Chapter 1

The diagnostic process

The purpose of this book *2*
When and how to use it *3*
'Intuitive' reasoning *4*
'Transparent' reasoning *5*
Differentiating between diagnoses *6*
Changing diagnostic leads *7*
Confirming a diagnosis *8*
Evidence that 'suggests' a diagnosis *9*
Confirmatory findings based on general evidence *10*
Findings that suggest diagnoses based on general evidence *11*
Explaining a diagnostic thought process *12*
An evidence-based diagnosis and plan *13*
Medical and surgical sieves *14*
Diagnoses, hypotheses, and theories *16*
Imagining an ideal clinical trial *17*
Diagnostic classification, pathways, and tables *18*
A diagnostic pathway for jaundice *19*
Dynamic diagnoses *20*
Explaining diagnoses to patients *21*
Informed consent *21*
Minimizing diagnostic errors *22*

The purpose of this book

This book explains how to interpret symptoms, physical signs and test results during the differential diagnostic process. There are many books that provide lists of differential diagnoses. However, this book also explains how you should use them. Each page describes

- The main differential diagnoses of a single diagnostic 'lead'
- How to 'differentiate' between these differential diagnoses
- How to confirm the diagnosis and to begin treatment

Making diagnostic reasoning and decisions transparent

The book explains how to outline your diagnostic reasoning on paper. It does this by showing you how to write a list of differential diagnoses and established diagnoses, each with its supportive evidence so far and proposed management (see 📖 p.13). This can be used as a draft management plan and later as a hospital discharge summary. The differential diagnoses in the pages of this book with their evidence and initial management are described in the same format and can be used as example entries when writing out an outline of the diagnoses, evidence and management for a patient.

Understanding the reasoning of others

This book helps you to understand the diagnostic reasoning and decisions of others. In order to do so, you (and patients, carers, nurses and other health professionals) have to ask:

- What problem findings have been identified (including the presenting complaint)?
- What diagnoses are being considered (provisional and final)?
- What is the evidence for each diagnosis (how it presented, how it was confirmed and how its progress or outcome is being assessed)?
- What is the management of each diagnosis (the treatments, the tests being requested and plan)?

Look up the 'problem findings' and diagnoses in this book so that you know what type of answers to expect to the above questions. You can write them out in a similar format (see 📖 p.13). After hearing these answers, you may wish to add new information to the pages of this book. You will learn more quickly by doing this.

Checking a clinical impression and explicit reasoning

It is important to check all diagnoses and decisions. Reasoning alone using knowledge from a book of this kind is not enough. Such reasoning should be checked by discussing it with someone who is familiar with the situation from past experience and who can recognize if the reasoning makes sense. However, it is equally important to check that diagnoses and decisions made 'intuitively' make sense when checked with transparent reasoning of the type described in this book.

When and how to use it

This book can be used:

- When assessing a patient, e.g. after the history of presenting complaint, after completing the full history, after completing the examination, and when the test results come back
- In the same way during problem-based learning with case histories
- During private study to allow you to solve clinical problems later without having to refer to the book
- When asking someone else to explain a diagnosis and decision to you

If the presenting complaint is severe (e.g. pain or breathlessness), disabling (e.g. inability to move a limb or speak), or unusual (e.g. coughing or vomiting blood), then it will tend to be good lead with a shorter differential diagnosis. The most useful diagnostic leads are described in this book—look at the 'Contents' page of each section and the title of every page so that you can recognize them.

If the presenting complaint is not a good lead, then consider what systems (e.g. cardiovascular or respiratory) it came from and ask 'direct questions' directed at this system to try to find better leads. Also, focus on that system first in your examination. Note the speed of onset; this will suggest the underlying disease process. Onset within seconds suggests an 'electrical' cause, e.g. a fit or rhythm abnormality; onset over seconds to minutes suggests an embolus, a trauma, or rupture; onset over minutes to hours suggests a thrombotic process, over hours to days an acute infection, over days to weeks a chronic infection, weeks to months a tumour, and months to years a degenerative process.

Read the book during private study by covering the column of diagnoses on the left of the page with a bookmark and testing your ability to recognize the diagnoses when you read the nature of the diagnostic lead on top of the page, the suggestive and confirmatory findings. If you are able to do this successfully, you will soon learn to take a history and examine a patient without having to use this book. Do it first with the symptoms and physical signs that are common in your current (and next) clinical attachment so that you are prepared.

'Intuitive' reasoning

It is important to bear in mind that most of the time, experienced doctors use a non-transparent reasoning process. This seems to involve recognizing combinations or patterns of findings consciously or subconsciously which suggest or confirm a diagnosis, or indicate that some treatment should be given. This is a skill that is improved by experience. This book will encourage you to do this sooner. However, all doctors specialize and the information in this book will be of help to experienced doctors with patients outside their specialty.

