1 Diagnosis: the art of being a doctor

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CORE SKILLS AND KNOWLEDGE

Being a doctor is a privilege; our patients allow us to share in their troubles and triumphs, and place their faith in our judgement and skill.

In the 21st century, professional roles and responsibilities are rapidly changing, and so defining what it means to be a doctor is difficult. Healthcare is provided by teams, not individuals, and other healthcare professionals now perform tasks in assessing and managing patients that were previously carried out exclusively by doctors. The use of artificial intelligence, which applies vast, unsorted clinical and academic datasets to produce algorithms that can inform diagnostic and management questions, provides a fundamental challenge to the historically crucial role of doctors in these areas.

While the process of diagnosis will be increasingly augmented by artificial intelligence, it is unlikely ever to be replaced by it because at the centre of any diagnostic conundrum is a human being. Working out what is wrong with a patient involves not just technical skills, such as the assimilation of information, interpretation of data and use of clinical reasoning to reach a diagnosis. It also requires compassion, empathy, trust, respect and humour: the stuff that makes up a relationship between human beings. And it brings into play an ethical code developed to ensure that the great power of medical knowledge is used for the good of individual patients and of society as a whole.

Once a diagnosis is made, the human tasks continue. Doctors tailor their explanations to the needs and understanding of their patients. They negotiate a management strategy and share clinical decision-making with the person under their care. However medicine evolves in the future, at the centre of everything we do must remain the huge privilege and pleasure of caring for, supporting, empowering and helping people so that their health and social needs are optimally met.

DIAGNOSIS IN THE CLINICAL CONSULTATION

In many patient interactions, the forming of a diagnosis stands as a turning point in the therapeutic journey: the focus shifts from the gathering of information and the performance of tests to planning of treatment and discussion of outcomes. Forming a diagnosis is a complex process, which always starts with taking a history. The initial patient story is first developed into a set of problems, which in turn becomes a list of differential diagnoses. Data-gathering, further information, initial treatment and the passage of time help to form and confirm a definitive diagnosis.

Fig. 1.1 shows how diagnosis is often an iterative process, in which information is gathered, interpreted and integrated to form a working diagnosis, and then communicated and acted on through treatment; all the time the working diagnosis is refined or revised using newly gathered information, including the patient's response to treatment.

THE MEDICAL CONSULTATION

The initial interaction – forming a rapport

First impressions matter. A clinical interaction that begins badly often runs into problems later on. Patients need to be able to share their most intimate and worrying problems and this will be impossible if they cannot develop confidence and trust in their clinicians. The clinician should always ask, 'How would I wish my own doctor to behave?' The physical environment can also be improved to help the interaction go well: always ensure patients' privacy, dignity and comfort, treating them and their carers with respect.

A warm greeting is always worthwhile. Introduce yourself (and any other health professionals present), explaining your role and the purpose of the encounter. A healthcare system is generally busy and confusing, so help patients understand where they are in their journey through it. The 'Hello, my name is ...' campaign, supported by the Department of Health in the UK, highlights the importance of basic introductions on the part of staff members (**Box 1.1**). Further

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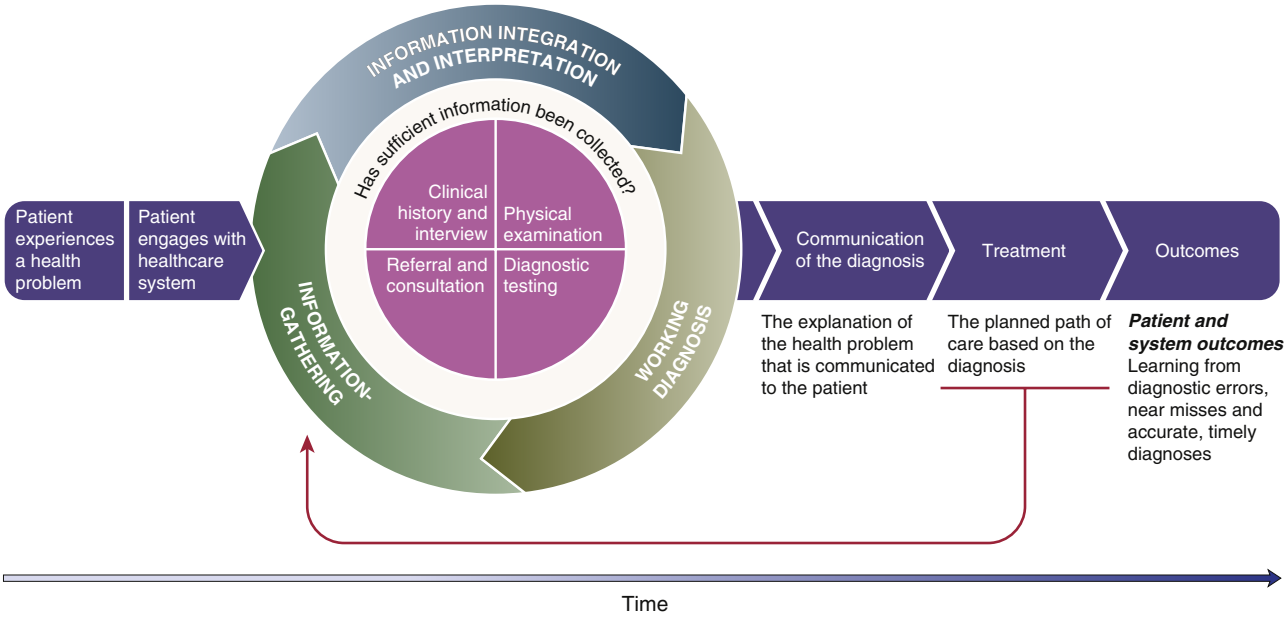

Fig. 1.1 A conceptualization of the diagnostic process. *(From National Academies of Sciences, Engineering, and Medicine.* Improving Diagnosis in Health Care. *Washington, DC: The National Academies Press; 2015. https://doi.org.10.17226/21794.)*

i Box 1.1 The 'Hello, my name is ...' campaign

In 2011, geriatrician Dr Kate Granger was diagnosed with metastatic sarcoma, from which she eventually died in 2016. Noticing that many of the healthcare staff treating her during her illness failed to introduce themselves, she created the 'Hello, my name is ...' campaign. Describing why an initial introduction is so important, she said, 'I firmly believe it is not just about common courtesy, but it runs much deeper. Introductions are about making a human connection between one human being who is suffering and vulnerable, and another human being who wishes to help. They begin therapeutic relationships and can instantly build trust in difficult circumstances.'

The campaign encourages healthcare staff to greet and introduce themselves to patients and encourages the wearing of easy-to-read name badges like this one.

comments expressing empathy, support or care for the patient at this point can be very helpful in ensuring that a good rapport is established, before proceeding to talk about the problem that has led the patient to seek medical help (**Box 1.2**).

Information-gathering

Once a rapport has been established, the consultation is generally divided into three parts: ***history***, ***examination*** and ***investigations/management***. Depending on the context and the acuity of the situation, these may occur in isolation, follow on from one another or, in severely ill patients, happen synchronously.

