DIAGNOSIS IN GENERAL PRACTICE

Iterative diagnosis

Geoff Norman,† Kevin Barraclough,² Lisa Dolovich,¹ David Price¹

Strategies for improving the pattern recognition involved in making a correct diagnosis amount to forcing yourself to use analytical reasoning

What is iterative diagnosis?
The traditional model of diagnosis is one of initial collection of information in the history and examination, followed by deductive steps to reach a diagnosis. We suspect that most clinicians do not recognise or use this process.

A more realistic model was formulated by Elstein and Schwarz 25 years ago.¹ It was called the hypothetico-deductive model, but we will call it the process of iterative diagnosis. This model recognises that clinical reasoning usually involves the clinician generating one or more possible hypotheses early on in the consultation (often, but not always, by pattern recognition) and then recurrently—iteratively—testing these. Clinicians use many such shortcuts (heuristics) in clinical reasoning. This is not a fault: the shortcuts are typically correct and allow them to arrive at a working diagnosis with the minimum of delay, while avoiding excessive testing and anxiety. Exhaustive data collection without hypotheses—the medical student's history and examination—usually does not improve diagnostic accuracy and may make it worse.

The initial steps in the process of making a diagnosis are therefore often non-analytical or intuitive.² The initial hypothesis (the limited list of possible diagnoses) is often formulated before much data collection has occurred—from the “eyeball” impression as the patient walks in or as he or she is speaking.³ The process of testing of the hypothesis then proceeds by careful and systematic gathering of data and weighing the elicited information against the mental rules that are referred to in the literature as analytical reasoning.

Research in clinical reasoning is moving to a consensus that both analytic and non-analytical processes operate simultaneously in problem solving and that the clinician relies to a greater or lesser degree on one or the other, depending on experience, familiarity with the problem, and the stage of the diagnostic process.⁴ It is clearly not always a case of pattern recognition and, faced with difficult problems, clinicians may revert to “basic principles,” where they reason the problem out from a mechanistic, physiological, model.⁵

When is iterative diagnosis used?
General practitioners will recognise that they often formulate one or more presumptive hypotheses as the patient walks into the room or when they start speaking: a “hang dog” demeanour suggests depression; a unilateral stiff arm gait suggests parkinsonism; the acute onset of vertigo when rolling over in bed suggests benign positional vertigo. The general practitioner then listens to the history through the “filter” of the initial hypothesis: does the patient describe low mood, agitation, sleep disturbance? Does he or she get stuck as they roll over in bed; have they noticed that the fingers of the non-swinging arm are clumsy; does the vertigo come on with head movement and become less severe over a minute or so?

The general practitioner’s examination will usually be directed towards supporting or refuting a hypothesis: on direct questioning does the patient admit to anhedonia and pessimism? Does he have lead pipe rigidity of the arms? Does the Hallpike test have positive results?

Sometimes intuition applies a brake to the reasoning in the recurrent (iterative) testing: despite the coherent illness narrative, something doesn’t fit. Although the doctor has seen a hundred febrile children with sore throat, something “just doesn’t feel right” about this one. This may prompt referral or early review.

How does iterative diagnosis go wrong?
This process (of simultaneous intuitive and analytical clinical reasoning) is usually invisible. It is so inherent to the clinician that he or she will be scarcely be aware of using it. That it is occurring at all is usually apparent only when it fails.

When diagnoses are missed it is usually assumed that they have been missed because of inadequate data collection. No doubt, some errors are a consequence of poor data gathering. One study in particular found that the dominant cognitive bias that resulted in diagnostic error was premature closure.⁶ However, the missed diagnosis may not be a consequence of sloppiness or inadequate attention to detail; instead, the critical data are often missed simply because the clinician was not thinking of the correct diagnosis. Although clinicians
gather less data as they gain experience, this does not seem to have a negative effect on diagnostic accuracy. The “error” may more often be due to one or more common, recurrent cognitive biases. We clinicians, like other people, use cognitive heuristics that may occasionally lead us astray by biasing our weighting of evidence; these include confirmation bias—gathering information that will confirm rather than refute the diagnosis; availability bias—relating the case to easily recalled examples; premature closure—arriving at a conclusion before gathering the critical data and not revisiting it. Other examples of cognitive biases are the framing bias—being swayed by the way in which the problem is phrased, and base rate neglect—forgetting that common diseases are common. Many of these biases may affect the hypotheses that are initially considered, or the data gathered to support these hypotheses.