If you were told that a patient had suffered sudden onset of sharp chest pain over seconds to minutes, then this lead will make you think consciously or subconsciously of a pneumothorax, pulmonary infarction, etc. If another patient has suddenly started coughing up blood, then this lead would suggest acute bronchitis, pulmonary infarction, bronchial carcinoma, pulmonary tuberculosis, etc. However, if both happened in the same patient, your mental links would 'intersect' mentally on pulmonary infarction and it would surface to consciousness.

If you were to come across this combination of features and had read in this book during private study that they 'suggested' pulmonary infarction, then you might think of this diagnosis directly. If you came across these findings many times and a diagnosis of pulmonary infarction was usually confirmed on CT-pulmonary angiogram, then you would soon recognize that the combination of findings as suggesting pulmonary infarction (like recognizing someone's face).

If a diagnosis or small number of differential diagnoses do not come to mind readily in one of these ways, then it is important to use a 'transparent' reasoning process. You will always come across unfamiliar situations, however experienced you become, so the 'transparent' approach will always be important.

'Transparent' reasoning

'Transparent' reasoning involves assembling a combination of features that identifies a group of patients within which the frequency of those with a diagnosis would be high. This can be done by first selecting a diagnostic lead. It can be a symptom, sign, or any test result (e.g. acute abdominal tenderness in the right lower quadrant, see 📖p.480).

One of the lead's differential diagnoses is then chosen (e.g. appendicitis on p.480), and a finding is looked for that occurs often in the chosen diagnosis (e.g. guarding occurs often in appendicitis), but less often in others (e.g. guarding occurs less often in non-specific abdominal pain or NSAP). Appendicitis will thus occur more frequently (and NSAP less frequently) in a group of patients with right lower quadrant pain and guarding.

If a new finding becomes available which is a better lead with fewer differential diagnoses (e.g. a CT scan result), then this can be seized upon instead. You can select any finding as a lead from the total evidence. It does not have to be the first finding you come across such as the presenting complaint. But this does not mean that you can ignore other findings.

A single diagnosis will only become final if it can explain all the patient's findings. For example, in some cases, a CT scan might also show an absent kidney shadow on the left side. None of the differential diagnoses of this finding would explain acute right sided abdominal tenderness. Therefore, it would be wrong to focus on the lead of an absent renal outline and ignore the other findings, so at least two diagnoses will be needed.

Differentiating between diagnoses

Eddy and Clanton analyzed the thoughts processes of senior doctors participating in the Clinico-Pathological Conferences at the Massachusetts General Hospital[1]. They pointed out that choosing a diagnostic lead, e.g. right lower quadrant pain (which they called a 'pivot') was central to these experienced doctors' explanations when solving diagnostic problems. They also noted that during diagnostic reasoning, other findings (e.g. guarding) were used to 'prune' some of the differential diagnoses (e.g. pruning away NSAP).

If a finding (e.g. being male) occurs often in a diagnosis being pursued (e.g. appendicitis) but cannot happen in a differential diagnosis (e.g. ectopic pregnancy), then that diagnosis can be ruled out. However, if a finding such as guarding occurs commonly in the diagnosis being chased (e.g. appendicitis) and less frequently in another diagnosis (e.g. NSAP), the other diagnosis will become less probable, not ruled out. That is, the diagnostic lead together with the new finding will form a combination within which the frequency of the diagnosis being chased becomes more frequent; the diagnosis in which the finding occurs less often thus becomes less frequent.

The frequency with which a finding occurs in a diagnosis is often described as its 'sensitivity' by epidemiologists, i.e. the frequency with which the finding 'detects' the diagnosis when screening a population. Statisticians also call the 'sensitivity' the 'likelihood' of the finding being discovered when the patient is known to have the diagnosis. If the finding is 'likely' to occur in a diagnosis being chased and is 'unlikely' to occur in one of its differential diagnoses, then the ratio of the two likelihoods represents the finding's ability to differentiate between those two diagnoses. This makes one more probable and the other less probable. This book describes such findings under the headings of 'Suggested by' and 'Confirmed by'.

Changing diagnostic leads

A patient presenting with breathlessness will have a long list of differential diagnoses. A circular shadow on a chest X-ray (CXR) will have a much shorter list of differential diagnoses and a CT scan showing a lesion contiguous with a bronchus an even shorter one. A biopsy might provide a diagnostic criterion for a bronchial carcinoma. However, this may only be a working diagnosis. All the diagnoses applicable to that patient will not become final until the patient's symptoms have been cured, stabilized, or predicted correctly.