The history

In the construction of an accurate diagnosis, nothing is more important than taking a full history from the patient. It is estimated that up to 80% of the diagnosis can be made on the basis of a careful history alone. In addition to information-gathering, eliciting the history is key to the therapeutic relationship. If it goes well, the patient knows they have been listened to, has had an opportunity to convey all of their concerns, and knows that the clinician cares for them and will act as their advocate. This builds mutual trust and respect, and helps the patient to undergo invasive or intimate procedures and adhere to therapeutic interventions in the future.

A meeting of two experts

It has been said that a medical consultation is a meeting of two experts. A good consultation is based on mutual respect, rejecting a traditional paternalistic view of medicine ('doctor knows best'), and assisting joint exploration of the biomedical and the patient perspectives on the problem. This will lead to shared understanding, where clinician and patient jointly grasp what is wrong, what impact it is having on the patient's life, what the patient expects from medical intervention, and which options would be best for investigating and treating the problem (see **Fig. 5.1**).

The golden minute

At the start of the consultation the clinician should avoid interrupting the patient for as long as possible. Patients often mentally rehearse a script relating to the symptoms or problems they wish to describe. An early interruption may throw them off course and cause them to forget key points, leaving them feeling dissatisfied with the interaction. The term '***the golden minute***' has been coined to encourage clinicians to allow patients to tell their story uninterrupted for at least 1 minute. This gives them time to describe their symptoms as they have experienced them, including information that might be missed if the clinician jumps in too soon with closed, focused questions.

Allowing the patient freedom to tell their own story aids clinicians in achieving their primary objective of making a diagnosis, but ultimately forms and reinforces the therapeutic relationship and increases both patient and clinician satisfaction. **Box 1.3** summarizes some techniques that can be used to help patients share information in a way that is accurate and comprehensive. Some will

Box 1.2 Good practice for using the medical history to build a therapeutic relationship

- Allow the patient to tell their story, without jumping in prematurely with questions.
- Ask the patient specifically about their ideas, concerns and expectations (ICE):
  - ***Ideas:*** What do they think might be going on? Have they done any reading about their symptoms or asked anyone they know?
  - ***Concerns:*** Are they feeling anxious or worried about their symptoms? What is causing them concern? Are there any particular areas where their symptoms might be making life difficult?
  - ***Expectations:*** What are they hoping for from this consultation?
- Try to develop an understanding of who the patient is as a person. Where, and with whom, do they live? What is their occupation? What things do they enjoy? Moments of human connection, such as a shared interest in a place or activity, can be powerful in building a relationship between clinician and patient.
- Try to convey empathy and concern, reinforcing to the patient that you are their advocate and will do your best to help them.

Box 1.3 Strategies in history-taking

- ***Begin with open questions*** ('Could you tell me more about the pain?'): do this before moving to closed questions to help rule certain key problems in or out ('Did the pain get worse after eating?').
- ***Emphasize your active listening***: maintain eye contact, nod, acknowledge key points, and respond to comments the patient might make that are humorous or sad.
- ***Respond to the patient's body language***: note whether the patient becomes distressed or embarrassed. If so, acknowledge this and look for ways to address it.
- ***Empathize***: try to show the patient that you care about what they are going through. Put yourself in their shoes: how might they be feeling? Communicate this: 'Thanks for sharing this – it must be difficult not knowing what is going on.'
- ***Summarize***: run through what the patient has told you, to make sure nothing has been missed out.
- ***Signpost***: explain what you have just covered and what you are now going on to explore, and why.
- ***Use plain English***: avoid medical jargon or complicated vocabulary, unless it is clear that the patient is able to understand this.

Box 1.4 A structured approach to information-gathering in the medical history

- Presenting complaint: why has the patient sought medical advice?
- History of the presenting complaint: further information about the patient's main problem
- Past medical and surgical history
- Drug history and allergies
- Family history
- Social history: information on the patient's present living arrangements and relevant risk factors

Box 1.5 Using the time course of symptoms to suggest an underlying cause

Onset of symptoms	Possible causes
Immediate (seconds to minutes)	Vascular – thrombotic or embolic Anatomical – e.g. perforation of a viscus Electrical – e.g. dysrhythmias, seizures
Hours to days	Bacterial or viral infections Inflammatory and autoimmune diseases
Weeks to months	Malignant disease Inflammatory and autoimmune diseases Chronic infections, e.g. mycobacterial
Months to years	Degenerative conditions Fibrotic diseases

have given a lot of thought to their symptoms and come with a well-thought-through story; others may not have reflected much on what has been going on, and key information may need to be drawn out by sensitive questioning.

Structuring the medical history

Over the last 150 years a formal structure for the recording of the patient's history has evolved. This has several subsections (**Box 1.4**). While facts may be recorded in this very stylized manner, the patient will rarely, if ever, present them in this structure. It is up to the interviewing clinician to assimilate and interpret the information fully and to form a considered diagnostic narrative, so that when it is presented to others, either verbally or in written form, sense can be made of the diagnostic reasoning and conclusions.

History of the presenting complaint

The aim is to provide a thorough account of the symptoms that led the patient to seek medical attention. It is vital to listen closely to how the symptoms are described and not to miss any clues that can be followed up with direct questioning. For each symptom the patient presents with, additional questioning should be used to identify:

- ***Time course***: When did the problem begin? Does it come and go? Is there anything that triggers it? Is there any variation in the symptoms during the day or night? Has the patient ever had anything like this in the past? Establishing the pattern in which symptoms have developed is often one of the most helpful parts of the history in helping to form a diagnosis (**Box 1.5**).
- ***Associated symptoms***: What else has the patient noticed? Begin with an open question, and then proceed to asking about the presence or absence of relevant symptoms that may help to determine the cause of the problem.
- ***Severity, site, radiation and character*** of any pain: How would the patient rate it on a scale from 1 to 10? Do they describe it as tight, dull, electric or burning? Is it getting worse, staying the same or starting to improve?
- ***Responses***: What has the patient done about the symptoms? Have they sought medical advice or used medication that they have at home?

The 'Clinical skills' sections at the beginning of many of the chapters in this book present additional questioning techniques relevant to particular medical specialties. At this point in the history, asking questions to form a review of systems can be valuable in eliciting further symptoms that the patient may not have mentioned or not thought relevant (**Box 1.6**).

Past medical and surgical history

This should include all significant medical conditions, including hospital admissions, long-term conditions, life-threatening or life-changing conditions, and important investigations, procedures and therapeutic interventions (operations, endoscopies, biopsies and significant courses of treatment such as chemo- or radiotherapy).

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Box 1.6 Review of systems

Conducting a brief but comprehensive 'review of systems' may be a particularly useful schema when the patient has non-specific symptoms, e.g. weight loss, tiredness and weakness, or when someone reports 'I just don't feel right, doctor'.