Further, there is some evidence that age, independent of experience, leads to increasing use of non-analytical reasoning, which has the effect of increasing reliance on early data, and less willingness to re-examine the diagnosis in light of new, conflicting data.

How can we improve?

Awareness of these common pitfalls in our heuristic reasoning may help us to avoid them. Although some familiarity with the nature of these biases is probably useful, specific and simple strategies can lead to real improvement. The common denominator in these strategies is encouraging the clinician to re-examine the data and reconsider the formulation, a process we call iterative diagnosis. It amounts to deliberately forcing yourself to use analytical reasoning. Six strategies are useful.

Routinely second guess

We can remind ourselves to routinely consider alternatives to our initial diagnosis: “What can I not afford to miss? Am I sure that this person’s red painful foot is cellulitis rather than critical ischaemia?”

Seek data that would not fit with the hypothesis

We can specifically go after signs or symptoms that would be inconsistent with the diagnosis and suggest alternatives—such as facial weakness in benign positional vertigo, or an explosive onset of headache in migraine.

Reframe when recording

We can consciously re-examine the history as we write the notes. Do not merely record a history that fits in with the hypothesis—“pulsating unilateral headache with nausea and visual aura”. Consider that the “framing” maybe misleading—the history of nausea was elicited in response to a direct question (“yes, a bit”); the term pulsating was never used; the “aura” was cumulative visual “greying.”

Reconsider dissonant facts

We can review and re-examine facts that don’t quite fit—perhaps this headache is far worse than any migraine the patient has ever had and she reports slight neck stiffness.

Know about test accuracy

We can become more familiar with test accuracy: an earlier, normal cervical smear does not exclude cervical cancer; a raised serum urate concentration does not mean arthralgia is due to gout. This may also involve being aware of pretest and post-test probabilities, the subject of an article later in the series. Negative tests (for example, a D dimer blood test or an exercise electrocardiogram) often do not adequately rule out disease in patients with high pretest probabilities, whereas they may rule out disease in those with low pretest probabilities.

Use time as a diagnostic test

Appropriately timed follow-up (as occurred in the companion article on vertigo) may also allow the general practitioner to review the diagnosis and separate minor and time limited conditions from potentially more serious problems.

Conclusion

Iterative diagnosis is an essential component of medical expertise. It involves rapid, simultaneous generating and testing of hypotheses. It is usually fast, efficient, and accurate. When errors in diagnosis occur they are often due to one or more of a set of predictable cognitive errors, rather than carelessness or lack of knowledge. Simple strategies can increase awareness of potential pitfalls and reduce errors.

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Vertigo

Kevin Barraclough,1 Adolfo Bronstein2

The diagnosis of vertigo is used to show how analytic reasoning can augment pattern recognition

A 58 year old woman presented to her general practitioner stating that she woke that day feeling that the room was moving. She had vomited twice. She seemed anxious, was slightly unsteady on her feet, and was hyperventilating. She did not have fever but had a sore throat, slight difficulty swallowing, slight hoarseness, and a red throat. The Hallpike test induced vertigo and nystagmus; the nystagmus was sustained. The general practitioner thought the likely diagnosis was either viral labyrinthitis with pharyngitis, or benign paroxysmal positional vertigo.

The diagnostic dilemma

In the 1970s a classification of dizziness was developed, based on an analysis of 125 patients attending a dizziness clinic.1 2 Vertigo was defined as the illusory sense of movement or orientation, and indicates disorders of the labyrinth or brainstem; presyncope was defined as a sense of near faint, typically due to transient hypotension. Disequilibrium of the elderly is often described as a non-specific slight “unsteadiness,” particularly on turning, and indicates poor balance and strength.3 4 and lightheadedness is often associated with dysfunctional breathing or anxiety.

To distinguish vertigo from non-rotatory dizziness, this validated question may be asked: “Did you just feel light headed or did you see the world spin around as though you had just got off a playground roundabout?”

