejection:* Occurs more than 1 year after transplantation.

Examiner: **What factors determine prognosis/success in renal transplantation?**

Candidate:

- Type of transplant (HLA matched from live related donors carries the best prognosis)
- Age of the donor and recipient
- Serum creatinine and glomerular filtration rate (GFR) after transplantation
- Number of acute rejection episodes
- Kidney preservation methods (prolonged preservation carries worse prognosis)
- Reason for transplantation (diseases that tend to recur).

Examiner: **What did you notice about this patient that tells you why he has had a renal transplant?**

Candidate: The patient is using a hearing aid and this suggests that the cause might be Alport syndrome.

Examiner: **What is the mode of inheritance in Alport syndrome?**

Candidate: The most common mode of inheritance in Alport syndrome is X-linked dominant and rarely autosomal recessive or dominant

Examiner: **What is the basic abnormality in Alport syndrome?**

Candidate: Defective synthesis of type IV collagen that is responsible for the synthesis of basement membrane (basement membrane is present in kidneys, eyes and inner ear).

Examiner: **How would you manage this patient?**

Candidate:

- Immunosuppressant medications
- Regular monitoring of kidney function
- Osteoporosis prophylaxis
- Vaccination
- Patient education and counseling
- Family history and counseling in case of Alport syndrome

HEPATOSPLENOMEGALY

Common Pitfalls

- Failure to feel the liver because of improper technique
- Misdiagnosing hepatosplenomegaly as PKD
- Mistaking the contracted rectus abdominis as organomegaly
- Failure to recognize stigmata of CLD, particularly spider nevi over the shoulders.

Examiner Instructions

- Examine this patient's abdomen
- Feel this patient's abdomen
- Examine the gastrointestinal system

Common causes of hepatosplenomegaly in clinical exams:

- *Myeloproliferative disorders:*
  - Chronic myeloid leukemia
  - Myelofibrosis
  - Polycythemia vera
- *Lymphoproliferative disorders:*
  - Lymphoma
  - Chronic lymphocytic leukemia
- *Portal hypertension (From liver cirrhosis or Budd-Chiari)*
- *Do not forget hemolytic diseases such as thalassemia*

Other causes:

- *Infections:*
  - *Viral:* EBV, CMV, HBV, HCV, HIV, etc.
  - Brucellosis
  - Malaria
  - Leishmaniasis
  - Tuberculosis
- *Autoimmune:* Systemic lupus erythematosus (SLE), rheumatoid arthritis (RA)
- Sarcoidosis
- *Metabolic:* Gaucher's disease and amyloidosis
- *Hemolytic anemia's:* Autoimmune, thalassemia and hemoglobinopathies

MASSIVE SPLENOMEGALY

Causes of massive splenomegaly:

- Chronic myeloid leukemia
- Myelofibrosis
- Visceral Leishmaniasis (kala-azar)
- Malaria

HEPATOMEGALY WITHOUT SPLENOMEGALY

Causes of isolated hepatomegaly:

- *Infections:* CMV, EBV, viral hepatitis, brucellosis
- *Malignancy:* Hepatocellular carcinoma or metastases
- Autoimmune hepatitis and SLE
- Alcoholism
- Non-alcoholic fatty liver disease (NAFLD)
- Hemochromatosis
- Sarcoidosis
- Congestive heart failure.

Examiner: **What are the weight, location and size of the spleen?**

Candidate: The weight of the spleen is 150 grams, length 11 cm and it is located between the 9th and 11th ribs.

Examiner: **How much does the spleen need to be enlarged to be palpable?**

Candidate: 3 times its size or more.

Examiner: **How would you define massive splenomegaly?**

Candidate: When the size of the spleen > 1000 g or > 20 cm

Examiner: **What are the functions of the spleen?**

Candidate:

- Plays a part in the immune system (produces immunoglobulin's and lymphocytes)
- Removal of old RBCs
- Removal of bacteria

Examiner: **Which types of organisms commonly cause infections in a splenectomized patient?**

Candidate: Bacteria particularly polysaccharide encapsulated bacteria and protozoa.

Examiner: **What are the common vaccinations given to splenectomized patients?**

Candidate: Pneumococcal vaccine, Hib vaccine, meningococcal vaccine and influenza vaccine.

PRIMARY MYELOFIBROSIS

Important Clues

- Probably the most common cause of massive splenomegaly in the exam.
- The patient usually has huge splenomegaly, hepatomegaly and anemia.

- The enlarged spleen in myelofibrosis may even be appreciated by abdominal inspection (**Figure 3.10**)

Common Pitfall

Missing the presence of anemia.

Examiner Instructions

- This patient complains of fatigue, please examine his abdomen
- Examine this patient's abdomen

Examiner: **Which diseases are included under the term myeloproliferative neoplasms?**

Candidate:

- Primary myelofibrosis
- Chronic myelogenous leukemia
- Polycythemia vera
- Essential thrombocytosis
- Chronic neutrophilic leukemia
- Chronic eosinophilic leukemia
- Systemic mastocytosis

Examiner: **What are the mechanisms of anemia in myelofibrosis?**

Candidate:

- Ineffective erythropoiesis and bone marrow malfunction
- Splenomegaly
- Bleeding from thrombocytopenia

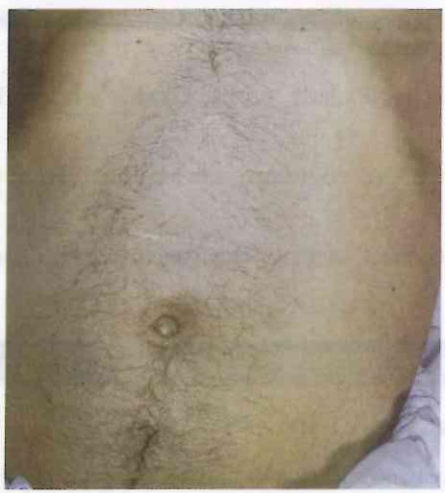

FIGURE 3.10 Massive splenomegaly in primary myelofibrosis (the enlarged spleen may be appreciated by abdominal inspection)

Examiner: **What are the complications of myelofibrosis?**

Candidate:

- Thrombotic episodes such as portal vein thrombosis
- Hyperuricemia and gout
- Anemia
- Infections
- Acute leukemic transformation

Examiner: **How would you investigate this patient?**

Candidate:

- *CBC:* Anemia with thrombocytopenia and leukopenia (initially there may be leukocytosis and thrombocytosis)
- Peripheral smear shows tear drop cells and a leukoerythroblastic picture
- High ALP, vitamin B_{12} and LDH
- JAK2 mutation
- *Bone marrow examination:* Often dry tap, increase in megakaryocytes and presence of fibrosis with a hypocelluar marrow on biopsy

Examiner: **What are the frequencies of JAK2 (Janus kinase) mutations in polycythemia vera, essential thrombocythemia and myelofibrosis?**

Candidate: The frequency is 95% in PV, 60% in ET and 60% in PMF

Examiner: **What is the prognosis of primary myelofibrosis?**

Candidate: The prognosis is generally poor with a median survival of 5–7 years after diagnosis.

Examiner: **What factors determine the prognosis in primary myelofibrosis?**

Candidate:

- Age > 65 years
- *Anemia:* Hb < 10 g/dL
- Presence of constitutional symptoms
- WBC > 25000
- Blasts of 1% or more

Examiner: **This patient has severe abdominal discomfort. What treatment may be used to reduce the size of spleen and relieve his abdominal discomfort?**

Candidate:

Treatment options used to reduce the size of the spleen include:

- *JAK2 inhibitors:* Ruxolitinib (recently the treatment of choice)
- Hydroxyurea
- *Splenic irradiation:* If drugs fail
- Splenectomy as the last option

Examiner: **What are the other treatment options for patients with primary myelofibrosis?**

Candidate:

- Correct anemia (erythropoietin)
- Aspirin to prevent thrombosis
- JAK2 inhibitors
- Hydroxy
- Stem cell transplantation.

POLYCYTHEMIA VERA

Important Clues

If you are asked to examine the abdomen of a patient with pruritus, the three main diagnoses to be suspected are primary biliary cirrhosis, polycythemia vera or cholestatic jaundice from other causes.

Polycythemia vera may present in the exam as a stroke, skin examination for generalized itching, retinal vein thrombosis or rarely portal vein thrombosis or Budd-Chiari syndrome.

Common pitfall

Failure to recognize the plethoric face and skin of the patient

Examiner Instructions

This patient complains of itching. Please examine his abdomen

Important points to consider when examining a patient with suspected PV: Observe for plethoric face, cherry red tongue, conjunctival injection, red nose, ear lobes and palmar erythema. Examine the abdomen for splenomegaly, signs of Budd-Chiari or portal hypertension and pulse oxymetry. Inform the examiner that you would like to measure the blood pressure.

Examiner: **What are the diagnostic criteria for PV?**

Candidate:

- Major criteria:
  - Hb > 18.5 g/dL in men and > 16.5 in women
  - Presence of JAK2 mutation
- Minor criteria
  - Bone marrow (BM) trilineage myeloproliferative
  - Subnormal serum erythropoietin level
  - Endogenous erythroid colony growth
- Diagnosis of polycythemia vera (PV) requires
  - 2 major criteria and one minor criterion
  - The first major and two minor criteria.

Examiner: **What might cause abdominal pain in a patient with PV?**

- Budd-Chiari syndrome
- Mesenteric thrombosis
- Peptic ulcer disease
- Splenomegaly

Examiner: **What are the complications of PV?**

Candidate:

- *Hyperviscosity symptoms:* Leading to dizziness, headache and visual blurring
- Thrombotic episodes
- Hyperuricemia and gout
- *Bleeding:* Due to abnormal platelet function
- Acute leukemic transformation

Examiner: **How would you manage this patient?**

Candidate:

- CBC
- ALP, LDH, uric acid level and B_{12} level
- Serum erythropoietin level
- JAK2 mutation
- US abdomen
- *Phlebotomy:* Keep hematocrit below 45%
- Aspirin to prevent thrombosis
- Ruxolitinib (JAK2 inhibitor)
- Hydroxyurea, interferon-alfa or busulfan.

ASCITES

Common Pitfalls

- Improper technique for performing shifting dullness
- Missing coexisting organomegaly.

Examiner Instructions

- Examine this patient's abdomen
- Examine the gastrointestinal system

Examiner: **What is the most common cause of ascites?**

Candidate: Liver cirrhosis and portal hypertension

Examiner: **What are the 5 most common causes of ascites?**

Candidate:

- Liver cirrhosis and portal hypertension
- CHF
- Malignancy
- Tuberculosis

Examiner: **Can you name some other causes of ascites?**

Candidate:

- Constrictive pericarditis
- Portal vein thrombosis
- Budd-Chiari syndrome
- Parasites (strongyloidosis)

Examiner: **What is the importance of Serum-Ascites Albumin Gradient (SAAG) in investigating the cause of ascites?**

Candidate:

- SAAG > 1.1 g/dL indicates that the cause of ascites is portal hypertension (accuracy 97%)
- SAAG < 1.1 g/dL indicates the cause is not portal hypertension. Examples of low SAAG ascites are malignant ascites and TB ascites.

Examiner: **Do you know some other conditions that may cause high SAAG ascites other than portal hypertension?**

Candidate: In general high SAAG ascites is suggestive of portal hypertension (accuracy 97%). However, occasionally high SAAG ascites may be seen in patients with CHF treated by diuretics and patients with myxedema.

Examiner: **How would you diagnose chylous ascites?**

Candidate: Chylous ascites has a milky appearance with a triglyceride level > 110 mg/dL.

Examiner: **Which conditions cause chylous ascites?**

Candidate: Chylous ascites occurs due to disruption of the lymphatics. The causes include:

- *Malignancies (the most common):* Lymphoma, pancreatic, breast, colonic, ovarian, testicular and renal malignancies
- Tuberculosis
- *Abdominal surgery:* Repair of abdominal aortic aneurysm and deep lymph node dissection
- Abdominal trauma
- Peritoneal dialysis

Examiner: **Which conditions may cause ascites without pedal edema?**

Candidate: Ascites in the absence of pitting pedal edema can be seen in:

- Malignancy
- Tuberculosis
- Constrictive pericarditis
- Sometimes cirrhosis

Examiner: **Which conditions commonly cause ascites with pitting leg edema?**

Candidate: Ascites with pitting pedal edema is usually seen in:

- Nephrotic syndrome
- Liver cirrhosis

Examiner: **How does malignancy lead to ascites?**

Candidate: The mechanisms by which malignancy can lead to ascites include:

- Lymphatic obstruction
- Peritoneal metastasis
- Metastasis to the liver and portal vein compression.

Examiner: **What is the mechanism of ascites in liver cirrhosis?**

Candidate:

- Portal hypertension leading to splanchnic vasodilatation
- Hypoalbuminemia reduces plasma oncotic pressure and leads to extravasation of fluid from the plasma to peritoneal fluid

Examiner: **What is the portal venous pressure at which ascites starts to develop?**

Candidate: > 12 mm Hg

Examiner: **What is the most sensitive clinical sign that suggests the presence of ascites?**

Candidate: Dull percussion note over the flanks (present in about 90% of cases of ascites) and shifting dullness

Examiner: **What is the most specific clinical sign for ascites?**

Candidate: Fluid thrill is the most specific sign but is less sensitive

Examiner: **In which conditions other than ascites can you illicit fluid thrill or wave sign?**

Candidate:

- Tense ascites
- Large ovarian cysts
- Large hydrated cysts
- Pregnancy with polyhydramnios

Examiner: **How much fluid should be present in the peritoneal space to be able to detect shifting dullness?**

Candidate: >500 mL

Examiner: **Do you know any other technique to detect ascites clinically if the fluid is less than 500 mL?**

Candidate: The puddle sign (may be present when the amount of ascites is as little as 120 mL) but this sign has a low sensitivity

Examiner: **What is the meaning of puddle and how would you perform the puddle sign?**

Candidate: A puddle is a small collection of water on the ground after rain. Put the patients in a prone position for few minutes, and then ask him/her to raise themselves on his/her elbows and knees. Start percussion over the flanks and then over the most dependent part of the abdomen (center of the abdomen). The puddle sign is said to be positive if you can hear a resonant note over the flanks and a dull note over the dependent part of the abdomen (the puddle).

***Examiner:* What clinical features may suggest that the cause of ascites is malignancy rather than liver cirrhosis?**

Candidate:

- Presence of abdominal pain
- Absence of pedal edema
- Absence of stigmata of chronic liver disease (**Figure 3.11**)
- Presence of supra-clavicular lymph node enlargement
- SAAG < 1.1 g/dL

***Examiner:* How would you diagnose SBP?**

Candidate: Ascitic fluid polymorphonuclear leukocyte count > 250 cells/mm^3.

***Examiner:* Which organisms commonly cause SBP?**

Candidate: The most common bacteria causing SBP are gram-negative bacteria such as *Escherichia coli* and *Klebsiella pneumoniae* and some gram-positive bacteria such as *Streptococcus pneumoniae*. Culture negative SBP is seen in up to 50% of cases.

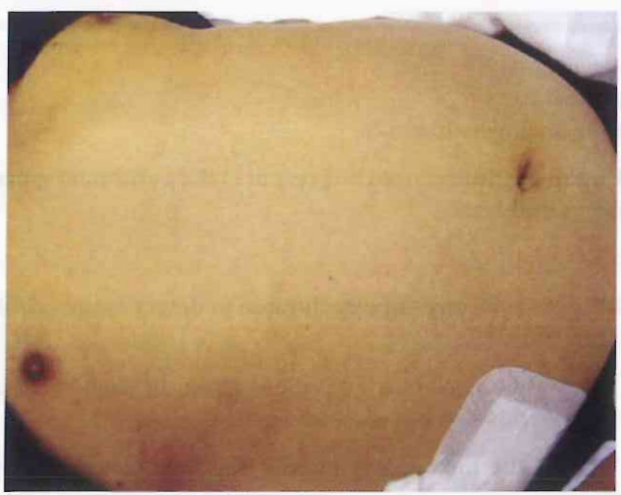

FIGURE 3.11 Ascites from CLD. Observe the yellow skin and scarce hair. Candidates may miss paracentesis marks

Examiner: **What are the most reliable signs that determine the prognosis of cirrhotic patients with ascites?**

Candidate:

- Presence of hyponatremia
- Low arterial pressure
- Increased serum creatinine
- Low urine sodium

Examiner: **How would you manage this patient?**

Candidate:

- Obtain a proper history to determine the etiology such as liver disease, blood transfusion, sexual history, drug abuse, old TB, malignancy, etc.
- Ultrasound of the abdomen
- CBC, LFT, urea and electrolytes and urine protein
- *Paracentesis:* Look for appearance, cell count, protein, albumin, glucose, SAAG, gram stain and culture, cytology, triglyceride and bilirubin level.
- Treat underlying cause
- Salt restriction 80–120 mmol/day
- Spironolactone combined with furosemide
- Therapeutic paracentesis with albumin replacement
- TIPS and liver transplantation in refractory ascites due to liver cirrhosis
- Prophylaxis for SBP in cirrhotic ascites.

Examiner: **What are the indications to start prophylactic antibiotics in patients with cirrhotic ascites?**

Candidate:

- Any patient who has recovered from an episode of SBP should receive long-term antibiotic prophylaxis
- If the ascites protein level is low < 15 g/L

Examiner: **What is the preferred antibiotic used for SBP prophylaxis?**

Candidate: Norfloxacin

Examiner: **Which drugs should be avoided in ascites due to liver cirrhosis?**

Candidate:

- *NSAIDs:* Cause renal failure and hyponatremia
- *ACE inhibitors:* Cause hypotension and renal failure
- *Alpha blockers:* Prazosin
- Dipyridamole
- Aminoglycosides

LIVER TRANSPLANT

Important Clues

- The presence of a Mercedes-Benz incision scar in the abdomen is suggestive of liver transplantation **(Figure 3.6)**
- In the initial few weeks after transplantation or if there are biliary complications from liver transplantation, you may see also a T-tube for biliary drainage
- Hepatomegaly and signs of CLD may be present

Common Pitfalls

- Some candidates, particularly from countries with no facilities for liver transplantation have not seen the Mercedes- Benz scar before the exam
- Some candidates may not know the T-tube and think it is an Ascitic drain.

Examiner Instructions

- Examine this patient's abdomen
- Why does this patient have such a big scar in his abdomen?

Examiner: **What are the indications of liver transplantation?**

Candidate:

- Acute liver failure from conditions like hepatitis A or B, drugs and toxins
- End stage CLD from any cause
- *Malignancies:* Primary such as hepatocellular carcinoma and cholangiocarcinoma and secondary from carcinoid or islet cell tumors
- *Miscellaneous:* Polycystic liver disease and Budd-Chiari syndrome

Examiner: **Among these indications, which are the most common?**

Candidate: Hepatitis C and alcohol induced liver diseases are the most common indications for liver transplantation.

Examiner: **What are the absolute and relative contraindications for liver transplantation?**

Candidate:

- *Absolute contraindications:*
  - Active extra hepatic malignancy
  - Hepatic malignancy with macrovascular or diffuse tumor invasion
  - Uncontrolled infection
  - Active substance or alcohol abuse
  - Severe comorbid conditions
  - Non-compliance or insufficient motivation
  - Technical impediment
  - Brain death

- *Relative contraindications:*
  - Advanced age
  - HIV infection
  - Cholangiocarcinoma
  - Portal vein thrombosis
  - Psychosocial problems

Examiner: **Why is HIV not currently considered an absolute contraindication?**

Candidate: Because of the availability of effective highly active anti-retroviral medications.

Examiner: **What is the prognosis of liver transplantation?**

Candidate: The prognosis is good as the 10 year survival exceeds 70%.

Examiner: **Which indications for liver transplantation carry the most favorable prognosis?**

Candidate: Primary biliary cirrhosis and autoimmune hepatitis.

Examiner: **Which two indications carry the worst prognosis and why?**

Candidate: Malignancies and hepatitis C virus due to a tendency for the primary disease to recur in the transplanted liver.

Examiner: **What are the complications associated with liver transplantation?**

Candidate:

- Acute rejection
- Increase risk of infections
- Biliary strictures and leak
- Hepatic artery thrombosis
- Portal vein thrombosis
- Malignancies
- Cardiovascular disease
- Renal failure
- Side effects of immunosuppressive medications
- Recurrence of disease in the transplanted liver

Examiner: **Why is this patient jaundiced? What causes jaundice in a patient with a liver transplant?**

Candidate: Important causes of jaundice in a liver transplant patient include:

- Development of biliary stricture
- Graft rejection
- Portal vein or hepatic artery thrombosis
- Recurrence of disease (HCV and neoplasms)
- Drug induced

Examiner: **What are the causes of mortality in liver transplant patients?**

Candidate: Early in the course after transplantation infections are the most common cause, but later cardiovascular disease, renal failure and malignancies.

Examiner: **Why are patients with a liver transplant at risk of cardiovascular disease?**

Candidate: Because of the increased risk of development of metabolic syndrome, diabetes, hypertension and hyperlipidemia.

Examiner: **Which malignancies is a liver transplant patient at risk of?**

Candidate: Most common are the skin malignancies followed by lymphoma, kidney and others.

Examiner: **What is peculiar regarding graft rejection and use of immunosuppressive medication for liver transplantation compared to other solid organs?**

Candidate: In some patients with a liver transplant a tolerance of the graft to the recipient immune system may develop and graft rejection may not occur even if immunosuppressive medications are discontinued

Examiner: **How would you manage this patient?**

Candidate:

- Immunosuppression
- Care of hygiene and diet
- Monitor regularly LFT, RFT, blood sugar and blood pressure
- Osteoporosis prevention and treatment
- Treat complications
- Vaccination.

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4

Neurology Cases

HOW TO PERFORM A NEUROLOGIC EXAMINATION OF THE LOWER (OR UPPER) LIMBS?

LFT-CPR-CSF

L : Look (inspection)
F : Feel (palpation)
T : Tone
C : Clonus (if hypertonia)
P : Power, Plantar
R : Reflexes
C : Coordination
S : Sensation
F : Foot walking

- Wash your hands
- Shake hands with the patient, introduce yourself and take permission
- Position the patient flat and ask to remove the lower clothes (cover the genitalia with a bed sheet)
- Ask the patient if he/she has any pain in the legs before touching his/her legs
- Start by a quick surveillance of the surroundings and the patient. Pay particular attention to the presence of a bladder catheter (suggests patient may have a neurogenic bladder), nearby walking aids, ongoing intravenous solutions such as IV immunoglobulin (may suggest Guillain-Barré syndrome or transverse myelitis) or methylprednisolone and presence of nasogastric or tracheostomy tube (may suggest the patient has bulbar involvement).
- Inspect the lower limbs. Look for the posture (external rotation of hemiplegia), presence of foot drop, inverted champagne bottle shape of the

legs (**Figure 4.1**), muscle wasting (**Figure 4.2**), fasciculations, loss of hair, diabetic foot ulcers, etc.

- Feel the lower limbs for any tenderness and tap the muscles to illicit fasciculations
- Examine the tone by passively moving different joints
- If you find hypertonia, do not forget to test for ankle and patellar clonus
- Examine the power by asking the patient to raise the lower limbs straight without and then with resistance and then examine the power of each group

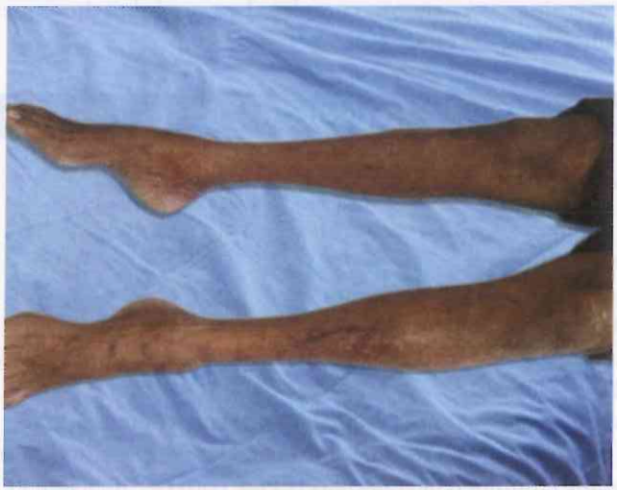

FIGURE 4.1 Charcot-Marie-Tooth disease. Observe the inverted champagne bottle shape of the left leg, foot drop and high arched foot

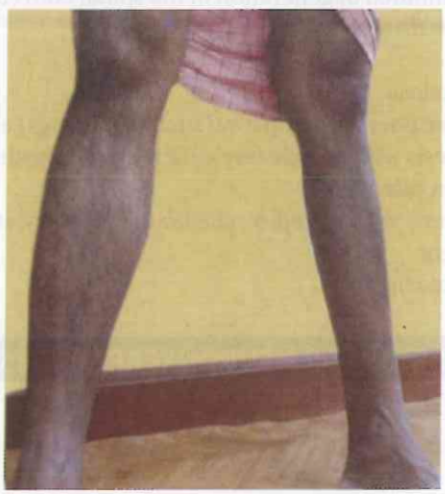

FIGURE 4.2 Asymmetry of the legs and wasting of the left leg muscles from old poliomyelitis

of muscles separately. Grade according to Medical Research council (MRC) grading system.

