

Diagnosis: an impossible but essential task?

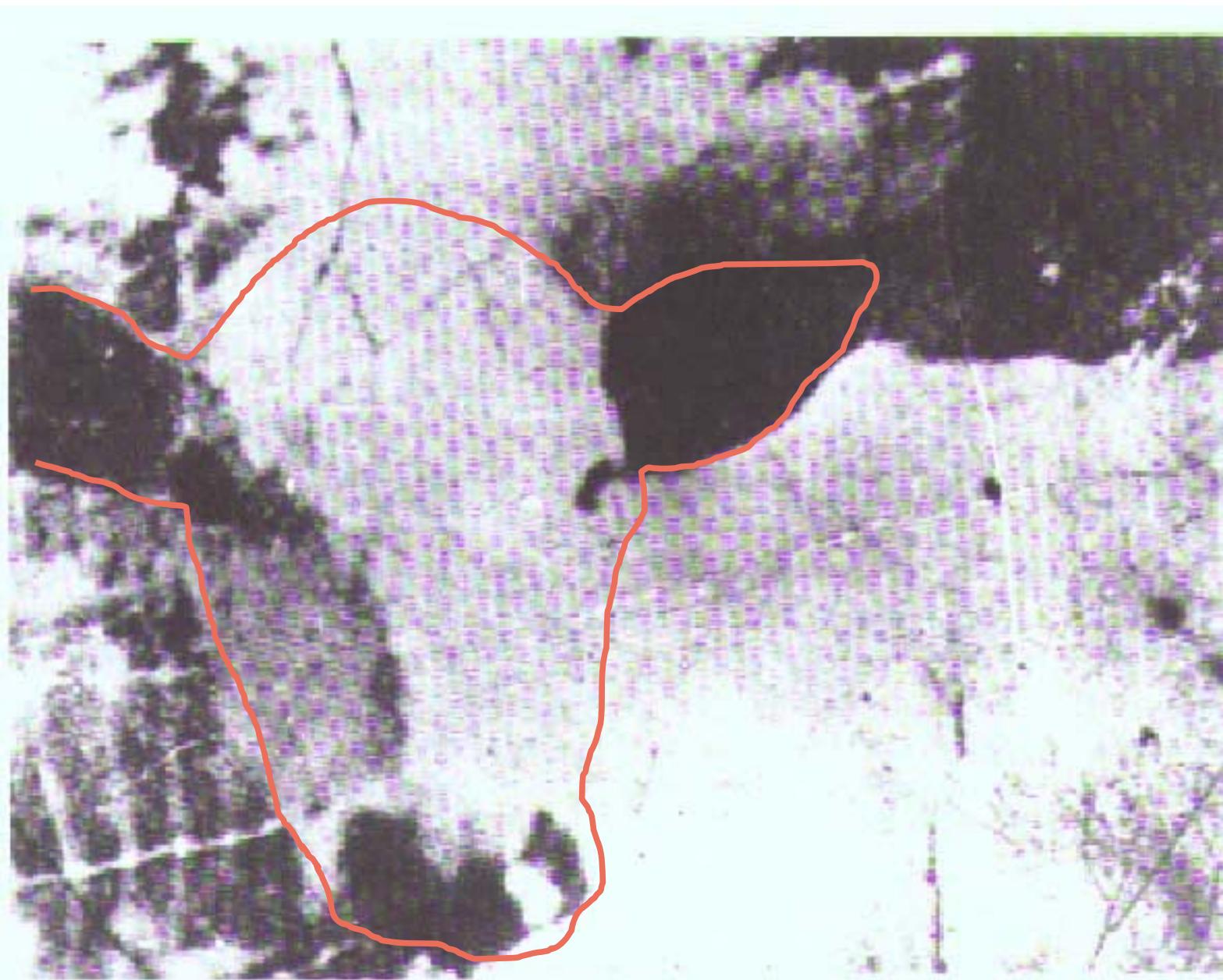
Professor Paul Glasziou
Bond University, Australia
Paul_Glasziou@bond.edu.au



WONCA Copenhagen 2016

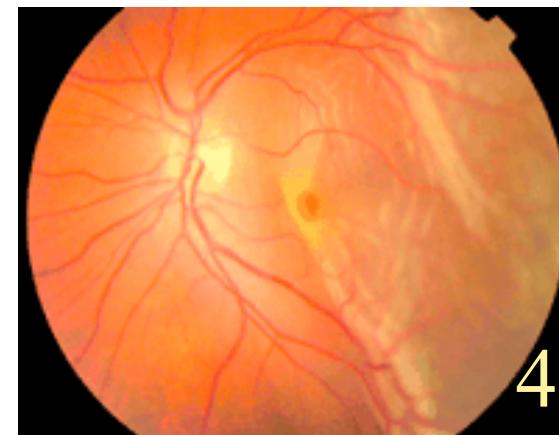


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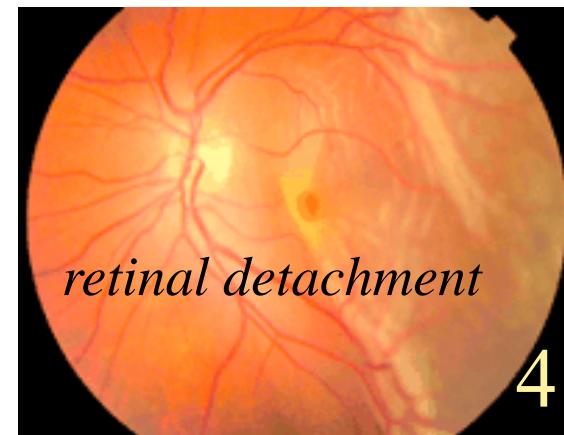
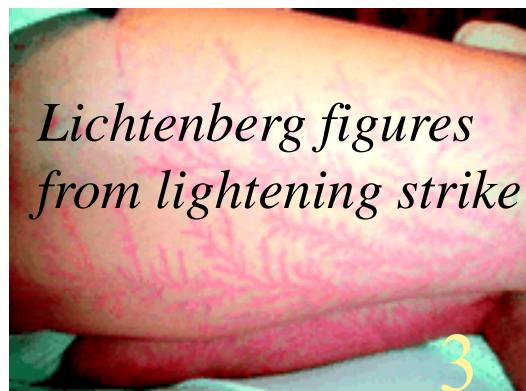
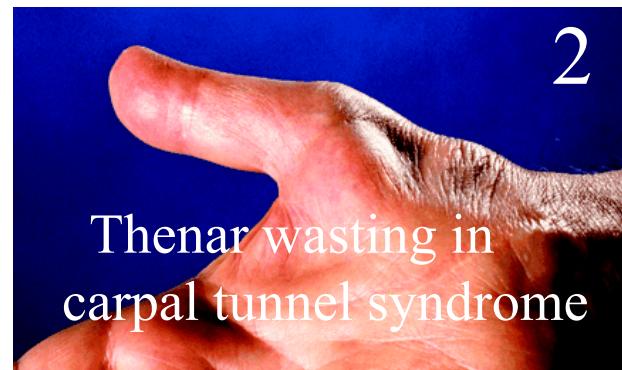
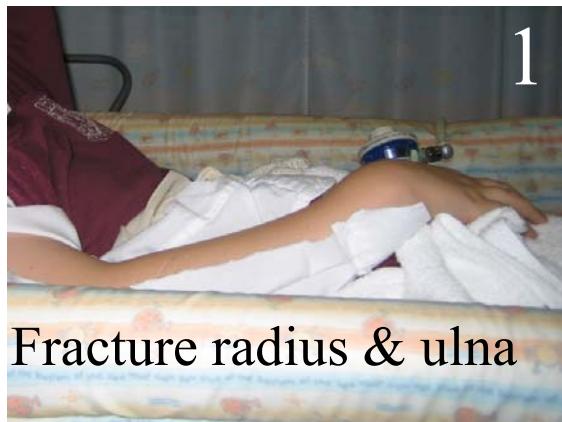


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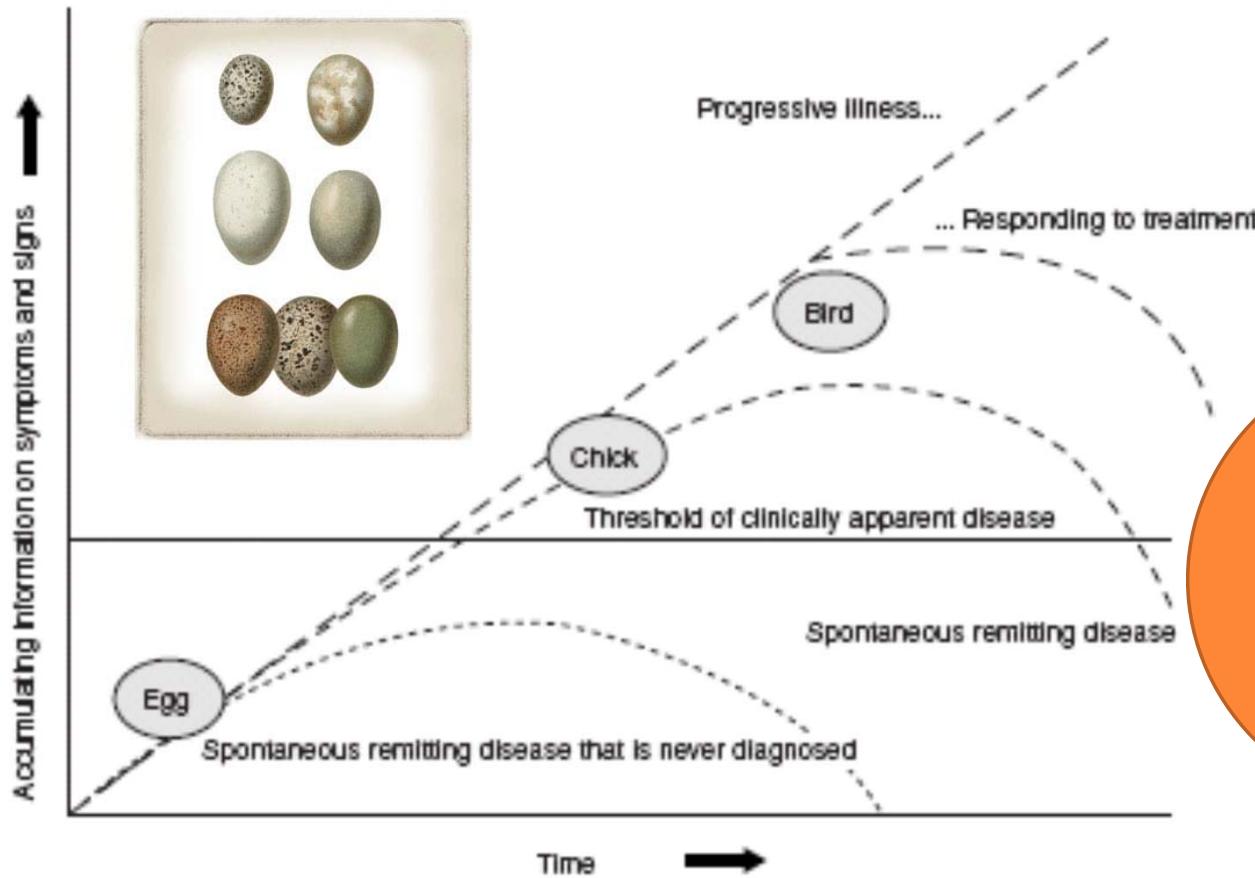
Diagnosis by visual pattern matching quick – but requires experience of pattern



