THE ART and SCIENCE of DIAGNOSIS

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September 2012
Medicine is a science of uncertainty and an art of probability.

Attributed to Osler
If only it were so easy...

Eight signs of cancer
Sydney Morning Herald August 31, 2010

• Scientists have identified eight key signs of cancer which should be investigated immediately.

• The symptoms, including rectal bleeding and difficulties swallowing, predict the risk of cancer with such accuracy in certain age groups that patients should be referred to a specialist straight away unless there is a plausible explanation.

• Identifying these symptoms could improve the early diagnosis of cancer, when people have the best chance of responding to treatment.
Expert Diagnosis: *Tacit Knowledge*

‘The analysis of a skilful feat in terms of its constituent motions remains always incomplete.

...Indeed, the identification of the constituent motions of a skill tends to paralyse its performance.

...We cannot identify, let alone describe, a great number of the particulars that we which are in fact noticing when we diagnose a case of the disease. Though we can identify a case of the disease by its typical appearance, we cannot describe it adequately.’

*Knowing and Being* Polanyi 1961
Dr: And it’s not to do with something that you could actually, point point point point point, it’s to do with the overall, and I think that’s where the art of medicine comes in.....Because there are lots of things that come of the way you just assess the interactions and the discussions, and things like that. That give you a – gut feeling. You know, gut feeling’s not the right word, but you know what I mean?

WH: Hmm. Intuition?

Dr: Yeah, intuition...intuition’s something that in fact, it’s just because you can’t quite articulate. You’re sort of in your mind, ticking off some points, without being able to know exactly what the points are?
What is a “diagnosis”? 
How we teach diagnosis
How diagnosis actually happens...

- Symptoms
- Physical signs
- Test results
- Revised working diagnosis
- Working diagnosis
- More tests
- Another opinion
- More symptoms
- More signs

TIME...
What *is* a diagnosis?

- Diagnoses are conceptual constructs based on professional judgement.
- Diagnoses are not a *disease*, or a *risk factor*.
- Diagnoses are a *label of convenience*.
- Pursuit of diagnostic accuracy has costs and risks

the “myth of certainty”
What is a diagnosis?

But a diagnosis is also:

• A socially accepted category that sanctions treatments and assistance
• A reassurance for patients, families - and doctors!
• A label that incurs obligations
A standard of practice

All Australian medical graduates should have on graduation:

The ability to interpret and integrate history and physical examination findings to arrive at an appropriate diagnosis or differential diagnosis.

AMC Standards for Accreditation of Medical Schools 2010
Who is this person?
Who is this person?
Who is this person?

Analytic or rule based diagnosis
Who is this person?

*Pattern recognition or intuitive diagnosis*
Two systems of reasoning

**Analytic, rule based**

- Good for novices
- Activated by unknown
- Compares options
- Requires time
- Not suited to distracting environments
- Easier to justify
- Affected by stress
- May lead to cognitive overload and procrastination

Adapted from Flin 2008
## Two systems of reasoning

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**Intuitive, experiential**
- Fast, uses emotions
- Little conscious thought or effort
- Good in routine situations
- Resistant to stress
- Needs experience
- May be difficult to justify - “implicit”
- May lead to premature closure

Adapted from Flin 2008
Why do we need both an art and a science of medical practice?
Medicine is inherently uncertain...
Medical uncertainties

A. Disease factors
   - Medically unexplained symptoms (MUS)
   - Poorly understood diseases, “functional” diseases
   - Natural and induced fluctuation over time
Medical uncertainties

B. Limits of diagnostic information

– Ability to elicit history and detect physical signs
– Borderline abnormal findings
– Limitations of laboratory tests: no “gold standard”
Laboratory investigations

“the doctor should have done more tests”

– Limitations of tests (sensitivity and specificity)
– Risks to patient, costs and bother
– Mayo Clinic Study (2006)
  • 50% diagnoses made by clinical judgement
  • 31% by imaging (X-ray, CT, MRI)
  • 9% blood test, 3% biopsy, 5% other tests