- Examine the reflexes. Make sure that you reinforce reflexes if they are absent by tapping them while you instruct the patient to clench his/her teeth
- Examine the coordination (heel-shin test in lower limbs or finger nose test in upper limbs).
- Examine the 5 modalities of sensation: touch, pinprick, vibration, position and temperature.
- Examine the gait. Examine also for tandem gait.
- Tell the examiner, you want to complete by examining the upper limbs and the eyes including the fundus.

COMMON LOWER LIMB NEUROLOGIC CASES

Spastic Paraparesis

- Spastic paraparesis with a sensory level
  - Multiple sclerosis
  - Spinal cord compression/lesion
  - Transverse myelitis
- Spastic paraparesis with cerebellar ataxia (impaired coordination)
  - Multiple sclerosis
  - Friedreich's ataxia (both cerebellar and sensory ataxia)
- Spastic paraparesis with sensory ataxia and loss of position sense
  - Subacute combined degeneration of the cord (B_{12} deficiency)
  - Friedreich's ataxia
  - Tabes dorsalis
- Spastic paraparesis with up going plantars and loss of ankle or knee reflexes
  - Subacute combined degeneration of the spinal cord (B_{12} deficiency)
  - Motor neuron disease
  - Syphilis
  - Friedreich's ataxia
  - Coexisting peripheral neuropathy (such as diabetes) and cord lesion
- Spastic paraparesis with muscle fasciculations and wasting
  - Motor neuron disease
- Spastic paraparesis with mixed cerebellar and sensory ataxia with pes cavus in a young patient
  - Friedreich's ataxia

FLACCID (LOWER MOTOR NEURON) PARAPARESIS

- Pure motor
- Pure sensory
- Sensori-motor

SPASTIC PARAPARESIS

Important clues in cases with spastic paraparesis

- The three main diseases that cause absent position sense in the exam or sensory ataxia are subacute combined degeneration of the cord, Friedreich's ataxia or tabes dorsalis (tabes dorsalis is very rare nowadays).
- When you find a patient with up going plantar reflexes and absent knee or ankle jerks the main differentials in the exam are Friedreich's ataxia in younger patients and B_{12} deficiency or syphilis in older patients.
- Typical findings in Friedreich's ataxia are combined cerebellar and sensory ataxia with positive finger nose test and positive Romberg's sign. In addition there is loss of position and vibration sense. Additional features that support the diagnosis are the presence of foot deformities such as pes cavus **(Figure 4.3)**.
- Candidates frequently misdiagnose Friedreich's ataxia as multiple sclerosis because both can affect young patients, cause cerebellar signs and paraparesis. The clue in the presence of sensory ataxia, absent ankle or knee jerks, absent position sense and pes cavus in Friedreich's ataxia.
- The hallmark of motor neuron disease cases in the exam is the presence of a combination of upper motor neuron signs such as spasticity, hyperreflexia and up going plantar response and lower motor neuron signs in the form of wasting and atrophy of the muscles and the presence of muscle fasciculations, claw hands or foot drop **(Figure 4.4)**. It usually occurs in male patients between 40–60 years old. (Remember motor neuron disease when you see fasciculations in the exam).
- Tabes dorsalis is very rare but examiners usually include it in the discussion of a patient with sensory ataxia. It is sometimes difficult to differentiate clinical

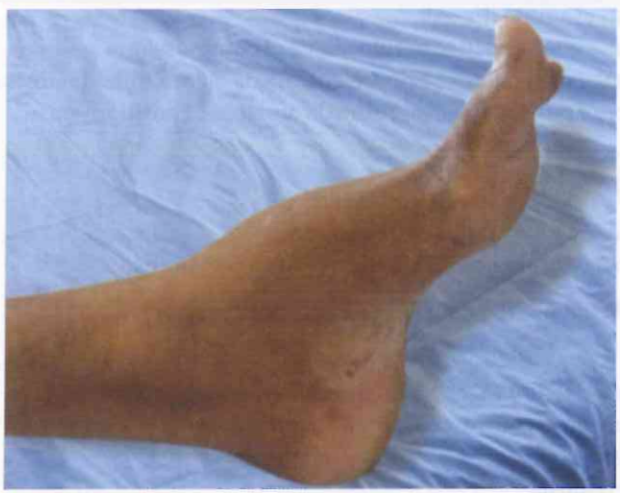

FIGURE 4.3 Pes cavus in a patient with Friedreich's ataxia

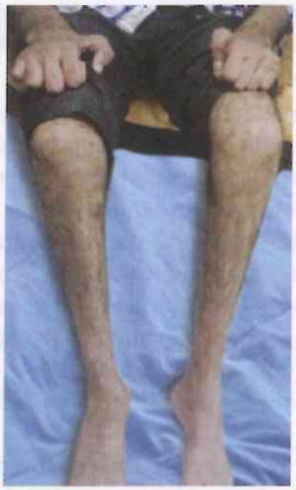

FIGURE 4.4 Severe muscle wasting of the hands and legs in a patient with motor neuron disease (observe bilateral claw hands)

subacute combined degeneration of the cord (B_{12} deficiency) from tabes dorsalis. They both can cause spastic paraparesis, absent posterior column sensation, sensory ataxia, up-going plantars with absent reflexes **(Figure 4.5)**. The main clinical features that differentiate these two diseases are the severe episodic lancinating pains, the presence of Argyll Robertson pupils (ARPs) or presence of Charcot's joint in tabes dorsalis.

Common Pitfalls

- Failure to observe the urethral catheter
- Failure to examine the gait
- It is always good practice to rule out cord compression
- Candidates misdiagnose Friedreich's ataxia as multiple sclerosis despite the presence of pes cavus, sensory ataxia and loss of position sense.

Examiner Instructions

- Examine the lower limbs of this patient
- Perform a neurologic examination of the lower limbs.

Examiner: **What are the causes of spastic paraparesis?**

Candidate:

- *Spinal cord lesion (5 Ts):* This should always be kept in mind as spinal cord compression is considered a medical emergency
  - Tumor
  - Tuberculosis

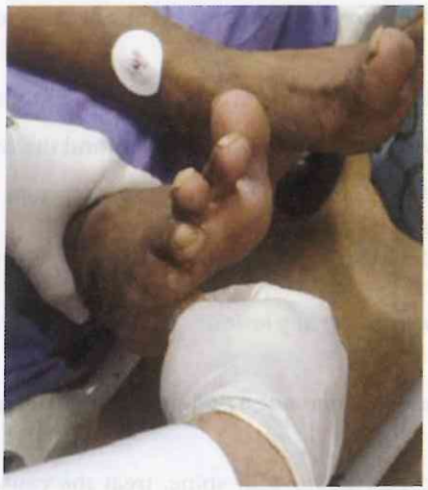

FIGURE 4.5 Typical positive Babinski sign. Observe the extension of the big toe and fanning of the other toes. If associated with loss of ankle or knee jerks, differential diagnoses becomes limited

  - *Trauma:* Due to accidents or from a disc or myelopathy
  - Transverse myelitis
  - Thrombosis (anterior spinal artery)
- Demyelinating disease (MS)
- Vitamin B_{12} deficiency
- Syringomyelia
- Hereditary spastic paraparesis (Friedreich's ataxia)
- Parasagittal meningioma
- Motor neuron disease

Examiner: **What are the causes of up-going plantars and absent ankle jerks?**

Candidate:

- Subacute combined degeneration of the spinal cord (B_{12} deficiency)
- Syphilis (Tabes dorsalis)
- Friedreich's ataxia
- Coexisting peripheral neuropathy (such as diabetic) and cord lesion
- Motor neuron disease

Examiner: **Which conditions give paraparesis and sensory ataxia (absent position sense)?**

Candidate:

- Tabes dorsalis
- Subacute combined degeneration of the spinal cord
- Friedreich's ataxia

Examiner: **What else do you want to examine in a patient with spastic paraparesis?**

Candidate: The upper limbs and the eyes including the fundus.

Examiner: **Why do you want to examine the eyes and the fundus?**

Candidate: Eye and fundus examination can give important clues to the cause of spastic paraparesis and examples are:

- *Optic atrophy:* Multiple sclerosis, B_{12} deficiency, Friedreich's ataxia and neurosyphilis
- *Papilledema:* Space occupying lesion
- *ARP:* Syphilis

Examiner: **How would you manage a patient with spastic paraparesis?**

Candidate:

- *Suspected cord compression:* MRI spine, treat the cause, DVT prophylaxis, physiotherapy and occupational therapy.
- *Suspected MS:* MRI brain/spine, lumbar puncture, visual evoked potential, IV steroids, interferon therapy and the use of new MS drugs
- *Suspected B_{12} deficiency:* B_{12} level, methylmalonic acid level, IM B_{12} injection, anti-intrinsic factor and anti-parietal cell Abs
- *Suspected tabes dorsalis:* CSF for VDRL
- *Suspected Friedreich's ataxia:* Cardiac monitoring, treat heart failure and arrhythmia, manage diabetes, physiotherapy and speech therapy, family counseling and advice to join disease societies.
- *Suspected MND:* Nerve conduction study (main), nerve or muscle biopsy, care of respiratory muscles, non-invasive ventilation, speech therapy, occupational therapy, Riluzole, join MND society, discuss with the patient regarding tracheostomy, tube feeding and DNAR.

MULTIPLE SCLEROSIS

Examiner: **Are demyelinating plaques seen in MRI specific for MS?**

Candidate: No. Demyelinating and hyperintense lesions can also be seen in other diseases such as:

- SLE
- Behçet's disease
- Syphilis
- Sjögren's syndrome
- Sarcoidosis
- Acute disseminating encephalomyelitis

Examiner: **What are the types of MS?**

Candidate:

- Relapsing remitting (most common form)

- Primary progressive
- Secondary progressive
- Progressive relapsing

Examiner: **What criteria are used in the diagnosis of multipe sclerosis (MS)?**

Candidate: The criteria used for the diagnosis of MS are the revised McDonald criteria. It requires demonstration of dissemination of the lesions in time and in space based on MRI and clinical findings

Examiner: **What are the typical areas in the central nervous system (CNS) where MS lesions are seen?**

Candidate: Periventricular, juxtacortical, infratentorial and spinal cord

Examiner: **Is CSF analysis mandatory in the diagnosis of MS?**

Candidate: The diagnosis of MS depends mainly on clinical and MRI findings. However, positive findings in the cerebrospinal fluid (CSF) of elevated IgG or 2 or more oligoclonal bands can be important to support the presence of inflammation, to exclude other diagnosis and to predict clinically definite MS.

Examiner: **What are the ocular complications of MS?**

Candidate:

Optic neuritis and retrobulbar optic neuritis

- Internuclear ophthalmoplegia (**Figure 4.6**)
- Nystagmus

Examiner: **What is the difference between optic neuritis and retrobulbar neuritis?**

Candidate: Optic neuritis (or papillitis) means inflammation of the optic disc head whereas retrobulbar neuritis implies inflammation of the posterior portion of the optic nerve.

Examiner: **How does optic neuritis manifest?**

Candidate:

- Sudden visual loss
- Pain on moving the eyes
- In retrobulbar neuritis the examination of the fundus is usually unremarkable (patient sees nothing and doctor sees nothing)
- Usually unilateral in adults
- Recovery is usually spontaneous.

FIGURE 4.6 Right internuclear ophthalmoplegia. The patient is attempting to look to the

Examiner: **What conditions other than MS may cause optic neuritis?**

Candidate: SLE, syphilis, sarcoidosis, Lyme disease, vitamin B_{12} deficiency, toxins and neuromyelitis optica.

Examiner: **Which differential diagnosis of MS constitutes a nightmare to neurologists, as it may be difficult to differentiate from MS?**

Candidate: Neuromyelitis optica (NMO) or Devic syndrome. This disease may present with sudden attacks of optic neuritis and spastic paraplegia. Sometimes it is very difficult to differentiate from MS. The main differentiating points are:

- Usually leads to bilateral optic neuritis (MS in adults typically unilateral)
- The attacks of optic neuritis tend to be more severe and last longer
- Spinal cord MRI shows larger lesions that extend 3 or more vertebral segments
- Presence of anti-aquaporin 4 (AQP4) antibodies
- Attacks of unexplained hiccups and vomiting.

Examiner: **What are the poor prognostic factors in MS?**

Candidate:

- Male gender
- Late age onset
- Frequent relapses
- Sphincter involvement at onset
- Motor or brainstem involvement
- Cerebellar involvement at onset.

Examiner: **What are the good prognostic factors in MS?**

Candidate:

- Female gender
- Age of onset before the age of 40 years
- Few early relapses
- Only sensory involvement at the start.

Examiner: Can you name new drugs approved for MS treatment?

Candidate: Interferon-β, glatiramer, dimethyl fumarate, fingolimod and natalizumab, alemtuzumab.

SUBACUTE COMBINED DEGENERATION OF THE CORD (B_{12} DEFICIENCY)

Examiner: **What are the causes of vitamin B_{12} deficiency?**

Candidate:

- Pernicious anemia
- Strict vegetarians
- Gastric and bariatric surgery
- Chronic atrophic gastritis

- *H. pylori* infection
- Metformin and proton pump inhibitors
- *Terminal ileum diseases:* Crohn's disease, tuberculosis (TB) and lymphoma.

Examiner: **How long do you expect for neurologic manifestations of B_{12} deficiency to improve after starting parenteral vitamin B_{12}?**

Candidate: Neurologic manifestations of vitamin B_{12} deficiency are the last to improve (usually take 3–6 months).

Examiner: **What precautions should you take when giving large doses of vitamin B_{12}?**

Candidate: Monitor potassium levels for hypokalemia.

Examiner: **If the hemoglobin level is normal, can B_{12} deficiency cause this patient's neurologic abnormalities?**

Candidate: Yes, neurologic manifestations of B_{12} deficiency can still occur in the absence of anemia.

TABES DORSALIS

Examiner: **What simple bedside signs can differentiate tabes dorsalis from subacute combined cord degeneration due to B_{12} deficiency?**

Candidate:

- Pupil examination for Argyll Robertson pupils (ARPs).
- Tabes dorsalis characteristically causes episodes of severe lancinating or lightning pain in the legs
- Presence of Charcot's joint.

Examiner: **What do you see in ARP?**

Candidate: Typically, these pupils are bilaterally small (meiotic) and fail to constrict to light but constrict to accommodation. (**ARP** can be read forward and backward as "**A**ccommodation **R**eflex **P**resent and **P**upillary **R**eflex **A**bsent").

Examiner: **Is ARP pathognomonic for neurosyphilis?**

Candidate: No, ARP can be seen in other conditions such thiamine deficiency (Wernicke encephalopathy) multiple sclerosis, neurosarcoidosis, brain tumors.

Examiner: **How long after exposure to *Treponema pallidum* does tabes dorsalis develop?**

Candidate: It takes up to 20 years for tabes dorsalis to develop after exposure to *T. pallidum*

Examiner: **How would you diagnose tabes dorsalis?**

Candidate: CSF serology—CSF VDRL if positive is very suggestive of Tabes dorsalis. CSF FTA-ABS can be used to confirm the diagnosis.

FRIEDREICH'S ATAXIA

Examiner: **What is the mode of inheritance in Friedreich's ataxia.**

Candidate: Autosomal recessive caused by a mutation of the frataxin gene on chromosome 9.

Examiner: **Which parts of the nervous system are commonly affected in Friedreich's ataxia?**

Candidate: Friedreich's ataxia causes degeneration and loss of the spinal cord and peripheral nerve myelinated fibers. The commonly affected tracts in the spinal cord are the posterior columns, corticospinal, and spinocerebellar tracts.

Examiner: **What is the prognosis in Friedreich's ataxia?**

Candidate: Friedreich's ataxia is a progressive disorder with an average life expectancy of 40–50 years.

Examiner: **What is the most common cause of death in Friedreich's ataxia?**

Candidate: Cardiomyopathy and arrhythmias.

Examiner: What are the other important complications of Friedreich's ataxia?

Candidate:

- Hypertrophic cardiomyopathy
- Optic atrophy
- Deafness
- Diabetes mellitus
- Scoliosis

MOTOR NEURON DISEASE

Examiner: **What is a motor neuron? And what is its function?**

Candidate: A motor neuron is a nerve cell with its body in the spinal cord and axons attached to the skeletal muscles. It's function is to carry electrical signals to the skeletal muscles to make them contract or relax.

Examiner: **What are the types of motor neuron disease?**

Candidate:

- *Amyotrophic lateral sclerosis:* The most common type is characterized by a combination of upper and lower motor neuron signs. Spastic paraparesis (or tetraparesis) with up going plantar response and hyperreflexia mixed with muscle fasciculation, wasting, claw hands or foot drop. Average life expectancy is 2–5 years
- *Progressive bulbar palsy:* Average life expectancy 6 months to 3 years

- *Progressive muscular atrophy:* Characterized by mainly lower motor neuron type of weakness with fasciculation and muscle atrophy. Patients may live beyond 5 years
- *Primary lateral sclerosis:* Life expectancy may be normal.

Examiner: **Where do you commonly see fasciculation in motor neuron disease?**

Candidate: Tongue and the limb muscles particularly deltoid.

Examiner: **What is the most common cause of death in MND?**

Candidate: Respiratory failure from respiratory muscle weakness and bulbar palsy.

Examiner: **How do you diagnose MND?**

Candidate: The diagnosis of MND requires the presence of combined upper motor neuron (UMN) and lower motor neuron (LMN) weakness along with characteristic electrophysiologic features on nerve conduction and EMG studies after excluding other causes.

Examiner: **What neurologic signs are not consistent with MND? (What are the 5 No's in MND?)**

Candidate: The 5 No's of MND are:

1. No sensory symptoms
2. No sphincteric involvement
3. No autonomic dysfunction
4. No cerebellar signs
5. No ophthalmoplegia

Examiner: **Which disease is considered the main differential diagnosis of MND? And how would you differentiate it from MND?**

Candidate: Cervical myelopathy from compression by a prolapsed disc or others causes. Cervical myopathy may also cause UMN signs in the lower limbs and LMN signs in the upper limbs at the level of the compression. In cervical myelopathy the LMN findings are seen only at the level of the compressed nerve roots while in MND they are seen in multiple areas of the body. Cervical myelopathy causes dermatomal sensory loss at the level of compression. Cervical myelopathy can cause the inverted supinator reflex sign. Cervical myelopathy can cause neck pain. Presence of tongue fasciculation favors MND. MRI of spinal cord is diagnostic in cervical myelopathy.

Examiner: **What do you mean by inverted supinator reflex sign in cervical myelopathy?**

Candidate: Inverted supinator reflex sign is seen when there is compression of cervical cord at C5–C6 level which is a common site for cervical myelopathy from disc compression. Normally when we illicit the supinator reflex it leads to flexion at the elbow. In inverted supinator reflex, this does not happen but rather finger

flexion occurs. In addition there will be hyperreflexia in the reflexes below C5-C6, which is the triceps reflex (C7).

PERIPHERAL NEUROPATHY

Common Pitfalls

- Misdiagnosing peripheral neuropathy and lower motor signs as spastic paraparesis
- Missing the clinical signs of Charcot-Marie-Tooth **(Figure 4.1)**
- Failure to mention nerve conduction studies in the investigations.

Examiner Instructions

- Examine the lower limbs of this patient
- Perform neurologic examination of the lower limbs.

The causes of peripheral neuropathy can be memorized as:

A: Autoimmune vasculitis
B : B_1, B_6, B_{12} deficiency
C : Cancer
D : Diabetes/Drugs
E : Ethanol
F : Failure (Renal and liver)
G : Guillain Barré
H : Hereditary sensorimotor (CMT)
I : Infections (Lyme, HIV, hepatitis, leprosy)

Examiner: **What are the causes of peripheral neuropathy?**

Candidate:

Causes of peripheral neuropathy include:

- Diabetes mellitus **(Figure 4.7)**
- Alcoholism
- Guillain Barré syndrome and chronic inflammatory demyelinating polyneuropathy (CIDP)
- Charcot-Marie-Tooth (Hereditary sensory motor neuropathy)
- *Nerve injuries:* Needle injection and trauma
- *Malignancies:* Multiple myeloma, paraneoplastic
- *Infections:* Lyme disease, hepatitis C, leprosy **(Figure 4.8)**, diphtheria and HIV
- *Vasculitis and CT diseases:* Lupus, rheumatoid arthritis, polyarteritis nodosa (PAN) **(Figure 4.9)**
- *Drugs:* INH, vincristine, metronidazole, nitrofurantoin
- Chronic liver and kidney diseases
- *Vitamin deficiency:* B_1, B_6, B_{12}, E

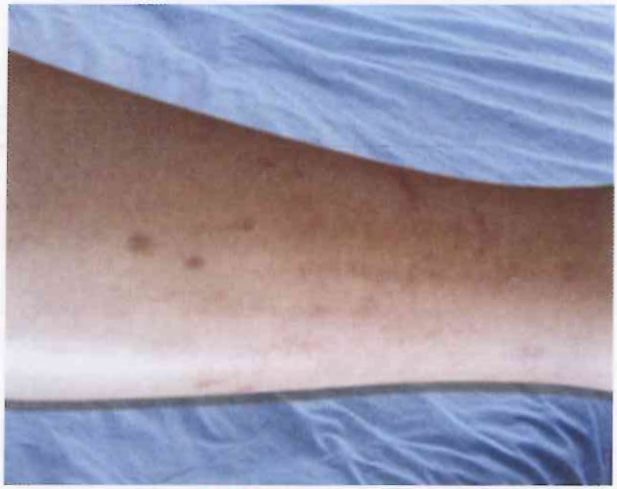

FIGURE 4.7 The presence of diabetic dermopathy may suggest diabetes as the cause of peripheral neuropathy

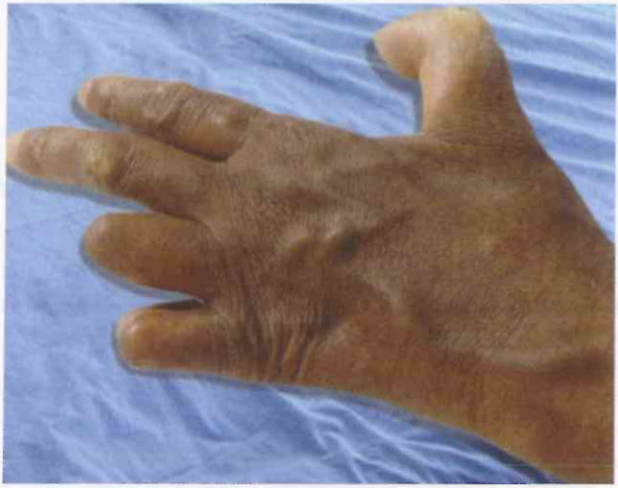

FIGURE 4.8 Typical claw hand due to peripheral neuropathy in leprosy. Observe the multiple ulcerations of the fingers

Examiner: **Which conditions cause predominantly sensory neuropathy?**

Candidate:

- Diabetes
- *Drugs:* INH, metronidazole
- Alcohol (thiamine deficiency)
- Chronic kidney disease/chronic liver disease

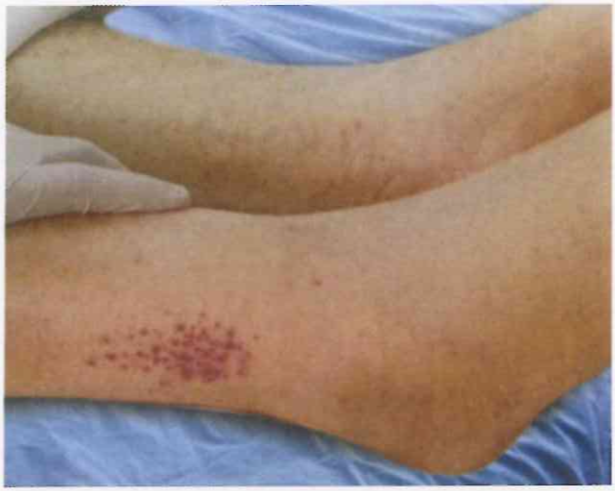

FIGURE 4.9 Presence of a vasculitic rash suggests the diagnosis of vasculitis as the cause of this patient's severe numbness and right foot drop

- Paraneoplastic (Lung/ovarian cancer)
- *Malignancy:* Multiple myeloma
- Vasculitis (PAN, SLE)
- Leprosy
- HIV

Examiner: **Which conditions cause predominantly motor neuropathy?**

Candidate:

- Guillain-Barré syndrome and chronic inflammatory demyelinating polyneuropathy (CIDP)
- Charcot-Marie-Tooth **(see Figure 4.1)**
- Poliomyelitis **(see Figure 4.2)**
- Porphyria
- Lead poisoning
- Dapsone

Examiner: **Which conditions cause sensori-motor neuropathy?**

Candidate:

- Alcohol
- Diabetes
- Charcot-Marie-Tooth
- Drug-related neuropathy (Vincristine, Nitrofurantoin)
- Vitamin B_{12} deficiency
- Critical care polyneuropathy

Examiner: **How would you investigate this patient?**

Candidate:

- Blood sugar
- Full blood count and erythrocyte sedimentation rate (ESR)
- Liver and renal function tests
- Vitamin B_{12}
- Para protein screen
- Vasculitic profile
- Nerve conduction study
- Nerve biopsy
- Skin biopsy
- Corneal confocal microscopy

HEREDITARY SENSORI: MOTOR NEUROPATHY (CHARCOT-MARIE-TOOTH)

Common Pitfalls

See peripheral neuropathy.