Diagnosis by visual pattern matching quick – but requires experience of pattern



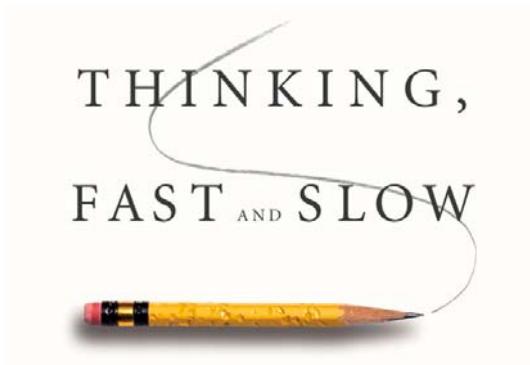
Patterns change with time



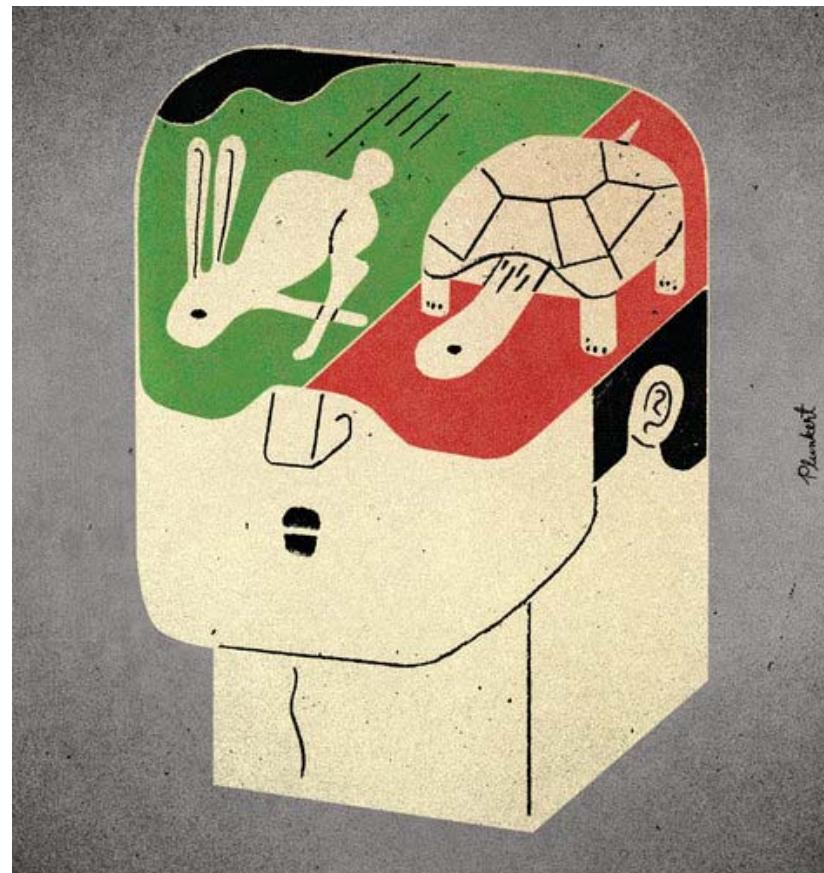
Del Mar, Doust, Glasziou
Clinical Thinking: Evidence, Communication and Decision-Making

Diagnosis not
essential for
spontaneously
remitting
diseases

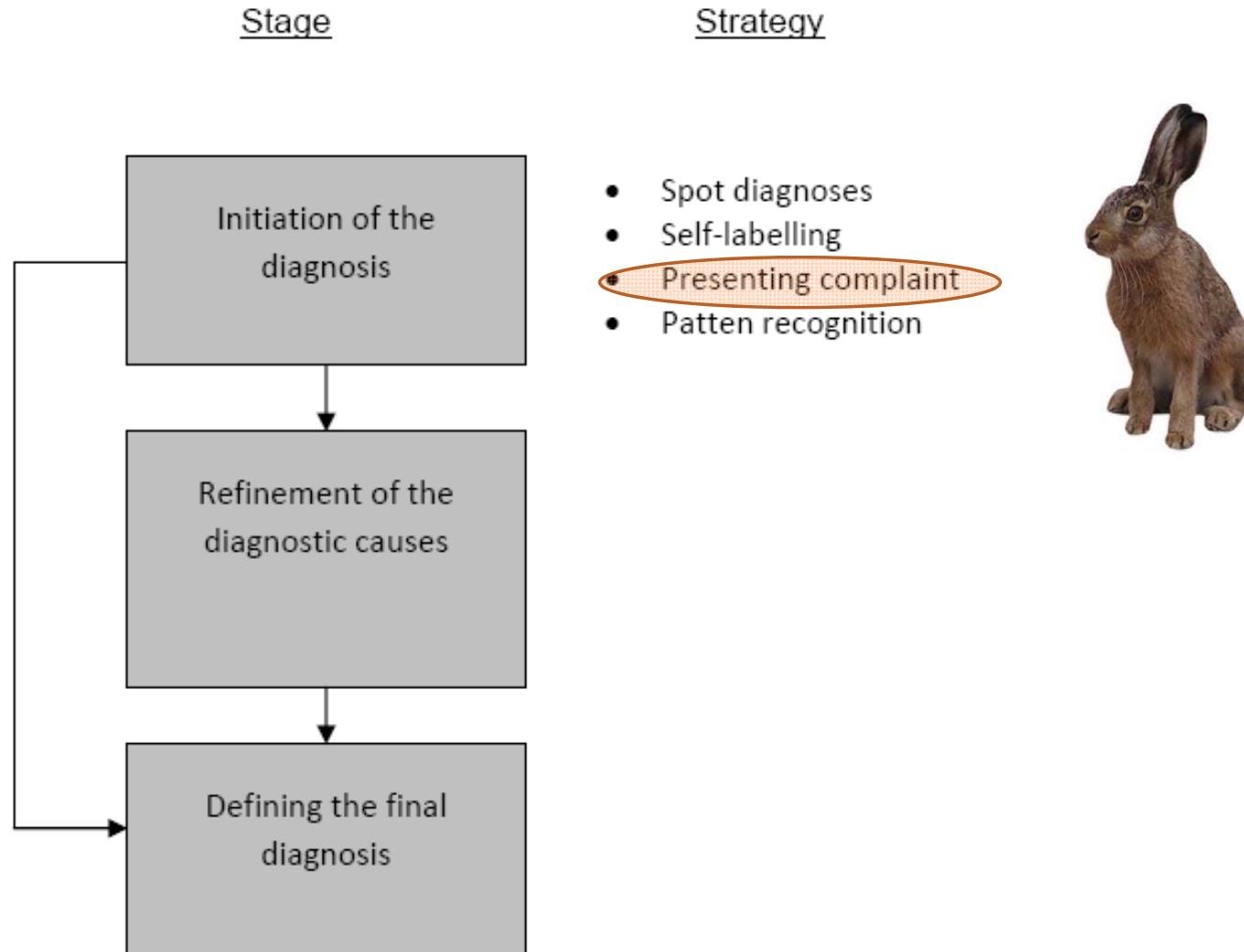
Types of Diagnostic Thinking processes



- Fast thinking:
Intuitive, heuristic
- Slow thinking:
analytical, deliberate



Diagnosis: 3 Stages and 14 Skills



Heneghan , Glasziou, Thompson et al. Diagnostic strategies used in primary care BMJ 2009

Initiation: Presenting Complaint

Lately I feel **tired** all the time ...

Patient 1: Few specific features,

- But Beck DI suggested depression
- Inquiry -> **Depressed**

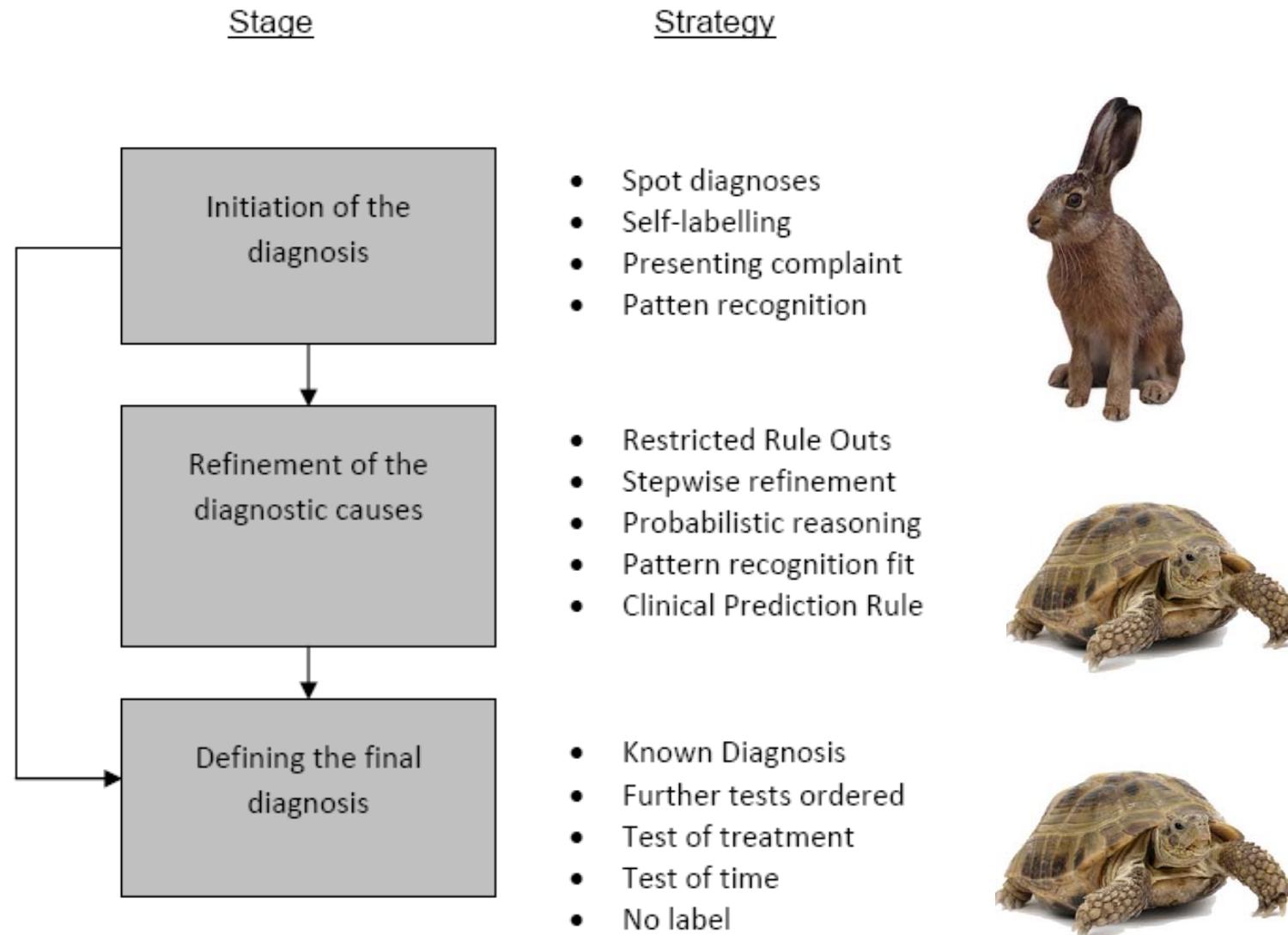


Patient 2: Few specific features,

- But on taking BP, noted pulse of 30
- Complete Heart Block**



Diagnosis: 3 Stages and 14 Skills



Heneghan, BMJ 2008

Tired: Is the patient anaemic?