In our case scenario, the patient has vertigo, a common condition in the community. In a postal questionnaire study of general practice patients aged 18 to 65, 7% of the 2044 respondents reported true vertigo in the past year.5 A full time general practitioner may expect to see 10-20 cases of vertigo a year.6

Eliciting the symptom of vertigo narrows the differential diagnosis to disorders of the labyrinth or its central connections. With new onset of vertigo, the diagnostic challenge for the general practitioner is to distinguish the (common and easily treatable) peripheral causes of vertigo from the (very uncommon and more serious) central causes such as a brainstem stroke. The key causes of vertigo are outlined in the table; the clinical features to be elicited are shown in box 1.

In primary care we often identify the patient who has clinically important disease because he or she feels very unwell, or we recognise that within our professional lifetime of experience, this patient “does not fit” with our initial hypothesis of minor disease.9 However, this “intuitive” approach may fail with vertigo. The symptom is often alarming for both the patient and the assessing doctor, yet it is usually due to self limiting disease. Equally, the features that differentiate between major and minor illness may be subtle unless verified by analytical reasoning.
Vertigo: the red flags

Other neurological symptoms or signs

Because structures in the brainstem are closely packed together, vertigo in the absence of any other cranial nerve feature (such as diplopia, facial weakness, facial numbness, dysphagia, dysphonia) or long tract symptom (such as weakness or numbness of the limbs) is unlikely to have a central cause. This reasoning is partly confirmed by a large study of patients with vascular brainstem disease. Fewer than 1% of 407 patients with posterior circulation strokes in the New England Medical Centre posterior circulation registry presented with a single isolated symptom. Another study of 1666 patients aged over 44 presenting to a US emergency department with “dizziness” found that 53% (3.6%) were due to stroke or transient ischaemic attacks, and among patients with dizziness without other symptoms or signs, only 0.7% had had a stroke or transient ischaemic attack. Although case series indicate that 5% of new presentations of multiple sclerosis do have vertigo, a first episode of demyelination is unlikely to be present with vertigo only. Acoustic neuroma is also a very rare cause of isolated acute vertigo.

However, vertigo associated with any other neurological features such as visual blurring, diplopia, facial weakness or numbness, dysphagia, dysphonia, limb weakness or ataxia is likely to be due to brain stem disease such as lateral medullary or cerebellar stroke. These patients should be referred urgently.

Headache

Vertigo in association with a headache may be due to migraine. Vestibular migraine (migrainous symptoms associated recurrently with unsteadiness or vertigo) may be commoner than previously recognised in patients with recurrent symptoms. Two large secondary care studies found that 7% of a total of 544 chronically dizzy patients attending dizziness clinics had vestibular migraine. A population questionnaire study of 8318 participants found that 29.5% reported an episode of vertigo or severe dizziness. Of 243 participants with vertigo who were studied further, 27 women and 6 men (14%) had vestibular migraine. Case studies indicate that vertigo does not necessarily follow the normal time course of other migrainous “auras.” The vertigo may occur before, during, or after the characteristic headache and vomiting. The symptoms may last many days, rather than the usual 5-60 minutes of a typical migrainous aura.

The diagnosis of vestibular migraine needs to be made with caution, particularly in the acute setting, because brainstem vascular events can present in an almost identical way. Prospective studies indicate that between 10% and 34% of all stroke patients experience headaches around the time of the stroke. Headaches are commoner in patients with posterior circulation strokes.

Vestibular migraine is largely a diagnosis of exclusion. Most patients with new onset headache and clear vertigo will need admission to hospital, unless there is a history of recurrent similar episodes.

Fig 1 | Diagnostic strategy with acute vertigo
The normal state
Head movement towards a canal (yellow in figure) will cause activation of that canal, and reflex movement of the eyes in the opposite direction - that is, away from the canal.

The pathological state and the basis of the head thrust test
Head movement towards a defunct canal (blue in figure) will result in failure of activation of the vestibular ocular reflex and thus the visual target will be lost from fixation during sudden head movements. In the head thrust test, the examiner turns the patient’s head with a high acceleration but low amplitude head thrust, in this case to the patient’s left. The test is positive when the patient makes a catch-up saccade to refixate the visual target (usually towards the examiner’s nose).

Vertical nystagmus
Vertical nystagmus indicates brainstem or cerebellar disease. The nystagmus of benign positional vertigo is torsional (“rotatory”) and not sustained. The nystagmus of acute vestibular neuritis is unidirectional (fast phase to one side) and horizontal.