So if we come across a powerful finding or combination of findings (e.g. a dense round shadow within an organ on a CXR), this will form a stronger lead with a shorter list of differential diagnoses. It is easier to make a fresh start with such a powerful new finding than to try to work out which of the original diagnostic possibilities are being made more probable or less probable. Therefore, another measure of a powerful finding is the number of differential diagnoses required to explain, say 99% of patients with that finding. The better the lead the fewer the differential diagnoses.

Confirming a diagnosis

A diagnosis can be confirmed in different ways, the different confirming (or 'sufficient') findings taken together form the 'definitive criteria' of the diagnosis. The definitive criteria thus identify all those and only those with the diagnosis. Such criteria can be based on symptoms, signs, and test results (and in some cases, on the result of treatment). Ideally, the 'pre-treatment' criteria should identify all those who respond to the various treatments available for patients with that diagnosis or those for whom such a label is of practical value in other ways (e.g. prognosis alone). In some cases, the diagnostic criteria are proposed by experts set up by official bodies.

In many cases, a diagnostician will start treatment when a diagnosis is probable or suspected strongly without waiting for formal criteria to be fulfilled (e.g. a treatment given on suspicion of meningitis or of inhalation pneumonia). In such a situation, the diagnostician might imagine the existence of a large number of identical patients who were randomized into different treatment limbs of a randomized clinical trial. The treatment chosen would be the one imagined to produce the best outcome, bearing in mind the benefits and adverse effects. If the patient responds to treatment, then this may also be regarded as confirmation of the diagnosis in some circumstances.

There may be no formal criteria that are suitable for use in day-to-day clinical care and it is up to the individual doctor to use what he or she considers reasonable. One such subjective approach is to provide a trial of therapy, and if the patient improves, to regard this as a confirmatory result. If the treatment is successful, then no other explanation is looked for. The confirmatory findings in this book are based on all of the above approaches. They reflect typical approaches used by doctors in the authors' experience. However, none of these approaches are ideal; future medical research may improve matters.

Looking at the situation in a different way, the group of patients with a probable or confirmed diagnosis encloses other subgroups of patients for which different actions are indicated. For example, some patients with a diagnosis have mild conditions so that treatment is not necessary, others may be so severe that it is too late to treat while others are treatable ('triage' in emergency situations is a special case of this principle). The group with a diagnosis may also contain subgroups with causes and complications that also require treatment. Therefore, diagnoses (probable or confirmed) may be thought as 'envelopes' that enclose subgroups of patients for which different actions are indicated. The way in which symptoms, signs, and test results can be chosen as diagnostic gold standard criteria is described in the Appendix (see 📖p.751).

Evidence that 'suggests' a diagnosis

It is important to remember what 'evidence' means. Evidence is made up of facts, which are records of observations and actions that took place at a place and time. A fact becomes evidence when it is used to persuade someone else to accept an opinion—a diagnosis and what should be done in the context of this book. A diagnosis is the title to what we picture is happening to a patient. This will include causes and complications. This may be pictured with certainty or with a degree of probability, depending on the available evidence.

Evidence may be based on facts such as symptoms, signs, and test results recorded in a particular patient. This is 'particular' evidence by analogy with a 'particular' proposition in logic. In contrast to this, 'general' evidence will be based on facts related to groups of patients such as the result of a clinical trial, which is analogous to a 'general' proposition in logic. In order to practice evidence-based medicine, we have to relate the 'particular' evidence from a particular patient to 'general' evidence about groups of similar patients published in the medical literature.

The opinions supported by 'particular' evidence are diagnoses with different degrees of probability about what is wrong with patients and what to do. If the listener is going to accept such an opinion on the basis of the evidence, there has to be agreement as to what is acceptable as evidence. This book contains typical evidence that is used to 'suggest' and 'confirm' diagnoses as accepted at present by most doctors in their day-to-day work. These conventions will no doubt change as more 'general' scientific evidence is published.

Each differential diagnosis on every page is followed by the evidence that 'suggests' the presence of the diagnosis, the diagnosis being considered to be present when the 'confirmatory' findings is present. The confirmatory evidence for each diagnosis is provided under another subheading, followed by the initial management.

For example, acute abdominal tenderness localized to the right lower quadrant in combination with guarding 'suggests' that the diagnosis will probably be appendicitis (see 📖p.446). The diagnosis of appendicitis is 'confirmed' by the appearances at laparotomy and by histological examination. It is important to note that not all the findings have to be used in the reasoning process at one time; this is discussed in more detail later in this chapter.