302 patients scored by 3 clinicians vs Hemoglobin
Scores for agreement between paired observers were
 κ appa = 0.75 between observers A and B and
 κ appa = 0.54 between observers A and C.

Conjunctival pallor for anaemia

- Sensitivity = 55%; Specificity = 75%

Is this a good test?

Sensitivity = 50%; Specificity = 50%



Positive test



Negative test

Table 3. Likelihood Ratios for Conjunctival Pallor with Anemia Defined as Hemoglobin ≤ 90 g/L

Pallor	Hemoglobin ≤ 90 g/L (n)	Hemoglobin ≥ 91 g/L (n)	Likelihood Ratios (95% CI)
Present	8	8	4.49 (1.80, 10.99)
Borderline	22	55	1.80 (1.18, 2.62)
Absent	25	184	0.61 (0.44, 0.80)



Clinical Rules: combine several weakly predictive symptoms, signs, and tests

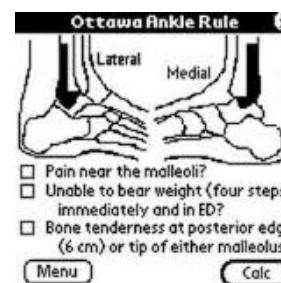
- Charchot's triad – 1880's
 - jaundice; fever; RUQ pain



- Apgar score – 1953
 - 0-10 Scale

	0	1	2
Appearance	Pale	Blue	Pink
Pulse	Absent	<100	>100
Grimace	Absent	Grimace	Cry Active
Activity	Limp	Some tone	Active
Respiration	Absent	Irregular	Reg & Cry

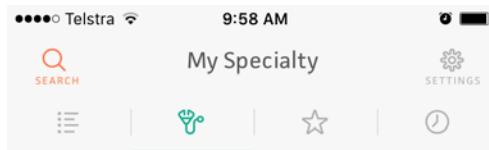
- Ottawa Rules
 - Ankle
 - Knee
 - Cervical spine



- Well's DVT & PE rules

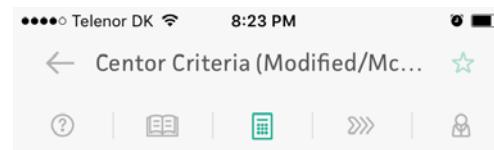
Clinical Prediction Rules as ‘instant’ experience

MDCalc – free App, 170 prediction rules; 53 for GPs



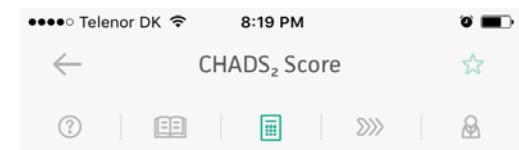
- C**
 - ⌚ Canadian CT Head Rule ★
Very sensitive, rules out imaging in many.
 - ⌚ Centor Criteria (Modified/McIsaac) ★
Strep throat diagnosis and treatment.
 - ⌚ CHADS₂ Score ★
Stroke risk in AFib.
 - ⌚ CHA₂DS₂-VASc Score ★
Stroke risk in Afib, better than CHADS2.
 - ⌚ Corrected QT (QTc) ★
Corrects QT interval for heart rate extremes.
 - ⌚ COWS Score ★
Severity of opiate withdrawal.
 - ⌚ Creatinine Clearance ★

A
B
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W



- ⌚ Age Range (Group A) ★
Streptococcus(GAS) Rare Under 3 3-14... +1 ▾
- ⌚ Exudate or Swelling on Tonsils +1 Y N
- ⌚ Tender/swollen anterior cervical lymph nodes 0 Y N
- ⌚ Fever (T > 38°C, 100.4°F) +1 Y N
- ⌚ Cough Coug... +1 ▾

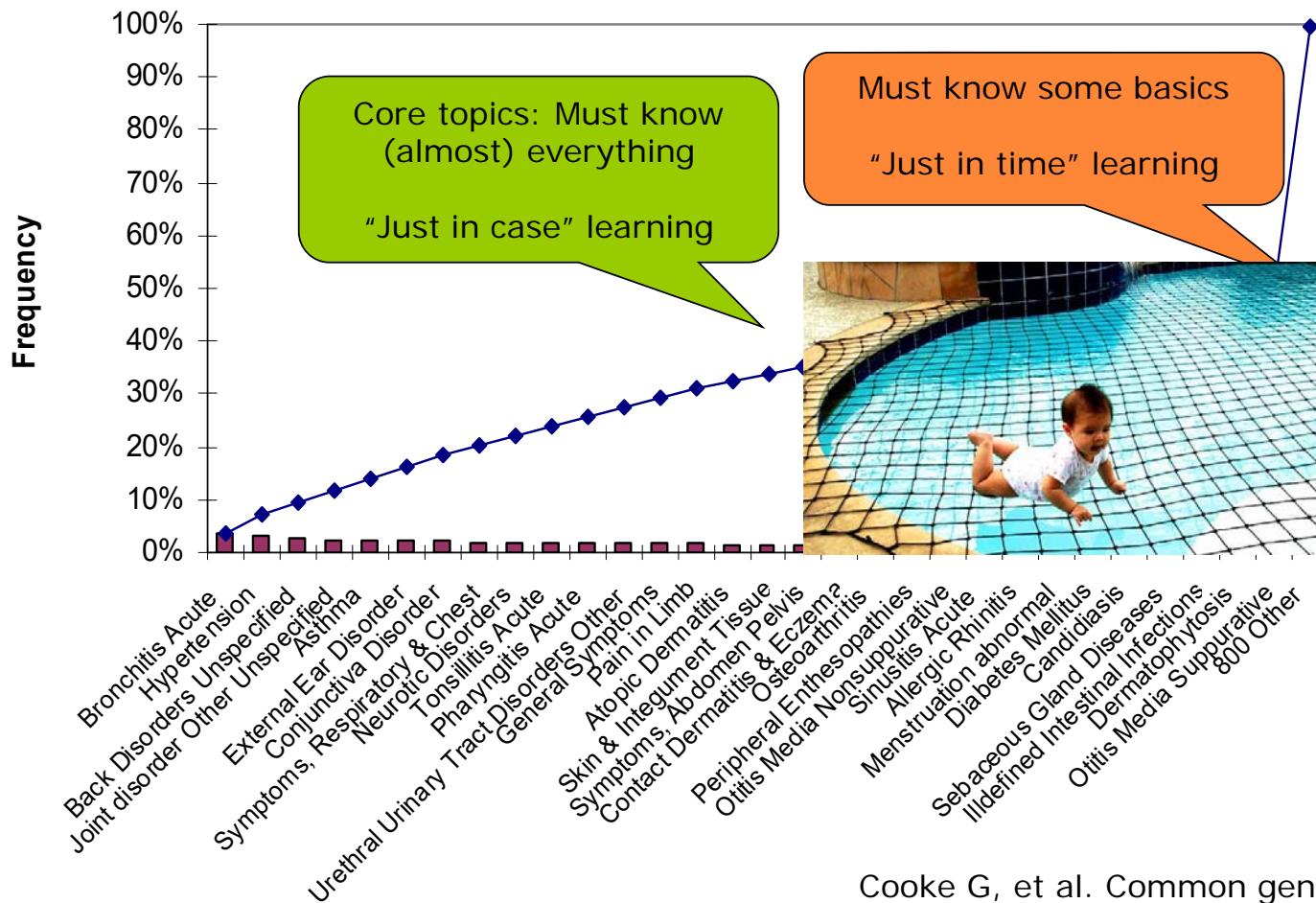
4
points 51% - 53% Probability of Strep Pharyngitis ↑



- ⌚ Congestive Heart Failure History 0 Y N
- ⌚ Hypertension History +1 Y N
- ⌚ Age ≥ 75 years 0 Y N
- ⌚ Diabetes Mellitus History 0 Y N
- ⌚ Stroke or TIA Symptoms Previously +2 Y N

3
points 5.9% risk of event per year. ↑

Half GP consultations are for 30 conditions Other half are for over 800 conditions



The number of diseases/diagnoses

NLM MetaThesaurus - 875,255 concepts

Diagnosis Pro (www.diagnosispro.com) 1 disease per day for 40 years

- 13,000 diseases;
- 30,000 abnormalities (symptoms, signs, tests)



How many rare diseases are there ?