Respiratory
Cough, sputum (volume, frequency, consistency, colour, offensive taste or smell), haemoptysis (volume, frequency, consistency, colour, freshness, altered nature, clots), shortness of breath, exercise tolerance, orthopnoea, wheeze, chest pain

Cardiovascular
Chest pain, shortness of breath, cough, sputum, orthopnoea, swelling of ankles (peripheral oedema) or abdomen (ascites), paroxysmal nocturnal dyspnoea, palpitations

Gastrointestinal
General – normal and present weight, appetite, oral intake
Upper – dysphagia (level: high – base of neck, mid and lower chest), consistency of food tolerated/not tolerated), dyspepsia, odynophagia, upper abdominal pain, early satiety, nausea/vomiting (volume, frequency, consistency – unaltered food (regurgitation), altered food, blood), haematemesis (volume, frequency, consistency, colour, freshness, altered nature, clots)
Lower – lower abdominal pain, altered bowel habit, constipation, diarrhoea (volume, frequency, consistency, colour), blood per rectum, mucus per rectum, anal pain

Hepatobiliary and pancreatic
Jaundice, associated pain, pruritus, symptoms of encephalopathy, abdominal swelling (ascites)

Renal
Urine (frequency, volume, colour, offensive odour), dysuria, haematuria (volume, frequency, colour, freshness, clots), symptoms of bladder outflow tract obstruction (hesitancy, frequency, small volume, terminal dribbling)

Musculoskeletal
Bone pains, back pain and stiffness, joint stiffness, swelling, pain, erythema, patterns of joints involved, muscular pain, weakness, acute pain suggesting pathological or fragility fractures

Dermatological
Rashes, blisters, ulcers

Endocrine
Diabetic symptoms and complications, sexual function, menstruation, symptoms of thyroid dysfunction

Neurological
Seizures, muscle weakness, involuntary movements, loss of sensation, altered gait, speech and swallowing dysfunction

Ophthalmic
Eye pain, redness, dryness or grittiness, changes in vision, flashing lights

Ear, nose and throat
Changes in smell, taste or hearing, pain in ears, nose, throat or sinuses, nasal discharge or crusting

Haematological
Easy bleeding or bruising, tiredness, lymph node swelling, abdominal fullness (splenomegaly)

Mental health
Mood (suicidality when relevant), anxiety, altered perceptions (hallucinations), abnormal beliefs (delusions)

Genitourinary
Urethral or vaginal discharge, pain or itching, pain during sexual intercourse, sexual function, in women – menstrual cycle, use of contraception, history of pregnancies and childbirth

Box 1.7 Five questions for the patient with a long-term condition

1. ***Diagnosis*** – HOW, WHY, WHERE and WHEN was your condition first diagnosed?
2. ***Progression*** – How has it progressed since?
3. ***Control*** – How do you monitor your condition? Which healthcare professionals are involved with your care? When did you last have a check-up? What medications do you take? What are your biggest challenges in controlling your condition?
4. ***The good and the bad*** – What is the BEST and WORST your condition has ever been? How does it have an impact on your life?
5. ***Today/recently*** – How have you been? If unwell, have you ever been this unwell before? What happened when you were last this unwell?

If the patient has had any surgical interventions a comprehensive anaesthetic history should be sought and recorded. Five good questions to ask about chronic conditions are listed in **Box 1.7**.

Drug history

Document all medications the patient has been taking, including prescribed, over-the-counter and herbal or traditional treatments. Record when each was started, along with the dosing regimen; ask about side-effects and adherence to treatment. When you are unsure or ignorant of a drug, it is essential to research and record the class, common side-effects and interactions. Medication error is a common cause of morbidity and mortality, and ignorance is no defence. Ask about and record ***drug allergies***, including the timing and nature of any reactions.

Family history

This is particularly relevant when assessing younger patients or when the differential diagnosis includes possible genetic conditions. It is essential to record the structure of the patient's family in detail, including the patient's and parents' siblings, any 'half-siblings' (genetically related to only one of the patient's parents), and where relevant, a history of consanguinity. Once this is completed, confirm which of these relatives have been affected by a given condition or by premature death (see p. 13).

Social history

The social history has two key purposes:

- ***Establishing whether there are any environmental factors*** that may be causative or exacerbating the patient's symptoms. Always ask about housing, occupation, tobacco smoking, alcohol intake, and use of recreational and illicit drugs. Where relevant, draw up a travel history, including animal and insect bites, a sexual history (if a sexually transmitted disease, including HIV infection, is suspected), and hobbies, leisure activities and pets.
- ***Understanding more about the lifestyle*** of the patient. Where do they live? How active are they? Are they limited in any daily activities by physical or mental health problems? Do they have informal or formal carers? If so, how often do the carers attend and what do they do for the patient?

Physical examination

The physical examination, including any objective observations, should be used to confirm or refute the initial diagnosis/diagnoses made from the history. It is a key part of all medical interactions but its duration and extent will be guided by the patient's history and the acuity of the presentation.

Even in the era of complex technological investigations a careful physical examination performs a number of key functions, including:

- Providing objective evidence (physical signs) to complement subjective evidence (symptoms) from the history. Sometimes a