Examiner: **What is your diagnosis?**

Candidate: This patient has foot drop, pes cavus, claw toes and inverted champagne bottle shape of the leg (due to wasting of the distal muscles of the leg). There is obvious wasting of muscles of the hands. He has weakness of the legs and absent touch and pin prick sensation in the legs. This patient has mixed motor and sensory neuropathy most likely CMT **(Figure 4.1)**.

Examiner: **Which type of neuropathy does Charcot-Marie-Tooth cause?**

Candidate: It causes both motor and sensory neuropathy (also named Hereditary motor and sensory neuropathy) but predominantly motor.

Examiner: **What is the mode of inheritance in CMT?**

Candidate: Mainly autosomal dominant but can be autosomal recessive or X-linked.

Examiner: **What is the genetic abnormality in CMT?**

Candidate: Duplication of the *PMP 22* gene

Examiner: **How many variants of CMT are there?**

Candidate: There are many variants but the most important and most common are CMT1 and CMT2.

Examiner: **What are the most commonly involved peripheral nerves in CMT?**

Candidate: Peroneal and ulnar nerves but can involve other peripheral nerves as well.

Examiner: **How would you manage this patient?**

Candidate:

- Nerve conduction study (demyelinating in CMT1and axonal in CMT2)
- Genetic testing and counseling
- Daily stretching exercises and referral to orthopedic if pes cavus develops
- Ascorbic acid (increase myelination)
- Avoid drugs that may worsen neuropathy (such as vincristine, Dapsone, Nitrofurantoin and Metronidazole)

GUILLAIN-BARRÉ SYNDROME

Important Clues to the Diagnosis

Presence of lower motor neuron weakness in the lower limbs with absent lower limb reflexes.

Common Pitfall

- Candidates find difficulty in diagnosing Miller Fisher variant in the exam
- Missing tracheostomy scar or bilateral facial weakness.

Examiner: **What are your findings?**

Candidate: This gentleman has lower motor neuron type of weakness in the lower limbs with hypotonia, power of grade 1/5 and loss of knee and ankle reflexes. Sensation is intact. I see the patient has a tracheostomy tube. I would suggest a diagnosis of Guillain-Barré syndrome (GBS) with respiratory muscle involvement.

Examiner: **What conditions might precipitate GBS?**

Candidate:

- Viral infections
- Campylobacter gastroenteritis
- Immunization
- Upper respiratory tract infection (URTI)
- Mycoplasma infection
- Surgery

Examiner: **What is Miller Fisher variant of GBS?**

Candidate: It comprises a triad of ataxia, areflexia and ophthalmoplegia in addition to the lower limb weakness?

Examiner: **Which test is considered very specific for the Miller Fisher variant of GBS?**

Candidate: Presence of anti-GQ1b antibodies in the CSF is very specific for Miller Fisher variant.

Examiner: **What serious complications can this patient develop?**

Candidate:

- Respiratory muscle weakness
- Autonomic neuropathy leading to hypotension, hypertension or arrhythmias.
- Bilateral facial nerve weakness.

Examiner: **What are the poor prognostic factors in GBS?**

Candidate:

- Age
- Preceding diarrhea
- Positive *C. jejuni*
- Rapid disease progression
- Severe disease indicating by GBS disability score
- Absence of preceding upper respiratory infection.

Examiner: **How would you investigate this patient?**

Candidate:

- *CSF analysis:* Characteristically shows high protein with normal cell count (protein-cellular dissociation)
- Nerve conduction studies
- *Lung function testing:* Forced vital capacity (FVC), maximal inspiratory pressure (MIP), maximal expiratory pressure (MEP).

Examiner: **How would you treat this patient?**

Candidate:

- Plasma exchange
- IV immunoglobulin
- Care of respiratory muscles
- Monitoring of blood pressure and heart rhythm.

CRANIAL NERVE PALSIES

How to Perform a Cranial Nerve Examination?

- Wash your hands
- Introduce yourself and take permission from the patient
- You may ask the patient to sit on the bed side or on a chair
- *Olfactory nerve:* Ask the patient if he/she has problems with smelling
- *Optic nerve:*
  - Visual acuity
  - Visual field
  - Pupils (reaction to light and accommodation)
  - Fundus
- *Oculomotor, trochlear and abducent:*
  - Test eye movements

  - Test pupil size and reaction
- *Trigeminal:*
  - Test sensation over the face
  - Test muscles of mastication
  - Corneal reflex
- *Facial nerve:*
  - Ask the patient to raise the eyebrows, close eyes tightly, show the teeth, blow, whistle
- *Acoustic:* Check hearing
- *Glossopharyngeal and vagus:*
  - Perform the "ah test" by asking the patient to say "ah", check the soft palate on both sides (should be at the same level) and the uvula movement and direction
  - Check the gag reflex on each side (remember there are 2 gag reflexes, one on each side). Some candidates check the gag reflexes only in the center. This is not correct
  - Check the voice for dysphonia
  - Ask the patient about choking during feeding
- *Accessory nerve:* Ask the patient to shrug the shoulder and check the muscle strength and movement.
- *Hypoglossal nerve:*
  - Look at the tongue for wasting and fasciculation's
  - Ask the patient to protrude the tongue, check for deviation to one side
  - Ask the patient to push the tongue against each cheek and compare the power on both sides.

Common Pitfalls

- Improper technique of cranial nerve examination
- Failure to complete examination within the allocated time due to inadequate prior practice.

Examiner Instructions

Examine the cranial nerves.

THIRD CRANIAL NERVE PALSY

Examiner: **What are the causes of third nerve palsy?**

Candidate:

- *Medical causes:*
  - *Ischemia (infarction) of the nerve:* Diabetes mellitus (DM), hypertension and vasculitis
  - *Basal meningitis:* TB, sarcoidosis, nasopharyngeal tumors
  - Cavernous sinus thrombosis

  - Multiple sclerosis
- *Surgical causes:*
  - Posterior communicating artery aneurysm or brain tumors compressing the nerve
  - Uncal herniation

Examiner: **How would you differentiate third nerve palsy due to nerve ischemia from that due to compression by aneurysm or tumor?**

Candidate: In ischemic causes, usually the peripherally located parasympathetic nerve fibers are spared and therefore the pupils are spared. In compressive lesions usually there is complete third nerve palsy with a dilated pupil and the condition is often painful **(Figures 4.10 and 4.11)**.

Examiner: **What are the manifestations of cavernous sinus thrombosis and which cranial nerves are involved?**

Candidate:

- Headache
- Periorbital edema
- Chemosis
- 3, 5, 6 cranial nerve palsies (ophthalmoplegia and ptosis with dilated pupil)

FIGURE 4.10 Complete ptosis due to left third nerve palsy from a compressive lesion

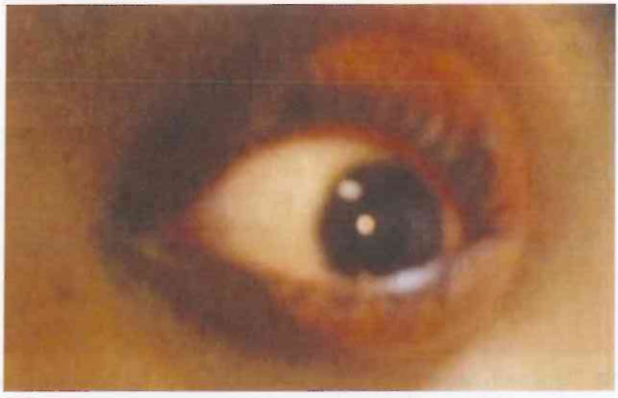

FIGURE 4.11 Same patient in Figure 4.10. Observe the left eye globe deviation to the left side due to paralysis of the left medial rectus muscle

Examiner: **How would you manage this patient?**

Candidate:

- Control DM and blood pressure if the cause is ischemic
- Magnetic resonance imaging (MRI)/magnetic resonance angiography (MRA) brain
- Neurosurgical referral if the cause is a compressive lesion.

SIXTH CRANIAL NERVE PALSY

Examiner: **What are the causes of sixth nerve palsy?**

Candidate:

- *Ischemia to the nerve:* DM, hypertension and vasculitis
- Raised intracranial pressure
- Cavernous sinus thrombosis
- Multiple sclerosis
- Brain stem meningitis/tumors

Examiner: **Why is the sixth cranial nerve affected in raised intracranial pressure? What do we call this and why?**

Candidate: The sixth cranial nerve has the longest intracranial course among all the cranial nerves. When the intracranial pressure increases, the nerve gets stretched and compressed against the petrous ligament or the ridge of the petrous temporal bone. This is called a "false localizing sign". A false localizing sign means that the pathology causes dysfunction distant or remote from the expected anatomical site of the pathology and therefore may give a false impression as to the site of origin of the pathology **(Figure 4.12)**.

Examiner: **How would you manage this patient?**

Candidate:

- Control DM and blood pressure
- MRI/MRA brain
- Treat the underlying cause

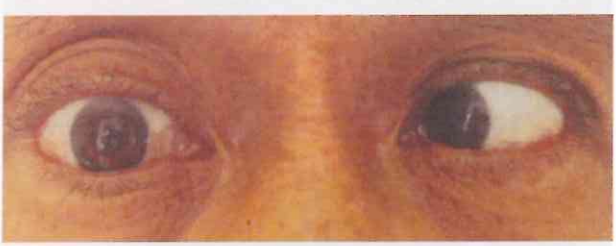

FIGURE 4.12 Right sixth nerve palsy. The patient is attempting to look to the right side. Observe the paralyzed right lateral rectus

LOWER MOTOR NEURON FACIAL PALSY

Examiner: **What are the causes of LMN facial palsy?**

Candidate:

- Bell's palsy (most common cause)
- Herpes Zoster (Ramsey-Hunt syndrome)
- Parotid tumor
- Middle ear infection
- Lesion at the nucleus (pontine infarction/tumors)
- DM

Examiner: **Which conditions can cause bilateral seventh nerve palsy?**

Candidate:

- Guillain-Barré syndrome **(Figure 4.13)**
- Sarcoidosis
- Lyme disease
- HIV
- Melkersson-Rosenthal syndrome (facial palsy, granulomatous cheilitis, and fissured tongue)

Examiner: **Why in upper motor neuron facial palsy are the forehead muscles spared?**

Candidate: Motor fibers of the facial nerve originate from the frontal lobe. Unlike the motor fibres that supply the muscles below the eyebrows, not all the motor fibers supplying the forehead muscles cross the midline to reach the facial nerve nucleus in the lower pons. Some of these fibers do not cross the midline and travel to the ipsilateral motor nucleus. Thus, muscles of the forehead receive innervation from both sides of the motor cortex, and sparing of forehead muscles in facial palsy indicates a central etiology.

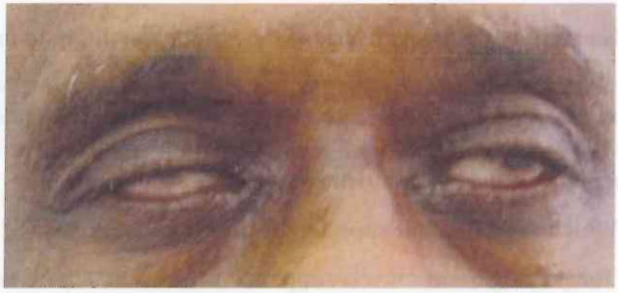

FIGURE 4.13 Bilateral Bell's phenomenon from bilateral lower motor neuron facial palsy (patient is attempting to close the eyes tightly)

Examiner: **In which part of the course of the facial nerve does Bell's palsy happen and why?**

Candidate: Bell's palsy happens in the labyrinthine part of the facial canal. This is because the canal is very narrow in this part and any swelling of the nerve may lead to nerve compression.

Examiner: **What is the cause of Bell's palsy?**

Candidate: Herpes simplex virus.

Examiner: **What is the prognosis of Bell's palsy?**

Candidate: Prognosis is good if recovery begins within 3 weeks of onset.

Examiner: **What are the complications of Bell's palsy?**

Candidate:

- Corneal dryness and abrasion
- Persistent weakness
- Synkinesis (cross innervations upon healing) leading to lacrimation when eating (crocodile tears) winking on smiling, etc.
- Contracture

Examiner: **How would you manage this patient?**

Candidate:

- Artificial tears and ointment to avoid corneal dryness and abrasion
- Eye cover
- Early initiation of steroids
- Acyclovir/Valacyclovir
- Nerve stimulation
- Botulinum toxin injection for synkinesis
- Surgery (rarely)

HYPOGLOSSAL NERVE PALSY

Examiner: **Name the extrinsic muscles of the tongue and their actions.**

Candidate: Genioglossus responsible for protrusion of the tongue, styloglossus muscle responsible for retraction and elevation of the tongue, hyoglossus muscle responsible for depression of the tongue and palatoglossus muscle responsible for elevation of the posterior part of the tongue and swallowing.

Examiner: **Which muscle is not supplied by the hypoglossal nerve?**

Candidate: The palatoglossus muscle supplied by the accessory nerve.

Examiner: **Why the tongue is deviated to the paralyzed side?**

Candidate: Due to unopposed action of the opposite genioglossus muscle **(Figure 4.14)**

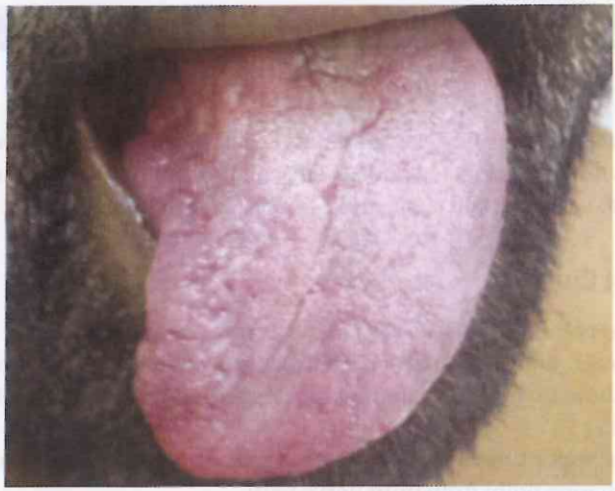

FIGURE 4.14 Right hypoglossal nerve palsy. Observe the atrophy of the right side of the tongue and tongue deviation to the right side

Examiner: **How would you differentiate between lower motor neuron and upper motor neuron lesions of the hypoglossal nerve?**

Candidate: LMN hypoglossal palsy leads to atrophy and fasciculation of the affected side whereas UMN lesion leads to spastic tongue.

Examiner: **What are the causes of hypoglossal nerve palsy?**

Candidate:

- *Lesions in the medulla*
  - Medullary infarction (Medial medullary syndrome)
  - Hemorrhage
  - Syringobulbia
  - Medullary tumor (Glioma)
  - Demyelination
  - Arnold-Chiari malformation
  - *Bulbar palsy:* Motor neuron disease
- *Lesions in the base of skull (hypoglossal canal)*
  - Metastatic carcinoma
  - Nasopharyngeal carcinoma
  - Meningioma
  - Basal meningitis (such as TB)
- *Lesions in the carotid space:* Carotid aneurysm
- *Cerebral lesions:* Ischemic stroke.

INTERNUCLEAR OPHTHALMOPLEGIA

Common Pitfalls

- Candidates diagnose INO as third nerve palsy despite vertical movements of the eyes not being affected and absence of ptosis or pupillary dilatation
- Failure to suspect brain stem stroke as the cause in older patients.

Important Clues

- The most common cause of INO in the exam is multiple sclerosis
- The typical findings are total or partial failure to adduct one eye with nystagmus in the abducting eye on lateral gaze **(Figure 4.6)**. It may be unilateral and bilateral.
- It is very important to note that convergence in INO is normal and this is a differentiating point from third nerve palsy.

Examiner Instructions

- Examine this patient's cranial nerves
- Examine the eyes of this patient.

Candidate

This patient has failure of adduction in the left eye associated with nystagmus in the right eye. Convergence of the eyes is normal. INO is the most likely diagnosis. I would like to examine her limbs and cerebellar system for evidence of multiple sclerosis

Examiner: **What are the causes of INO?**

Candidate:

- Multiple sclerosis is the most common cause, particularly in young patients
- Brain stem infarction (the second most common cause and probably the most common in older patients)
- CNS tumors
- *CNS infections:* Toxoplasmosis, encephalitis, acquired immune deficiency syndrome (AIDS)
- Head injury
- Arnold-Chiari malformation
- Wernicke's encephalopathy
- Vasculitis (SLE)
- Miller Fisher syndrome

Examiner: **What happens to eye convergence in INO?**

Candidate: Eye convergence is preserved in INO which signifies normal medial rectus muscle. The convergence is mediated by a pathway that is separate from

Examiner: **Which part of the brain is involved in INO?**

Candidate: INO is caused by injury in the MLF, a tract of nerve fibers that connect the abducens nucleus to the oculomotor nucleus and are responsible for the conjugate eye movements.

Examiner: **What makes the medial longitudinal fasciculus vulnerable in MS?**

Candidate: The MLF is a heavily myelinated nerve tract, which makes it more likely to be affected by demyelinating diseases such as MS.

Examiner: **What is one and half syndrome?**

Candidate: One and half syndrome is characterized by conjugate horizontal gaze palsy in one direction plus an internuclear ophthalmoplegia in the other.

CEREBELLAR SYNDROME

How to Examine the Cerebellar System?

- Wash your hands
- Introduce yourself to the patient and ask permission
- Perform a quick surveillance of the surroundings and the patient. Check for walking aids, urinary bladder catheter and intravenous drug infusions such as methylprednisolone that may suggest MS
- It is preferred to ask the patient to sit on the bedside rather than lying in the bed. This may be useful to unmask the presence of truncal ataxia and also allows the examination for pendular knee jerks
- Ask the patient his/her name and to repeat certain phrases like "British constitution", "west register street" and "hippopotamus" to demonstrate the presence of scanning dysarthria
- Check for nystagmus
- Check finger to nose test and dysdiadochokinesis
- Check for rebound hypotonia by asking the patient to push his/her extended arms up against your hands. Be ready to hold patient's arms before reaching his/her face
- Check pendular knee reflexes
- Check the heel to shin test
- Ask the patient to walk and be ready to prevent his/her fall and check for ataxic gait
- Check tandem gait
- Check for Romberg sign
- Tell the examiner you would normally examine the fundus for papilledema or optic atrophy.

Common Pitfalls

- Failure to perform a proper examination of the cerebellar system

Examiner Instructions

- Examine this patient's cerebellar system
- This gentleman has difficulty in walking, please examine his gait and proceed accordingly.

Candidate

This gentleman has scanning dysarthria, nystagmus, impaired finger nose test with past pointing, dysdiadochokinesis, pendular reflexes and broad based ataxic gait. Heel to shin test and tandem walking are abnormal.

Examiner: **What is the differential diagnosis?**

Candidate:

- Cerebellar stroke (hemorrhage/ischemia)
- Cerebellar tumor
- Multiple sclerosis
- Alcohol induced cerebellar degeneration
- *Drugs:* Phenytoin
- Friedreich's ataxia
- Paraneoplastic

Examiner: **What is the importance of Tandem walking in testing cerebellar function?**

Candidate: Tandem walking is a very sensitive test when cerebellar disease is mild and other cerebellar signs are negative particularly when the lesion is in the vermis of the cerebellum.

Examiner: **How would you differentiate a lesion in the vermis from that in the cerebellar hemispheres?**

Candidate: A lesion in the vermis of the cerebellum typically gives truncal ataxia. Finger to nose and heel to shin test may appear normal. The patient usually has abnormal gait particularly the tandem gait.

Examiner: **What name is given to dysarthria caused by cerebellar disease? How would you test for it?**

Candidate: In cerebellar disease the words are usually jerky and broken into syllables. To test for scanning dysarthria, the patient is asked to pronounce certain phrases that have multiple syllables such as "British constitution", "Hippopotamus", "West Register Street" and "Walking Happily"

Examiner: **How would you differentiate nystagmus due to a cerebellar cause from that due to a peripheral cause (inner ear)?**

Candidate: Nystagmus from a peripheral cause is typically horizontal and unidirectional with the fast phase being away from the affected side. Nystagmus

due to cerebellar cause may be horizontal or vertical with the fast component towards the site of the lesion.

Examiner: **How would you manage this patient?**

Candidate:

- History of drugs, epilepsy or alcohol ingestion
- MRI of the brain and posterior fossa
- Lumbar puncture and visual evoked potential if MS is suspected
- Chest X-ray if paraneoplastic syndrome is suspected.

MYASTHENIA GRAVIS

How to Examine a Case of Myasthenia Gravis?

- Wash your hands
- Introduce yourself and request permission from the patient
- Perform a quick surveillance of the surroundings and the patient. Check for non-invasive ventilation machine, nasogastric tube, intravenous drugs such as IV immunoglobulin and steroids. Inspect the patient for ptosis and myopathic face **(Figure 4.15)**
- Ask the patient about his/her name and other questions to demonstrate the dysphonia and nasal tone of speech
- Ask the patient to smile to demonstrate myasthenic sneer
- Examine the eye movement and ask the patient to inform you if they see double vision at any time.
- Test for fatigability by asking the patient to count loudly up to "50" or by elevating the eyebrows continuously for a minute or so
- Ask the patient to push his/her head against your hand to check for neck muscle power
- Examine the gag reflex and undertake the "ah test"
- Examine for proximal muscle weakness by asking the patient to stand from a squatting position or abduct and adduct shoulders against resistance
- Tell the examiner you would normally test for respiratory muscle function.

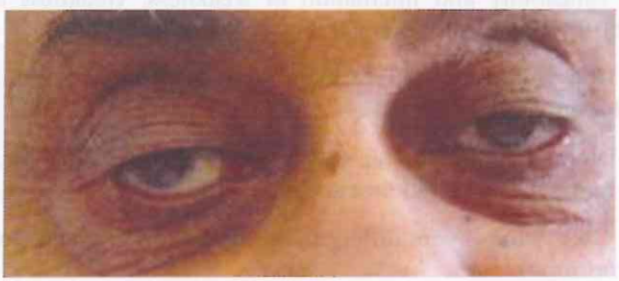

FIGURE 4.15 Bilateral ptosis in a patient with myasthenia gravis

Common Pitfalls

- Failure to recognize ptosis
- Failure to consider myasthenia when diplopia is multidirectional and cannot be explained by specific cranial nerve palsy
- Failure to test for fatigability (asking patient to maintain upward gaze to demonstrate increasing ptosis or count continuously for voice fatigue)
- Failure to examine for proximal myopathy.