There are thousands of rare diseases. To date, six to seven thousand rare diseases have been found and approximately five new diseases are described every week in the medical literature. This number also depends upon the accuracy of the



RARE DISEASE | UK

The National Alliance for people with rare diseases & all who support them

- 6-8,000 rare* diseases
- 7% of population
- 68% saw 3+ doctors before diagnosed

*Rare = less than 5/10,000



Syndrome Without A Name



[Support & Information](#) [Get Involved](#) [News & Events](#) [About us](#) [Contact Us](#)

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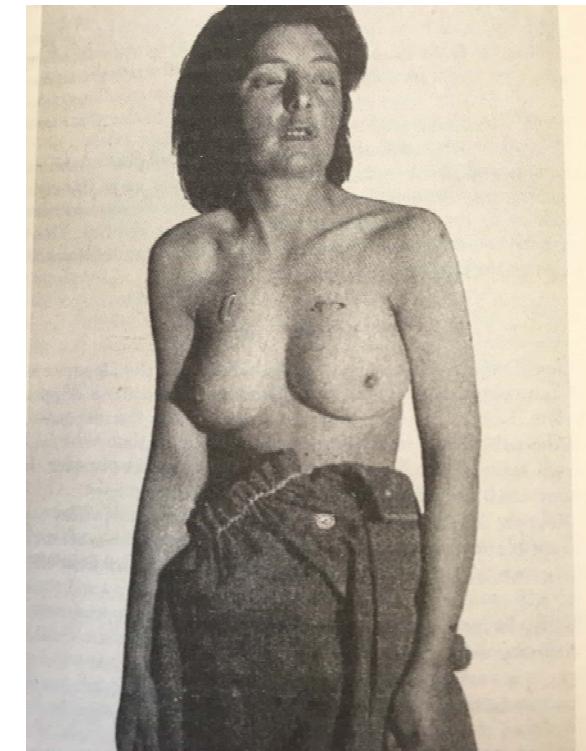
**We support families
and those affected
by a syndrome
without a name**



[How can I get a diagnosis for my child?](#)

[How can I make contact with other families?](#)

[How can I fundraise for SWAN UK?](#)



Hysteria or syringomyelia?
(From: Toscanini's Fumble)

Summary so far

- Diagnosis uses both fast & slow processes
- Fast (“intuition”) requires experience
- CPG’s provide “experience”
- Over 10,000 diseases
- ½ consults involve 30 conditions

Diagnosis in general practice

Latest from The BMJ

Practice

[Clinical prediction rules](#)

Published 07 August 2009

Practice

[Acute diarrhoea in adults](#)

Published 15 June 2009

Practice

[Test of time](#)

Published 15 June 2009

Practice

[Diagnosis using “test of treatment”](#)

Published 24 April 2009

Practice

[Chronic cough in adults](#)

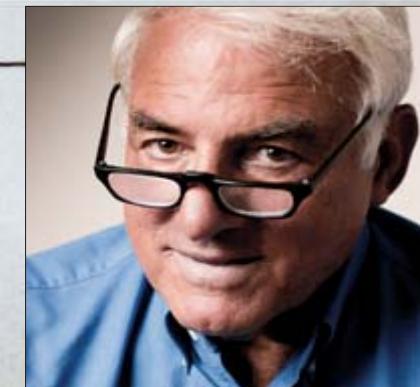
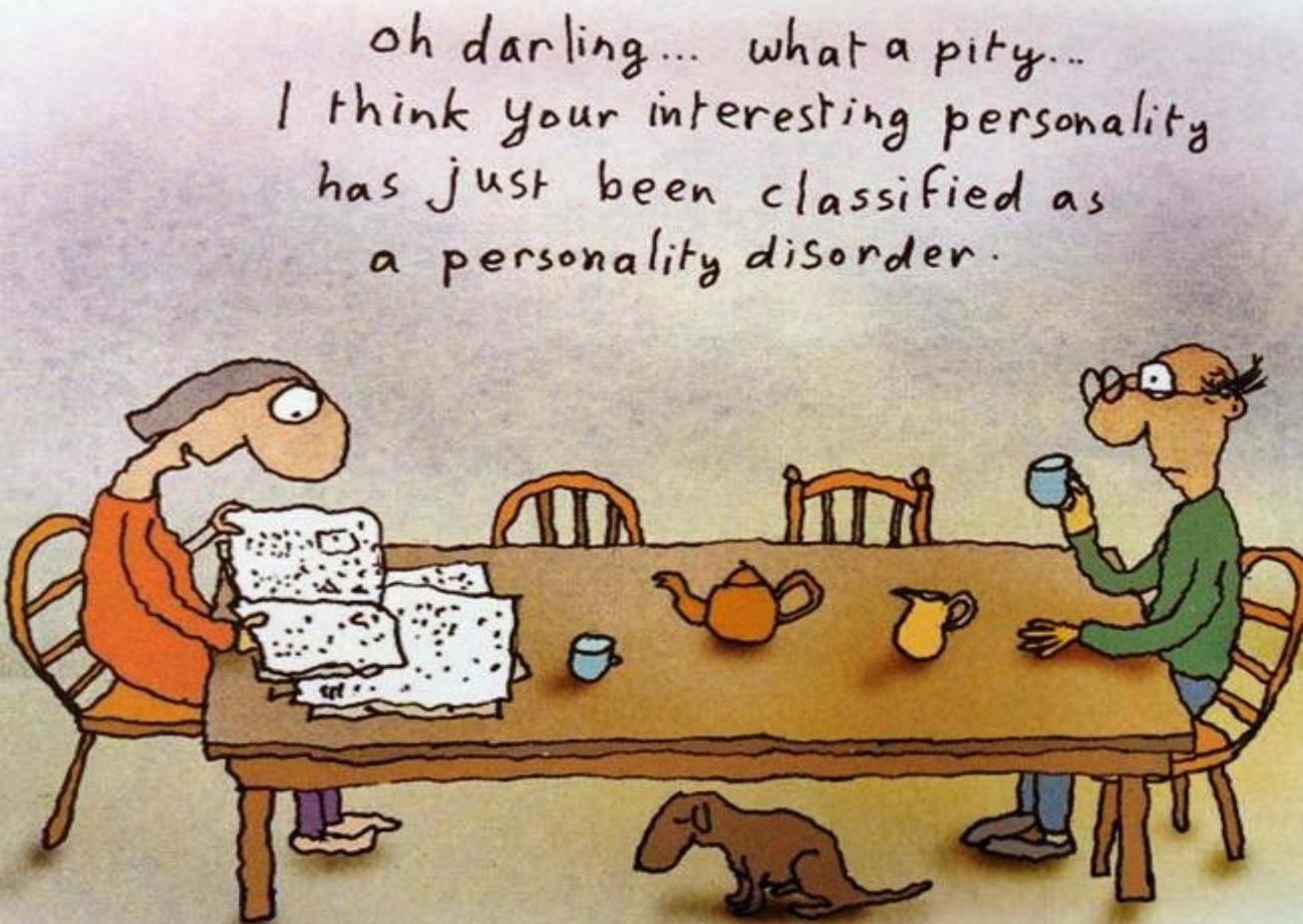
Published 24 April 2009

Practice

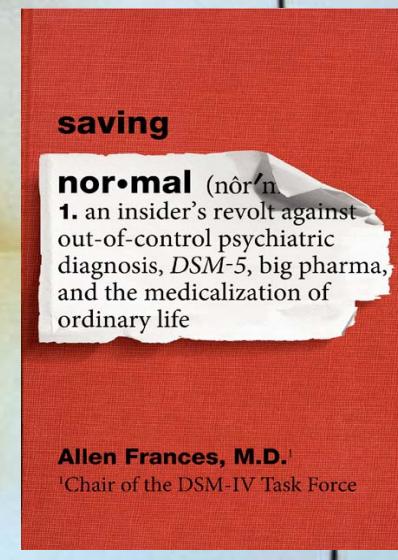
[Diagnostic strategies used in primary care](#)

Published 20 April 2009

Overdiagnosis



Allen Frances
Chair DSM IV

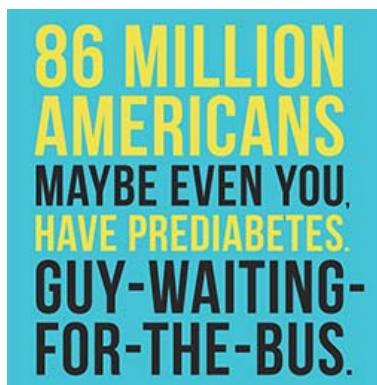


Three types of Overdiagnosis



News Home

Early detection key to treating thyroid cancer, experts say



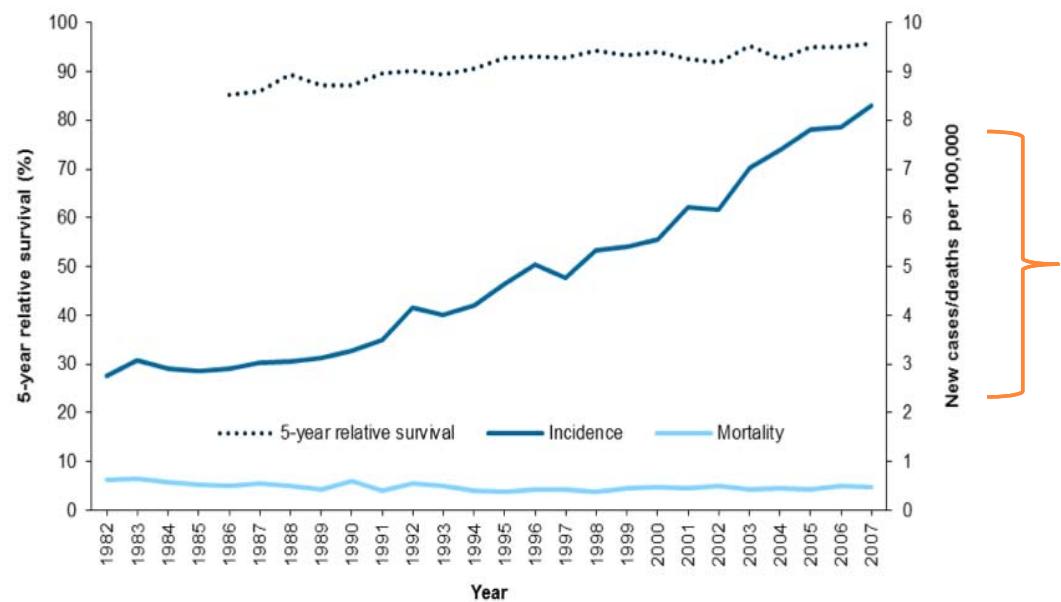
1. Overdetection

2. Overdefinition

3. Medicalization

A. Overdetection: a thyroid cancer 'epidemic'?

Thyroid cancer tripled in 25 years; no more deaths



Notes

1. Incidence and mortality rates are age standardised to the Australian population as at 30 June 2001 and are expressed per 100,000 population.
2. Survival data for this figure are presented in online Table S26.3.

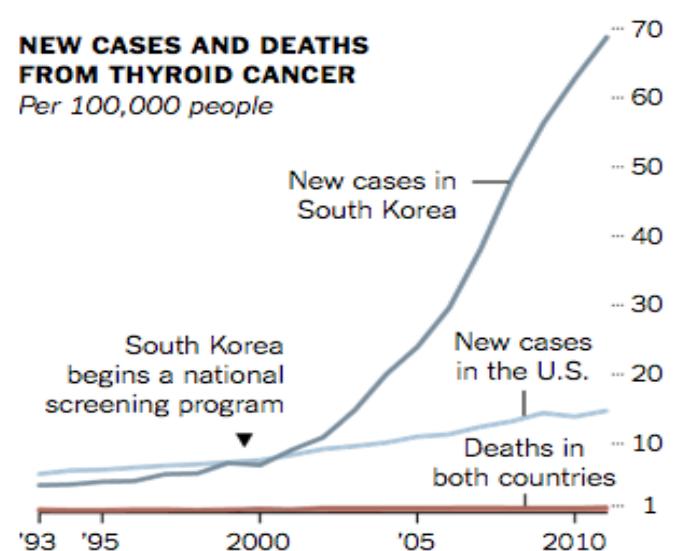
Source: AIHW Australian Cancer Database (2007); AIHW 2010b.