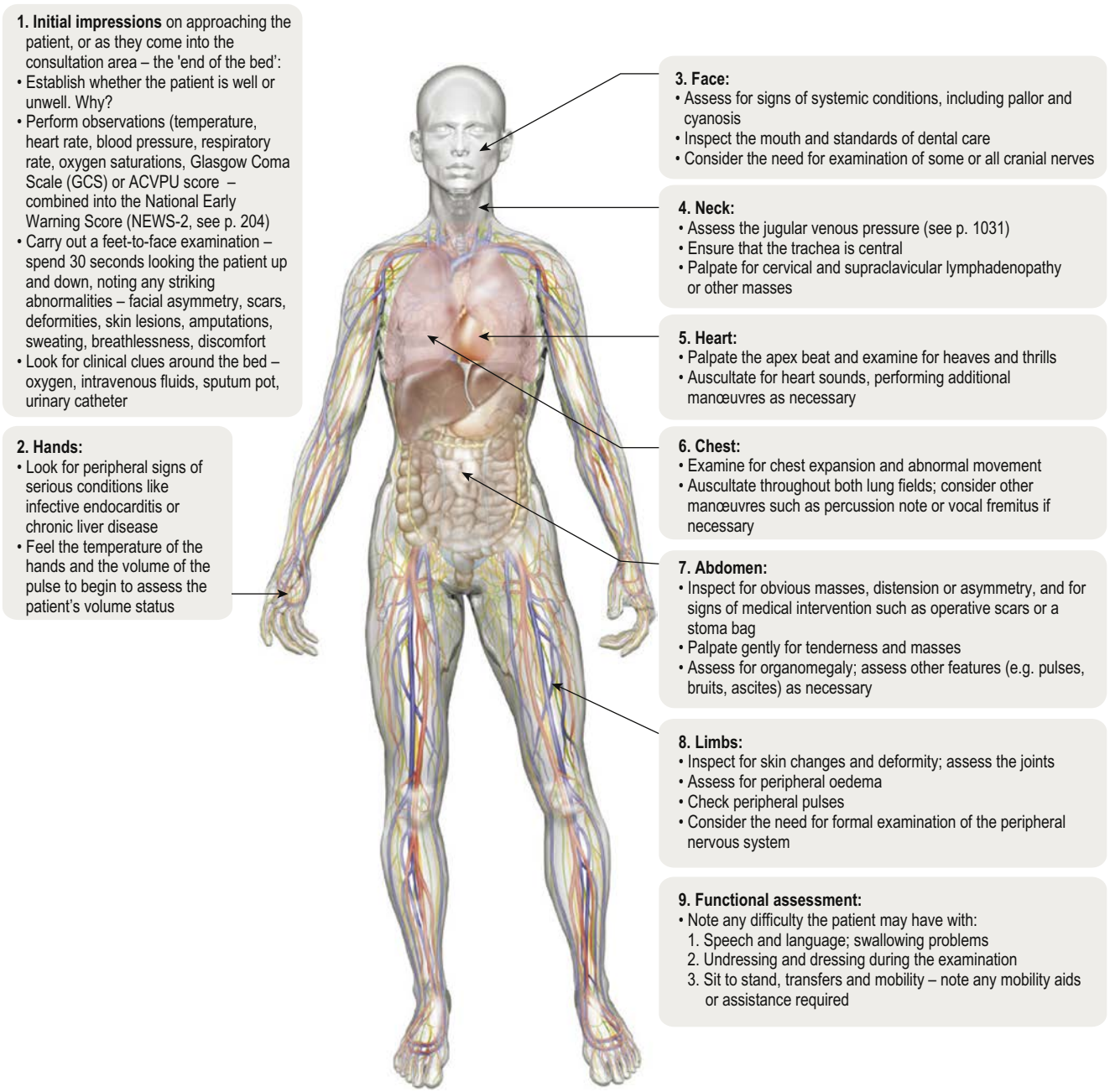

Fig. 1.2 **A basic approach to clinical examination.**

firm diagnosis can be made almost solely on the basis of examination findings, such as in a number of skin disorders.

- Assessing the severity or extent of problems.
- Identifying unexpected findings that patients themselves have not noticed.
- Building rapport with patients. The value of performing a physical examination can be significant in reinforcing to patients that they have been dealt with thoroughly and compassionately.

Typically a general assessment will be made as the history is being elicited (Is the patient well or unwell? Are there any obvious clinical signs?). This will be aided by a set of formal observations (blood pressure, heart rate, oxygen saturation, respiratory rate, level of consciousness, capillary blood glucose and temperature). A more detailed assessment is then carried out, including a 'general examination' (hands, upper limbs, face and neck), examination of the likely affected system(s) as suggested by the history, and finally a wider examination of other organ systems.

Fig. 1.2 outlines a typical general and systematic examination routine suitable for use in patients presenting with a wide range of medical conditions. The 'Clinical skills' sections at the beginning of many of the chapters in this book offer tailored versions of this basic routine relevant to patients with specific types of complaint.

Discussion and negotiation around investigations

In the modern era, there are a huge number of investigations available to the clinician, ranging from simple bedside tests such as spirometry or urine dipstick analysis, through to complex radiological

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imaging and invasive procedures such as endoscopy or angiography. Some tests may combine both diagnostic and therapeutic potential. Choosing appropriate and cost-effective interventions that maximize diagnostic yield, while minimizing the burden on the patient and the cost to providers, can be challenging, and each chapter in this book will provide guidance in specific contexts.

For any test that is being considered, a number of questions are relevant:

- ***What question will this test help to answer?*** Only the most basic of tests should be performed routinely. For all others, it is helpful to have clear diagnostic questions in mind, and often specialists, such as diagnostic radiologists, can help with choosing the most appropriate investigation to answer the relevant question in a particular patient context.
- ***What is the sensitivity and specificity of the test?*** A highly sensitive test will correctly identify a high proportion of patients with a given disease (true positives); a highly specific test will correctly identify a high proportion of those who do not have the disease (true negatives). For example, in the diagnosis of venous thromboembolism (VTE, see Ch. 29), measurement of serum D-dimer has a high sensitivity (a positive result picks out almost all patients with VTE) but a low specificity (many of those with a positive D-dimer do not have VTE). Since D-dimer measurement is cheap, it is a useful screening test (because a negative test effectively rules out VTE); a positive test is followed up by a high-sensitivity test, such as venous Doppler ultrasound or computed tomography pulmonary angiography (CTPA).
- ***What are the risks of the test to the patient?*** All ionizing radiation carries a small risk of future malignant disease, and invasive procedures may cause bleeding, infection or injury to internal organs. These dangers, along with the benefits of the investigation result, need to be discussed with patients to help them make a good decision.
- ***How much certainty is needed?*** Where highly burdensome treatment is contemplated (such as surgery or chemotherapy for cancer), it is usually necessary to obtain a formal histological diagnosis by tissue biopsy before starting treatment. If such treatment would not be appropriate, then undergoing invasive diagnostic procedures may not be appropriate either.

Further reading

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CLINICAL DIAGNOSTIC REASONING

Forming a diagnosis involves a complex process of reasoning. Large amounts of information gathered from the history, examination and available investigation results need to be assimilated and synthesized into a working diagnosis. Each piece of evidence should be weighed according to the degree of confidence you have in its accuracy, and no significant findings should remain unexplained. If you, as the treating clinician, are unable to make sense of the information presented to you, you should be humble and insightful enough to seek help from others.

Models of diagnosis

A traditional model of medical diagnosis suggests that a clinician should begin by considering all possible causes of a particular presenting symptom, and use information gathered from the diagnostic process to include and exclude likely causes gradually, using probabilistic reasoning, until only one remains (**Fig. 1.3A**). According to the insights of the Nobel Prize-winning psychologist and economist Daniel Kahneman, this is 'type 2 thinking', and the kind of thinking we like to imagine that we carry out all the time: logical, deductive and rational.

An alternative model suggests that diagnosis proceeds instead primarily by pattern recognition, the kind of 'type 1 thinking' that allows humans to make quick judgements of new situations by comparing them with similar situations encountered in the past. In this form of diagnostic reasoning, clinicians rapidly compare the patient presenting to them with many other patients they have seen previously, subconsciously drawing on similarities and differences to form an initial impression that is then tested as further information becomes available (**Fig. 1.3B**).

In reality, a combination of these two approaches generally occurs: an initial rapid impression is formed, chiefly by type 1 thinking, which is subsequently revised by the slower, more reason-based type 2 thinking where the initial diagnosis proves inadequate. This 'back to the drawing board' approach, which draws on the strengths of both types of thinking, has been suggested as the best model for safe diagnosis – where a working diagnosis is continually re-evaluated as new evidence becomes available (**Fig. 1.3C**).

Type 1 thinking is prone to bias (see later), where it is assumed that everything new must be similar to things seen before. It is crucial for doctors to re-evaluate an initial diagnosis actively in situations where things 'don't quite fit' rather than persisting with a hastily formed assumption that may prove incorrect.

Diagnostic error and patient safety

It has been estimated that 10–15% of medical diagnoses are wrong. Misdiagnosis or delayed diagnosis can be the cause of significant patient harm, including adverse effects from unnecessary treatment or failure to receive an appropriate timely intervention. While misdiagnosis may occur as a result of inadequate clinical knowledge or (particularly in resource-poor settings) through a lack of diagnostic resources, it also arises as a direct result of one or more cognitive biases. **Box 1.8** lists a number of common cognitive biases, applied to the field of diagnostic reasoning, which together form a key avoidable source of medical harm, termed 'human error'.