Examiner Instructions

- Have a look at this patient's face and tell me the diagnosis
- Examine this patient's eyes
- This patient complains of easy fatigability, what is the cause?
- What is the cause of this patient's difficulty in breathing?

Candidate

This lady has ptosis, diplopia, dysphonia or nasal speech, dysphagia, expressionless face, myasthenic sneer, weakness of the neck muscles (dropped head syndrome) and proximal muscle weakness. She most likely suffers from myasthenia gravis.

Examiner: **What do you mean by myasthenic sneer?**

Candidate: A sneer means to smile at someone but with an expression on your face that shows a dislike. When a patient with myasthenia attempts to smile the angles of the mouth fail to move and there will be slight rise in the mid upper lip.

Examiner: **What are the types of myasthenia gravis?**

Candidate:

- *Ocular:* Weakness is limited to eyelid and extra-ocular muscles
- *Generalized:* Weakness involves ocular muscles, limbs, bulbar and respiratory muscles.

Examiner: **What symptom differentiates myasthenic weakness from other causes of weakness and fatigue?**

Candidate: Fatigability and fluctuation of weakness. Weakness and fatigue worsen in the evening or after exercise.

Examiner: **How would you differentiate diplopia of myasthenia from other causes of diplopia?**

Candidate: Diplopia in myasthenia usually cannot be explained by a particular nerve palsy and is usually multidirectional.

Examiner: **How would you differentiate myasthenia gravis from Eaton-Lambert syndrome?**

Candidate: In Eaton-Lambert syndrome:

- Symptoms are worse in the early morning,
- Leg weakness is more than arm weakness,
- There may be associated autonomic dysfunction
- Incremental pattern on EMG (decremental pattern in MG, particularly in the proximal muscles).

Examiner: **Botulism can be very difficult to differentiate from myasthenia. What clinical sign may differentiate the two?**

Candidate: Botulism typically causes fixed dilated pupils due to paralysis of the muscles of the pupils. In addition, the course is typically acute.

Examiner: **How would you differentiate oculopharyngeal dystrophy from myasthenia on physical examination?**

Candidate: Oculopharyngeal dystrophy is a genetic disease that presents in adult life with symptoms similar to myasthenia such as dysphagia, bilateral ptosis and proximal muscle weakness. It is difficult to differentiate it from myasthenia by clinical examination alone. However, the presence of tongue wasting and weakness is characteristic of oculopharyngeal dystrophy.

Examiner: **Can you name some drugs that can cause myasthenia?**

Candidate: Penicillamine, statins.

Examiner: **Can you name some drugs that may exacerbate myasthenia?**

Candidate:

- Penicillamine
- Statins
- Aminoglycosides, fluor quinolones, macrolides
- Phenytoin
- Gabapentin
- Chlorpromazine, magnesium, steroids.

Examiner: **What factors would make you consider endotracheal intubation in a myasthenic crisis?**

Candidate:

- Vital capacity < 20 mL/kg
- Signs of respiratory distress
- Inability to clear secretions
- Inability to complete sentences because of breathing difficulty
- Dyspnea, particularly on lying supine

Examiner: **What is a cholinergic crisis and how do you differentiate it from a myasthenic crisis?**

Candidate: A cholinergic crisis is due to an over dose of anticholinesterase drugs. It is very rare in patients with myasthenia gravis at the usual doses. It can lead to weakness and respiratory muscle involvement that may be difficult to differentiate

diarrhea, sweating, pinpoint pupil, bradycardia and bronchoconstriction are prominent features in a cholinergic crisis.

Examiner: **What important point should you consider during intubation of myasthenic patients?**

Candidate: Non-depolarizing muscle relaxants such as rocuronium or vecuronium may be preferred over succinylcholine during intubation of myasthenic patients. If succinylcholine is to be used then higher doses may be required because of lack of acetylcholine receptors, which may lead to prolonged paralysis.

Examiner: **How would you confirm the diagnosis of myasthenia in this patient?**

Candidate:

- *Bedside tests:* Ice pack test and Tensilon test
- EMG
- Serum anticholinesterase antibodies (sensitivity > 80%, specificity > 90% in generalized myasthenia)
- Muscle specific tyrosine kinase antibodies
- CT scan of the mediastinum for thymus hyperplasia or thymoma (present in about 70% and 10% of patients with myasthenia, respectively)

Examiner: **How would you treat this patient?**

Candidate:

- Anticholinesterase inhibitors (pyridostigmine and neostigmine) used in mild cases and as maintenance therapy
- Steroids and immunosuppressant drugs such as Azathioprine for more difficult cases
- Thymectomy for all patients with thymoma and for patients aged 10–55 years without thymoma but with generalized MG.

Treatment of myasthenic crisis

- ICU admission
- Endotracheal intubation if necessary
- IV immunoglobulin
- Plasmapheresis
- Steroids

MYOTONIC DYSTROPHY

How to Examine a Case of Myotonic Dystrophy?

- Wash your hands
- Introduce yourself to the patient and request permission for examination. Check for the presence of a non-invasive ventilation machine beside the patient
- When you shake hands with the patient when introducing yourself, observe that the patient does not release you hands easily

- Inspect the patient for a apathetic monk face, ptosis, wasting of the temporalis muscle and frontal balding
- Ask the patient to squeeze your hand with his/her hand grip and keep holding for sometime and then release it
- Illicit percussion myotonia using the tendon hammer
- Examine for proximal muscle weakness
- Tell the examiner you would normally test for respiratory muscle weakness and ask the patient about dysphagia and family history

Clues: Suspect MD when you see a 'monk' in your exam or the patient does not want to release your hand after a handshake.

Common Pitfalls

- Missing the characteristic facial appearance of MD is common
- Failure to recognize the presence of myotonia after a handshake.

Examiner Instructions

- Have a look at this gentleman, what is the diagnosis?
- This patient complains of generalized body pain, please do a general examination
- Can you shake hands with this patient and proceed accordingly?

Candidate

This gentleman has frontal balding and wasting of the temporalis and masseter muscles. He has ptosis with a monk face appearance. There is slow release of hand grip and percussion myotonia.

Examiner: **How can you illicit percussion myotonia?**

Candidate: Percussion on the thenar eminence with a tendon hammer results in prolonged adduction of the thumb.

Examiner: What are the cardinal manifestations of myotonic dystrophy?

Candidate: "the six Ds"

1. **D**ominant inheritance (AD)
2. **D**ystrophy
3. **D**isturbance of conduction *and* dilated cardiomyopathy
4. **D**iabetes mellitus
5. **D**ysphagia
6. **D**ecreased fertility

Examiner: **What is the most common cause of poor vision in patients with MD?**

Candidate: Cataract.

Examiner: **What are the two most common causes of death in patients with MD?**

Candidate:

- Respiratory muscle weakness
- Sudden cardiac death from arrhythmia

Examiner: **How would you manage this patient?**

Candidate:

- EMG
- Serum immunoglobulin level (Hypogammaglobulinemia)
- Serum testosterone, follicle-stimulating hormone (FSH) and luteinizing hormone (LH) (primary hypogonadism)
- Genetic counseling
- Physiotherapy
- Regular monitoring of cardiac rhythm
- Swallowing therapy
- Regular testing and care of respiratory muscle function.

PARKINSON'S DISEASE

How to Examine a Case of Parkinson's Disease?

- Wash your hands
- Introduce yourself to the patient and take permission for examination
- Quick surveillance of the surroundings for a walking aid
- Inspect the patient for a mask like face, presence of pill-rolling tremor, dribbling of saliva, slow movement and monotonous speech
- Perform the glabellar tap sign and observe continuous blinking
- Passively move the wrist joint and elbow to check for cogwheel rigidity (exacerbated by voluntary movement of other arm) and lead pipe spasticity (ask the patient about any pain in these joints before you do that)
- Ask the patient to stand and walk. Observe the stooped posture, lack of arm swinging and the short shuffling gait
- Tell the examiner that you would normally assess the patient's handwriting and examine eye movement and for postural hypotension (to exclude Parkinson-plus syndromes).

Common Pitfalls

- Failure to recognize clinical features of Parkinson's disease
- Failure to differentiate Parkinson's disease from Parkinson-plus syndromes

Examiner Instructions

- This patient has had frequent falls, perform a general examination

Examiner: **What is the pathologic mechanism behind Parkinson's disease?**

Candidate: The pathological mechanism behind the development of Parkinson's disease is loss of the dopamine-producing neurons in the substantia nigra and development of Lewy bodies in dopaminergic neurons. This in turn leads to reduction in the neurotransmitter "dopamine" and an increase in "gamma-aminobutyric acid (GABA)" release in the basal ganglia which in turn suppresses the cortical motor system.

Examiner: **What are the four main clinical features of Parkinson's disease?**

Candidate: Tremor, Rigidity, Akinesia and Postural instability. In addition a significant proportion of patients also develop sensory symptoms such as allodynia, hyperalgesia and pain.

Examiner: **What clinical features can differentiate Parkinson's disease from Parkinson-plus syndromes?**

Candidate:

- Parkinson's disease tends to be asymmetric (tremor of one side of the body)
- Early falls or postural instability (Shy-Drager syndrome)
- Involvement of autonomic nervous system such as urinary or fecal urgency, incontinence or retention and postural hypotension (Shy-Drager syndrome)
- Presence of tongue fasciculation's (progressive supranuclear palsy)
- Presence of ophthalmopathy (progressive supranuclear palsy)
- Presence of pseudobulbar palsy (progressive supranuclear palsy)
- Dementia and hallucinations suggest Parkinson-plus syndromes
- Presence of pyramidal signs (multiple system atrophy)
- Good response to levodopa suggests Parkinson's disease.

Examiner: **How would you differentiate Parkinson's disease from essential tremor?**

Candidate:
Essential tremor:

- Usually bilateral
- Affects also head and neck (head and neck tremor is not seen in Parkinson's disease)
- Increases with activities and decreases with rest
- Absence of bradykinesia.

Examiner: **What is the mechanism of cogwheel rigidity in Parkinson's disease?**

Candidate: Cogwheel rigidity is caused by the combination of tremor and hypertonia.

Examiner: **Do you know of any new imaging modalities that can help in diagnosing Parkinson's disease?**

Candidate:

- Magnetic resonance volumetry, diffusion weighted MRI and MR spectroscopy

- Positron emission tomography (PET) scan
- DaTscan- Ioflupane iodine-123 injection with SPECT for detecting dopamine transporters (DaT)

Examiner: **How would you treat this patient?**

Candidate:

- *Non-pharmacological alternative therapies:* Exercise, physiotherapy, occupational therapy speech therapy and nutrition
- *Pharmacologic therapy:*
  - *Carbidopa/levodopa (Sinemet):* A dopamine precursor that crosses the blood brain barrier gets converted to dopamine in dopaminergic terminals by dopa-decarboxylase. Levodopa is peripherally converted to dopamine by a peripheral decarboxylase enzyme before it can reach the blood-brain barrier. Carbidopa is added as a peripheral decarboxylase inhibitor
  - Dopamine agonists such as bromocriptine
  - Injectable dopamine agonist such as Apomorphine
  - Monoamine oxidase-B (MAO-B) inhibitors such as Selegiline
  - *Catechol O - Methyltransferase (COMT) inhibitors:* Increase synthesis and release of dopamine
  - *Anti-cholinergics:* Benztropine
- *Surgical:* Deep brain stimulation (DBS) where an electrode is surgically implanted in the subthalamic nucleus to provide continuous electrical stimulation for dopamine.

Examiner: **What is the "wearing-off phenomenon"?**

Candidate: The "wearing-off" phenomenon is the most common motor complication noted during the treatment of Parkinson's disease with levodopa. It refers to the recurrence of tremor and rigidity before the next dose of Carbidopa/levodopa is due. It happens as a result of the short half-life of levodopa. Increasing the frequency of doses of levodopa or the addition of a COMT or MAOI inhibitors may help in alleviating this phenomenon.

Examiner: **What is tardive dyskinesia?**

Candidate: Tardive dyskinesias are repetitive involuntary movements such as facial grimacing, tongue thrusting or repetitive chewing that occur as a complication of long-term levodopa use.

PTOSIS

Important causes of ptosis in clinical exams:

- *Bilateral ptosis in the exam*
  - Myasthenia gravis **(Figure 4.15)**
  - Guillain-Barré syndrome
  - Myotonic dystrophy

- Facioscapulohumeral muscular dystrophy (rare in exam)
- Oculopharyngeal muscular dystrophy (rare in exam)

- *Unilateral*
  - Third nerve palsy (**Figure 4.10**)
  - Horner's syndrome
  - Myasthenia may be unilateral

HORNER'S SYNDROME

How to Examine a Patient with Horner's Syndrome?

- Look at the patient in general for evidence of brain stem stroke such as presence of nasogastric feeding tube, dysphonia, weakness, etc.
- Check for ptosis, meiosis and reaction to light and enophthalmos
- Feel both sides of face to check for absence of sweating on the affected side (anhidrosis)
- Check the pupil size and reaction to light (dilated pupil means third nerve palsy and meiotic pupil suggest Horner's syndrome)
- Examine the neck for scars, central lines and masses
- Percuss and auscultate the lung apex for evidence of apical lung lesion
- Inspect the hand for wasting of the small muscles (**Figure 2.1**).

Common Pitfalls

- Failure to examine the hands (wasting) and lungs (Pancoast lesion)
- Failure to observe neck scars.

Examiner Instructions

- Have a look at this patient's face and proceed accordingly
- This patient has a cough, examine his eyes.

Examiner: **What are the causes of Horner's syndrome?**

Candidate:

- Cervical rib
- Pancoast tumor of the lung
- Lateral medullary syndrome (brain stem stroke)
- Neck tumors
- Trauma, e.g. central line or chest tube insertion.

Examiner: **How would you investigate this patient?**

Candidate:

- Chest X-ray
- CT scan chest and neck
- MRI brain if suspected brain stem lesion.

FURTHER READING

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5

Endocrine, Rheumatology, Connective Tissue and Skin Cases

GRAVES' DISEASE

How to Examine a Patient with Graves' Disease?

- Wash your hands
- Introduce yourself to the patient and take permission for examination
- It is better that the patient sits on the side of the bed or a chair. A goiter may not be obvious when the patient lies flat or semi-sitting in the bed with the head relaxed on a pillow
- Begin by general inspection of the patient. The patient may appear anxious with a staring look and may be thin. Observe for the presence of exophthalmos, lid retraction or goiter **(Figure 5.1)**
- Hold the hand of the patient, examine the pulse rate, rhythm and check for the presence of a collapsing pulse. Feel the palm of the hand for sweating and warmth. Ask the patient to extend his/her hands and spread the fingers to look for fine tremor. Examine the fingers for clubbing and nail onycholysis
- Examine the eyes for the presence of chemosis, redness, lid retraction, exophthalmos, lid lag and ophthalmoplegia
- Follow the standard four steps for thyroid gland examination.
  Inspection: Look for swelling of the gland **(Figures 5.2 and 5.3)**. Ask the patient to swallow and see the movement of the gland. Ask the patient to protrude his/her tongue "for exclusion of thyroglossal cyst". Feel the thyroid gland anteriorly and posteriorly for surface, nodularity, temperature and adjacent lymph nodes. Feel for tracheal deviation. Ask the patient to swallow some water and feel for movement with swallowing. Percuss below the gland for retrosternal extension of the goiter. Listen over the thyroid gland for a bruit.
- Examine the legs for pretibial myxedema **(Figure 5.4)**
- Examine for proximal muscle weakness

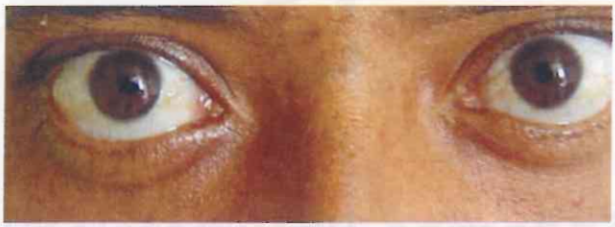


FIGURE 5.1 Bilateral exophthalmos in Graves' disease. Observe the sclera between the lower eyelid and the cornea

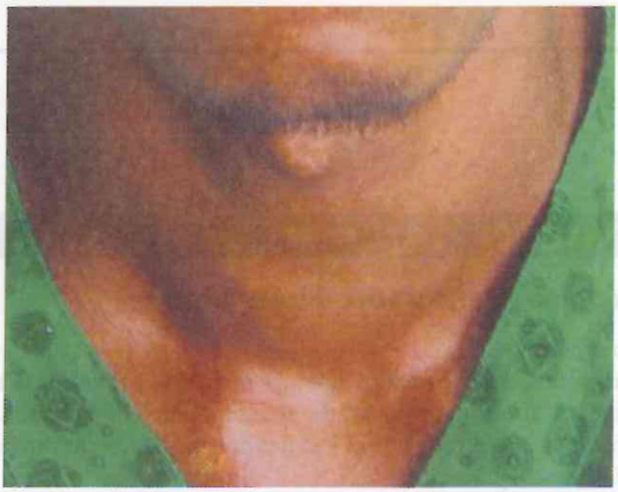


FIGURE 5.2 Diffuse goiter in Graves' disease

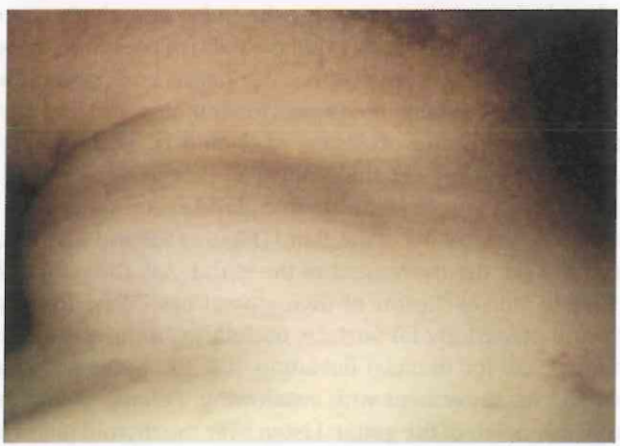


FIGURE 5.3 Neck swelling caused by cystic hygroma. Not all neck swellings are goiter

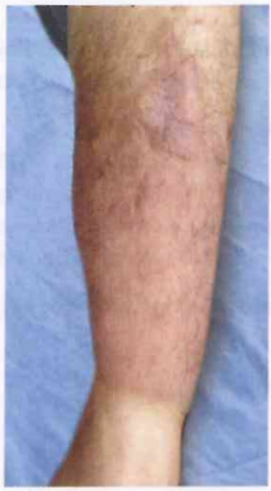


FIGURE 5.4 Pretibial myxedema is suggestive of Graves' disease as the cause of thyrotoxicosis

- Tell the examiner you would like to ask the patient about appetite, weight loss and heat intolerance.

Common Pitfalls

- Failure to recognize the presence of atrial fibrillation
- Failure to properly assess the thyroid status.

Examiner Instructions

- This patient complains of palpitation; please have a look at her
- Assess the thyroid status of this patient
- Examine the neck.

Candidate

This patient is clinically hyperthyroid. She appears thin and anxious, has a staring look, exophthalmos, lid retraction, lid lag, chemosis, ophthalmoplegia, with warm sweaty hands and tachycardia. She has a diffuse goiter with a bruit. There is no ophthalmoplegia, pretibial myxedema or nail onycholysis.

Examiner: **What do you think the cause of this patient's hyperthyroidism is and why?**

Candidate: The cause in this patient is most likely Graves' disease. Features that differentiate Graves' disease from other causes of hyperthyroidism such as toxic multinodular goiter are:

- Presence of eye signs (Graves' ophthalmopathy)

- Diffuse goiter

In general, the presence of these three features along with signs and symptoms of hyperthyroidism can establish the diagnosis of Graves' disease.

Other features that can help in differentiation:

- Assays for thyrotropin-receptor antibodies "TRS-Ab" (particularly thyroid stimulating immunoglobulin "TSIs") is confirmatory for Graves' disease. Detection of TSIs is diagnostic for Graves' disease.
- Diffuse uptake of iodine on Radioactive iodine uptake scanning.

Examiner: **What clinical features suggest a patient is hyperthyroid?**

Candidate:

- Tachycardia or atrial fibrillation
- Warm and moist skin
- Presence of lid lag
- Stare
- Hand tremor
- Proximal muscle weakness
- Weight loss despite increased appetite
- Thyroid bruit

Note: Lid retraction is not a good sign to indicate hyperthyroid status.

Examiner: **What other causes of hyperthyroidism do you know?**

Candidate:

- Graves' disease (most common 60–90% of all causes of hyperthyroidism)
- Toxic multinodular goiter
- Toxic adenoma
- Subacute thyroiditis
- Drug induced—Amiodarone
- Factitious thyrotoxicosis (Taking too much thyroxin)

Examiner: **How would you manage this patient?**

Candidate:

- *Investigations:*
  - Thyroid function test
  - Complete blood count (CBC) and liver function test (LFT) as baseline before starting antithyroid drugs
  - Radioactive iodine uptake thyroid scan
  - Thyrotropin-receptor antibodies
- *Treatment:*
  - *Radioiodine ablation:* The most commonly used therapy, radioactive iodine ablation therapy is preferred in the following situations:
    - *Severe forms of hyperthyroidism:* A large thyroid gland, multiple symptoms of thyrotoxicosis, high levels of thyroxin, and high titers of TSI.

- Younger patients, due to the high relapse rate (>50%) associated with antithyroid therapy.

  Pretreatment with antithyroid drugs: Patients who cannot tolerate hyperthyroidism such as the elderly or patients with heart diseases must be premedicated with antithyroid drugs to make them euthyroid, as radioactive iodine therapy may result in a transient exacerbation of hyperthyroidism. However, antithyroid drugs should be discontinued a few days before radioiodine treatment as pretreatment with thioamides reduces the cure rate of radioiodine therapy in hyperthyroid diseases.

  Contraindications: Pregnancy and severe ophthalmopathy (may worsen Graves' ophthalmopathy)

- *Antithyroid drugs:* Usually indicated in mild thyrotoxicosis or patients who cannot take radioiodine therapy. Methimazole is preferred because of its long duration of action (given once daily) and rapid onset of action. Propylthiouracil is preferred in the first trimester of pregnancy because of potential teratogenic effect of Methimazole. TFT should be assessed six weeks after starting the treatment. TFT monitoring should be mainly by T3 and T4 values as thyroid-stimulating hormone (TSH) may remain suppressed for several months despite normalization of T3 and T4 levels. Duration of therapy varies but usually one to two years.
- *Surgery:* Is not popular therapy nowadays. Indicated mainly for obstructive goiter or patients who cannot tolerate other therapies.

***Examiner:* How would you minimize the risk of worsening of ophthalmopathy by radioactive iodine?**

Candidate: Administration of steroids before and during radioactive iodine.

***Examiner:* What advise would you give to this patient if she receives radioactive iodine therapy?**

Candidate:

- Patients who receive radioactive iodine therapy can expose household contacts via saliva, urine, body fluids or emission from their bodies. Pregnant women, children and sexual contacts are vulnerable. They should avoid sharing cups, sleeping in the same bed, close and sexual contacts for up to one month.
- Pregnancy should be postponed for up to six months post-treatment
- Monitor thyroid function for hypothyroidism
- Monitor for worsening of ophthalmopathy

***Examiner:* What causes pretibial myxedema in Graves' disease? Is it specific for Graves' disease?**

Candidate: It results from accumulation of glycosaminoglycans (hyaluronic acid) in the dermis. It is not specific for Graves' disease as it may be rarely seen in normal patients and in patients with autoimmune thyroiditis. Treatment includes control of thyroid function, local steroid use and pentoxifylline in resistant cases

Examiner: **How would you assess proptosis in thyroid ophthalmopathy?**

Candidate: By using an exophthalmometer one measures the distance from the lateral angle of the orbit to an imaginary line drawn from the most anterior part of the cornea. The normal limit is usually up to 20 mm.

Examiner: **How would you treat thyroid ophthalmopathy in this patient?**

Candidate:

- *Avoid smoking:* Smoking is a proven risk factor for worsening ophthalmopathy in Graves' disease
- Control hyperthyroidism
- Artificial tears
- Oral steroid therapy
- Orbital irradiation
- Decompressive orbital surgery
- Rituximab.