Figure 4.73: Yearly trends in incidence, mortality and 5-year relative survival of thyroid cancer, 1982 to 2007

Screening for Thyroid Cancer

Since South Korea adopted widespread cancer screening in 1999, thyroid cancer has become the most diagnosed cancer in the country. But if this early detection were saving lives, the already-low death rate from thyroid cancer should have fallen, not remained steady.

NEW CASES AND DEATHS FROM THYROID CANCER Per 100,000 people



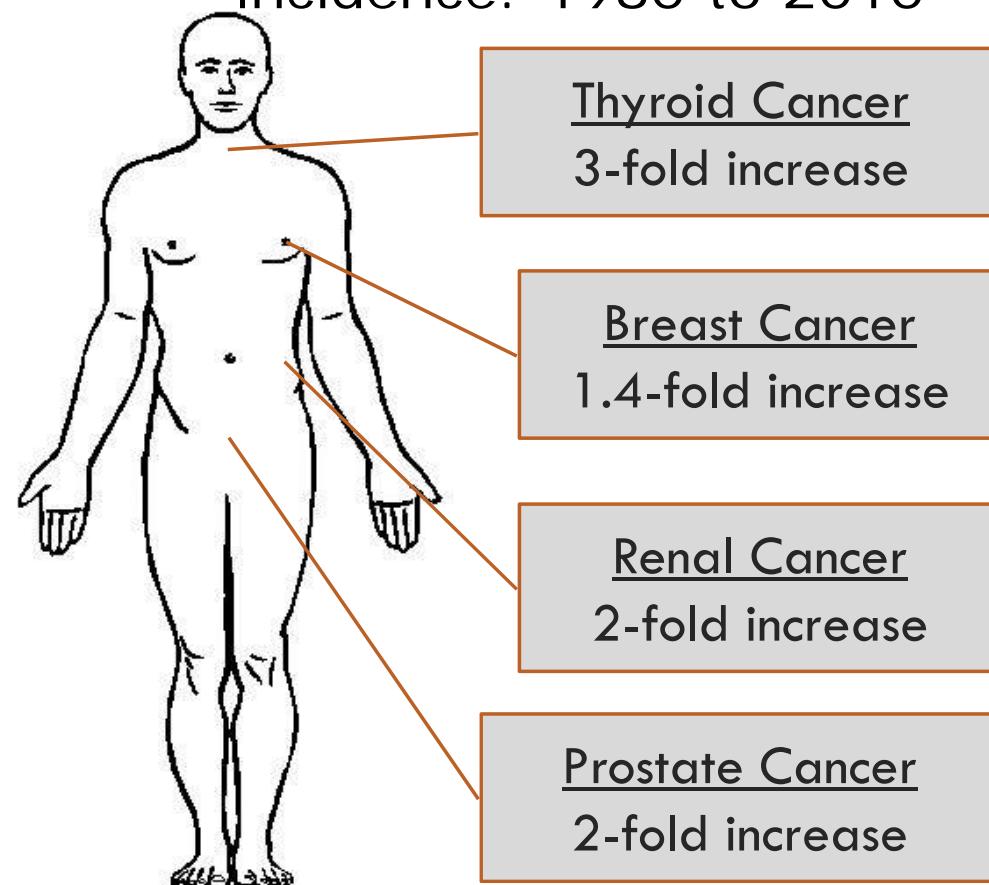
Sources: New England Journal of Medicine; National Cancer Institute

By The New York Times

Overdetection

CANCER

Increase in adult cancer incidence: 1980 to 2010



Overdetection

MUSCULOSKELETAL

Imaging findings in 50-60 yr old asymptomatic adults

Ultrasound shoulder[8]

Rotator cuff tear – 13%

MRI Spine[10]

Disc bulge – 78%

Disc protrusion – 28%

MRI Knee[9]

Meniscal damage – 20%

Any abnormality – 84%

CANCER

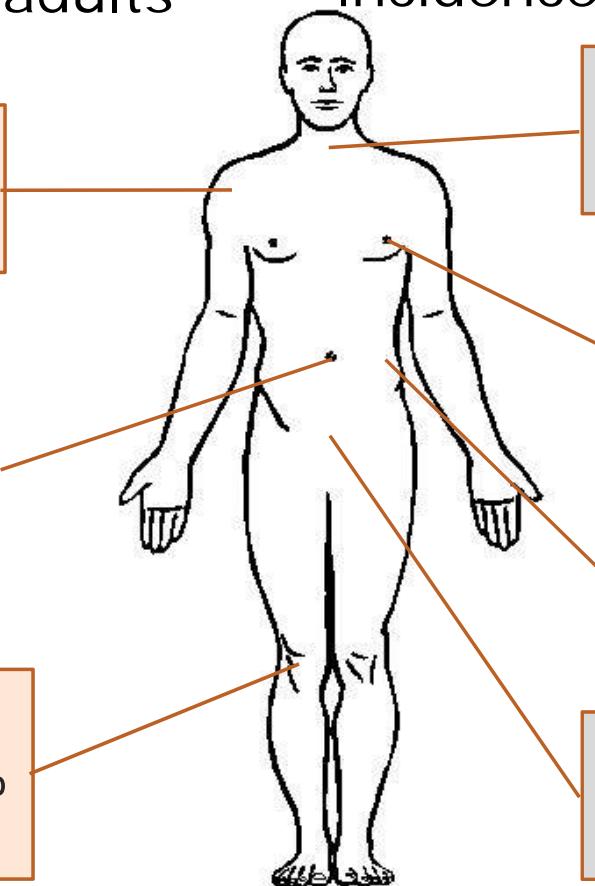
Increase in adult cancer incidence: 1980 to 2010

Thyroid Cancer
3-fold increase

Breast Cancer
1.4-fold increase

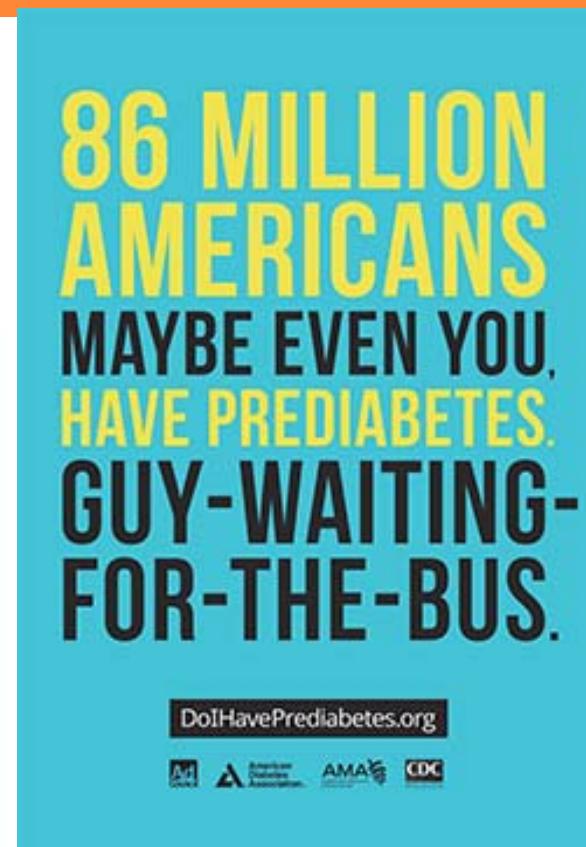
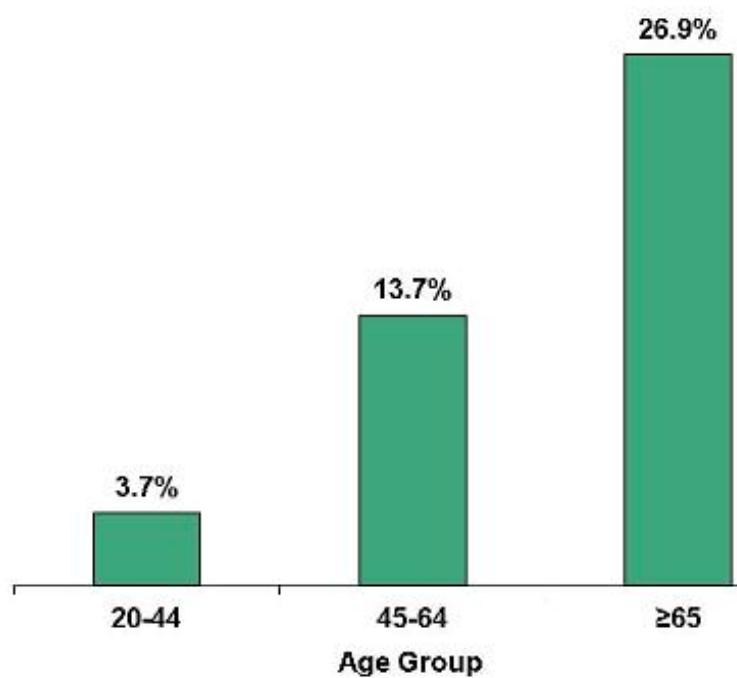
Renal Cancer
2-fold increase