Confirmatory findings based on general evidence

A confirmatory finding identifies a group of patients that envelops all those with indications for treatment explained by the diagnosis. If new treatment indications are discovered that are explained by the diagnostic theory, then 'the envelope' may need to be expanded. For example, it was discovered some years ago that many patients with features of diabetic retinopathy requiring treatment had blood sugars outside the criteria for diabetes mellitus. Because of this, meetings were convened by the World Health Organization and the American Diabetes Association, and the 'envelope' for diabetes was expanded by lowering the diagnostic cut-off point of fasting blood glucose.

It is also possible that new tests may be discovered in future that select patients more efficiently for treatment. If these new treatable patients lie outside the diagnostic group that was previously considered for treatment, then it might be appropriate to use the new test to identify patients who should be deemed to have the diagnosis. So if 'confirmatory' tests are to be chosen in an evidence-based way, then they should be shown to be superior to rival tests by including more patients who respond to the treatments directed at the diagnosis and/or excluding more patients with no prospect of responding.

Many diagnoses are based on test results that are 'abnormal', i.e. above or below two standard deviations of the test result in the general population. This means that the 2.5% of patients above and 2.5% of those below these two standard deviations could be regarded as 'abnormal'. The use of two standard deviations is arbitrary and not 'evidence-based'. For example, patients with diabetes mellitus are 'diagnosed' as having 'diabetic microalbuminuria' if their albumin excretion rates (AER) are above two standard deviations of the mean (i.e. >20mcg/min).

In a clinical trial on patients with type 2 diabetes mellitus where their blood pressures had been controlled, there was no difference between those on treatment and placebo in the proportion of patients developing nephropathy within two years if they had an AER between 20 and 40mcg/min.[2] This suggests that the cut-off point should be 40mcg/min. However, before changing the definition, it would be important to ensure that the patients inside the envelope with an AER between 20 and 40mcg/min might not benefit in other ways.

Ruling diagnoses in and out

A diagnosis is ruled in if at least one of its confirming (or sufficient) criteria is present. A diagnosis is ruled out if it can be shown that the patient lies outside the diagnostic envelope. One way of doing this is to show that not one of the possible confirming (or sufficient) features is present. Another way is to show that a single feature is absent which must occur in those with the diagnosis, e.g. that the patient is not female and therefore, cannot have an ectopic pregnancy. Such a constant diagnostic finding is called a 'necessary' criterion.

Findings that suggest diagnoses based on general evidence

The best findings for 'suggesting' probable diagnoses are those which, when used alone or in combination with others, predict the presence of 'confirmatory' test results with the highest frequency of success. The general evidence for the ability of findings to do this during population screening is usually offered in the form of indices such as sensitivity, specificity, and likelihood ratios. However, in order to assess the usefulness of tests during the differential diagnostic process, other indices have to be used. One index is the number of diagnoses required to explain most (e.g. 99%) of the differential diagnoses of a diagnostic lead—the fewer the better.

Another index is the ability of a test to differentiate between pairs of diagnoses in such a lead. If a test result occurs commonly in patients with confirmatory findings of one diagnosis and uncommonly in patients with another diagnosis, then that test will help to differentiate between them. The difference in these frequencies of occurrence can be measured by their ratio.

Statisticians describe the frequency of a finding that occurs in those known to have a diagnosis as the 'likelihood' of it occurring (the 'likelihood' is also known to epidemiologists as the 'sensitivity'). The difference between these 'likelihoods' for two different diagnoses can be represented by the ratio of the two likelihoods. As this ratio refers to a pair of differential diagnoses, we can call it a 'differential likelihood ratio'. This is different to the plain 'likelihood ratio' which is the frequency of a finding in patients with a confirmed diagnosis divided by the frequency of the same finding in ALL those confirmed NOT to have that diagnosis. This 'non-differential' likelihood ratio is more useful when screening populations by using one test to detect one diagnosis.

Explaining a diagnostic thought process

You may well have arrived at differential diagnoses by using intuitive, non-transparent, pattern recognition and not considered in an explicit way how it was done. Alternatively, you may have recorded your team's consensus opinion. However, you may be asked by a patient, student, nurse or doctor to explain your thinking. In fairness, the way that your own mind (let alone someone else's mind) has actually worked subconsciously may be impossible to explain.

The first step is to write a summary of the positive findings, diagnoses, evidence, and management as shown on p.13. The original evidence for established diagnoses (e.g. type 2 diabetes) may not be available. However, for new diagnoses, choose from the evidence the best lead with the shortest differential diagnosis. Use the other findings to show that the one (or some) diagnoses are more probable or confirmed, and others less probable or ruled out.