ACROMEGALY

How to Examine a Case of Acromegaly?

- Wash your hands
- Introduce yourself to the patient and take permission for examination
- Inspect the patient for coarse facial features, prominent supraorbital ridge, cutis verticis gyrata **(Figure 5.5)** large nose and lips and tall stature
- Ask the patient to show his/her teeth and open the mouth. Observe for prognathism, large tongue and wide spacing of the teeth **(Figure 5.6)**
- Hold the patient's hands. Observe the spade shape, tight rings and the doughy-feeling of hands **(Figure 5.7)**
- Examine for carpal tunnel syndrome by tapping over the median nerves and testing hand sensation
- Examine the axilla and the neck for evidence of acanthosis nigricans and increased number of skin tags **(Figure 5.8)**
- Examine the visual field for bitemporal hemianopia
- Tell the examiner that you would normally examine the blood pressure, test for blood sugar and ask the patient about a change in shoes or ring size.

Common Pitfalls

- Failure to recognize acromegalic features
- Failure to examine for visual field defects
- Failure to examine for carpal tunnel syndrome.

Examiner Instructions

- Have a look at this gentleman, what is the diagnosis?
- This man complains of headache, what is the diagnosis?

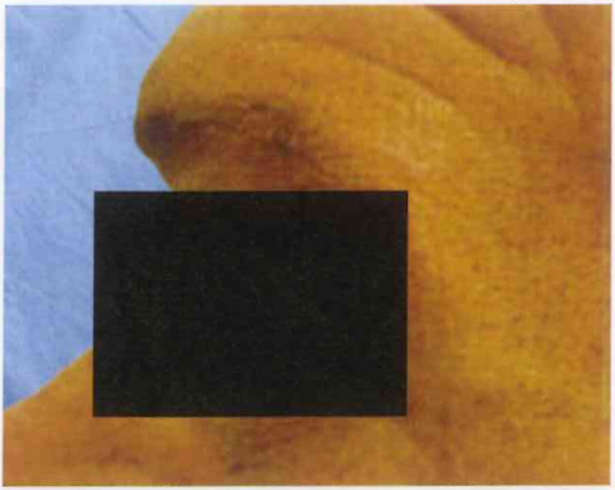


FIGURE 5.5 Prominent supraorbital ridge and cutis verticis gyrata in a patient with acromegaly

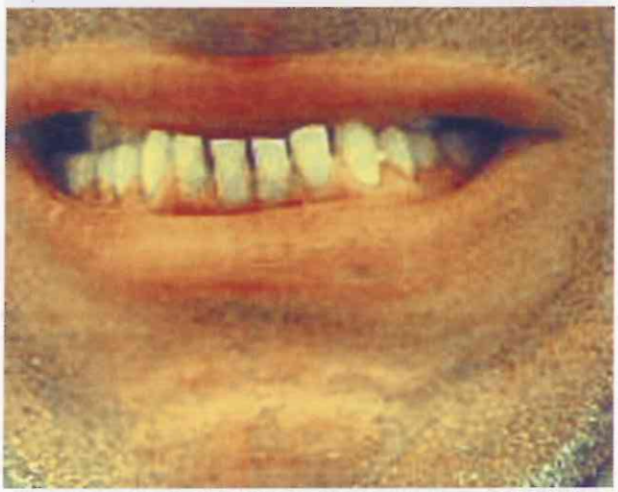


FIGURE 5.6 Prognathism and wide spacing of the teeth in acromegaly

Candidate

This gentleman has features suggestive of acromegaly. He has tall stature, coarse features, frontal bossing, Wide spacing of the teeth, prognathism, large lip and nose, cutis verticis gyrata (furrows resembling gyri of the scalp), spade shaped hands with doughy-feeling and tight ring, acanthosis nigricans, high blood pressure, carpel tunnel syndrome and bitemporal hemianopia.

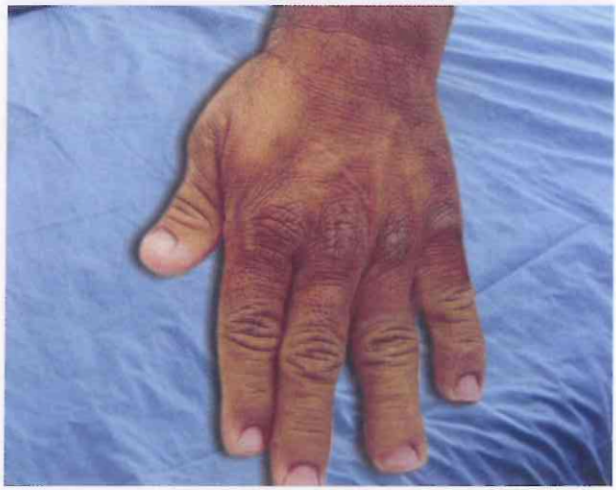


FIGURE 5.7 Spade shaped hand and tight ring in a patient with acromegaly

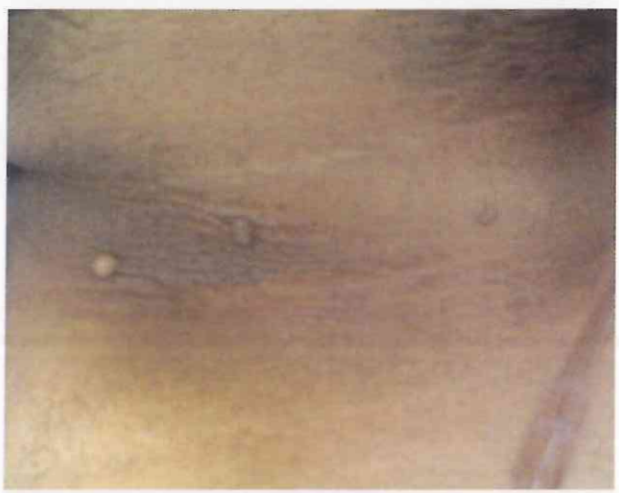


FIGURE 5.8 Acanthosis nigricans and skin tags in acromegaly

Examiner: **How would you confirm the diagnosis of acromegaly?**

Candidate:

- *Measurement of growth hormone (GH) after administration of 75 g oral glucose:* Clearly elevated GH levels (>10 ng/mL) after oral glucose in the presence of clinical features of acromegaly confirms the diagnosis
- Elevated IGF-I values is the best single test for diagnosis of acromegaly.
- Magnetic resonance imaging (MRI) to image pituitary adenomas

Examiner: **Why is IGF-I better than GH in establishing the diagnosis of acromegaly?**

Candidate: Serum GH levels vary during the day according to the food intake, sleep, exercise, etc. Serum IGF-I levels do not vary during the day and is elevated in almost all patients with acromegaly.

Examiner: **Do you know any other causes of acromegaly other than pituitary tumors?**

Candidate:

- Hypothalamic tumor secreting growth hormone-releasing hormone (GHRH)
- Ectopic GH or GHRH secretion from pulmonary carcinoid or small cell lung cancer.

Examiner: **Can you name some conditions that mimic acromegaly?**

Candidate:

- Familial tall stature
- *Pseudoacromegaly:* Acromegalic features with normal hormonal tests in patients with insulin resistance
- McCune-Albright syndrome
- Cerebral gigantism (Sotos syndrome)

Examiner: **How would you treat this patient?**

Candidate:

- Pituitary surgery is the main stay treatment of acromegaly
- Medical therapy with drugs such as octreotide and bromocriptine is indicated for patients who cannot undergo surgery or fail surgical treatment
- Radiotherapy for patients whose disease is not controlled by surgery and medical therapy.

Examiner: **Which of the clinical features of acromegaly are expected to improve with surgery?**

Candidate:

- Clinical features that may improve after surgery
  - Diabetes
  - Soft tissue abnormalities (gradual)
- Clinical features that do not improve with treatment
  - Bony abnormalities
  - Joint disease

Examiner: **Which type of malignancy are patients with acromegaly at risk of?**

Candidate: Colon cancer and polyps.

Examiner: **What should you suspect if this patient complains of nocturnal breathlessness?**

Candidate:

- Obstructive sleep apnea

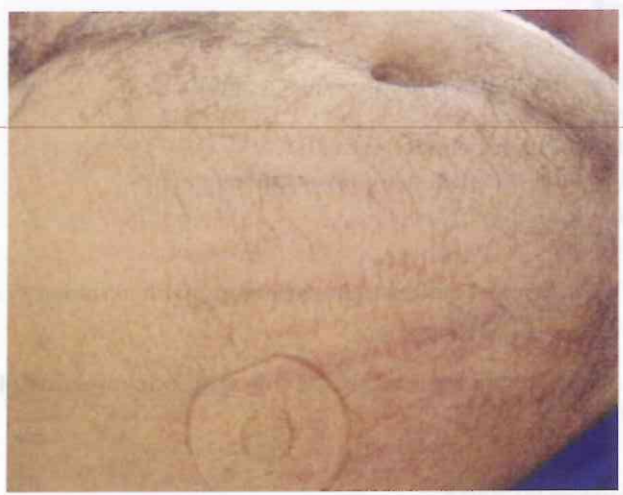
Examiner: **What is the most common cause of death in patients with acromegaly?**

Candidate: Cardiovascular disease.

CUSHING'S SYNDROME

How to Examine a Patient with Suspected Cushing's Syndrome?

- Wash your hands
- Introduce yourself to the patient and request permission for examination
- Inspect the patient for rounded "moon like" face, red cheeks, truncal obesity with limb wasting.
- Examine the eyes for cataract
- Examine the mouth for oral thrush
- Look at the back of the neck and the back for "buffalo hump" and acne
- Examine the skin for ecchymosis and thin skin and the presence of pink striae over the abdomen **(Figure 5.9)**
- Test for proximal muscle weakness
- Tell the examiner that you would like to measure the blood pressure and check the blood sugar
- **Important clue:** In clinical exams, Cushing's syndrome is most frequently encountered as an iatrogenic type from long-term exogenous steroid use. During the examination observe carefully for features of the underlying disease for which steroids are being used. Examples are rheumatoid arthritis, systemic lupus erythematosus (SLE), interstitial lung disease (patient may be using oxygen, with dyspnea or has finger clubbing) and a renal transplant patient.



Common Pitfalls

- Failure to recognize features of Cushing's syndrome
- Failure to suspect fracture of the spine or avascular necrosis as causes of low back pain.

Examiner Instructions

- This patient complains of low back pain, what could be the cause? (This is a very common instruction)
- This patient has a high blood pressure; please have a look at her
- This patient complains of generalized fatigue; examine her to find the cause.
- This patient has systemic lupus erythematosus, what is the cause of her arm weakness?

Candidate

This patient has central obesity, moon like face, thin skin, acne, multiple bruises, proximal muscle weakness, high blood pressure, neck hump, facial plethora and pinkish striae over the abdomen. She has Cushing's syndrome.

Examiner: **Why does this patient have low back pain?**

Candidate: Patients with Cushing's syndrome are at risk of osteoporosis and vertebral fractures. In addition, avascular necrosis of the head of femur may give hip or low back pain

Examiner: **What is the most common cause of Cushing's syndrome?**

Candidate: Iatrogenic due to use of steroids

Examiner: **What are the other causes of Cushing's syndrome?**

Candidate:

- *ACTH dependent Cushing's syndrome:*
  - Pituitary adenoma "Cushing's disease"
  - Ectopic adrenocorticotropic hormone (ACTH)
- *ACTH independent Cushing's syndrome:*
  - Adrenal adenoma
  - Adrenal carcinoma
- *Pseudo-Cushing's syndrome:*
  - Alcoholism
  - Depression

Examiner: **Can you name some conditions associated with ectopic ACTH secretion?**

Candidate: Small cell lung cancer, carcinoid tumors, medullary carcinoma of thyroid, islet cell tumors.

Examiner: **What biochemical tests suggest ectopic ACTH-related Cushing's syndrome?**

Candidate:

- Presence of profound hypokalemia
- Markedly elevated plasma ACTH levels not suppressed by a high-dose (8 mg) dexamethasone suppression test.

Examiner: **How would you investigate this patient?**

Candidate:

- Ask about exogenous use of corticosteroids
- Tests to differentiate true Cushing's from pseudo-Cushing's syndrome
  24 hours urinary free cortisol
  Overnight 1 mg dexamethasone suppression test
  Late night salivary cortisol
- *Serum ACTH level:*
  High: ACTH dependent Cushing's syndrome
  Low: ACTH independent Cushing's syndrome
- *ACTH dependent Cushing's syndrome:*
  Do high dose dexamethasone suppression test
  Suppressed: Pituitary adenoma
  Not suppressed: Ectopic ACTH
  Do MRI pituitary
  Do chest and abdominal MRI
- *ACTH independent Cushing's syndrome:*
  Do adrenal MRI.

Examiner: **How would you manage this patient?**

Candidate:

- Investigations as mentioned above
- Gradually stop or decrease the dose of steroids if iatrogenic
- Trans-sphenoidal surgery in pituitary related Cushing's disease
- Adrenalectomy in adrenal tumors.

Examiner: **What other important points do you consider when managing such patients?**

Candidate:

- Monitor and treat diabetes and hypertension
- Bone densitometry and use of bisphosphonates
- Ophthalmologic check up for cataract.

PSEUDOHYPOPARATHYROIDISM

How to Examine a Case of Pseudohypoparathyroid Hormone (PTH)?

- Wash your hands

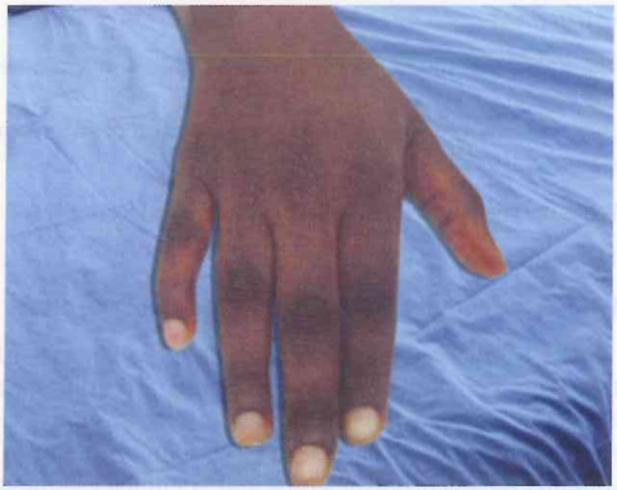


FIGURE 5.10 Pseudohypoparathyroidism. Observe the short fifth finger

- Perform a general inspection of the patient. Observe the short stature, moon like face, short neck and obesity
- Ask the patient to open the mouth to see hypoplasia and missing teeth
- Examine the hands for short fourth and fifth metacarpal bone resulting in shortening of these fingers **(Figure 5.10)**
- Perform Chvostek's and Trousseau's sign for hypocalcemia
- Tell the examiner that you would like to ask about family history and symptoms of paraesthesiae and that you would like to examine the feet for shortened metatarsal bones.

Important Clues

The most common two scenarios in the exam are either the patient is presenting with paraesthesiae in the hands (tingling and numbness) or referred by her GP for investigation of short stature.

A less common scenario is referral for investigation of recurrent seizure episodes. Findings particularly of a short fourth and fifth fingers are usually classical but many candidates tend to miss them. It is also important to notice that almost all cases seen in the exam are females. This is because the disease is rare and the male to female ratio is almost double.

Therefore, when you have a female patient in an exam referred for investigation of short stature, the three most likely possibilities are Turner syndrome, pseudo-hypo-PTH or congenital hypothyroidism.

Common Pitfalls

Candidates frequently miss the short fourth and fifth fingers particularly when the scenario is of paraesthesiae of the hands and think the case is carpal tunnel

Examiner Instructions

This patient complains of tingling in her hands. Could you examine the hands and tell me the cause?

This patient was referred by her GP for investigation of short stature. Could you perform a general examination?

Examiner: **What do you think is the cause for her hand numbness?**

Candidate: Hypocalcemia

Examiner: **What is the pathophysiology behind pseudohypo-PTH?**

Candidate: Type 1 is inherited as AD. Patients with pseudohypo-PTH develop resistance to PTH action leading to hypocalcemia, hyperphosphatemia and increased PTH. The elevated PTH in the blood results from stimulation by hypocalcemia. Due to tissue resistance to PTH, administration of exogenous PTH fails to produce adequate phosphaturia and to stimulate kidneys to produce c-AMP.

Examiner: **What are the other causes of short fourth and fifth metacarpal bones?**

Candidate:

- Turner syndrome
- Idiopathic
- Pseudopseudohypo-PTH
- Sickle cell anemia
- Trauma
- Homocystinuria

Examiner: **What is the difference between pseudohypo-PTH and pseudopseudohypo-PTH?**

Candidate: In pseudopseudohypo-PTH, there will be morphological features of pseudohypo-PTH but the biochemical parameters are normal.

Examiner: **What other complications may happen?**

Candidate: Elevation of PTH in the blood may lead to tissue calcification, cataract and band keratopathy.

Examiner: **How would you manage this patient?**

Candidate:

- Identify low serum calcium, high phosphorus and high PTH
- Failure of the c-AMP to increase after administration of PTH
- Thyroid function test (TFT) (hypothyroidism may occur)
- The treatment is administration of calcium and vitamin D to correct hypocalcemia and suppress the level of PTH (this is the main treatment)
- Genetic and family counseling.

TURNER SYNDROME

How to Examine a Case of Turner Syndrome?

- Wash you hands
- Introduce yourself to the patient and get permission for examination
- *Start by inspection:* Observe the patient's short stature and webbed neck (note that you have to obtain proper exposure in order to see the webbed neck)
- Observe the epicanthic folds and ask the patient to open her mouth to look for a high arched palate
- Observe the presence of a wide carrying angle (cubitus valgus)
- Look at the back of the head for low hair line
- Examine the hands for the short fourth and fifth metacarpal bones, hypoplastic nails and the pulse for radio femoral delay
- Tell the examiner that you would like to check the blood pressure, examine the heart and the chest for widely spaced nipples and shield chest.

Important Clues

The three most likely diagnoses if you face a patient with short stature in the exam are Turner syndrome, pseudohypo-PTH or rarely congenital hypothyroidism.

Common Pitfalls

- Webbing of the neck may not be observed if the neck is covered by upper clothes
- Candidates tend to forget the risk of osteoporosis associated with Turner.

Examiner Instructions

- This patient complains of back pain, please do a general examination.
- This patient was referred by her GP for investigation of short stature
- This patient was referred by your gynecology colleague for infertility or amenorrhea

Examiner: **What is the cause of low back pain in this patient?**

Candidate: Patients with Turner syndrome are at risk of osteoporosis because of primary hypogonadism and this may lead to vertebral fractures.

Examiner: **How common is coarctation of the aorta in adults with Turner syndrome?**

Candidate: Almost one-third of adults with Turner syndrome will have coarctation of aorta.

Examiner: **What should you suspect if you see a male with clinical features of Turner syndrome?**

Candidate: Noonan syndrome. It may occur in males and females and is considered the male version of Turner syndrome.

Examiner: **What renal abnormality may be found on US of this patient?**

Candidate: Horse-shoe kidneys

Examiner: **Can a patient with Turner syndrome get pregnant?**

Candidate: About 98% of patients with Turner syndrome have ovarian dysgenesis and are infertile. However, 2%, those with the mosaic type of Turner syndrome may still produce follicles and can get pregnant.

Examiner: **How would you manage this patient?**

Candidate:

- Regular cardiac evaluation
- Regular blood pressure monitoring
- Check LH, FSH and TFT
- *Hormone replacement therapy:* For development of secondary sexual characteristics and osteoporosis prevention
- Growth hormone therapy for short stature (children).

DEFORMING ARTHRITIS OF THE HANDS

How to Examine a Patient with Deforming Arthritis of the Hands?

- Wash your hands
- Introduce yourself to the patient
- Avoid shaking hands with the patient as it may be painful
- Ask the patient if he/she has any pain in the hands and to alert you in case he/she feels any pain during the examination
- Ask the patient to put his/her hands on a pillow for his/her comfort
- Inspect the hands for redness, different types of deformities, muscle wasting, skin rash, pitting of the nails, onycholysis, gouty tophi or nail fold infarcts **(Figures 5.11 to 5.13)**.
- Feel the joints for hotness, tenderness and synovial thickening
- Ask the patient to move his/her hands at all joints
- Check hand function. Ask the patient to button or unbutton clothes, write with a pen and hold a bottle
- Ask the patient to turn his/her hands to keep the palms up and inspect for wasting
- Check for carpal tunnel syndrome by tapping over the median nerves and examining the sensation
- Move up to the extensor surface of the arms and elbows to check for rheumatoid nodules and psoriatic skin rash **(Figures 5.14 and 5.15)**

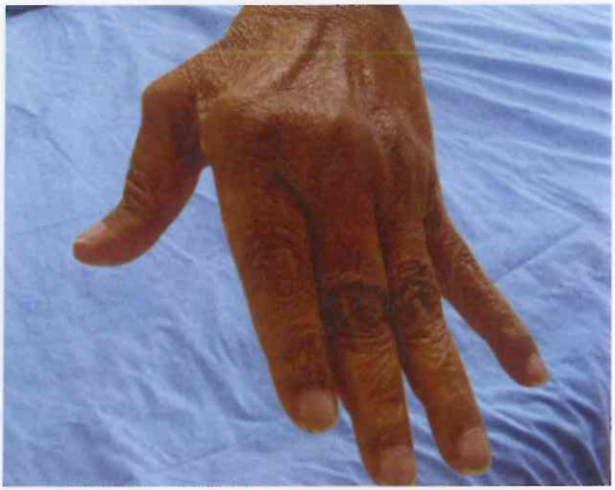


FIGURE 5.11 Rheumatoid hand: Observe the hand deformities, ulnar deviation, MCP subluxation and muscles wasting

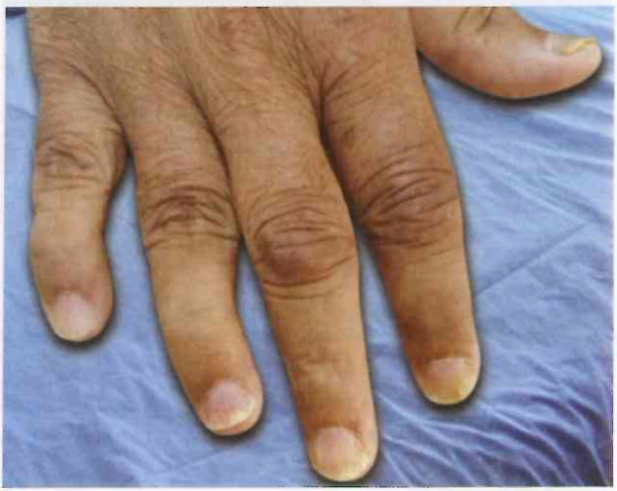


FIGURE 5.12 Psoriatic arthritis. Observe deformity of DIP, onycholysis and nail pitting

- Check the ears for tophi (**Figure 5.13**) and behind the ears and scalp for psoriasis
- Tell the examiner that you would normally examine the other joints in the body including the sacroiliac joints and the eyes for episcleritis or uveitis.