Prostate Cancer
2-fold increase

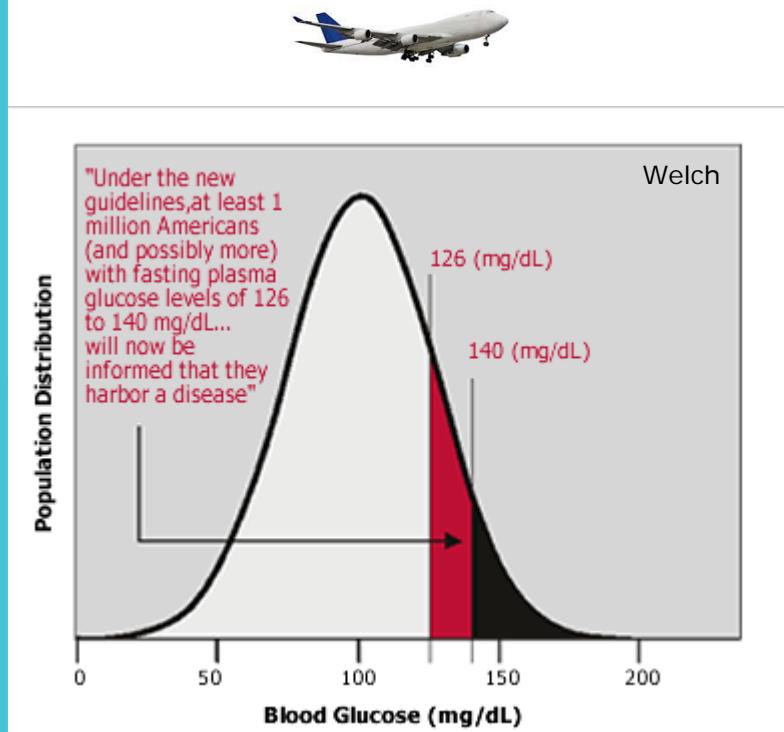


B. Expanding definitions of Diabetes

US Prevalence by age



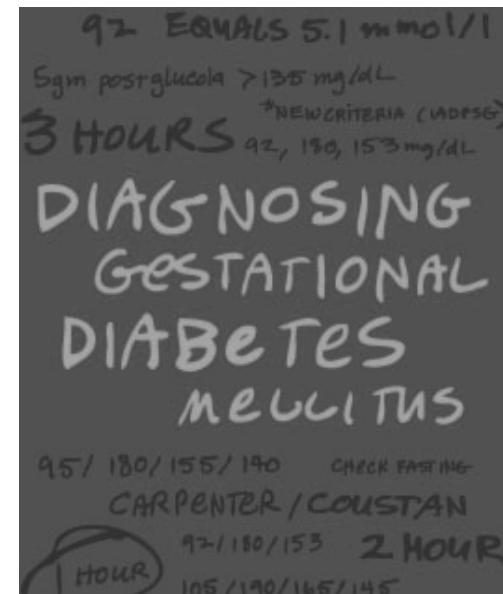
Change of Definition



2003 ADA update

A Tale of Two GDM Definitions

- **2008:** New criteria for Gestational Diabetes (GDM) by *International Association of Diabetes in Pregnancy Study Group (IADPSG)*
 - Increase GDM diagnoses from **6% (WHO)** to **18% (IADPSG)**
 - **Cost** extra \$US2.5 billion/year
- **2013: NIH** NO evidence treatment of new cases improve outcomes



NIH Development Conference:
Diagnosing Gestational
Diabetes Mellitus

March 4–6, 2013
Bethesda, Maryland

Expanding Disease Definitions in Guidelines and Expert Panel Ties to Industry: A Cross-sectional Study of Common Conditions in the United States

Raymond N. Moynihan^{1*}, Georgia P. E. Cooke¹, Jenny A. Doust¹, Lisa Bero², Suzanne Hill³, Paul P. Glasziou¹

1 Bond University, Robina, Australia, **2** University of California, San Francisco, San Francisco, California, United States of America, **3** Australian National University, Acton, Australia

- Of 16 publications on 14 common conditions, **10 widened** and 1 narrowed definitions.
- Widen by 3 methods: (i) “pre-disease”; (ii) lowering thresholds; (iii) earlier or new diagnostic methods.

CONCLUSION:

“research and policy attention might be directed at designing new processes for reviewing disease definitions, free of financial conflicts of interest and informed by rigorous analysis of benefits and harms.”

C. Medicalization

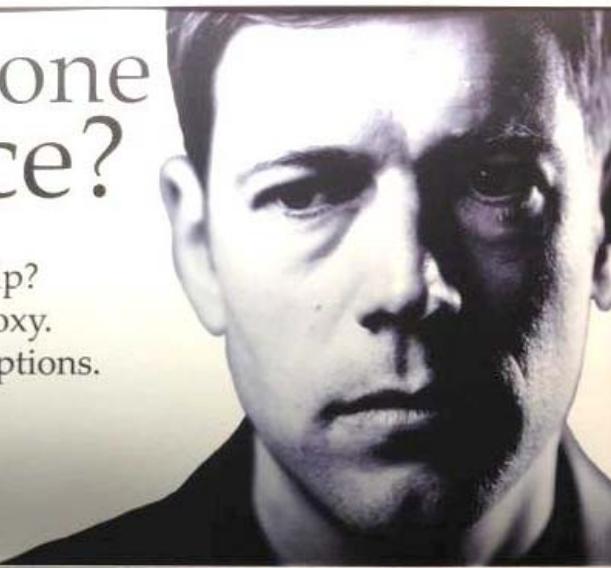
Do you hate someone else's appearance?

Is it making you miserable? Ruining a relationship?
You may have Body Dysmorphic Disorder by proxy.
Contact us to learn about research and therapy options.



(617)643-6204

mghocd.org/bddbp



Then you might have
"Body Dysmorphic Disorder by Proxy"

Medicalization of normal aging

ADVERTISEMENT

BREAKTHROUGH

Hope Has Arrived For Men Over 30 With Low Testosterone

Now, as part of a New Health Drive, Australian Male Hormone Clinic is offering FREE MEDICAL ASSESSMENTS - limited number of places available. Before 29/05/2016.

Finally! After 5 years of intensive research, this much awaited breakthrough is available to the public, for the first time.

Medical experts believe this breakthrough could potentially be the most effective way ever to restore testosterone balance, increase stamina and improve libido.

Patients are already reporting more desire, more passion, and harder, longer-lasting erections. Others are building solid muscle, losing soft belly fat, feeling stronger and more powerful - in a few short weeks.

As part of a public health drive, the Australian Male Hormone Clinic is offering FREE MEDICAL ASSESSMENTS - for the next week only.

Why Will This Work For You - No Matter What You've Tried Before

Many existing treatments are ineffective because they use artificial chemical testosterone. These chemicals are foreign to your body, so they are not as effective - even potentially dangerous.

The Australian Male Hormone Clinic's groundbreaking treatment on the other hand stimulates natural testosterone production.

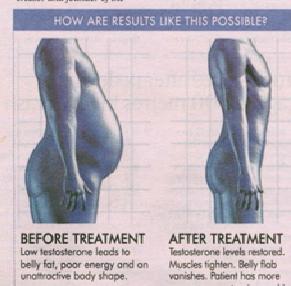
"Natural testosterone is better because your body recognises it. It's almost like turning back the clock to your teenage years again," explains Jowett...

After creating individually tailored programs based on patient's blood results and testosterone levels, the results were truly dramatic.

The wife of one patient sent flowers as a thank you for saving her marriage. Another patient started dating again for the first time in years. And a Sydney man packed on muscle so quickly his family didn't know what to think.

"This treatment helps patients take their manhood back, even when nothing else has worked," explains Geoff Jowett, creator and founder of the

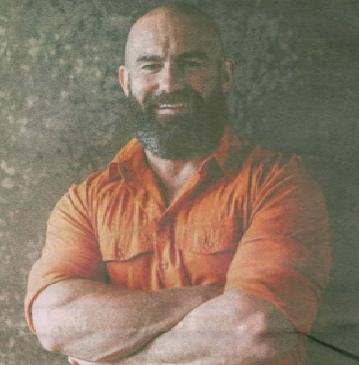
HOW ARE RESULTS LIKE THIS POSSIBLE?



BEFORE TREATMENT
Low testosterone leads to belly fat, poor energy and an unattractive body shape.

AFTER TREATMENT
Testosterone levels restored. Muscles tighten. Belly fat vanishes. Patient has more energy, more stamina and is more attractive to opposite sex.

WHO'S BEHIND THIS BREAKTHROUGH?



Geoff Jowett is passionate about helping Aussie men be their best. He is a Sports Scientist, champion bodybuilder and weight loss guru and knows the critical importance of healthy testosterone levels in men over 30. Jowett has assembled a team of doctors, hormone specialists, research scientists and nursing staff to make this breakthrough treatment available to aussie men tailored to the individual. Jowett believes male hormone therapy is critical for optimal health in men over 30.

**Feel Stronger, Last Longer
Than Men Half Your Age**

"Our love life was dead in the water. But thanks to AMHC my libido is much better and my wife runs up and down at night," says one patient.

"My energy and stamina are up, and I think it's even helping my overall health," explains another.

"I couldn't satisfy my wife. I felt like a dad. But thanks to AMHC, every night feels like our honeymoon again," says another.

**Be The Alpha Male
In The Room**

Research shows men with high testosterone are more dominant and attractive. In studies women subconsciously sense High Testosterone and are magnetically drawn to it. If you have ever seen women flock around a guy, it's

nothing to do with his personality, his body balance or looks. It's because his body is releasing alpha testosterone hormones.

**You Are Just One
Confidential Phone Call
Away From A Better Life**

Great results. Clinically supported data. Natural treatment

that works with your body instead of against it. It's easy to see why this treatment is the smart choice for men with low testosterone.

You can call if it will help you with a FREE MEDICAL ASSESSMENT, but we can only accept a limited number of people," says Jowett.

From 8am today, the FREE MEDICAL ASSESSMENT Hotline will be open. Simply call 1300 285 802. We will fill the spaces on a first-come, first-served basis. There is no obligation if you call and every call is confidential.

SAY GOODBYE TO LOW T TODAY!

- Increases muscle size
- Wakes up in the morning bursting with powerful energy
- Stay razor sharp and focused* all day long
- Feel happier, younger, more alive
- Get sexual partners
- Command more respect
- Burn off pudge fat
- Restore masculinity and strength

Be your best
amhc.com.au

AMHC
AUSTRALIAN MALE HORMONE CLINIC

Overdiagnosis is hidden in
the garden of diagnosis



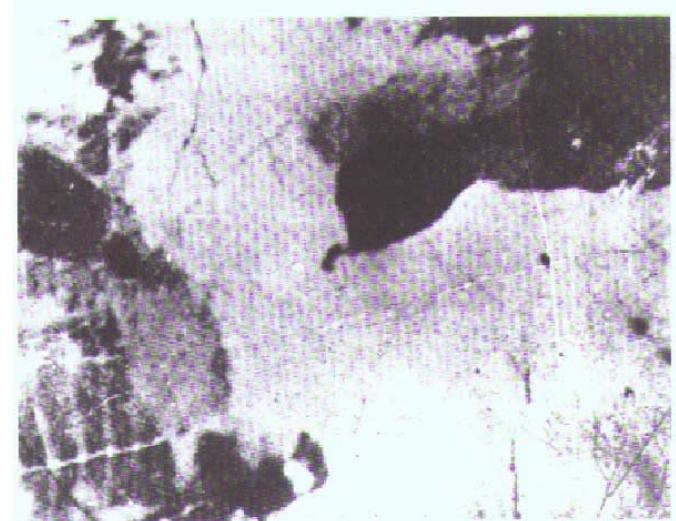
<http://natassatriviza.com/pepperdine/weekfive.html>