⇒ Will the result change the diagnosis or treatment?
Human variation & tests

**False positives** = Don’t have disease but have a positive test result

**False negatives** = Have the disease but have a negative test result
Medical uncertainties

C. Patient factors

– Incomplete communication  
  e.g. CALD patients, children, cognitively impaired
– Difficult interactions (not “difficult patients”!)  
  e.g. “Heartsink” patients
“Heartsink” patients

- Frequent attenders
- Many complaints but little improvement
- No clear diagnosis or treatment
- Induce feelings of anger, frustration or despair
- Doctor’s response ➔ “heartsink” patient
Medical uncertainties

E. Doctor factors
- Failure to seek advice and information
- Ignorance (rare) or carelessness (more common)
- Influence of bias, personal values

F. System factors
- Poor communication between health professionals
- Lack of continuity of care, patient information
- Lack of supervision
How can doctors respond to diagnostic uncertainty?
Tolerance of uncertainty

• Responses to uncertainty depend on the doctor’s capacity to tolerate uncertainty

• Tolerance of uncertainty is associated with choice of specialty career:

  Psychiatry > General Practice > General Medicine > Paediatrics > O &G > Surgery

(Gerrity 1992)
Diagnostic uncertainty

GPs manage “undifferentiated” or imprecise symptoms often at early stages in the development of an illness. This means that important decisions for patients have to be taken on the basis of limited information, with early signs of disease often non-specific to a particular condition. Having excluded an immediately serious outcome, the decision may well be to await further developments and review later.

“Characteristics of a GP” Royal College of General Practitioners UK
1. **What is the probability diagnosis?**
   - Common things occur commonly

2. **What serious conditions must not be missed?**
   - “Red Flags”, “Alarm bells”, commonly missed conditions

3. **Could this patient have one of the “masquerades”?**

4. **Is this patient trying to tell me something else?**
   - Hidden agendas

After Murtagh, 2007
What we teach students

• Listen to the patient - and the family
• Take a comprehensive history
• Examine the patient thoroughly
• Stop and think about missed diagnoses
• Order tests only if the result will make a difference
• Explain your thinking
• Offer to get another opinion
• Use safety nets; ensure follow up
• Reflect on your own response
What we teach students

- Listen to the patient - and the family
- Take a comprehensive history
- Examine the patient thoroughly
- Stop and think about missed diagnoses
- Order tests only if the result (normal or abnormal) will make a difference
- Explain your thinking
- Offer to get another opinion
- Use safety nets; ensure continuity and follow up
- Reflect on your own response

Don’t abandon the patient
THE ART and SCIENCE of DIAGNOSIS – a rheumatologist’s emphasis

Neil McGill
Rheumatologist
Royal Prince Alfred Hospital, Uni of Sydney
History

• **Most important step to diagnosis**
  – Patient listening (for a while)
  – Value of “off target” questioning (social, work, general health etc)
  – Disallow use of diagnostic labels
  – Beware terms that mean different things to doctors compared to the rest of the world
    • Hip, shoulder, numbness…
Examination

- Starts in the waiting room
- Asymptomatic regions / side first
- Negative control essential for interpretation of provocative tests
Investigations

• **Need to be interpreted in light of history & examination**
  – Ideally the images should be described by the radiologist or nuclear physician etc
  – Clinical relevance should be determined by the clinician

• **Prevalence of abnormal findings in asymptomatic people should be considered**
A tennis injury?