Common Pitfalls

- Causing pain to the patient by rough manipulation of hands

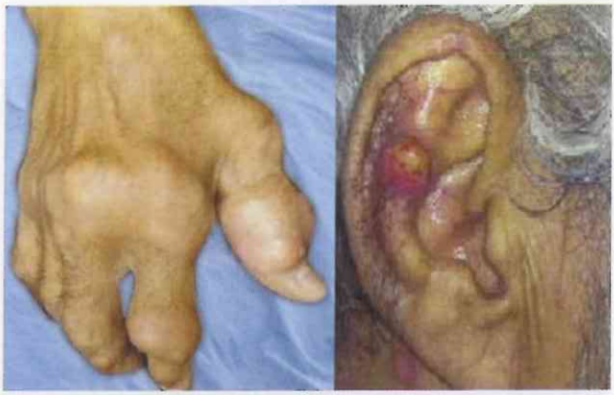


FIGURE 5.13 Chronic tophaceous gout. Observe the tophi in the hands. Ear lobes should also be inspected for tophi

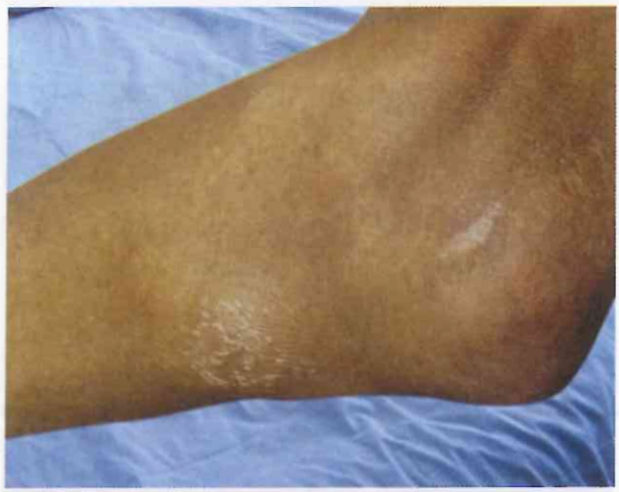


FIGURE 5.14 Examination of the extensor surface of the forearm and elbow may reveal rheumatoid nodules

- Failure to recognize presence of nail pitting, onycholysis or psoriatic skin rash over the extensor surfaces and scalp
- Failure to examine the ears for tophi, the elbows for rheumatoid nodules and the scalp (hair lines) and extensor surfaces for psoriatic rash
- Mistaking gouty tophi as rheumatoid nodules
- *The four likely causes in exam are*—Rheumatoid arthritis, psoriatic arthritis, gouty arthritis and osteoarthrosis. Remember, the hand deformities caused by these 4 conditions might look similar and you have to look carefully during examination for clues to the correct diagnosis

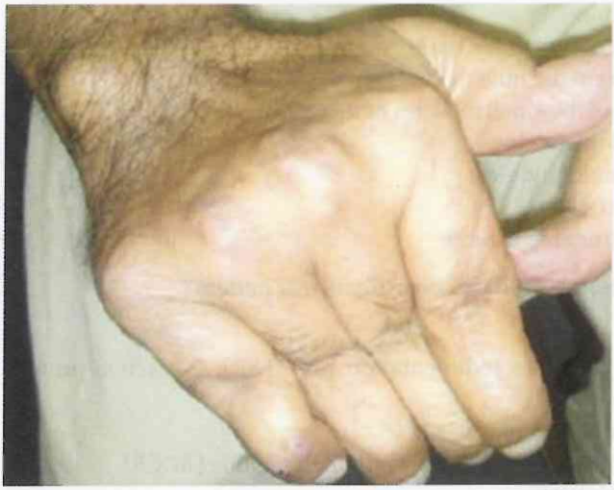


FIGURE 5.15 Severe psoriatic arthropathy may mimic rheumatoid hand. (Observe the psoriatic rash)

Examiner: **How do you differentiate rheumatoid from other causes of hand arthritis?**

Candidate:

- *Rheumatoid hands:* Symmetrical arthritis, involvement of proximal and metacarpophalangeal (MCP) joints with sparing of distal interphalangeal (DIP) joints, presence of rheumatoid nodules and presence of nail bed or digital infarcts **(Figure 5.11)**.
- *Psoriatic arthritis:* Usually asymmetrical involvement, presence of nail pitting or onycholysis, involvement of DIP joints and presence of psoriatic skin rash. In as many as 15–20% of patients, arthritis appears before the psoriasis **(Figures 5.12 and 5.15)**
- *Gouty arthritis:* Asymmetrical and presence of tophi **(Figure 5.13)**
- *Osteoarthrosis:* Involvement of distal IPJ with sparing of the wrist and the MCP joints and the presence of Heberden's nodes.

Examiner: **What are the patterns of psoriatic arthritis?**

Candidate:

- Asymmetrical oligoarticular arthritis
- Symmetrical polyarthritis
- Distal interphalangeal arthropathy
- Arthritis mutilans **(Figure 5.15)**
- Spondylitis with or without sacroiliitis

Examiner: **What are the deformities and changes seen in rheumatoid hands?**

Candidate:

- Boutonniére's deformity
- Swan neck deformity

- Trigger finger
- Ulnar deviation of the hand
- Spindling of the fingers
- Wasting of the small muscles of the hands
- Synovial thickening
- Rheumatoid nodules
- Palmar erythema
- Carpal tunnel syndrome

Examiner: **How would you investigate this patient?**

Candidate:

- CBC, erythrocyte sedimentation rate (ESR), C-reactive protein (CRP) and serum urate
- Rheumatoid factor
- Antibodies against cyclic citrullinated peptide (ACCP)
- *Joint aspiration:*
  - *RA findings:* WBC 1500–25000/cubic mm predominantly PMN and low glucose
  - *Gout:* Crystals
- *Hand X-ray/MRI:*
  - Radiological features of RA include:
    - Joint space narrowing
    - Bone erosions (cardinal feature)
    - Soft tissue swelling

Examiner: **How would you treat rheumatoid arthritis affecting the hands?**

Candidate:

- Early administration of nonbiologic and biologic disease—modifying antirheumatic drugs (DMARDs) alone or in combination is currently recommended as they induce remission and prevent disease progression (anti-TNF agent, methotrexate, hydroxychloroquine, steroids, leflunomide, azathioprine, sulfasalazine, etc.).
- Nonsteroidal anti-inflammatory drugs (NSAIDs), and pain killers
- *Cold therapy to relieve pain and inflammation:* Ice packs or ice water.
- Occupational therapy
- *Rehabilitation:* Orthotic and splint devices
- Surgical referral for deformity correction.

Examiner: **How would you treat gouty arthritis?**

Candidate:

- **Acute arthritis treatment**
  - NSAIDs
  - Colchicine
  - Corticosteroids
  - Adrenocorticotropic hormone (ACTH)
  - Intra-articular steroids

- *Prevent recurrent attack (lowering serum uric acid level):* Allopurinol (should not be used alone in an acute attack as it can exacerbate gout)
- *Nonpharmacologic measures:*
  - Restrict high-purine food consumption
  - Adequate hydration
  - Avoid excess of alcoholic drinks, particularly beer
  - Avoid sodas and beverages sweetened with high-fructose corn syrup
  - Weight reduction.

ANKYLOSING SPONDYLITIS

How to Examine a Patient with Suspected Ankylosing Spondylitis?

- Wash your hands
- Introduce yourself to the patient
- It is better that you ask the patient to stand in order to see the typical posture and perform Schober test and occiput to wall test.
- Inspect the patient for question mark posture with loss of lumbar lordosis and protruding abdomen **(Figure 5.16)**
- Ask the patient to move his neck in all directions and observe that the patient cannot tilt his/her head without moving the whole trunk and there is limitation of all neck movements
- Perform the Schober test
- Check the occiput to wall distance
- Examine for sacroiliac joint tenderness
- Look at the eyes for the presence of acute uveitis **(Figure 5.17)**
- Tell the examiner that you would like to examine the chest and heart for evidence of lung fibrosis and aortic regurgitation.

Common Pitfalls

- Candidates fail to consider ankylosing spondylitis in a patient who cannot tilt his head
- Failure to recognize the typical posture of a patient with ankylosing spondylitis
- Failure to consider sacroiliitis in the differential diagnosis of back pain
- Failure to perform bedside tests for ankylosing spondylitis.

Examiner Instructions

- This patient complains of low back pain, examine his back
- Have a look at this patient and till me the diagnosis.

Candidate

This patient has features suggestive of ankylosing spondylitis as evidenced by a

FIGURE 5.16 Ankylosing spondylitis. Observe the typical question mark posture and loss of lumbar lordosis

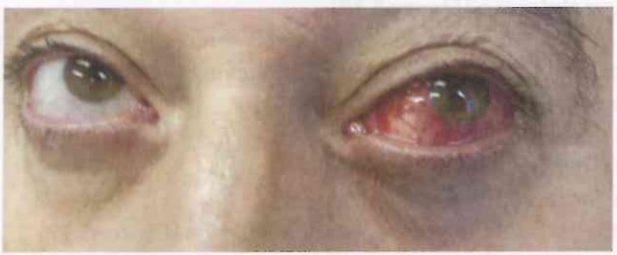


FIGURE 5.17 Anterior uveitis as a complication of ankylosing spondylitis

There is restriction of the spinal movement in all directions. Occiput wall distance is about 8 cm and Schober's test is positive.

Examiner: **How do you perform the Schober test?**

Candidate: With the patient standing erect, make a mark over a line joining the posterior superior iliac spines (or over the spinous process of the 5th lumbar vertebra or over the line joining the dimples of venus). Make another mark 10 cm above it in the midline. When the patient bends maximally forward, the distance between the two points normally exceeds 15 cm.

Occiput to wall distance testing: The patient should stand with the back to the wall. Ears and nose should be at the same horizontal level. Normally the distance between the occiput and the wall should be zero. This distance increases in ankylosing spondylitis.

Examiner: **How would you test for sacroiliac joint tenderness at the bedside?**

Candidate: By the Faber test and Gaenslen's test

- *Faber test:* With the patient supine, flex, abduct and externally rotate the hip joint by placing the heel on the opposite leg. Then push down on the knee, while stabilizing the opposite pelvis. The test is considered positive if the patient feels lower back or sacroiliac pain.
- *Gaenslen's test:* The patient lies supine at the edge of the bed with one leg hanging off the bed, while using his/her hands to bring the other knee to the chest. The test is positive if the patient feels pain in the back or buttock.

***Examiner:* What are the other complications of ankylosing spondylitis?**

Candidate: (Ankylosing spondylitis is the eight "A" disease)

- Ankylosis
- Aortitis and Aortic regurgitation
- Amyloidosis
- IgA nephropathy
- Anterior uveitis
- Apical fibrosis
- Arthritis
- Achilles tendinitis (peripheral enthesitis)

***Examiner:* What is enthesitis?**

Candidate: It is inflammation of the region of attachment of tendons and ligaments to the bones. It can occur in ankylosing spondylitis and other spondyloarthrititis. The most common site is the calcaneal attachment of the Achilles tendon.

***Examiner:* What are the causes of renal disease in ankylosing spondylitis?**

Candidate:

- Drugs (NSAID)
- Amyloidosis
- IgA nephropathy.

***Examiner:* What are the causes of low back pain in a patient with ankylosing spondylitis?**

Candidate:

- Sacroiliitis
- Spinal cord compression and cauda equina syndrome
- Fracture of the ankylosed spine.

***Examiner:* How would you manage this patient?**

Candidate:

- *Investigations:*
  - ESR/CRP
  - *X-ray of the spine:* Particularly the lumbar and cervical spine. Findings include: loss of lumbar lordosis, squaring of the vertebra and bamboo spine (calcification of ligaments, fusion of the spinal facets and syndesmophytes)

- *Imaging of the sacroiliac joint such as X-ray and MRI:* Look for erosions, sclerosis, narrowing or ankylosis of the sacroiliac joints.

- *Treatment:*
  - Nonpharmacologic:
    - Exercise
    - Hydrotherapy
    - Stop smoking
  - Pharmacologic:
    - NSAID
    - Sulfasalazine
    - Biologic agents and anti-TNF, e.g. etanercept, adalimumab, etc.
- *Surgical:*
    - Total hip arthroplasty, spinal surgery for severe flexion deformities of spine and cervical fusion for atlantoaxial dislocation.

SYSTEMIC SCLEROSIS

How to Examine a Patient with Suspected Systemic Sclerosis?

- Wash your hands
- Introduce yourself to the patient and request permission for examination
- Avoid shaking hands with the patient as it might be painful for the patient
- Inspect the patient particularly the face for the presence of telangiectasia, beaked nose, small mouth with puckered appearance and tight skin of the face.
- Ask the patient to open her/his mouth and ask them to insert three of their hand fingers into their mouth
- Examine the hands for pulse (Raynaud's phenomenon), presence of tight and shiny skin, fingertip ulceration and atrophy, contractures and nail fold infarcts **(Figures 5.18 and 5.19)**. Think of calcinosis if you find hard swellings in the fingers **(Figures 5.19 and 5.20)**
- Tell the examiner that you like to ask the patient about dysphagia, symptoms of Raynaud's phenomenon, examine the chest for evidence of lung fibrosis and Jugular venus pressure (JVP) for pulmonary hypertension.

Common Pitfalls

- Failure to include Raynaud's phenomenon in the differential diagnosis of hand pain
- Failure to include systemic sclerosis in the differential diagnosis of a patient with dysphagia
- Failure to recognize the facial or hand manifestations of systemic sclerosis
- Failure to recognize the presence of associated keratoconjunctivitis sicca when the scenario given is for gritty eye sensation
- Failure to recognize features of scleroderma in a chest examination of a patient with lung fibrosis

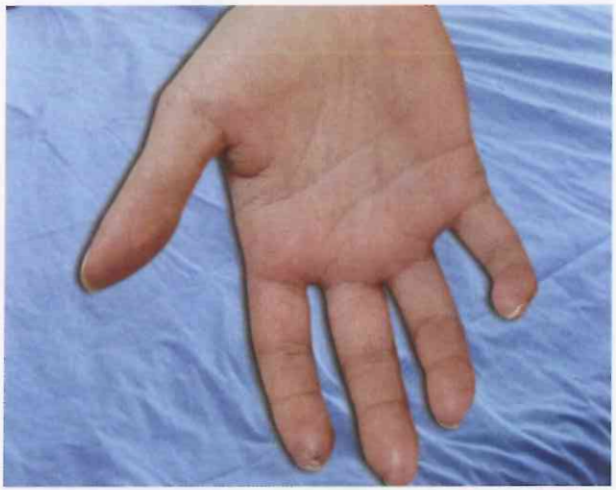


FIGURE 5.18 Systemic sclerosis. Observe the tight skin, contractures of fingers and finger tip ulceration

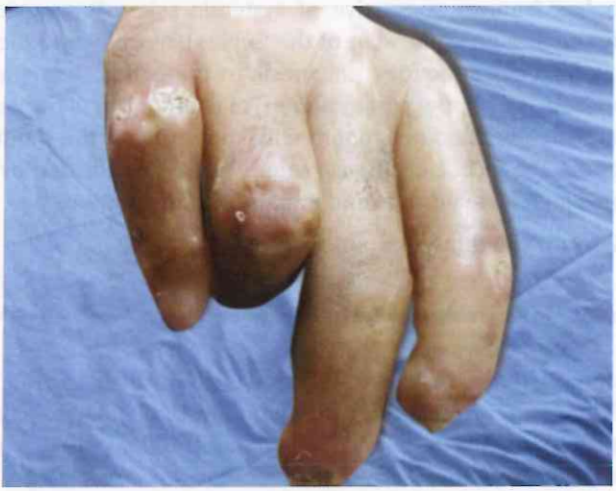


FIGURE 5.19 Severe systemic sclerosis. Very tight skin, contractures, finger tip ulcerations. Observe also the presence of calcinosis

Examiner Instructions

- This patient complains of hand pain, examine her hands
- This patient complains of difficulty in swallowing, have a look at her face/hands
- Have a look at this patient's face and tell me the diagnosis
- This patient complains of gritty eye sensation, what do you think is the cause?

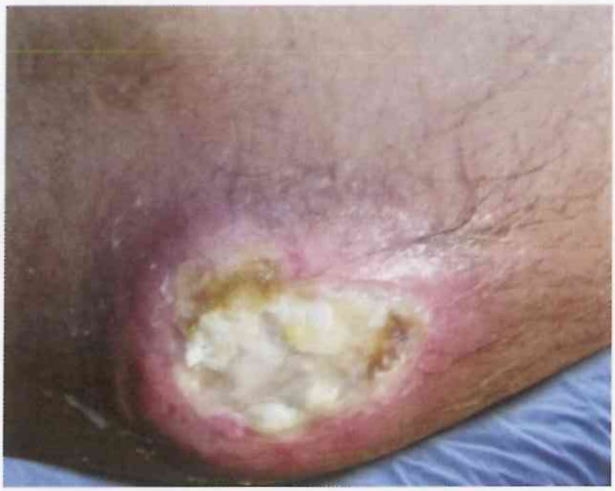


FIGURE 5.20 Calcinosis in systemic sclerosis

Candidate

This woman has features suggestive of systemic sclerosis as evidenced by: Sclerodactyly (tightening of the skin of the fingers), atrophy and ulcerations of the fingertips, finger contractures, calcinosis in the hands. Telangiectasia and pigmentation (salt and pepper pigmentation) of the skin, small beaked nose and inability to sufficiently open her mouth with puckering of the mouth and loss of wrinkling over the face. Her disease seems to be complicated by Raynaud's phenomenon, Sjögren's syndrome and impaired hand function.

Examiner: **Why do you think this patient is pale?**

Candidate:

Anemia in systemic sclerosis may be due to:

- Anemia of chronic disease
- Low vitamin B_{12} due to malabsorption and bacterial overgrowth
- Iron deficiency anemia secondary to gastrointestinal blood loss due to:

  Recurrent esophagitis and gastroesophageal reflux disease (GERD) or angiodysplasia.

Examiner: **What causes interstitial lung disease in scleroderma patients?**

Candidate:

- Pulmonary involvement by the disease itself
- Recurrent aspiration due to dysphagia and GERD
- Bronchiectasis
- Drugs used in treatment of scleroderma, e.g. methotrexate

Examiner: **What are the other serious pulmonary complications of scleroderma?**

Candidate:

- Pulmonary hypertension

Examiner: **What are the gastrointestinal manifestations of scleroderma? How common are these manifestations?**

Candidate:
The gastrointestinal tract is the second most common site of involvement by scleroderma after skin and can be affected in 50–90% of patients to manifest as:

- Dysphagia
- GERD
- Delayed gastric emptying leading to food intolerance and vomiting
- Small bowl malabsorption secondary to bacterial overgrowth
- Colonic telangiectasia
- Fecal incontinence from anal canal involvement

Examiner: **If this patient was found to have a blood pressure of 220/120 what would you suspect?**

Candidate:
Scleroderma renal crisis which is a medical emergency characterized by:

- Acute renal failure
- Sudden onset of severe hypertension
- Normal or only mild proteinuria

Treatment of a renal crisis is by prompt control of blood pressure using ACE inhibitors with frequent monitoring of blood pressure and kidney function.

Examiner: **How would you manage Raynaud's phenomenon in this patient?**

Candidate:

- Avoid cold exposure and keep hands warm (use gloves)
- Stop smoking and caffeine
- Avoid certain drugs such as beta-blockers and migraine medications
- Use dihydropyridine-type calcium antagonists, such as oral nifedipine (first line drugs)
- Sildenafil, nitroglycerine
- Iloprost, or other prostanoids for severe cases
- *Surgical treatment:* Sympathectomy

Examiner: **Which diseases may give skin manifestation similar to systemic sclerosis?**

Candidate:

- *Nephrogenic systemic sclerosis:* In patients with ESRD receiving gadolinium
- Amyloidosis
- Eosinophilic fasciitis
- Chronic graft versus host disease
- Drug induced sclerosis (Bleomycin)

Examiner: **How would you manage this patient?**

Candidate:

- *Investigation:*

- *CBC:* Thrombocytopenia, microangiopathic hemolytic anemia
- Regular follow-up of kidney function
- *CPK level:* Associated polymyositis
- *Serum immunoglobulin:* Hypergammaglobulinemia
- Antibodies against topoisomerase I (anti-Scl 70) are highly specific for systemic sclerosis but are only detected in two-thirds of patients
- Anticentromere antibodies are most commonly detected in patients with limited cutaneous sclerosis
- Echocardiography
- High resolution CT lungs
- Pulmonary function testing

- *Treatment:*
  - Treat specific manifestations such as Raynaud's phenomenon and Sjögren's syndrome
  - *ILD:* Cyclophosphamide
  - *Skin manifestations:* Methotrexate
  - PPI for GERD
  - *Vasodilator drugs for pulmonary hypertension:* Bosentan, Sitaxsentan, Sildenafil, Epoprostenol
  - As malabsorption in scleroderma is caused by bacterial overgrowth, rotating antibiotics may be considered in such cases
  - *Corticosteroids use should be limited:* Steroids are associated with a higher risk of scleroderma renal crisis. Patients on steroids should be carefully monitored for blood pressure and renal function.

TAKAYASU'S ARTERITIS PULSELESS DISEASE

How to Examine a Patient with Suspected Takayasu's Disease?

- Observe the patient for evidence of hemiplegia/stroke
- Examine the radial pulses in both hands
- Examine other pulses in the body.

Common Pitfalls

- Failure to recognize the absence of a radial pulse
- Failure to consider Takayasu's arteritis in the differential diagnosis of absent radial pulse in a patient with stroke.

Examiner Instructions

- In this patient with ischemic stroke, please examine his pulse
- Perform a general examination for this patient
- This young patient has hypertension, please undertake a general examination to establish the cause

Examiner: **Which vessels are most commonly involved in Takayasu's arteritis?**

Candidate: Takayasu's arteritis tends to affect large and medium sized vessels. The most commonly affected vessels are:

- Subclavian and innominate arteries
- Carotid
- Renal arteries
- Pulmonary

Examiner: **How would you manage this patient?**

Candidate:

- *Investigations:*
  - ESR and CRP
  - MRA/FDG-PET scans have replaced the gold standard arteriography for the diagnosis of the Takayasu's disease as it can also assess disease activity
  - Angiography.
- *Treatment:* Steroids, methotrexate, anti-TNF therapy, surgical
  - *Pregnancy:* Increases risk of hypertension complications, maternal and fetal mortality.

Examiner: **What complications are patients with Takayasu's arteritis at risk of?**

Candidate:

- Renovascular hypertension
- Stroke
- Myocardial infarction
- Intracranial hemorrhage
- Limb ischemia
- Retinal ischemia
- Aortic regurgitation.

MARFAN'S SYNDROME

How to Examine a Patient with Suspected Marfan's Syndrome?

- Wash your hands
- Introduce yourself to the patient and request permission for examination
- Inspect the patient for tall stature, long arms and fingers
- Measure the arm span and the height and take the ratio
- Check the wrist sign and the thumb sign **(Figures 5.21 and 5.22)**
- Check for hypermobility of the joints
- Examine the eyes for dislocated lens
- Examine the mouth for high arched palate and crowding of the teeth **(Figure 5.23)**
- Tell the examiner that you would like to examine the heart and check for pectus excavatum

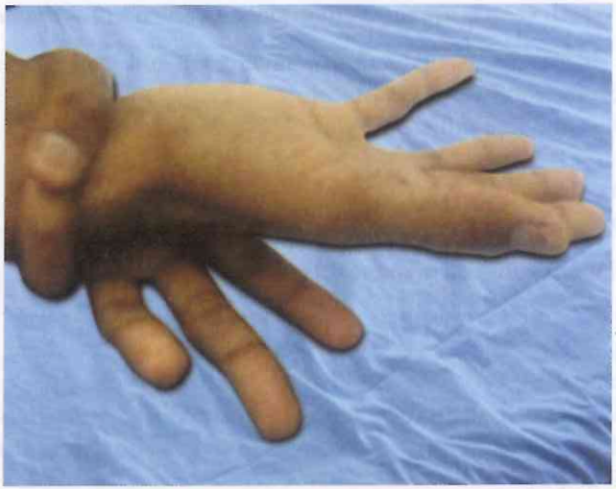


FIGURE 5.21 Wrist sign in Marfan's syndrome

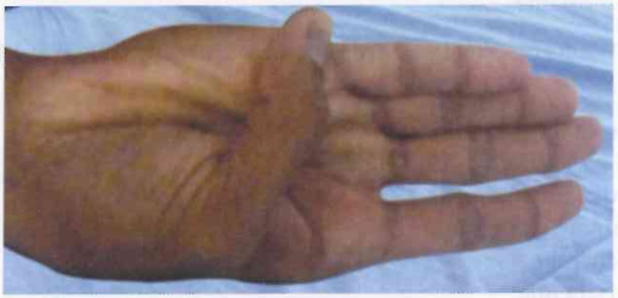


FIGURE 5.22 Thumb sign in Marfan's syndrome

Common Pitfalls

- Failure to recognize features of Marfan's syndrome
- Failure to correlate diplopia to lens dislocation
- Failure to recognize features of Marfan's syndrome in a patient with aortic regurgitation.

Examiner Instructions

- This patient has diplopia, please examine him
- This patient was referred by his GP for a heart murmur, please do a general examination.