Diagnosis: impossible but essential task

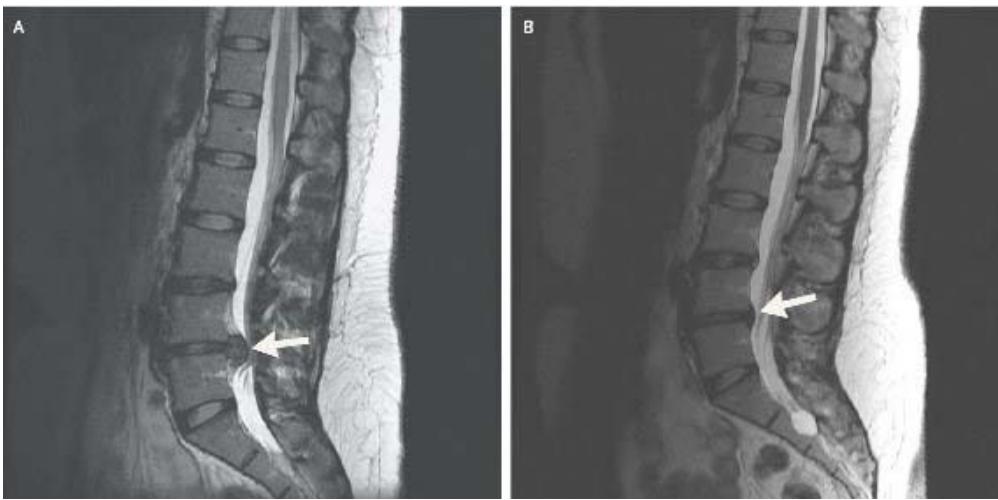
Diagnosis:

- Requires fast and slow thinking
- An essential skill; but not essential for every presentation
- Half consultations involve 30 conditions; but other half involves 1,000s
- Overdiagnosis is a growing problem

www.bmjjournals.org/bmj/2013/06/07/diagnosis-general-practice
www.preventingoverdiagnosis.net

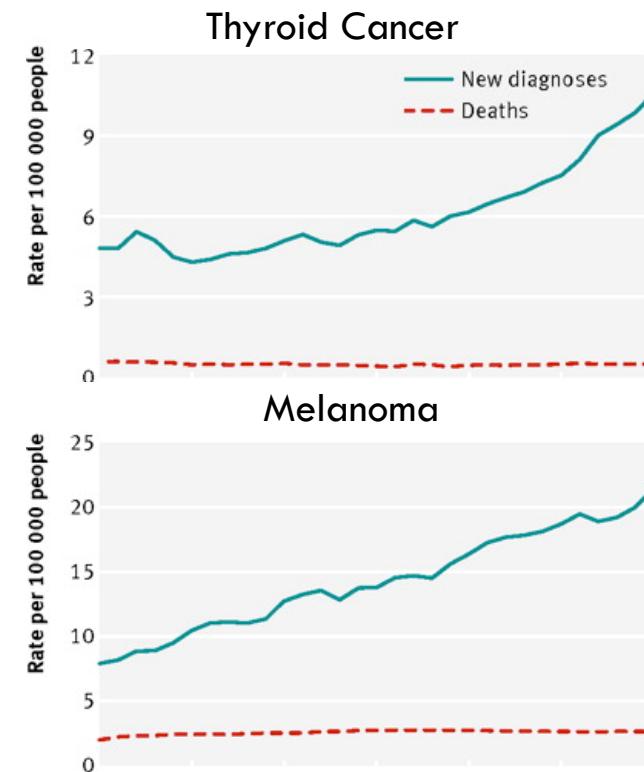
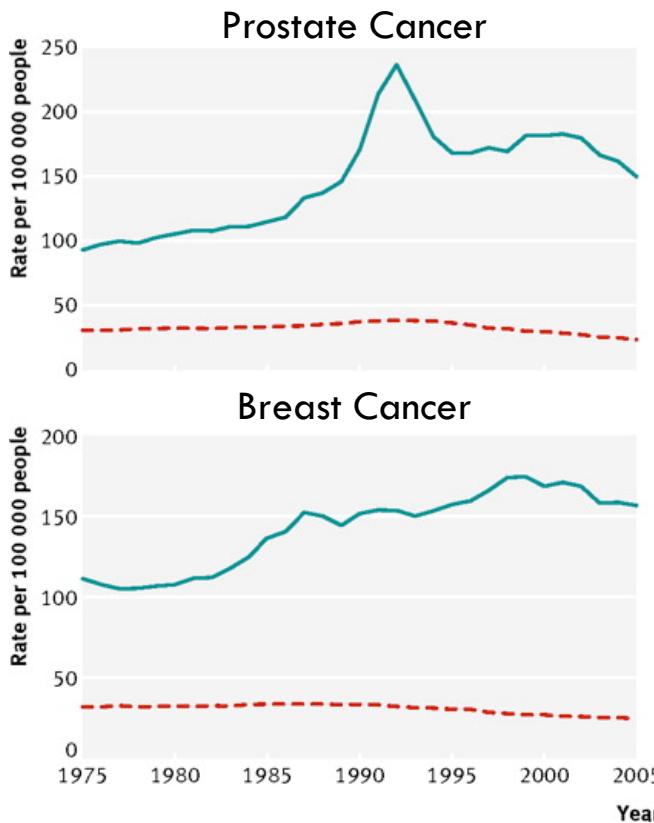


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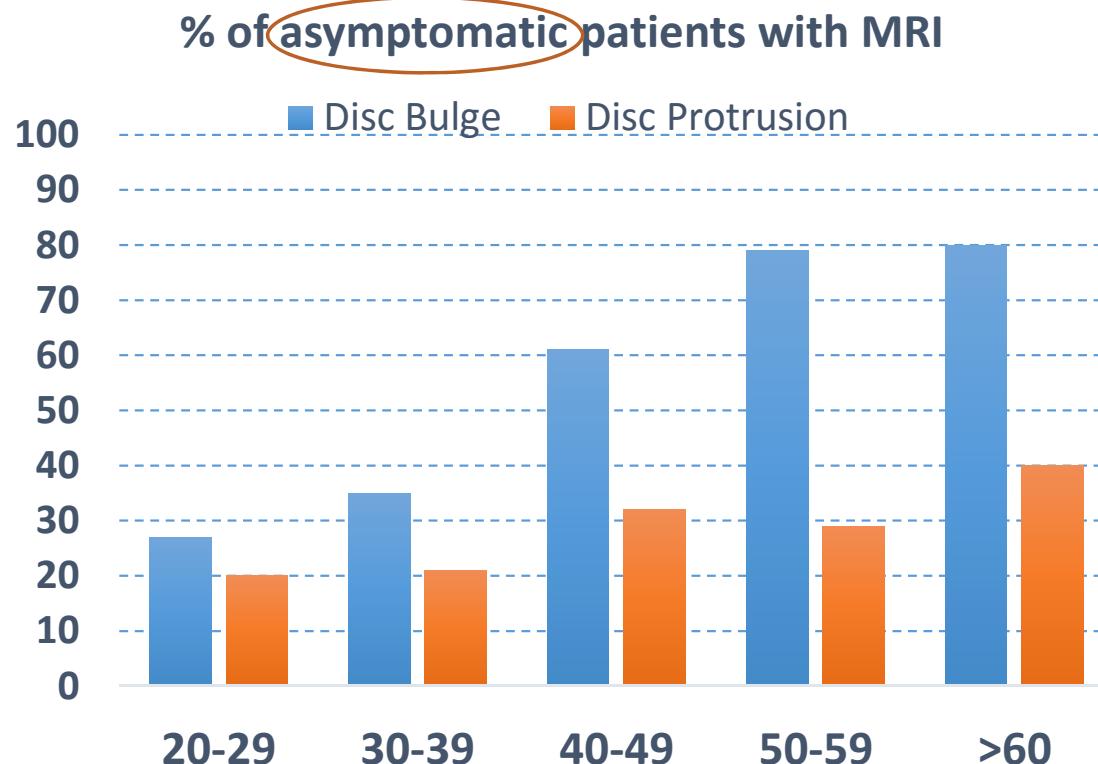
A. Overdetection: 4 cancers

An epidemic of diagnosis, not an epidemic of cancer!



Source: AIHW

Incidentalomas in MRI of spine



Disc protrusion in 24 year old with no back pain

Jensen MC, NEJM, 1994

Incidental findings with MRI of head

Frequency of incidental findings with MRI of head: a meta-analysis

	Prevalence (%)	NNS
Arachnoid cyst	0.5	200
Aneurysm	0.35	286
Meningioma	0.3	345
CCM	0.16	625
AVM	0.05	2,000
Low grade glioma	0.05	2,000

Victims of Modern Imaging Technology *BMJ 2009; 339: b3016*

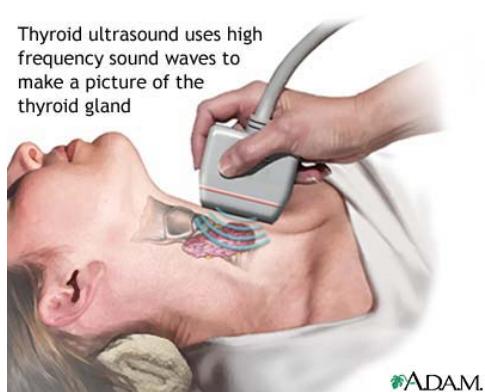
For most, treatment is not needed or is harmful.

SIGN Guidelines suggest: "DO NOT do MRI unless ..."

Too much testing?