Strategies for avoiding bias

Various strategies can be employed in order to reduce the chances of making an incorrect diagnosis:

- ***Adopting an iterative approach***, in which all previous diagnoses are subjected to appropriate re-evaluation, especially if evidence appears that brings them into question.
- ***Team discussions***, where all team members are empowered to challenge the reasoning of more senior clinicians. Multidisciplinary team meetings bring clinicians from different specialties together with allied healthcare professionals to ensure that all relevant

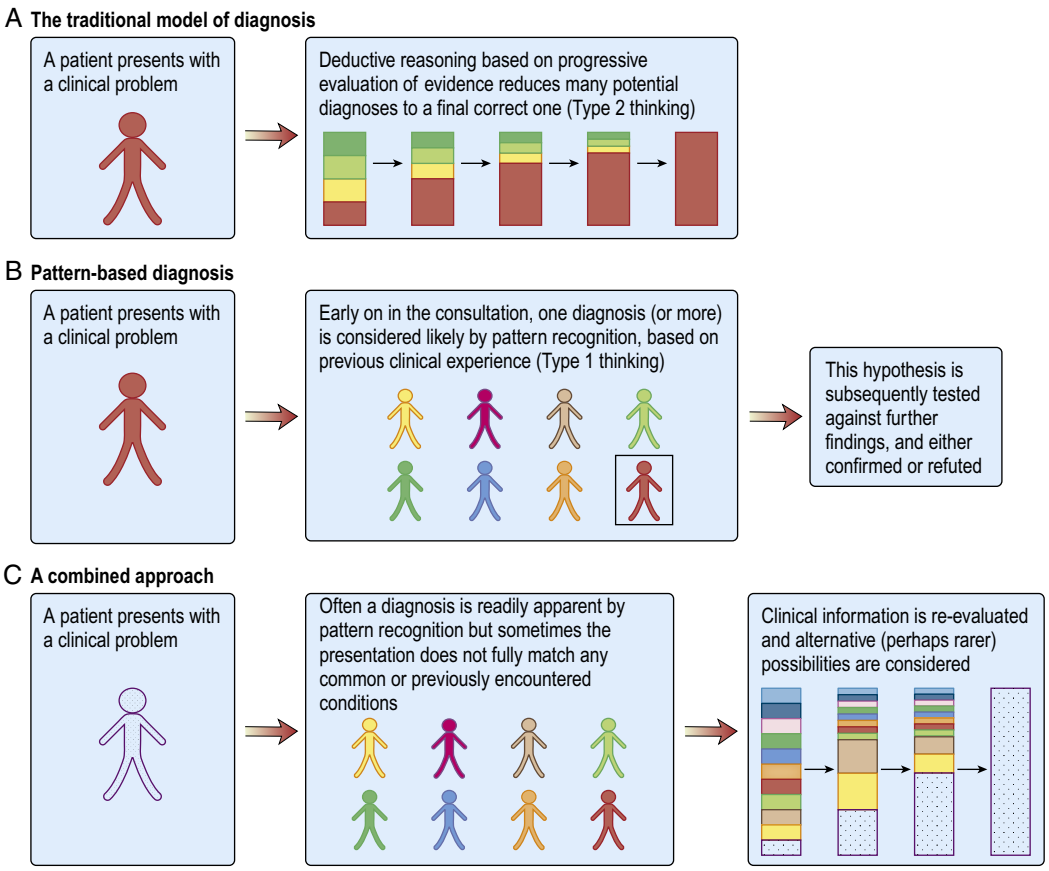

Fig. 1.3 Approaches to diagnosis. **(A)** The traditional model of diagnosis. **(B)** Pattern-based diagnosis. **(C)** A combined approach.

knowledge and expertise is shared in a collaborative environment. Leaders of healthcare teams have a particular responsibility to 'flatten the authority gradient' and empower more junior members to contribute knowledge and ideas to team discussions.

- ***Diagnostic criteria and guidelines***, where robust standards are outlined to ensure that potential diagnoses are correctly assigned and similar conditions are differentiated from each other.
- ***Considering patient problems in language devoid of assumption***, as far as possible, seeking to describe a patient's problems succinctly and objectively without recourse to previous diagnostic reasoning.
- ***Using lists of disease classes to avoid jumping to conclusions***: simple *aides-mémoire* encourage clinicians to consider, for example, malignant, infectious, vascular, metabolic, inflammatory and degenerative causes of a particular problem.

Levels of diagnostic depth

A clinical impression is formed by taking a comprehensive history and undertaking a relevant physical examination. Drawing on the information gathered, clinicians may typically assign a widely understood umbrella term to categorize a collection of signs and symptoms into a clinical entity: for example, 'acute coronary syndrome', 'delirium', 'upper respiratory tract infection' or 'sepsis'. Some patient presentations do not fall neatly into boxes, and so sticking with a narrative description of the patient's problems is appropriate in some cases. However, naming a clinical impression in a few simple words is often vital in moving towards a final diagnosis.

An impression formed at the end of a clinical consultation can often be refined by simple, quick and relatively non-invasive tests; but it might require complex, expensive or potentially hazardous investigations to produce a definitive diagnosis. Likewise, additional tests might be needed in order to demonstrate the extent, severity or treatment-responsiveness of the condition (Fig. 1.4).

Often, it is appropriate to stop at the level of a syndrome or clinical impression rather than continuing with investigations to demonstrate a precise histopathological cause. This might be the case if:

- the patient is satisfied with this level of explanation and declines further diagnostic work-up
- the problem is mild or self-limiting
- further investigation is unlikely to yield a specific cause, e.g. where a patient with acute but resolving diarrhoea or vomiting is diagnosed with likely viral gastroenteritis without a specified pathogen
- further investigation is unlikely to influence management, e.g. if none of the specific pathological processes that cause the condition is amenable to active management
- treatment is possible, but the patient is very frail and the proposed investigations or treatments are felt, after discussion with the patient, to be inappropriate.

The role of watchful waiting

All diseases have a natural history. Some progress inexorably, some are self-limiting and some relapse and remit. Where there is diagnostic uncertainty, there can be a role for waiting to see how events develop before reaching for a diagnostic label. Indeed, this is sometimes

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Box 1.8 Cognitive biases leading to misdiagnosis

- ***Anchoring***: Relying too heavily on a piece of information offered early as a potential explanation. For example, an elderly patient is sent to hospital by their GP with a 'suspected chest infection' and the doctor in the emergency department fails to consider other possible causes of breathlessness, such as heart failure or pulmonary embolus.
- ***Availability***: Assuming that because certain explanations spring easily to mind, they are likely to be correct. For example, a doctor assumes that a patient presenting with acute kidney injury is dehydrated and administers intravenous fluids, even in the absence of evidence to support this, because of unfamiliarity with less common causes of this presentation, such as renal parenchymal disease.
- ***Framing***: Making decisions in different ways, depending on whether a choice is presented in positive or negative terms. For example, a doctor may respond differently to a radiology report of an incidentally discovered adrenal mass that is reported as 'probably benign', compared to the same mass if the report reads 'unable to exclude malignancy'.
- ***Optimism***: Assuming that negative outcomes will occur at a lower rate than they really do. For example, a junior doctor may fail to call for senior help with a clearly deteriorating patient because of an unrealistically positive expectation that the interventions they have instituted will improve the situation.
- ***Recency bias***: Remembering most easily things that have happened most recently. For example, a doctor may be distracted away from a common diagnosis by a much rarer one, which was covered in a recent teaching session they attended.
- ***Substitution***: An easier (and related) question is solved in place of a more difficult one. For example, a patient with a history of intravenous drug use presents with worsening headache, although a CT scan of their head is normal. Rather than address a computationally difficult question (why does this patient still have a headache?), a simpler question is solved (might they simply be seeking opiate analgesia?).
- ***The sunk-cost fallacy***: Continuing to invest in a failing idea because of significant prior investment. For example, a cardiologist recommends that a patient undergo high-risk elective valve replacement, despite colleagues suggesting conservative management because of the patient's co-morbidities. The procedure is unsuccessful and the patient suffers significant complications, but the doctor subsequently recommends a second procedure rather than conservative management, in order to try to salvage something from the situation.