Examiner: **What is the mode of inheritance in Marfan's syndrome?**

Candidate: Autosomal dominant occurring in 1 in 5000. The basic abnormality in Marfan's syndrome is mutation in the Fibrillin 1 gene located on chromosome 15.

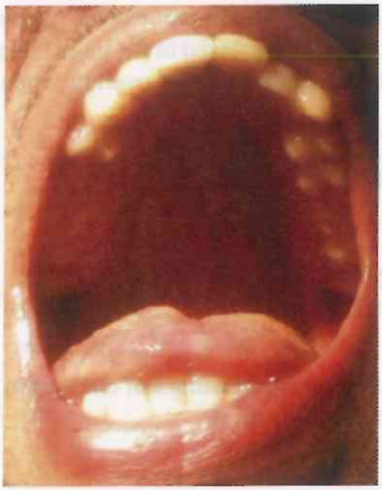


FIGURE 5.23 High arched palate and teeth changes in Marfan's syndrome

Examiner: **What could be the cause of this patient's diplopia?**

Candidate: The cause is usually lens dislocation (60% of cases), which results in uniocular diplopia. The dislocation is typically upward.

Examiner: **How can you differentiate Marfan's syndrome from homocystinuria?**

Candidate:

- *Inheritance:* Marfan's—autosomal dominant and homocystinuria—autosomal recessive
- *Mental retardation:* In homocystinuria, not in Marfan
- *Lens dislocation:* Marfan's—upward lens dislocation and homocystinuria—downward
- *Cardiac manifestations:* Common in Marfan and rare in homocystinuria
- *Recurrent thromboembolism:* Common in homocystinuria.

Examiner: **What are the cardiac manifestations of Marfan's syndrome?**

Candidate:

- Mitral valve prolapse and or MR
- Dilatation of the ascending aorta which may lead to AR
- Aortic dissection.

Examiner: **What is the most common cause of sudden death in Marfan's syndrome patients?**

Candidate: Aortic dissection.

Examiner: **What are the skeletal abnormalities associated with Marfan's syndrome?**

Candidate:

- *An increased arm span:* Arm to height ratio that is greater than 1

- Thumb (Steinberg) sign (i.e. the thumb extends beyond the ulnar border of the hand when the digit is held flexed in the palm) **(Figure 5.22)**.
- *Wrist sign:* (thumb and index fingers overlap when encircling the wrist) **(Figure 5.21)**
- Pectus carinatum **(Figure 1.4)**
- Pectus excavatum **(Figure 1.3)**
- Incisional hernias
- Scoliosis
- Joint hypermobility
- High arched palate **(Figure 5.23)**
- Dental crowding

Examiner: **What are the important causes of chest pain you must consider in this patient with Marfan's syndrome?**

Candidate:
- Aortic dissection
- Spontaneous pneumothorax.

Examiner: **How would you manage this patient?**

Candidate:
- Regular ophthalmology follow-up
- Regular cardiology follow-up
- Echocardiography or MRI of the thoracic and abdominal aorta at diagnosis and then annually
- Control blood pressure
- Elective replacement of the aortic root when there is dilatation or a family history of dissection
- Pregnancy is risky in women with Marfan's syndrome (risk of dissection).

PAGET'S DISEASE OF THE BONE

How to Examine a Case of Paget's Disease?

- Wash you hands
- Introduce yourself to the patient and get permission for examination
- *Inspect the skull:* Observe the large skull, frontal bossing and notice the presence of hearing aids.
- Observe for any cranial nerve palsy
- Inspect the tibia for deformities and bowing
- Feel the tibia or any deformed bone for increased warmth due to increased blood flow. Note that the affected bones in these patients are usually tender so make sure that you do not cause pain to the patient
- Examine the pulse for a water hammer pulse (collapsing character). Be careful not to cause pain
- Tell the examiner you would like to examine the heart and ask the patient about an increase in hat size

Examiner Instructions

- Have alook at this patient, what is the diagnosis?
- This patient complains of generalized body aches, please perform general examination
- Examine the legs of this patient

Examiner: **What is the pathophysiologic mechanism underlying Paget's disease?**

Candidate: The etiology of Paget's disease is unknown. Individuals from the same family may be affected. The disease begins with focal areas of osteoclast induced bone resorption that is followed by osteoblast induced abnormal bone formation. The newly formed bones are less well organized than normal. This leads to enlarged affected bones and skeletal deformity, particularly in weight-bearing bones.

Examiner: **How common is Paget's disease?**

Candidate: Paget's disease is common in Europe, North America, Australia, and New Zealand. The disease may affect up to 2–4% of population older than 50 years and the prevalence increases with age in these countries.

Examiner: **Which bones are commonly affected in Paget's disease?**

Candidate: Pelvic bones, vertebrae, skull, femur and tibia are the most commonly affected

Examiner: **What are the complications of Paget's disease?**

Candidate: The complications of Paget's disease result from new bone formation and include:

- Hearing loss,
- *Spinal stenosis:* Paraplegia, quadriplegia
- Cranial nerve deficits
- Osteoarthritis
- High output cardiac failure
- Hypercalcemia from immobilization
- Pathological fractures
- Osteosarcoma in < 1% of patients

Examiner: **Which type of hearing loss occurs in Paget's disease?**

Candidate: Hearing loss is common in patients with Paget's disease occurring in up to half of these patients. The deafness in Paget's disease can be conductive deafness if the middle-ear ossicles are involved by the disease or sensorineural if the auditory nerve gets compressed by the enlarged petrous bone or it may be mixed.

Examiner: **How would you manage this patient?**

- *Serum ALP:* Very sensitive marker, levels are usually very high and correlate

- *Plain X-ray of the involved bones:* Will show osteolytic and osteosclerotic changes
- Radionuclide bone scan
- *Bisphosphonates are the best and the mainstay treatment:* The drug of choice is a zoledronate (single 5 mg dose IV). They are also used to treat complications of Paget's disease such as hearing loss, heart failure, paraplegia from spinal cord involvement.

***Examiner:* What is the best treatment for paraplegia caused by spinal cord compression from Paget's disease?**

Candidate: The best treatment is bisphosphonate

***Examiner:* What is the mechanism of action of bisphosphonate?**

Candidate: Bisphosphonates inhibit osteoclast activity; they have high affinity for the bones and can remain in the bones for years. They get incorporated into osteoclasts and inhibit the enzyme farnesyl pyrophosphate synthase responsible for maintaining osteoclast structure. This results in osteoclast death and apoptosis.

***Examiner:* What is the characteristic abnormality you may see in the fundus of this patient?**

Candidate: Paget's disease can cause angioid streaks in the retina. These are multiple irregular lines radiating around the optic disc. In addition optic atrophy from compression of the optic nerve may rarely be seen.

HENOCH-SCHÖNLEIN PURPURA

How to Examine a Case of HENOCH-SCHÖNLEIN (HS) Purpura?

- Wash your hands
- Introduce yourself to the patient, take permission and maintain patient dignity during exposure
- Ask the patient if he/she has leg or joint pain before starting the examination
- Make a quick surveillance of the patient and surroundings. Look for steroid infusions, intravenous immunoglobulin or dialysis machine
- Look for the typical vasculitic rash. The rash is elevated (unlike thrombocytopenic purpura); initially it is red then becomes purple and lastly rusty before fading. The typical distribution of the rash is over the legs and buttocks particularly the extensor surfaces **(Figure 5.24)**.
- Look for the site of skin biopsy and swelling of the knee or ankle joints
- Observe for the presence of livedo reticularis that may point to another diagnosis such as polyarteritis nodosa or SLE
- Press over the rash to confirm that it does not blanch with pressure and confirm that the rash is elevated above the normal skin
- Examine the knee and ankle joints
- Tell the examiner that you would like to examine the abdomen and check the blood pressure and urine dip stick

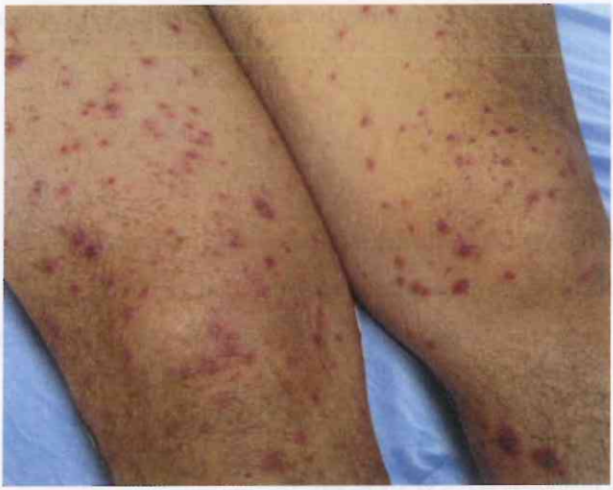


FIGURE 5.24 Typical skin rash in Henoch-Schönlein purpura

Common Pitfall

Candidates unable to differentiate vasculitic from thrombocytopenic purpura.

Examiner Instructions

- Examine the lower limbs of this patient
- This patient complains of abdominal pain, examine his legs.

Examiner: **Which type of vasculitis is HS purpura?**

Candidate: Immune complex-mediated, leukocytoclastic vasculitis affecting small vessels with dominant IgA deposits (IgA deposition is diagnostic).

Examiner: **Why is it called leukocytoclastic?**

Candidate: Because there is deposition of neutrophils in the small blood vessels

Examiner: **What is the main feature that differentiates HS purpura from other leukocytoclastic vasculitis?**

Candidate: IgA deposition in the small vessels.

Examiner: **What other causes of leukocytoclastic small vessel vasculitis do you know?**

Candidate:

- Henoch-Schönlein purpura
- Essential mixed cryoglobulinemia
- Drugs induced
- *Infections: Streptococcus*, hepatitis B, C, HIV, endocarditis
- *Connective tissue diseases:* SLE, RA

Examiner: **Which malignancies may be associated with HS purpura?**

Candidate:

- Non-small-cell lung cancer
- Multiple myeloma
- Prostate
- Non-Hodgkin's lymphoma

Examiner: **What is the classic presentation of HS purpura?**

Candidate: Palpable purpura, abdominal pain and joint pain

Examiner: **What factors predict the development of long-term renal disease (ESRD) in HS purpura?**

Candidate:

- Baseline renal function impairment
- Baseline proteinuria > 1 or 1.5 g/day
- Degree of interstitial fibrosis, sclerotic glomeruli and fibrinoid necrosis on renal biopsy.

Examiner: **What are the causes of abdominal pain in HS purpura?**

Candidate:

- Intestinal edema
- GIT hemorrhage
- Bowl ischemia
- Intussusception

Examiner: **What are the complications of HS purpura?**

Candidate:

- Glomerulonephritis
- Gastrointestinal hemorrhage
- Bowl ischemia
- Intussusception
- Duodenal obstruction
- Orchitis
- Testicular torsion
- Central nervous system (CNS) involvement: Seizure, ataxia
- Pulmonary hemorrhage

Examiner: **How would you diagnose HS purpura?**

Candidate:

- Mainly by the classic clinical picture
- *Skin biopsy:* Characteristic leukocytoclastic vasculitis and deposition of IgA is diagnostic of HS purpura
- Elevated serum IgA level
- Complete blood cell count
- Urine microscopy

- Kidney function test
- Erythrocyte sedimentation rate or C-reactive protein level
- Urinalysis
- Blood chemistry panel, with careful assessment of kidney function
- ANCA (cytoplasmic ANCA [cANCA], perinuclear ANCA [pANCA], atypical ANCA), and rheumatoid factor

Examiner: **What are the indications of systemic corticosteroids in HS purpura?**

Candidate:

- Significant arthritis,
- Severe abdominal pain
- Renal involvement (even early stages)
- Pulmonary hemorrhage.

DERMATOMYOSITIS

How to Examine a Case of Dermatomyositis?

- Look at the patient's face for heliotrope or lilac rash around the eyes and periorbital edema (make sure you do not miss Cushing's appearance from steroid use)
- Look for red or purple rashes over the cheeks, elbows and knees
- Look at the dorsum of the patient's hands for Gottron's papules and the nails for periungual telangiectasias and capillary loops **(Figure 5.25)**
- Look at the chest for the shawl sign and the V-sign. Shawl sign is a reddish rash over the area that is usually covered by the shawl (upper back, shoulders, and back of the neck) **(Figure 5.26)**. V-sign is a similar rash that appears over the anterior chest in a V-shaped pattern **(Figure 5.27)**
- Mechanic's hand. Fissured, cracked and roughened hands resembling those of manual laborers.
- Examine for proximal myopathy
- Tell the examiner that you would like to ask the patient about dysphagia, muscle pain and examine the chest and abdomen for signs of malignancy.

Common Pitfalls

- Candidates may mistake the skin rash over the dorsum of hands for psoriasis
- Candidates may easily miss the diagnosis when the patient has Cushing's appearance due to steroid use
- Candidates forget the association between dermatomyositis and malignancies

Examiner Instructions

- Look at this patient's face, what is the diagnosis?
- Examine this patient's hands

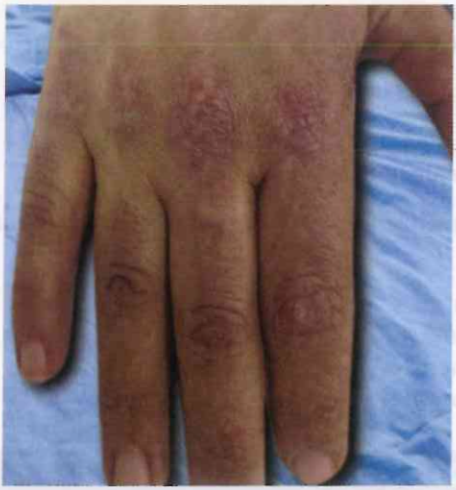


FIGURE 5.25 Gottron's nodules in dermatomyositis

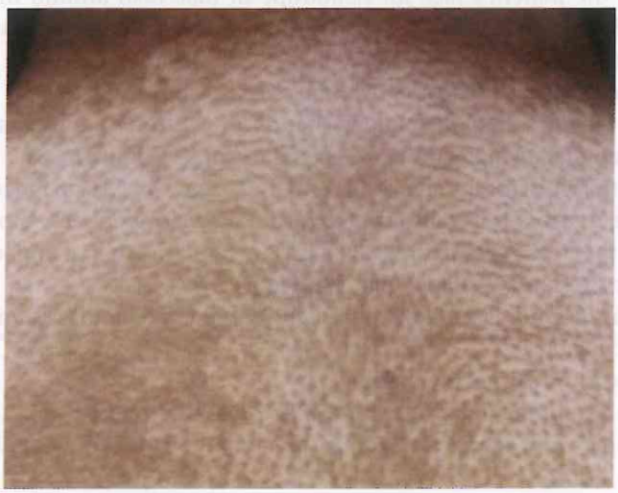


FIGURE 5.26 Shawl sign in dermatomyositis

- This patient complains of dysphagia (or muscle weakness), please examine the hands (or look at her face)

Examiner: **What systemic manifestations of dermatomyositis do you know?**

Candidate:

- Cardiomyopathy
- Interstitial lung disease
- Respiratory muscle weakness
- Association with malignancies

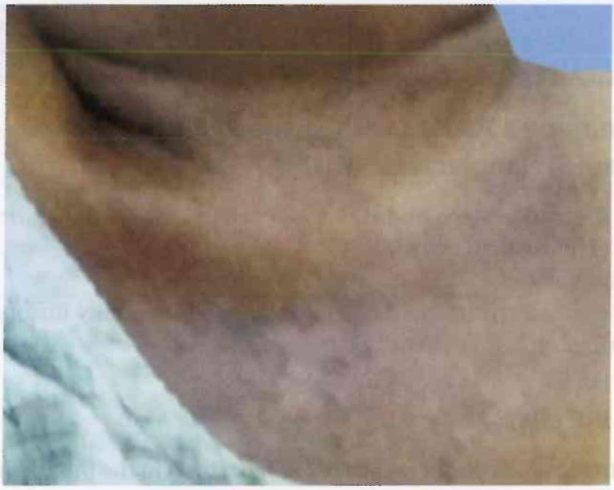


FIGURE 5.27 V-sign in dermatomyositis

- Association with other connective tissue diseases
- Cutaneous calcification (usually in children)

Examiner: **Which malignancies might be associated with dermatomyositis?**

Candidate:

- Ovarian
- Lung
- Lymphoma
- Stomach

Examiner: **How do you investigate this patient?**

Candidate:

- Serum CPK, aldolase and ESR
- ANA, anti-Jo-1 (most specific), anti-dsDNA, RF, anti-SCL70
- Chest X-ray
- Tumor markers
- Skin biopsy and/or muscle biopsy
- Lung function testing

Examiner: **How would you treat this patient?**

Candidate:

- Protection from the sun (avoidance of direct sun exposure, using sunscreens, creams, clothes, etc.)
- Systemic steroids
- Immunosuppressant drugs such as methotrexate, mycophenolate and azathioprine
- Rituximab
- IV immunoglobulin

HEREDITARY HEMORRHAGIC TELANGIECTASIA "OSLER-WEBER-RENDU DISEASE"

How to Examine a Patient with Suspected Hereditary Hemorrhagic Telangiectasia (HHT)?

- Inspect the patient face and mouth for telangiectasia (look also under the tongue) **(Figure 5.28)**
- Check for pallor
- Tell the examiner you would like to enquire about family history and check pulse oximetry.

Common Pitfalls

- Failure to recognize the presence of telangiectasia in the mouth or on the face
- Mistaking telangiectasia for other unrelated lesions
- Diagnosing hereditary hemorrhagic telangiectasia (HHT) but missing the presence of pallor.

Examiner Instructions

- This patient complains of epistaxis and melena; please do a general examination to establish the cause
- This patient was referred because of low Hb. Could you examine him to determine the cause?
- This patient complains of fatigue, could you perform a general examination to establish the cause?

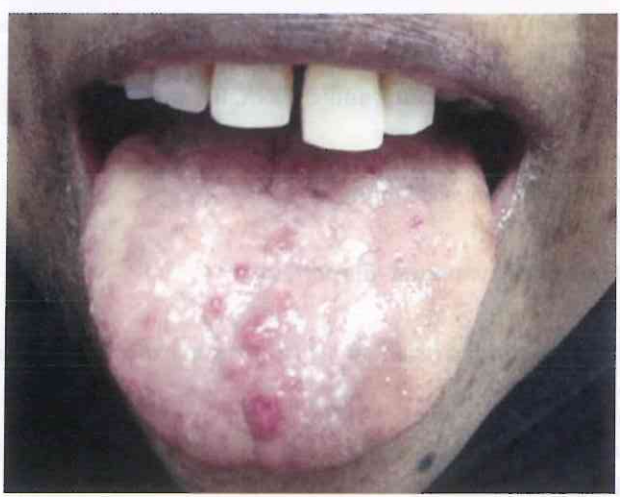


FIGURE 5.28 Hereditary hemorrhagic telangiectasia (observe the telangiectasia in the face and on the tongue)

Examiner: **What is the mode of inheritance in HHT?**

Candidate: Autosomal dominant

Examiner: **What question would you like to ask this patient to confirm your diagnosis?**

Candidate: Family history of HHT, epistaxis or bleeding

Examiner: **Which organs can be involved in HHT?**

Candidate:

- *Nasopharynx:* Recurrent epistaxis
- *Skin:* Telangiectasia
- *Liver and lungs:* Arteriovenous malformations
- *Gastrointestinal:* GIT bleeding
- *CNS:* Arteriovenous malformations

Examiner: **What serious complications are patients with HHT at risk of?**

Candidate:

- Brain abscess from right to left pulmonary shunting
- Hemorrhagic or ischemic stroke
- High-output congestive heart failure
- Chronic GIT bleeding and anemia
- Portal hypertension with esophageal varices
- Pulmonary hemorrhage
- Right to left shunt
- Pulmonary hypertension
- Liver cirrhosis
- Retinal telangiectasia

Examiner: **Is screening for cerebral AV malformations recommended in these patients?**

Candidate: This remains a controversial issue. Although cerebral AVM can develop in patients with HHT, intracerebral bleeding is rare and finding an AVM does not necessarily require intervention in most cases. Screening may be considered if there is family history of cerebral hemorrhage.

Examiner: **How would you manage this patient?**

Candidate:

- CBC and clotting profile
- Liver function
- Pulse oximetry (standing and supine)
- Chest X-ray
- MRI of the lungs
- Upper and lower GIT endoscopy
- *Treatment:* Cauterization, sclerotherapy, laser treatment, estrogen therapy, embolization

NEUROFIBROMATOSIS "TYPE 1" "VON RECKLINGHAUSEN'S DISEASE"

How to Examine a Patient with Suspected Neurofibromatosis?

- Wash your hands
- Introduce yourself to the patient and request permission
- Inspect the skin of the patient particularly the back for presence of multiple neurofibromas, plexiform neurofibroma (usually large and pedunculated mass), café-au-lait lesions
- Inspect the axilla for axillary flickering
- Inspect the iris for Lisch nodules (usually require slit lamp)
- Tell the examiner that you would like to measure the blood pressure, examine hearing and ask about family history

Note: Neurofibromatosis type 1 (Von Recklinghausen's disease) is the one that is seen in exams.

Common Pitfalls

- Missing the diagnosis (particularly when there are multiple large neurofibromas or a large plexiform neurofibroma in the skin some candidates may diagnose it as multiple skin warts or tumors)
- Poor examination techniques
- Poor discussion

Examiner Instructions

- This patient complains of skin rash/lesions. Could you examine his skin?
- This patient was referred because of high blood pressure and palpitations. Could you examine his back and tell me the cause of his hypertension?

Examiner: **What are the diagnostic criteria for neurofibromatosis type 1 (how many lesions are required for the diagnosis)?**

Candidate: Two or more of the following criteria in the absence of another diagnosis

- Six or more café-au-lait macules >5 mm in greatest diameter in prepubertal individuals and >15 mm in greatest diameter in post pubertal individuals;
- Two or more neurofibromas of any type or one plexiform neurofibroma
- Freckling in the axillary or inguinal regions
- Optic glioma
- Two or more Lisch nodules (iris hamartomas)
- A distinctive osseous lesion such as sphenoid dysplasia or tibial pseudarthrosis
- A first-degree relative with type 1 neurofibromatosis

Examiner: **What is the mode of inheritance of type 1 neurofibromatosis?**

Candidate: Autosomal dominant.

Examiner: **What is the prognosis and cause of mortality in patients with type 1 neurofibromatosis?**

Candidate: Life expectancy is reduced by 15 years in these patients. The most common cause of death in these patients is malignant peripheral nerve sheath tumors and vasculopathy.

Examiner: **What do you mean by neurofibromatosis vasculopathy?**

Candidate: Patients with neurofibromatosis are at risk of various vascular problems such as:

- Hypertension
- Arterial stenosis
- Arterial aneurysm formation
- Moyamoya (small vessels form around the stenotic area giving the appearance of a "puff of smoke")

Examiner: **How do you explain hypertension in these patients?**

Candidate:

- Essential hypertension is common in these patients
- Associated renal artery stenosis
- Associated pheochromocytoma
- Coarctation of the aorta

Examiner: **What other serious complications of type 1 neurofibromatosis do you know?**

Candidate:

- Optic nerve glioma. which may lead to progressive visual loss
- CNS tumors

Examiner: **How does type 2 neurofibromatosis differ from type 1?**

Candidate: Type 2 is a central type with more involvement of the CNS by tumors and less skin manifestations. Bilateral or unilateral acoustic neuromas are characteristic of this type.

Examiner: **How would you manage this patient?**

Candidate:

- Genetic counseling
- Regular monitoring of the blood pressure
- 24 hours urine for catecholamines
- MRI of the brain and optic nerves
- Surgery for the skin lesions.

DIABETIC FOOT, NEUROPATHY AND ARTHROPATHY

How to Examine the Diabetic Leg?