If these conclusions of the non-transparent and transparent thought processes are not the same, you may wish to revise your opinion and list of differential diagnoses. By doing this, you will be checking diagnoses by using a different mental process in the same way as you would check the answer to arithmetic addition by adding up the list of numbers in a different order.

In order to avoid overlooking diagnoses, jog your memory by using 'sieves' to use 'recognition' to and help 'recall' by listing the possible broad anatomical and physiological explanations (see 📖p.14).

An evidence-based diagnosis and plan

Positive findings summary

Central chest pain for 4h with jaw discomfort, sweating, and nausea (1/10/08). PMH of hypertension for 10y. History of mild jaundice during febrile illnesses for years. BP 146/88 on admission (1/10/08). ECG: T wave inversion S2, AvF, V4, and V5. Latest HbA1c=8.7% (5/8/08).

Assessment and plan

?Unstable angina

?Non-ST elevated myocardial infarction (NSTEMI)

Outline evidence: central chest pain for 4h with jaw discomfort, sweating and nausea (1/10/08). ECG: T wave inversion S2, AvF, V4 and V5.
Plan: for troponin I 12h after onset of pain. Aspirin 300mg stat, bisoprolol 5mg od, isosorbide mononitrate 10mg bd.

?Gilbert's disease

?Cholelithiasis

Outline evidence: jaundiced sclera, history of mild jaundice during febrile illnesses for years, none of liver disease (1/10/08).
Plan: check bilirubin, urobilinogen, AST, γGT.

Other active diagnoses

Essential hypertension

Outline evidence: history of raised BP for 10y. Current BP 146/88 on admission (1/10/08).
Plan: continue bendroflumethiazide 2.5mg od, perindopril 2mg od.

Type 2 diabetes mellitus

Outline evidence: latest HbA1c = 8.7% (5/8/08).
Plan: stop gliclazide 160mg bd. Start insulin sliding scale.

Medical and surgical sieves

Check that you have not forgotten something by using the following 'medical sieve'. Under each heading, think of the structures involved in flow (air, blood, food, etc.). Think of function in terms of feedback cycles (sugar, blood pressure (BP), etc.):

- Social system and environment
- Locomotor system
- Nervous system
- Cardiovascular system
- Respiratory system
- Alimentary system
- Renal and urinary tract
- Reproductive system
- Endocrine and autonomic system
- Haematological and immune system

Consider each of these systems by using the 'surgical sieve'. Is there a problem which is congenital, infective, traumatic, neoplastic, or degenerative?

The information in the pages of the OHCD is also set out in the same format as the Assessment and Plan (compare diagnoses of 'unstable angina' and 'NSTEMI' with those on p.216). The page on chest pain gives some differential diagnoses with typical suggestive and confirmatory evidence that could also be added to those opposite. You may refer to these as examples when writing your own assessments and plans.

Diagnoses, hypotheses, and theories

Although the findings used to confirm a diagnosis can be observed, all things pictured or imagined under the title of the diagnosis cannot be confirmed by observation, e.g. molecular changes in damaged tissue or what would have happened in a particular patient if a treatment had not been given. Not only does this apply to hypotheses for individual patients, it also applies to what is imagined about populations of patients in scientific hypotheses and theories. It is thus possible that something else will be imagined or pictured in future which is also compatible with findings previously explained by another theory.

This is why the philosopher of science, Karl Popper, argued that general hypotheses and theories cannot be proven or confirmed in their entirety. However, if a new observation is inconsistent with one aspect of the hypothesis, it will have been 'falsified'. It will thus have to be changed to some degree (perhaps completely or slightly) to take the new observation into account.

Raised ST segments on an ECG in someone with severe central chest pain were formerly part of the criterion for confirming MI, which suggested that a part of the myocardium was dead. However, one aspect of this theory has been 'falsified' because it has been discovered that some (or all) of the 'infarcted' myocardium is salvageable. With our new understanding, we use the same findings to 'confirm' an 'ST Elevated Myocardial 'Infarction'. We have modified the theory and now think that the process of infarction is not complete and can be stopped with treatment with reversal of many changes.

However, it is important to assess the reliability of the 'falsifying' fact. This is done by estimating the probability of the 'falsifying' observation being replicated by other scientists (or another doctor if the hypothesis is a diagnosis about an individual patient based on particular evidence). If the probability of replication of the evidence is high about a 'general' observation, then the observation may be accepted by the scientific community (but many may go to the trouble of repeating the study to make sure). If the P value is low or the 95% confidence intervals are narrow, then the probability of non-replication due to chance observations alone will be low.