Vertigo without red flags: confirmatory tests
Features characterising benign positional vertigo
In benign positional vertigo, the history is of a sudden onset of positionally induced vertigo which attenuates after 10 to 60 seconds and which is reproducible by head positioning. Often it first occurs when turning over in bed, and 20% of patients may have had a minor head injury. The vertigo is intense and nausea is common but vomiting is unusual. The Hallpike manoeuvre will reveal no other symptoms and no signs apart from torsional (rotatory) nystagmus that has a latency of a few seconds and disappears within about 30 seconds (fig 2; video).33-34

Benign positional vertigo is due to free floating crystals within the lumen of the semicircular canals (usually the posterior canal). It is easily and successfully treated by one of two simple repositioning manoeuvres: the popular Epley manoeuvre or the less well known but simpler Semont manoeuvre. These are illustrated by video clips on bmj.com.

Acute vestibular neuritis
Acute vestibular neuritis is a common diagnosis, defined by sustained (non-positional) vertigo with unidirectional, predominantly horizontal nystagmus (no hearing loss or tinnitus, and no other neurological symptoms or signs).35

The cause is uncertain; nausea and vomiting are common. The patient is usually young or middle aged and presents with sustained vertigo rather than positional vertigo. Some patients have symptoms of a preceding viral respiratory infection. Vertigo may have an abrupt onset (73% of cases) or increase over a few hours (27%), or it can occur on waking. The severity usually peaks within a day and resolves gradually over several days.

A problem for the general practitioner is that, rarely, cerebellar strokes can present with isolated, sustained vertigo. In one case series of 240 patients with cerebellar strokes, 25 had sustained vertigo as the sole feature.36 In elderly patients at high risk of vascular disease this may be a noteworthy possibility if the patient has sustained vertigo.

In recent years a simple test, the head thrust test, has been described (fig 3; video). This fast movement of the head tests the vestibular ocular reflex (which is also the basis of the doll’s eye test in comatose patients). The test always has abnormal results in patients with acute vestibular neuritis, and it was normal in 24 out of 25 patients with isolated vertigo due to cerebellar stroke.37 The vestibular ocular reflex fails with movement of the face towards the side of the affected labyrinth.

New hearing loss
Sudden onset of complete unilateral deafness associated with vertigo suggests acute ischaemia of the labyrinth or brainstem,31 and patients presenting in this way require immediate admission. Other rare causes of unilateral hearing loss with vertigo are an acoustic neuroma or (after straining or trauma) a perilymphatic fistula. All of these diagnostic possibilities should be referred to a specialist. Ménière’s disease may also present with hearing loss and vertigo.

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Practice

*No red flags, no confirmation of benign positional vertigo or acute vestibular neuritis*

If the vertigo is persistent and the head thrust test has negative results, consider admission to hospital for imaging.\(^{23}\) If the vertigo is not persistent and there is a history of recurrent episodes, consider the possibility of vestibular migraine. Ask specifically about photophobia and headache (even mild headache) and any history suggestive of migraine. Non-urgent referral or a trial of treatment would be reasonable.

If there is a suggestion of recurrent cochlear symptoms (“fullness” in the ear, transitory deafness, or tinnitus), consider the possibility of Ménière’s disease. Ménière’s is a rare cause of vertigo, but in the early stages it can be characterised by either paroxysmal vertigo or episodes of fluctuating aural symptoms without vertigo. In one case series of 243 patients with Ménière’s, vertigo was the only initial feature in 29%.\(^{13}\)

**Discussion of case scenario**

Vertigo is alarming for the patient, and anxiety can be expected. On review of this patient the general practitioner felt less confident that the diagnosis was “viral labyrinthitis” (acute vestibular neuritis) or benign positional vertigo. He had “prematurely anchored” the diagnosis, relied on a confirmatory test (the Hallpike manoeuvre), and ignored the data that did not fit—the hoarseness, difficulty swallowing with minimal signs of pharyngitis, and the fact that the positionally induced nystagmus was sustained. More careful questioning elicited facial numbness and slight clumsiness of the left hand. On admission to hospital the patient had Horner’s syndrome on the left side of the face and sustained nystagmus on positioning. Magnetic resonance imaging of the brain showed a left lateral medullary (brain stem) infarct. In this case review allowed the general practitioner’s cognitive errors to be corrected.

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