Adapted from Kahneman D. *Thinking, Fast and Slow*. New York: Farrar, Strauss and Giroux; 2012.

necessary, as the diagnostic criteria for some diseases stipulate that symptoms must have been present for a certain length of time before a diagnostic label can be assigned. For instance, in multiple sclerosis, where an initial presentation with symptoms suggestive of a demyelinating illness is usually termed a 'clinically isolated syndrome', some patients will suffer no further episodes while others will progress to multiple sclerosis. In other cases, it is wise to defer risky or burdensome investigations until the clinical course of the disease makes it clear that such investigations are justified. Watching and waiting can be a valid approach only if, firstly, the patient's clinical condition allows it; and secondly, it does not involve withholding treatment that would otherwise be of benefit (**Box 1.9**). Discussion with the patient is crucial.

When not to investigate

Decisions about whether and how to pursue a formal diagnosis are often complex and should be made in conjunction with the patient. Everyone is different and individuals differ in their willingness to tolerate uncertainty, with some wanting to seek a firm diagnosis at all costs, and others happy to accept a presumed diagnosis and run the risk that this may be wrong. Clinicians can guide patients in making complex decisions by helping them understand the likely or possible outcomes of different decisions; often, offering 'best- and worst-case scenarios' for different potential courses of action is helpful. In general, investigations should be avoided where:

- the patient is too frail to derive any benefit from confirming a diagnosis
- the patient agrees to have an initial investigation, but will not accept intervention if the result is positive, e.g. the patient agrees to a myocardial perfusion scan but would not want to undergo subsequent coronary angiography or stenting
- the treating clinician feels that investigation is not deemed to be in the patient's best interests – patients can refuse investigation or intervention, but cannot demand it.

Diagnostic criteria

Some conditions are diagnosed with a single pathognomonic investigation result: for example, the presence of urate crystals on microscopic examination of synovial fluid is diagnostic of gout. In other situations, confirming the diagnosis is far more complex and may require a combination of symptoms, physical signs and investigation results. For example, according to the 2018 diagnostic criteria for systemic lupus erythematosus, published jointly by the American College of Rheumatology and European League Against Rheumatism, patients need to score ten or more points from different domains, including symptoms, signs, haematological and immunological blood tests, and histology (see p. 440).

Diagnostic criteria have a range of functions beyond treating individual patients, including a role in public health (in the compilation of statistics for monitoring trends in the incidence and distribution of diseases), research (to allow study of diseases and treatments in well-defined disease populations), and remuneration or reimbursement (in many health systems, payment to healthcare providers is on the basis of diagnostic codes assigned to patients receiving care). The *International Statistical Classification of Diseases* (ICD), now in its 11th edition, is produced by the World Health Organization in order to provide a standardized set of coding and diagnostic criteria across the world. Although useful at a population level, and for the research and administrative purposes described, these criteria are rarely used in routine clinical practice.

Overdiagnosis

Overdiagnosis refers to a diagnosis that is correctly assigned on the basis of a screening programme, but is inappropriate because it is unlikely ever to cause harm to the patient in question. It is the inevitable result of population health screening and poses risks to patients because of the potential for unnecessary further diagnostic procedures, therapy, or insurance charges.

For example, a frail 93-year-old man with dementia is visited at home by his GP at the request of his daughter. A routine check reveals a heart rate of 66 bpm and a blood pressure of 168/96 mmHg. A diagnosis of hypertension is made and the elderly man is started on an antihypertensive. This diagnosis may be considered inappropriate on a number of grounds:

- While blood pressure rises with age, 'normal' blood pressure in a 93-year-old is not clearly defined; neither is an acceptable target blood pressure to guide intervention.

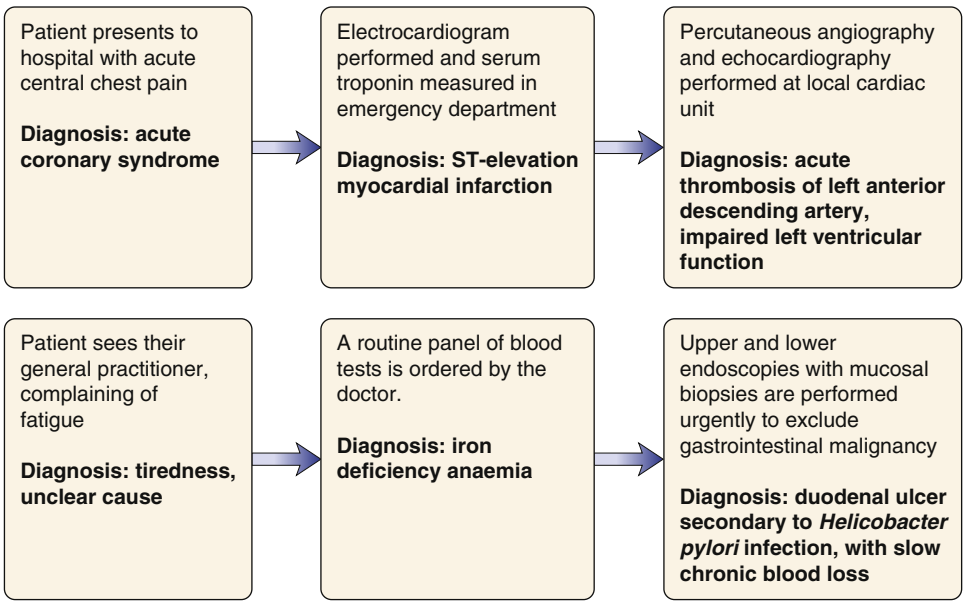

Fig. 1.4 Examples of different levels of diagnosis.

Box 1.9 The role of watchful waiting in diagnosis

- **A 77-year-old woman is undergoing CT scanning of the coronary arteries after an episode of chest pain.** A small (<10 mm) pulmonary nodule is incidentally noted. She is at high risk for lung cancer because of a long smoking history. In accordance with the British Thoracic Society's 'Guidelines for the Investigation and Management of Pulmonary Nodules', she is offered computed tomography screening at 3 months and 1 year. These reveal stable appearances with no increase in size, so she is reassured and discharged from further follow-up of this nodule.
- **A 34-year-old man is found to have abnormal liver function tests and is referred to a hepatology clinic.** A set of blood tests screening for common causes of liver disease, including viral hepatitis antibodies, are all normal. A liver biopsy is discussed with the patient, but because his liver function tests are not rapidly deteriorating, a decision is made in favour of watchful waiting. Two months later, his liver function tests return to normal ranges and remain normal after another 6 months. He is discharged from clinic without a diagnosis for his transient liver injury.
- **An 18-year-old woman with a history of presumed minimal change disease** as a child presents to the renal outpatient department with recurrent nephrotic syndrome. She is commenced on a loop diuretic and high-dose corticosteroids, but after 2 months remission has not been achieved. Because it is atypical for minimal change disease to fail to remit by this point, and because the diagnosis had never before been histologically proven, a renal biopsy is carried out. This revealed a different but related renal disorder: focal segmental glomerulosclerosis.