Imagining an ideal clinical trial

The findings used to define a 'diagnostic envelope' should enclose the best treatment indication criteria. These criteria should be chosen ideally from a number of candidate criteria. The chosen treatment criterion should be the one that produces the clearest outcome difference between the treatment and control in a comparative trial. For example, method A for measuring microalbumin in urine chose patients for a trial; 24% developed nephropathy on placebo and 12% developed nephropathy on treatment. However, with method B, 12% developed nephropathy on placebo and 12% developed it on treatment. This would suggest that method B was not identifying patients who benefited and would be inferior to method A.

In the absence of detailed trial data, a doctor may have to guess whether a patient's findings would identify a group of patients who would benefit from the treatment more than a placebo, bearing in mind side effects, costs, etc. If on balance, this would be the case, the doctor could apply a diagnostic term that would summarize his theoretical explanation as to why giving that treatment to a patient with that combination of findings would be better than not doing so.

Decision analysis

Decision analysis is a discipline that models mathematically what would happen if a detailed clinical trial were performed to compare the treatment options being considered for a particular patient. A 'decision tree' is constructed first to show all the possible diagnoses. The tree is extended to show the possible interventional limbs into which the patient could be randomized, followed by all the possible outcomes of each treatment. The branches would end with the effect that each outcome would have on the overall well-being of the patient.

An estimate is then made of the proportions of patients with each diagnosis, the proportions opting for each treatment and the proportions of those experiencing various degrees of well-being. These proportions are then multiplied together to estimate the average degree of well-being experienced by patients sharing each treatment outcome. Each of these average degrees of benefit are regarded as the 'expected' degree of well-being that would be experienced by an individual patient with each outcome. This is regarded as a representation of what an experienced doctor would do when he or she estimates the effect on the patient of the different interventions available.[3,4]

Medical science aims to provide diagnostic criteria, treatment indication criteria, and treatments that when used together will predict with a high degree of certainty which treatment will work best for each patient (or would not help at all). Such well designed diagnostic systems would make it easier to choose the best option and to justify it using evidence in the form of data. This will not be possible without a clear understanding of the diagnostic process and criteria for confirming diagnoses that also indicate the best treatment for that patient.

Diagnostic classifications, pathways and tables

A diagnostic pathway or algorithm is a way representing diagnostic reasoning processes or a diagnostic classification (see opposite 📖p.19). The same reasoning processes can be displayed using a table of the kind shown below. This is also how information in this book is displayed. It is flexible and also allows findings to be shown which do not form part of the diagnostic criteria. The reader can scan down such a table to find the diagnoses that are compatible with the findings so far. The entry can then be copied into a table in the patient's records as a draft entry for that diagnostic possibility.

Diagnostic table for the differential diagnoses of jaundice

Carotinaemia (not 'real' jaundice)
Suggested by: onset over months. Skin yellow with white sclerae, normal stools, and normal urine. Diet rich in yellow vegetables/fruits).
Confirmed by: no bilirubin, no **urobilinogen** in the urine, and normal **serum bilirubin.** Normal **liver function tests (LFT)**. Response to diet change.

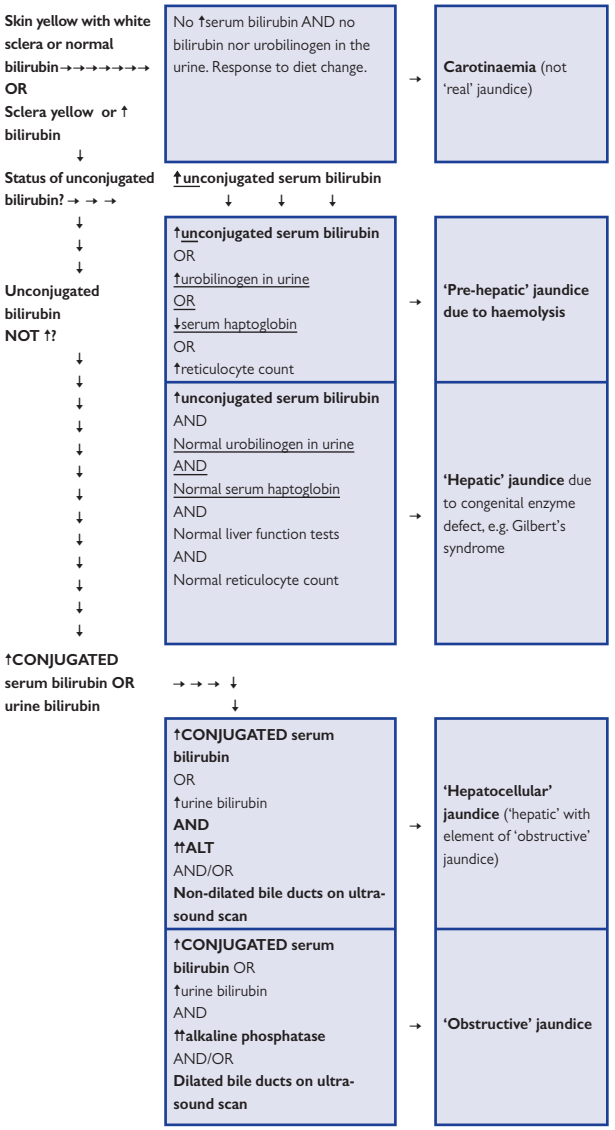
'Pre-hepatic' jaundice due to haemolysis
Suggested by: jaundice and anaemia (the combination seen as 'lemon' or pale yellow). Normal dark stools and normal-looking urine.
Confirmed by: ↑(unconjugated and thus insoluble) **serum bilirubin**, but normal (conjugated and soluble) bilirubin and thus no ↑bilirubin in urine. **↑urobilinogen in urine** and **↓serum haptoglobin**. Normal LFT. **↑reticulocyte count**.