- 78M “It started with a fracture….”
- Fell over playing tennis
- Developed groin pain
- 4 weeks later CT/SPECT = pubic ramus #
- Specialist → drug treatment for osteoporosis
- Progressive pain
History

- 78M
- Fell over playing tennis. *No pain.*
- 2 weeks later developed *bilateral* groin pain.
- 4 weeks later CT/SPECT = *unilateral* pubic ramus # (on low resolution CT, no abnormal radionuclide uptake)
- Specialist → drug treatment for osteoporosis
- Progressive pain *in both groins, both shoulders*
Cognitive Autopsy

Dr Romesh Singam
Report Card

• **ED physicians are accurate, could do better**
  – 98% ortho/83% surgery/78% medicine

• **Misdiagnosis assoc with bad outcomes**
  – 30% discrepancy btn diagnosis and post-mortem
  – Canada 50% closed malpractice due to misdiagnosis

• **Accurate diagnosis cornerstone of efficient ED**
“ED is a laboratory for errors”
Croskerry 2009

Disease factors

System factors

Patient factors

Doctor factors

Diagnostic information
Type 1 vs Type 2 Thinking

Fig 1 – The deterministic model

History taking

Physical examination

Investigations

Diagnosis

Treatment

Fig 2 – The tactical performance model

b) The tactical performance model

Immediate treatment  Monitoring

Unstable patient  Stable patient  Disposal

Physical examination  Problem Orientated History Taking
• Why not more Type 1 thinking?
  – ED grind to a halt
• Type 2, 30 types, heuristics
• metacognitive psychology suggest dual-process thinking is ideal i.e. cognitive autopsy
How do I think?

**Individual**
- Resuscitation
- Likely diagnosis
- ROWCS
- Rate limiting step?
- Try to document
- Assess data
  - More testing?
  - Home or admit?
  - Remember cognitive autopsy!

**Departmental**
- Is my department safe?
- Any staff sick, what are my resources vs demands
- Free beds, disposition ASAP
- Bottlenecks
- KPI!!!
- CQI
thankyou
THE ART and SCIENCE of DIAGNOSIS

Mark Burns
MBBS MSafSc FAFOEM(RACP) FAADEP

September 2012
Diagnosis in Medical Assessments
Vs
Medical Treatment

• In Medical Treatment information provided by patient is to best knowledge of patient to aid diagnosis, treatment and improvement.

• In medical assessments information provided by patient and/or parties is often biased to obtain a set outcome.
Factors affecting Diagnosis

The factors affecting diagnosis remain the same as those discussed by Dr Hu.

• Disease factors
• Patient factors
• Limits of Diagnostic Information
• Doctor Factors
• System Factors
Case - R

History
• 72 year old man with memory problems and word finding difficulties.
• Referred for assessment of “Solvent Neurotoxicity”
• Employed for 8 years in solvent recycling plant
• Memory problems commenced after he ceased work at age 65.
Case - R

- Very Poor Historian
- History from wife – “used to come home high after work”
- MRI scan of brain – no focal abnormality
- Psychometric testing (x2) – generalised cognitive deficits.

BUT
Case - R

Further History

• 2000 – Diagnosed as maturity onset diabetic (poorly controlled)

• 2001 – Cardiopulmonary bypass surgery with prolonged unconsciousness

• 2001/2 – Diagnosed with severe sleep apnoea – history of poor compliance with CPAP machine

• 2005 – legionella pneumonia with ICU admission, severe fever and delirium
Case - R

What is the correct diagnosis?
How doctors think?

An illustrative presentation

A/ Professor Hadia Haikal- Mukhtar
B Sc Hons; MBBS; FRACGP ; LLB Hons.
How doctors think

• Performance versus competence
• “A competent doctor performing poorly”
  –Why?
How doctors think

Work environment
  – Unsupportive
  – Lack of appropriate resources

Systems in place:
  • Hand-over and follow-up
  – Examples from Private practice
  – Examples from hospital practice
How doctors think

Doctor’s factors: Distractors

- Health: drug, alcohol, mental health, cognitive impairment and others
- Personal and family pressures
- Financial pressures
- Excessive work load and fatigue
- Job dissatisfaction
- Outlier practice
- Professional isolation
- Bias
- “Personality”/ “Disruptive behaviour”
- Cultural barriers
How doctors think

Patient factors:

– the VIP and special patient: Professional boundaries
– The “crying wolf patient”
– The “too hard basket” patient
– The “patient from hell”
– The “too familiar” patient
How doctors think?

Food for thought:

– screening or reactive performance assessment?