- A one-off high blood pressure measurement may have several confounding factors, including stress on being examined by the doctor. It would require further follow-up to corroborate this finding.
- Hypertension is known to increase the risk of cardiovascular disease and stroke significantly when left untreated for a period of years in younger patients, but it is far from certain that this pattern is seen in newly diagnosed hypertension in the elderly.
- All medications have side-effects: those of commonly used antihypertensive drugs include postural dizziness that may lead to falls, and increased susceptibility to acute kidney injury. These may significantly outweigh any benefits of treatment.

Some have argued that there is no such thing as overdiagnosis, only 'overtreatment'. Certainly, in this clinical scenario, little evidence exists to support the diagnosis or treatment. As any good geriatrician will confirm, there is art as well as science involved in intervening wisely in a complex situation such as this.

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COMMUNICATING A DIAGNOSIS

Towards the end of a clinical consultation, the clinician will need to explain their diagnostic reasoning to the patient, along with their plans for further investigation and intervention. Explaining and teaching is a key part of working in medicine, and this part of the consultation is crucial in laying the foundation on which decisions for the future can be made.

Explaining diagnoses to patients

Some patients will have little difficulty in understanding complex medical explanations; this should never be assumed, however. While it is wise to avoid patronizing patients with a good understanding, it is generally worth assuming that patients understand less rather than more about their condition, and explaining all parts of the reasoning process, at least in brief. Using a phrase like 'I know you understand lots about this already, but I'm just going to go over things right from the beginning so we both understand each other' can be a good way of giving simple explanations without causing embarrassment.

Explaining a patient's diagnosis is best done in chunks, waiting between each chunk to ensure they have understood (**Box 1.10**). It is wise to work chronologically through the symptoms, employing

Box 1.10 'Chunking' the diagnostic process

Beginning with a summary of the clinical presentation, the clinician explains their diagnostic reasoning before suggesting what should happen next, ensuring after each chunk that the patient understands and agrees.

- 'So you came in to see me because over the past month you've noticed pain, swelling and stiffness in your hands ...'
- 'When I examined your hands, I found that lots of the small joints in your fingers were hot, red and swollen, and I also noticed these new swellings behind your elbows you hadn't seen before ...'
- 'Now I don't know exactly what is going on, but when joints get red and hot like this, it often means there is inflammation in the joints – often the body's immune system is attacking something in the joint ...'
- 'I'd like to do some blood tests to look for evidence that the immune system is causing this, and also get an X-ray of your hands ...'
- 'Once the results are back, I'm going to talk to a specialist rheumatologist at the hospital ...'
- 'It might be that they suggest starting anti-inflammatory medication over the next few days to try to reduce the swelling and stop it causing any damage to the joints ...'
- 'In the meantime, let me prescribe you some painkillers to try to make things more comfortable.'

Box 1.11 Breaking bad news

- Choose a setting that is private, quiet and comfortable – perhaps a relatives' room off a main ward, after ensuring that you will not be disturbed.
- Ask the patient who they would like with them to discuss their test results – encourage them to bring along a trusted friend or relative. Try to have at least two members of clinical staff present, e.g. a senior treating doctor and a nurse caring for the patient.
- Set aside time – ideally, at least 20 minutes – to ensure that the discussion is not rushed and that questions can be dealt with.
- Introduce yourself and others present and explain the purpose of the meeting, e.g. to go through recent test results.
- Ask how the patient has been since the last time you spoke with them.
- Briefly (in not more than two or three sentences) recap their history – what symptoms they presented with and why certain tests were done.
- Fire a warning shot: explain that things are looking more serious than first thought.
- Pause.
- Clearly, briefly and in plain English, describe the findings, e.g. 'I'm afraid that the CT scan revealed there is a lump in your left lung, and the doctor reporting the scan thinks that this could be lung cancer.'
- Pause.
- Be prepared for any reaction from the patient – they might break down in tears, or argue with you, or become angry, or simply deny that what you have said is true. Avoid giving too much more information until they indicate they are ready.
- Briefly outline the likely diagnosis, any areas of uncertainty and the next steps (e.g. further investigations or referral to a specialist).
- Pause.
- Invite questions.
- Be ready to answer the question 'How long have I got?' It is often impossible to say at this stage, and it is generally misleading and inappropriate to give specific figures. It can sometimes be helpful to outline whether a condition is survivable, or to give best- and worst-case scenarios.
- Give hope: explain what can be done, even it is simply a matter of care, support and symptom control. Try to help patients to maintain some control over their life, but never make false promises.
- Empathize: e.g. some patients like to be touched, with a reassuring hand placed on theirs, while others do not. Always try to convey concern and support explicitly.
- Outline a brief plan: what happens now? What should they do if they think of questions over the next few hours?

causal explanations and tying symptoms to the underlying pathological processes. This helps patients to understand the clinician's diagnostic reasoning and future priorities in management.

Breaking bad news

Sometimes, clinicians have to tell patients that they have a severe, chronic or life-limiting disease. This might be expected news for the patient or it might come as a complete surprise. This is one of the most difficult things a clinician has to do. It is never a pleasant task but having a good structure enables a clinician to be sensitive and kind, and leaves the patient knowing that they are not on their own (**Box 1.11**).

Traditionally, it was commonplace to withhold bad news from patients, telling it only to their families. Such an approach will often cause far greater distress to the patient (as a result of being deceived) and is now considered unacceptable. Where patients do not wish to be told about their diagnosis, this should be respected, and information can be given to family members instead if the patient consents; often, in such cases, the patient may understand that the news is not good and may wish to be spared the full details. If major treatment decisions need to be made and the patient has mental capacity to make them, clinicians should do their best to persuade patients of the importance of understanding what is wrong with them.

Box 1.12 Example of a 'problem list' in an elderly patient admitted to hospital

1. Right lower lobe pneumonia (presumed aspiration pneumonia, no positive microbiology)
2. Impaired swallow, awaiting speech and language therapy assessment
3. Previous stroke, right-sided hemiparesis, bedbound
4. Acute kidney injury probably secondary to sepsis and hypotension – resolving
5. Previously struggling to cope at home despite four-times-daily care package; may require residential nursing care

Team communication

Communication of a diagnosis between members of the healthcare team should be succinct and accurate. Technical terms are appropriate, alongside abbreviations, as long as they are widely understood. Often, patients with complex medical histories may present with multiple problems, and so clinicians treating them may compile a '***problem list***' outlining currently unresolved issues (**Box 1.12**). This can help to highlight issues that need to be resolved, guide investigations and management, and be used in communicating a patient's needs between members of the healthcare team.

Box 1.13 Benefits of shared decision-making

Benefits for the patient

- Better understanding of the different treatment options available and their benefits and risks
- A feeling of empowerment as different options are explored
- A treatment plan tailored to the patient's individual needs

Benefits for the healthcare provider

- A better understanding of what things are most important to this individual patient
- Better patient engagement with the illness and its management
- Better adherence to prescribed medication

The SBAR tool

Reviews of serious incidents occurring in hospital often highlight poor communication between team members as playing a key role in subsequent patient harm. For example, a member of the nursing staff may notice a deteriorating patient and summon the on-call doctor, who may fail to appreciate the severity of the situation and therefore not respond in a timely manner.