'Hepatic' jaundice due to congenital enzyme defect
Suggested by: jaundice. Normal-looking stools and normal-looking urine. Jaundice worse during febrile illnesses.
Confirmed by: **↑serum bilirubin** (unconjugated), but no (conjugated) bilirubin in urine. No **urobilinogen in urine** and **normal haptoglobin**. Normal **LFT**.

'Hepatocellular' jaundice ('hepatic' with some 'obstructive' jaundice)
Suggested by: onset of jaundice over days or weeks, pale or stools but dark urine.
Confirmed by: ↑serum (conjugated) **bilirubin** and thus **↑urine bilirubin**. Normal **urine urobilinogen**. **LFT** all abnormal, especially **↑↑ALT**.

'Obstructive' jaundice
Suggested by: onset of jaundice over days or weeks with pale stools and dark urine. Bilirubin (i.e. conjugated and thus soluble) in urine.
Confirmed by: **↑serum conjugated bilirubin** and **urine bilirubin**, but no **↑urobilinogen** in urine. Markedly (**↑↑**) **alkaline phosphatase**, but less abnormal (**↑**) **LFT** and **↑γGT**.

A diagnostic pathway for jaundice



Dynamic diagnoses

It is important to understand that clinical diagnosis is not a static classification system based on diagnostic criteria or their probable presence. It is a dynamic process. Diagnostic algorithms 'classify' patients by following a logical pathway based mainly on diagnostic criteria. Other systems predict the probable presence of diagnostic criteria. All these methods can be regarded as 'diagnosing' a snapshot of what is happening at a particular time.

The diagnostician has to imagine the presence of a dynamic process that changes with time. There may be several processes taking place at the same time, some progressing over years (e.g. atheromatous changes), some over minutes to hours (e.g. a thrombosis in a coronary artery), some over minutes or seconds (e.g. ventricular tachycardia), and others instantaneously (e.g. a cardiac arrest).

A diagnostic process leading to treatment may have to happen repeatedly and for a number of diagnoses at the same time. It might be more appropriate to think of the process as one of 'feedback' control. In this way, the doctor would be acting as an external control mechanism to assist those of the patient who are failing. After the initial history and examination, the feedback information may come from electronic monitoring, nursing observations, ward rounds, hospital clinic, or primary care follow-up.

There are three types of mechanisms of interest to the diagnostician.

- Those that control the 'internal milieu' by keeping temperature, tissue perfusion, blood gases, and biochemistry constant.
- Those that control the body's structure by effecting repair in response to any damage.
- Those that control the 'external milieu' of day-to-day living.

These are all interdependent. If one mechanism fails, then it may unmask other weaknesses by causing other failures. It may not be enough to treat the main failure. It is often necessary also to treat the causes and consequences as they may be unable to recover on their own. For example, a coronary thrombosis may be treated with thrombolysis, but any resulting rhythm abnormalities may need to be treated and also the causative risk factors (e.g. smoking) that could result in recurrence. So when we explain our diagnostic thought processes, it helps to think of each diagnosis as a subheading with its own evidence and decision.

The whole patient

A 'diagnosis' does not imply that only one solution needs to be discovered. The complete diagnosis (or diagnostic formulation) may have to include various causes, consequences, interactions, and other independent processes. As well as internal medical processes, it has to include external factors such as circumstances at home and the effects on selfcare, employment, and leisure.

There may be many diagnoses which have been confirmed previously and for which the patient is on established treatment. Therefore, the diagnostician must imagine what is happening to the 'whole patient'. This requires a broad medical education that allows a range of phenomena to be pictured, from molecular events to events in the home and outside world.