- Wash your hands and introduce yourself to the patient
- Take permission and ask for any pain in the legs
- *The examination is composed of 3 parts:* Inspection of the legs, examination of the vascular system and neurological examination of the lower limbs with particular attention to sensation and reflexes
- *Inspection should include:* Presence of foot ulcers, callus, diabetic dermopathy, gangrene, shiny and cold skin with loss of hair suggestive of ischemia, amputation of toes and joint swelling or deformities (Charcot joint) **(Figure 5.29)**. Muscle wasting should also be noted. Particular attention should be paid to skin between the toes for wounds and fungal infection and the condition of the nails.
- Feel all the pulses in the legs and feet
- Perform a neurologic examination, paying particular attention to the sensory examination and reflexes
- Inform the examiner that you would like to perform a fundus exam and check the blood sugar and HbA_{1c}
- Charcot joint clinically presents with the triad of signs of inflammation (hotness and redness), joint swelling and loss of sensation in the foot with no or little pain relative to the degree of inflammation

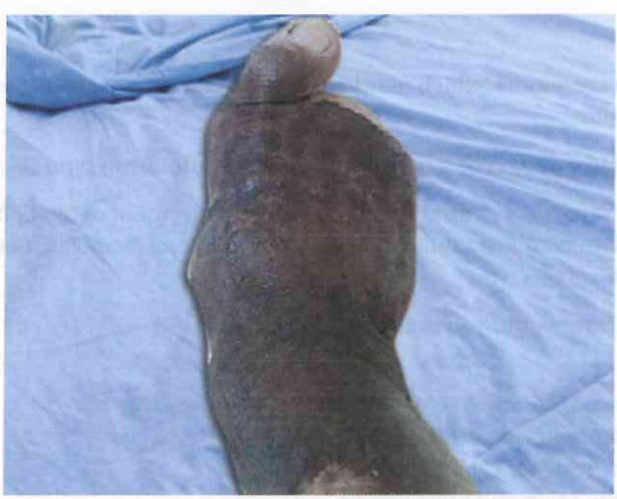


FIGURE 5.29 Charcot joint, toe amputation and healing skin ulcers in a diabetic foot

Examiner: **Which conditions may cause similar symptoms to diabetic neuropathy?**

Candidate:

- Drugs
- Vitamin B_{12} deficiency
- Alcohol
- Chronic renal disease
- Charcot-Marie-Tooth
- Vasculitis
- Heavy metal poisoning

Examiner: **Can you name a drug that is used in the treatment of orthostatic hypotension from autonomic neuropathy?**

Candidate: Midodrine

Examiner: **Which sensation is lost early in diabetic neuropathy?**

Candidate: Pain and temperature due to small fiber damage followed by touch (monofilament) and vibration due to large fiber damage.

Examiner: **What are the risk factors for a diabetic foot ulcer?**

Candidate:

- Poor glycemic control
- Peripheral neuropathy
- Peripheral vascular disease
- History of previous foot ulcer
- Visual impairment
- Chronic kidney disease (CKD)
- Smoking
- Presence of foot deformity

Examiner: **How would you manage a patient with diabetic neuropathy and diabetic foot?**

Candidate:

- Tight control of vascular risk factors (glucose, triglycerides, blood pressure) and stop smoking
- Regular examination of the feet
- Patient education about foot care
- Avoid tight shoes and wear diabetic shoes
- Aggressive antibiotics for diabetic foot infection
- Request X-ray/MRI or bone scan if there is suspicion of osteomyelitis or Charcot joint
- *Drugs for symptomatic relief:* Pregabalin, gabapentin, duloxetine, tricyclic antidepressants.

- Podiatrist, vascular surgeon and orthopedic consult if there are ulcers, suspected vascular insufficiency, osteomyelitis or Charcot joint
- Corneal confocal microscopy.

Examiner: **Do you know any noninvasive technique that can detect and predict development of diabetic neuropathy very early and monitor response to management of neuropathy?**

Candidate: Corneal confocal microscopy is a noninvasive ophthalmic technique, which can quantify small nerve fiber loss in the cornea. It may act as a surrogate marker for early diagnosis, stratification of severity, and assessment of therapeutic efficacy of new treatments in diabetic neuropathy.

Examiner: **When should patients with diabetes undergo neuropathy assessment?**

Candidate: According to the ADA all patients with Type 2 diabetes at diagnosis and those with Type 1 diabetes after 5 years of diabetes should undergo an annual assessment for neuropathy.

CHARCOT JOINT

Examiner: **What pathophysiologic mechanism is responsible for the development of a Charcot joint in diabetes?**

Candidate: Charcot joint results from peripheral neuropathy leading to painless recurrent trauma and injury to the joint, increased local bone resorption due to osteoclast formation and activation in a well-perfused foot.

Examiner: **What are the causes of a Charcot joint?**

Candidate:

- Diabetes is the most common cause **(Figure 5.29)**
- Leprosy
- Syphilis (tabes dorsalis)
- Chronic alcoholism
- Vasculitis
- Syringomyelia

Examiner: **Which joints are commonly affected in diabetic patients with Charcot arthropathy?**

Candidate: Mid-foot and ankle joints

Examiner: **If you find a Charcot joint in the upper limb, which disease should you suspect?**

Candidate: Syringomyelia is the most common cause of Charcot joint in the upper limbs. Usually shoulder (rarely elbow)

Examiner: **If you find a Charcot joint in the knee, what disease would you suspect?**

Candidate: Tabes dorsalis

Examiner: **How would you mange a patient with a Charcot joint?**

Candidate:

- *Investigations:*
  - X-ray and MRI to assess the degree of damage and to exclude osteomyelitis
  - Check HbA_{1c}, syphilis serology, vasculitic screen and B_{12} level
- *Treatment*
  - Off loading the foot and avoidance of weight bearing on the affected side and immobilization of the joint is the most important intervention
  - Surgery should only be considered in refractory cases with significant bone deformity (rocker bottom foot). It may also be considered in osteomyelitis to remove infected bones
  - Osteomyelitis should always be considered in any patient with a Charcot joint.

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6

Fundus Cases

FUNDUS CASES IN THE CLINICAL EXAMINATION

How to Perform Fundus Examination?

- Wash your hands and introduce yourself to the patient
- Decrease illumination in the room
- Remove your and the patient's glasses
- Instruct the patient to look at a fixed distant point behind you
- Use your right eye to examine the right fundus of the patient and your left eye for the left fundus
- First find the optic disc (How? when you see the retina, look for a vessel and follow it centrally, it will lead you to the optic disc). Check the optic disc color, edges and optic cup (central pale part of the disc)
- Second check the vessels starting at the disc. Observe that veins are larger and darker than arteries and arteries are lighter and narrower than veins
- Arteries have a central reflecting line—'silver-wire' appearance
- Check points of crossing of arteries and veins for arteriovenous (AV) nipping
- Check the retina for hemorrhage, microaneurysms and exudates. To check for the peripheral retinal you may angle the ophthalmoscope or simply ask the patient to look in the four quadrants (up, down, left and right) while examining the fundus
- *Check the macula:* By asking the patient to look directly at the light of the ophthalmoscope.
- Do not let an abnormal finding distract you from performing the complete retinal examination in a systematic manner, as one abnormal finding might be just the tip of the iceberg. Record all abnormal findings in your mind with their positions.

Common Pitfalls during Fundoscopic Examination

- Improper technique of examination because of lack of proper practice
- Candidates get distracted by the first abnormal finding and do not perform a complete fundoscopic examination
- Candidates get anxious as time passes, particularly if they are not able to identify an abnormality at the start of examination.

DIABETIC RETINOPATHY

Examiner: **What factors determine the development of diabetic retinopathy?**

Candidate:

- Poor glycemic control
- Duration of diabetes
- Presence of nephropathy (usually coexists with retinopathy)
- Presence of other factors such as hypertension.

Examiner: **What are the two earliest signs of diabetic retinopathy?**

Candidate:

- Microaneurysms
- Hard exudates.

Examiner: **Can visual loss occur in nonproliferative diabetic retinopathy?**

Candidate: Yes, usually due to macular edema.

Examiner: **What do you mean by the following terms?**

Candidate:

- *Microaneurysms:* The earliest clinical sign of diabetic retinopathy. They occur secondary to capillary wall out pouching due to pericyte loss and appear as small red dots in the superficial retinal layers
- *Dot and blot hemorrhages:* Occur as a result of microaneurysmal rupture. They may appear to be small if they are located in the inner nuclear and outer plexiform layers
- *Flame-shaped hemorrhages:* Larger hemorrhages than dots and blots that occur in the superficial nerve fiber layer
- *Hard exudates:* Caused by leakage of proteinaceous material and lipids from the vessels
- *Cotton-wool spots:* Represent ischemia and infarction of the nerve fiber layer from the occlusion of precapillary arterioles.

Examiner: **How is diabetic retinopathy classified?**

Candidate:

- Nonproliferative diabetic retinopathy (NPDR) **(Figure 6.1)**
  - *Mild:* At least one microaneurysm
  - *Moderate:* Hemorrhages, microaneurysms, and hard exudates

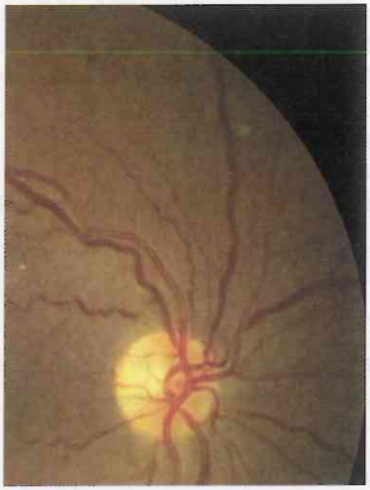

FIGURE 6.1 Nonproliferative diabetic retinopathy. Observe microaneurysms, dot hemorrhages and exudates

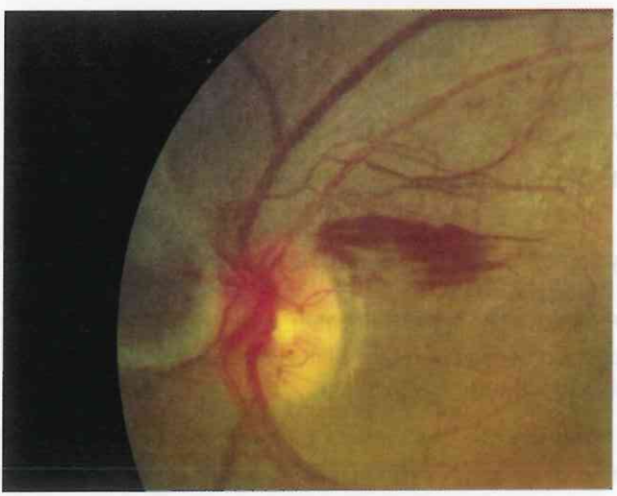

FIGURE 6.2 Proliferative diabetic retinopathy. Observe the new vessel formation (hallmark of proliferative retinopathy)

  - *Severe:* Hemorrhages and microaneurysms in 4 quadrants or venous beading in at least 2 quadrants or intraretinal microvascular abnormalities in at least 1 quadrant. To make it easier remember the numbers (4-2-1)
- Proliferative diabetic retinopathy (PDR) (**Figure 6.2**)
  - Neovascularization is the hallmark of PDR (can be near the disc or elsewhere in the retina)

  - Preretinal hemorrhages are pockets of blood (boat shaped) between the retina and the posterior hyaloid membrane
  - Vitreous hemorrhage may appear as a diffuse hazy blood clot within the vitreous
  - Traction retinal detachments
- Clinically significant maculopathy
  - Hard exudates and thickening of the macula near the fovea.

Examiner: **Can you name some factors that cause rapid worsening of diabetic retinopathy?**

Candidate:
- Intensive insulin therapy may lead to transient worsening of diabetic retinopathy during the first year of treatment
- Pregnancy.

Examiner: **How do you screen for diabetic retinopathy?**

Candidate:
- *Ophthalmoscopy:* Needs trained doctor
- *Digital fundus imaging:* More accurate.

Examiner: **When do you start screening for diabetic retinopathy?**

Candidate:
- *Type 1:* 3-5 years after diagnosis
- *Type 2:* At diagnosis.

Examiner: **How would you manage this patient?**

Candidate:
- Tight glycemic control (DCCT and UKPDS trials)
- Control blood pressure
- Focal photocoagulation for clinically significant macular edema
- Panretinal photocoagulation for PDR **(Figure 6.3)**
- Vitrectomy for vitreous hemorrhage.

Examiner: **What are the indications of laser photocoagulation for diabetic retinopathy?**

Candidate:
- Proliferative retinopathy
- Clinically significant macular edema
- In some cases of severe NPDR.

Examiner: **What is the latest treatment for diabetic macular edema?**

Candidate: Intravitreal injections of anti-VEGF drugs (ranibizumab and bevacizumab) or steroids [can raise intraocular pressure (IOP) and accelerate cataract formation]

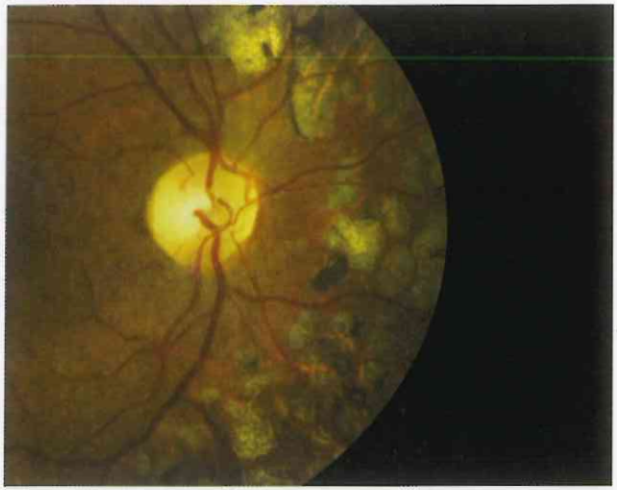

FIGURE 6.3 Typical laser burn (scar) appearance for diabetic retinopathy

HYPERTENSIVE RETINOPATHY

Examiner: **How would you classify hypertensive retinopathy?**

Candidate:

- *Mild:* AV nipping and silver wiring
- *Moderate:* Hemorrhages, cotton wool spots and hard exudates
- *Severe:* Papilledema.

Examiner: **What is the prognostic value of hypertensive retinopathy?**

Candidate: Hypertensive retinopathy is associated with increased risk of stroke, heart failure, coronary artery disease (CAD) and possibly chronic kidney disease (CKD).

Examiner: **How would you manage this patient?**

Candidate:

- Control blood pressure
- Check urea and electrolytes, lipid profile, blood sugar and chest X-ray
- Manage associated diabetes and hyperlipidemia.

OPTIC ATROPHY

Examiner: **What do you see in optic atrophy on fundoscopic examination?**

Candidate: The optic disc appears white (pale) and bright with well demarcated margins (full moon against a dark red sky) **(Figure 6.4)**

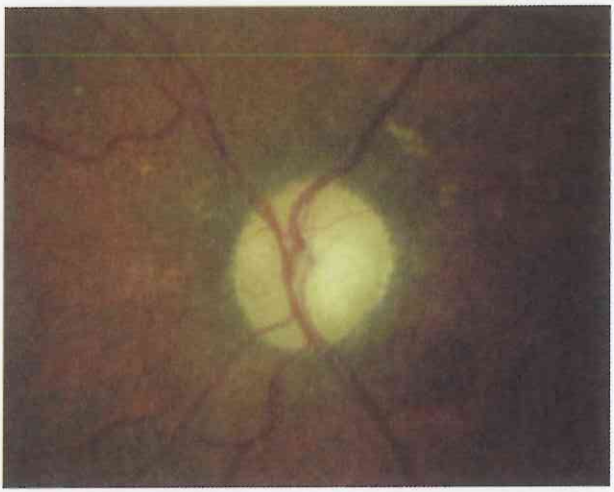

FIGURE 6.4 Optic atrophy. Observe the pale and bright looking disc with well demarcated margins

Examiner: **What are the causes of optic atrophy?**

Candidate:

- **Acquired causes (NITROGENS)**
  - *Nutritional:* Vitamin B_1, B_6 and B_{12} deficiency
  - Ischemia to the nerve
  - *Toxic:* Tobacco amblyopia, quinine, methanol, arsenic, lead
  - Retinitis pigmentosa
  - *Oncologic causes:* CNS tumors
  - Glaucoma
  - Ethanol
  - Neurosyphilis and neurosarcoidosis
  - Multiple Sclerosis (most common cause)
- **Congenital causes**
  - Friedreich's ataxia
  - DIDMOAD—the association of **D**iabetes **I**nsipidus, **D**iabetes **M**ellitus, **O**ptic **A**trophy and **D**eafness
  - Leber's optic atrophy.

PAPILLEDEMA

Examiner: **What is the earliest fundoscopic finding in papilledema?**

Candidate: Loss of venous pulsation.

Examiner: **What are the other fundoscopic findings in papilledema?**

Candidate:

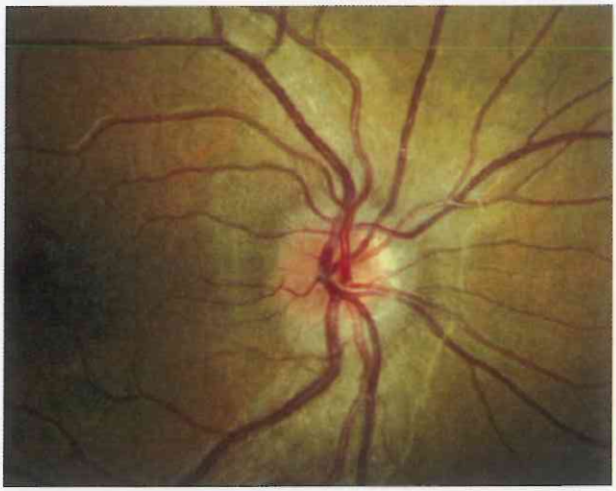

FIGURE 6.5 Papilledema

- Blurring of the disc margins
- Hyperemic disc
- Flame hemorrhages and cotton wool spots inside the disc due to infarction of the nerve fibers **(Figure 6.5)**.

Examiner: **What are the causes of papilledema?**

Candidate:

- Space occupying lesion in the brain (tumors, abscess, hematoma, granulomas, etc.)
- Choroid plexus papilloma with increased CSF production
- *Decreased cerebrospinal fluid (CSF) absorption:* Meningitis, Guillain Barré syndrome, thyroid eye disease
- *Obstruction of venous flow:* Cerebral vein thrombosis and occlusion of neck veins
- Cerebral edema (toxins, vascular stroke, trauma, hypoxic encephalopathy, CO_2 retention)
- *Unknown mechanism:* Idiopathic intracranial hypertension.

RETINITIS PIGMENTOSA

Important clue: If you see trabecular bone-like lesions that are pigmented in the periphery of retina, it is retinitis pigmentosa (RP) (*pieces of the femoral head in the retina*) **(Figure 6.6)**.

Examiner: **What are the typical symptoms caused by RP?**

Candidate: The most common symptoms include difficulty seeing at night (because the rods lie more peripherally) and a loss of peripheral vision (tunnel

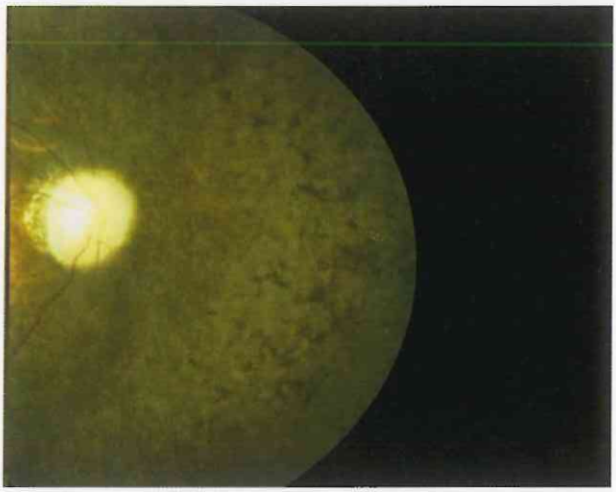

FIGURE 6.6 Retinitis pigmentosa

Examiner: **What is the prevalence of RP?**

Candidate: 1 in 5000.

Examiner: **What is the mode of inheritance in RP?**

Candidate: Autosomal dominant, autosomal recessive or X-linked.

Examiner: **What conditions are associated with RP?**

Candidate:

- Idiopathic
- Laurence-Moon-Bardet-Biedl syndrome (mental retardation, hypogonadism, obesity, short stature, polydactyly, deafness and renal cysts)
- Kearns-Sayre syndrome (ophthalmoplegia, ptosis and heart block)
- Refsum's disease (cerebellar ataxia, peripheral neuropathy and hearing loss)
- Usher syndrome
- Abetalipoproteinemia.

Examiner: **What is the prognosis for RP?**

Candidate: RP progresses gradually from the periphery of the retina towards the center. Most patients with RP are legally blind by age 40.

Examiner: **Do you know any recent treatments that improve the vision in RP?**

Candidate: The Argus II Retinal Prosthesis System (FDA approved). The system has three parts: a small electronic device implanted in the eye, a tiny video camera attached to a pair of glasses, and a video processing unit that is worn or carried by the patient. The video camera attached to the patient's glasses captures images of the surrounding area. The video processing unit then processes these images

the retina. This electrical stimulation of the retina is recognized by the brain as spots of light.

CENTRAL RETINAL VEIN OCCLUSION

Clue: Central retinal vein occlusion (CRVO) characteristically gives rise to diffuse retinal hemorrhages and dilated retinal veins **(Figures 6.7 and 6.8)**

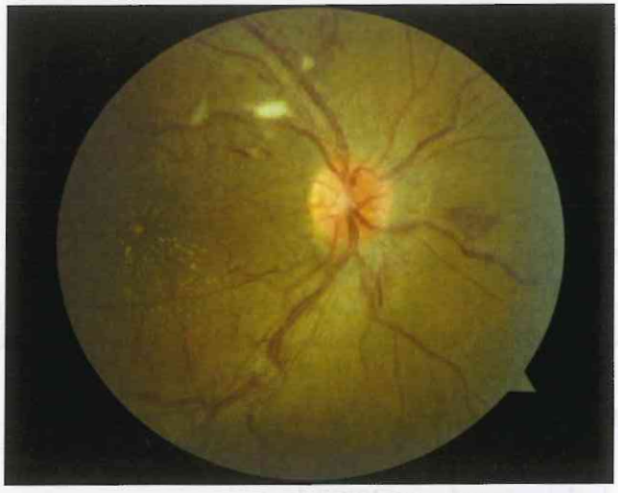

FIGURE 6.7 Central retinal vein occlusion

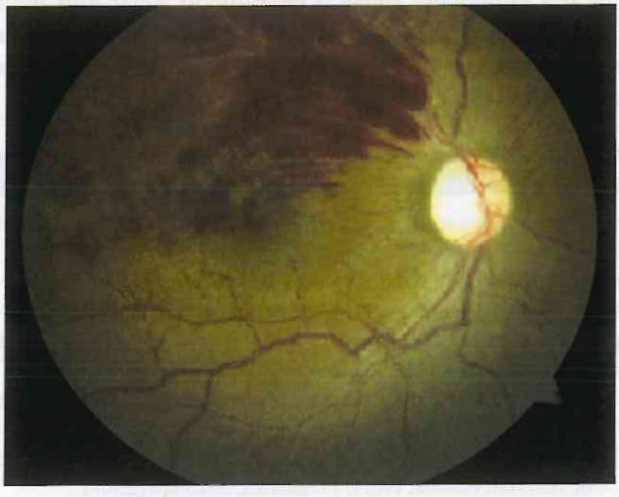

FIGURE 6.8 Branch retinal vein occlusion

Examiner: **What are the causes and risk factors of CRVO?**

Candidate:

- Age
- Diabetes
- Hypertension
- Smoking
- *Hypercoagulable states:* Factor V Leiden, Behçet's disease, polycythemia, etc.
- Vasculitis
- Glaucoma.

Examiner: **What are the complications of CRVO?**

Candidate:

- Neovascularization may lead to neovascular glaucoma and vitreous hemorrhage
- Macular edema
- Visual loss.

Examiner: **What is the prognosis in CRVO?**

Candidate: Prognosis depends on the initial visual acuity after occlusion. If the initial visual acuity is good the prognosis is good but if the initial visual acuity is worse than 20/200, it will remain at that level or worsen.

Examiner: **How would you manage this patient?**

Candidate:

- Intraocular injection of a anti-VEGF drug
- Intravitreal steroids dexamethasone triamcinolone to reduce macular edema
- Laser photocoagulation for neovascularization
- Search for the underlying cause. Check blood sugar and blood pressure and do a thrombophilia workup. Behçet's disease is easily forgotten by the candidates as a cause of CRVO or other kinds of thrombosis. Take a detailed history regarding recurrent mouth ulcers or joint pains.

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