To address this problem, the SBAR tool has been developed to help formalize such communication. Staff members are encouraged to use this to structure their communication, in order to emphasize the need for full attention and an adequate response from the professional receiving the call.

- ***S***ituation: who is making the call, which patient does it relate to and where are they in the hospital?
- ***B***ackground: why was the patient admitted and what have been the recent events?
- ***A***ssessment: why are you calling, what are the patient's current problems and what assessment have you made of them?
- ***R***esponse: what do you need the person receiving the call to do?

Shared decision-making

Generally a diagnosis will be reached that incorporates information from the history, physical examination and relevant investigations. This will be communicated to the patient by the clinician treating them, who should check their understanding and discuss the implications. As with history-taking, the patient must be regarded and treated as an equal partner. The concept of shared decision-making has been developed to emphasize just how important joint involvement is, with benefits for both the healthcare provider and the patient (**Box 1.13**).

For patients to participate as equal partners, it is crucial for them to be provided with all the necessary information about their condition and the various treatment options available, presented in language that is easily understood. Information leaflets and online materials, often produced by patient support groups and charities, are available to help achieve this.

Managing uncertainty

Sometimes, it is not possible to make a definitive, corroborated diagnosis. This may be because:

- the investigations available are unable to provide definitive proof, for example if medicine is being delivered in a resource-poor setting
- definitive investigation is avoided because of the dangers or burdens involved
- the patient declines investigation
- investigations have commenced but will not reach a conclusion for some time.

This can be very difficult for both patients and clinicians. Without a confirmed diagnosis, it is impossible to produce a confident prognosis, and this can be profoundly challenging for patients. It can also be unnerving to clinicians to be taken to the point at which science can offer nothing more and they are forced to acknowledge their limitations. At this point, the clinician should help the patient deal with this lack of closure and uncertainty. It might be relevant to establish what gives the patient's life meaning, to enquire about their religious or spiritual beliefs, or to ask what most concerns them about facing their current illness.

In the meantime, plans can be built on the available evidence relating to the patient's underlying illness, guided by key concerns expressed by the patient:

- Symptomatic treatment (such as pain relief or anti-sickness drugs) is always appropriate when required, and may be a way of restoring a sense of control in an uncertain situation (see Ch. 7).
- Empirical treatment can be instituted on the basis of a suspected underlying pathology, even if a firm diagnosis is not possible. However, this needs clear discussion with the patient about the likelihood that the proposed treatment may cause side-effects, and may be ineffective or even harmful if the suspected diagnosis is incorrect.
- If a rapidly life-limiting condition, such as advanced malignancy, is suspected, then discussions should begin early about end-of-life care. What is important to the patient? Where would they like to be cared for in their final months or weeks?
- Help from psychologists or chaplains can be of great benefit and is often readily available.

Further reading

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DIAGNOSIS, ARTIFICIAL INTELLIGENCE AND THE FUTURE OF MEDICINE

Artificial intelligence (AI) describes the ability of machines to perform tasks traditionally believed to require higher cognitive skills. Examples include natural language processing, learning, executive planning and pattern recognition. Although multiple predictions are made about the future powers of self-conscious computers and 'super-intelligences', all the AI technology available currently, and in the near future, may be described as 'narrow AI' or 'weak AI': that is, systems designed to augment specified tasks within limited and well-defined fields of activity. The basis of all AI systems lies in the ability of high-capacity computer systems to analyse and utilize trends in large datasets that correlate with predefined outputs, 'seeing' patterns in data that possess predictive significance but are invisible to human observers. AI systems are 'fed' data and use this to develop and refine rules that predict the outputs they are designed to detect.

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AI systems in healthcare

A number of such narrow AI systems have been trialled with encouraging results in different areas of medical practice:

- ***'Computer vision'.*** These AI systems review large numbers of medical images with appropriate diagnostic labels supplied by experts, and build diagnostic algorithms based on features of the image that correlate with the assigned diagnosis. Such systems have been shown to perform as well as, or better than, human experts in a number of clinical settings, including the interpretation of electrocardiograms, the diagnosis of skin cancers, the recognition of abnormalities on retinal screening photographs, and the reporting of chest X-rays and other radiological images.
- ***Risk prediction.*** These AI systems consider large sets of patient data (age, sex, ethnicity, environmental risk factors, physiological observations, results of blood tests and other investigations, administered medications) and correlate these with patient outcomes to form powerful predictive tools. Patterns are detected that may have previously been overlooked by human researchers, such as beat-to-beat heart rate variation, which correlates strongly with mortality and development of sepsis in patients in critical care settings. Similar algorithms have been shown to be effective in predicting in-hospital complications and mortality, out-of-hospital events (such as 10-year cardiovascular mortality) and even less obvious outcomes (such as suicide risk).
- ***Individualized treatment.*** No two patients suffer exactly the same disease and yet patients with the same diagnosis are generally, at present, offered the same treatment options. AI systems have been developed that analyse a large volume of patient data (relating, for example, to patients' whole-genome sequences or samples from tumours), allowing genetic variants to be identified that predict the likely responses to different types of treatment. These allow treatment regimens to be tailored to individual patients, maximizing efficacy and minimizing toxicity.

Future uses of AI

All of the currently and imminently available AI systems simply present clinicians with suggestions: probabilities that might suggest a diagnosis, or quantify a set of risks, or identify treatments most likely to be of benefit. As such, these AI systems provide doctors with a helpful '***second opinion***', from a unique, non-human perspective. The key ethical issue of deciding what to do next falls to the patient and doctor, working collaboratively.

However, other AI technologies are being developed that pose far greater ethical questions about the role that machines should play in healthcare. Although these are presently far off, some developers envisage robotic systems that would almost completely replace human doctors: patients would tell a computer system their symptoms, undergo whatever investigations were required, and receive an automated, algorithm-driven diagnosis and management plan. These systems would use natural language processing to review all relevant medical literature, and make use of this data to identify exactly which treatment would be most appropriate for the patient in question.

In the social care sector, robotic systems are already being designed that provide 'care' and 'companionship' for elderly, disabled or cognitively impaired people. Other systems administer talking-based therapy to patients with mood disorders such as depression. Does this represent a good use of technology, reducing the cost and improving the quality of care? Or is there something fundamental about caring for people that requires a human to do it?

The role and goals of medicine

Predictions about the future implications of technology are notoriously difficult to make and prone to embarrassingly high degrees of error. It is right for doctors to welcome rigorously tested narrow AI as it is currently available, and to appraise future developments critically to ensure that they maintain the safety and dignity of the patients they are designed to help.

However, technology can never replace the human-to-human interaction at the heart of every medical consultation. While we strive to use all available technology to refine diagnosis and improve management, this must never be at the expense of the relationship that has always formed the cornerstone of effective medical care. We are increasingly able to provide our patients with the most incredible therapeutic interventions that deliver invaluable improvements in quality and quantity of life. But we are also always able to give them our attention, comfort, compassion and care. There can be no greater privilege than having a patient trust you with their life and health, and we shoulder a heavy responsibility when seeking to act as their doctors.

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