Explaining diagnoses to patients

The patient may already be imagining with some trepidation what might be happening. It is important to find out what the patient is imagining and to use this as a starting point for your own explanation. The patient's own views are usually sought and documented at the end of the history of the presenting complaint.

Although patients may understand explanations at the time they are given them, even the most intelligent may forget unfamiliar technical terms and their meaning within a short time. Therefore, it is important to provide a written reminder of such terms and how they are related. This can be done by giving the patient a printed summary similar to that on page 12. This can also allow the patient to ask questions at his or her own pace.

Patients and relatives usually ask questions spontaneously or request an appointment for time to be set aside to do this. Some may be too shy and need encouragement to do so, in which case this important aspect of care will be omitted. Informed consent is also based on similar questions and discussion. The process is more effective if the patient is able to ask the questions (i.e. if the process is 'patient-centred'). Such a process may be facilitated if they refer to a summary such as that shown on p.13.

Ideally, patients should know the presenting complaint for their latest problems, the primary diagnosis or differential diagnoses, and what actions are being taken in terms of tests and treatments. They should also be aware of their past medical history: the various diagnoses, how they presented and were confirmed, their treatments, follow-up arrangements, and markers of progress. Again, the relevant technical terms and how they are linked can be summarized for them as shown on p.13.

Informed consent

In order for a patient to consent to treatment, he or she must understand what has been said and be able to retain that explanation. A basic understanding means the patient must know what actions have been agreed and the possible diagnoses in each case. In order to understand each diagnosis, it is essential to know which symptoms it explains and how these symptoms or some other markers are progressing. Few patients are able to retain all of this, especially if there are many technical terms that are unfamiliar to them. Therefore, it would be a sensible policy to provide the patient with a typed explanation setting out these basic relationships as shown on p.13. This would then become the next 'past medical history' when the patient is asked to provide it by another doctor or nurse. It would thus allow patients to ask a doctor or nurse to remind them of the meanings of the various terms.

Minimizing diagnostic errors

The diagnostic and decision-making process usually takes place in busy clinics, wards, operating theatres, and emergency rooms. Therefore, most diagnoses have to take place by some rapid conscious or subconscious pattern recognition, and there is usually little time for reflection. Mistakes are kept to a minimum by good training, especially listening carefully and writing out what has been observed, thought, and done.

Another important principle to bear in mind is that even the most expert and well-founded diagnoses and decisions can only be successful in a proportion of cases. Therefore, there must be a strategy to monitor their outcome and to change diagnoses and decisions, if possible.

Diagnostic errors can be classified in terms of cognitive psychology[5] into:

- Faulty triggering
- Faulty context information
- Faulty verification
- No fault errors
- Faulty information gathering and processing

Faulty triggering

This is a failure to consider appropriate diagnostic possibilities, often attributed to a weakness of medical education, which focuses on disease processes instead of the diagnostic processes. This type of error can be kept to a minimum by using the suggestions on pp.5–13, and by referring to the differential diagnoses on the other pages. Finally, this error can be reduced by not only writing down the differential diagnoses, but also by writing down the findings from which were chosen the leads that 'triggered' them as shown on p.13. This can be given to the patient to be shown to other doctors who might also spot any omissions.

Faulty context information

This is focusing on one diagnosis and failing to consider others that may also be present. It involves jumping to conclusions. This can be avoided by using the sieves on p.14, referring to the appropriate page in this book and writing out an overall plan as shown on p.13 so that other doctors might spot any errors. Again, this can be given to the patient (to show to other doctors who might spot any errors).

Faulty verification

This is failure to ensure that the patient's presenting symptom and other markers of poor health have been controlled or stabilized as well as possible. This is discussed on p.8. It also helps to set out each diagnosis with its evidence as shown on p.13, which includes the markers being followed and their latest results. Again, this summary can be given to the patient to be shown to other doctors who might spot such omissions.

No fault errors

Even the most expert and well-founded diagnoses and decisions can only be successful in a proportion of cases. This is why diagnoses and decisions are qualified with probabilities. Therefore, there must be a strategy to monitor the outcome of all diagnoses and decisions and to change them, if possible. If a summary of the kind shown on p.13 is given to the patient to be shown to other doctors, they will be able to understand the basis of previous decisions and take appropriate action.

Faulty information gathering and processing

This is poor use of leads and differentiators in appropriate settings. This book focuses on this process. It is important to know the differential diagnoses of leads and the frequency with which they occur in different clinical settings. It is also important to know the frequency with which findings occur in pairs of diagnoses. At present, this is gained from personal experience. Little research is done into leads and differential likelihood ratios because the main focus of research is currently on sensitivity, specificity, and overall likelihood ratios.

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