“A well person is someone who has not been completely worked up.”
- Clifton Meador

Thyroid ultrasound uses high frequency sound waves to make a picture of the thyroid gland



PROVEN
DIAGNOSTICS

PROVEN DIAGNOSTICS
252 Brodhead Road
Bethlehem, PA 18017
610.865.1334
marita.herrmann, MD, FRCR, Medical Director

Lab Administration
252 Brodhead Road
BETHLEHEM, PA 18017

PATIENT, TEST	OFFICE REC'D	PHONE	DOB	AGE, YRS	SEX
Specimen ID	Spec. Collected	Spec. Received	01/01/1970	39	M
M54454	06/29/2009 08:00	06/29/2009 08:05			
			DOC, TEST		
Test Description	Result	Abnormal	Reference Range	Units	Lab
BASIC METAB PANEL					
BUN	16		6-20	mg/dL	P1
CREATININE	0.9		0.7-1.5	mg/dL	P1
SODIUM	145		135-146	mmol/L	P1
POTASSIUM	3.5		3.5-5.1	mmol/L	P1
CHLORIDE	110		98-111	mmol/L	P1
CO2	22		22-32	mmol/L	P1
GLUCOSE RESULTS RECHECKED	250	H	70-120	mg/dL	P1
ANION GAP	13		7-15	mEq/L	P1
CALCIUM	9.5		8.8-10.5	mg/dL	P1
GFR ESTIMATED	>60.0		>60	mL/min	P1

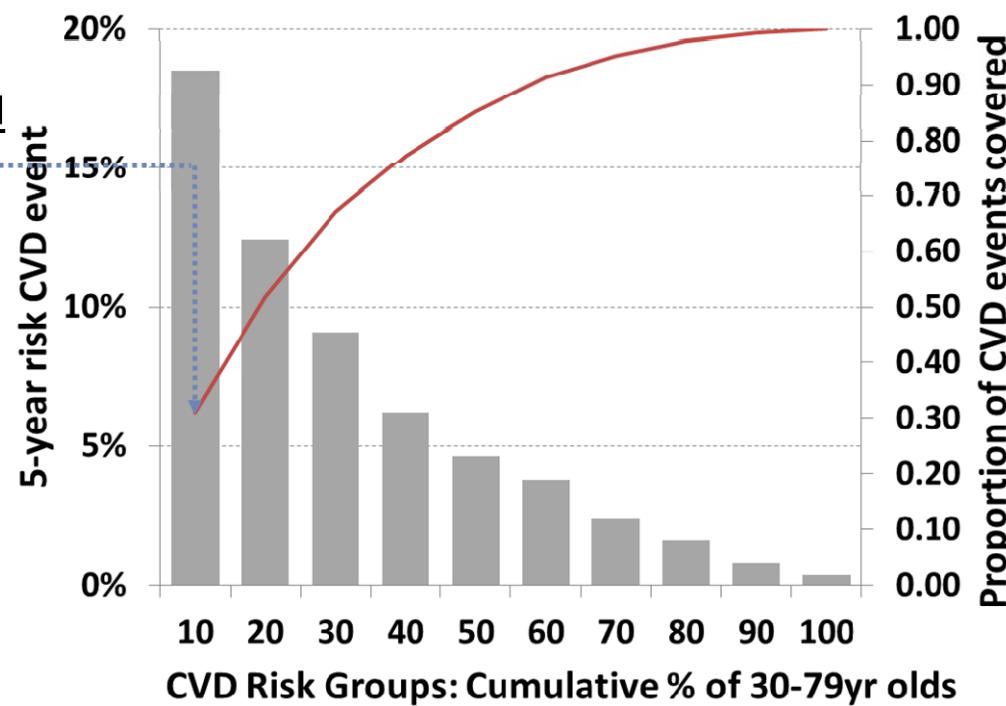
Performing Laboratory Information:
P1 - PERFORMED AT PROVEN DIAGNOSTICS LABORATORY 252 BRODHEAD RD BETHLEHEM, PA 18017

Declining thresholds; increasing prevalence

LOWERING OF THE CVD RISK THRESHOLD

5-year CVD risk threshold

1990's New Zealand 15%



Declining thresholds; increasing prevalence

LOWERING OF THE CVD RISK THRESHOLD

5-year CVD risk threshold

1990's New Zealand 15%

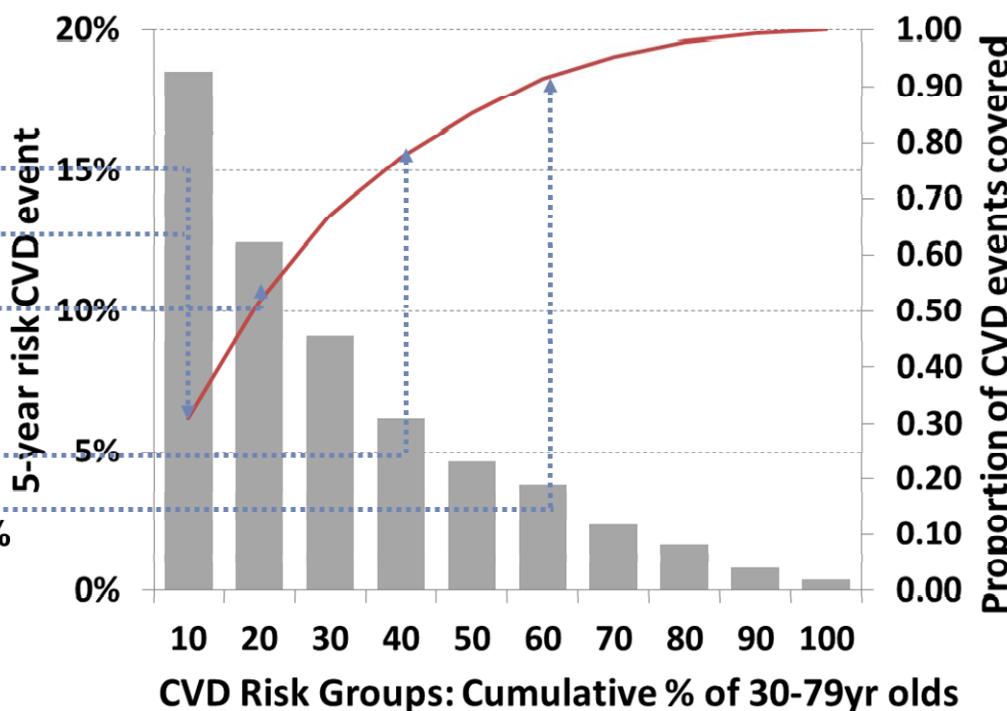
2001 - Australia: 10–15%

2006 - NICE (UK) 10%

2014 – NICE (UK) 5%

2014 – CC-AHA (USA) 3.75%

(* CC-AHA is 7.5% in 10-yrs)



Saving Normal

DSM 5 Is Guide Not Bible—Ignore Its Ten Worst Changes

APA approval of DSM-5 is a sad day for psychiatry.

Post published by Allen J Frances M.D. on Dec 02, 2012 in DSM5 in Distress



Like

11k

Diagnosis and screening

Sackett's "Normal"s

Table 5.1 Six definitions of normal

1. *Gaussian*: the mean ± 2 standard deviations (SD) – this one assumes a normal distribution for all tests and results in all "abnormalities" having the same frequency.
2. *Percentile*: within the range, say of 5–95% – has the same basic defect as the Gaussian definition. Implies a specificity of 95% but with unknown sensitivity.
3. *Culturally desirable*: when "normal" is that which is preferred by society, the role of medicine gets confused.
4. *Risk factor*: carrying no additional risk of disease; nicely labels the outliers, but does changing a risk factor necessarily change risk?
5. *Diagnostic*: range of results beyond which target disorders become highly probable; the focus of this discussion.
6. *Therapeutic*: range of results beyond which treatment does more good than harm; means we have to keep up with advances in therapy!

120

but also to defy common sense.



Allen Frances,
Chair DSM IV

saving

nor·mal (nôr'môl)
1. an insider's revolt against out-of-control psychiatric diagnosis, *DSM-5*, big pharma, and the medicalization of ordinary life

Allen Frances, M.D.¹
¹Chair of the DSM-IV Task Force

What can we do?

1. Raise awareness
2. Inform patients of pros & cons
3. Guidelines for disease definitions
4. Invest in research & policy initiatives
5. ...



1 Raise Awareness: #Overdiagnosis



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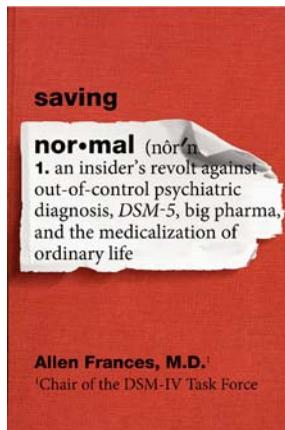
OVER-DIAGNOSED

MAKING PEOPLE SICK IN
THE PURSUIT OF HEALTH

DR. H. GILBERT WELCH,
DR. LISA M. SCHWARTZ, AND DR. STEVEN WOLOSHIN

"This brilliantly researched, well argued, and directly written book will help us recall the essential role, tragic, sanguine, and witty that are the heart and soul of our medical diagnosis and diagnosis."

—SIDNEY WOLFE, MD, author of *Worse Than Well*



Conference partners
THE DARTMOUTH INSTITUTE
BMJ BOND UNIVERSITY
Consumer Reports

2013 Dartmouth
2014 Oxford
2015 Washington

LESS IS MORE
- = + MEDICINE

HOME THE IDEA TOOLS BLOG [FRANÇAIS] ABOUT



An initiative of NPS MedicineWise

(Launch in April, 6 Colleges)

2 Inform patients of pros and cons

- Patients overestimate benefits and underestimate harms (of screening, treatments, etc)
- Informing patients often dampens enthusiasm
- Two processes
 - ▣ For individuals: shared decision making
 - ▣ For policy: community juries

3 Guidelines for disease definitions

OPEN  ACCESS Freely available online



Expanding Disease Definitions in Guidelines and Expert Panel Ties to Industry: A Cross-sectional Study of Common Conditions in the United States

Raymond N. Moynihan^{1*}, Georgia P. E. Cooke¹, Jenny A. Doust¹, Lisa Bero², Suzanne Hill³,
Paul P. Glasziou¹

1 Bond University, Robina, Australia, **2** University of California, San Francisco, San Francisco, California, United States of America, **3** Australian National University, Acton, Australia

- Guidelines being developed by GIN, GRADE, WHO collaboration
- 1-day Meeting: August 2014
- Position paper: 2015

4 Invest in research & policy initiatives

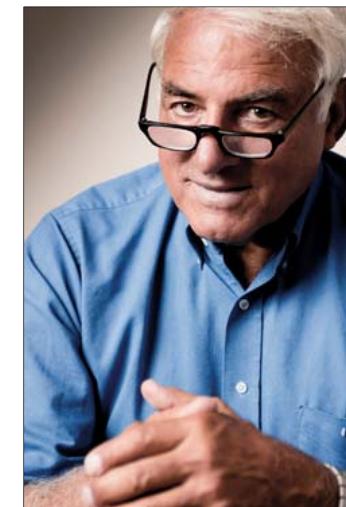
“Our combined resources are a few \$million, but for those promoting overdiagnosis it is \$billions. It is a David and Goliath struggle.”



The screenshot shows the homepage of the Preventing Overdiagnosis website. At the top left is the organization's logo with the text 'PREVENTING OVERDIAGNOSIS' and 'Winding back the harms of too much medicine'. To the right are 'Conference partners' logos for The Dartmouth Institute, BMJ, Bond University, and Consumer Reports. Below the logo is a navigation bar with links: Home, About, Readings, Conference, Blog, and Contact. A large red-bordered box in the center contains a photograph of a university building with a clock tower and people in the foreground. To the right of the photo is text: 'September 10-12, 2013 Conference, Abstract submission and Registration now open'. Below this is a detailed description of the conference.

September 10-12, 2013 Conference,
Abstract submission and Registration now open

The Preventing Overdiagnosis conference will take place on 10-12 September 2013 in the United States, hosted by The Dartmouth Institute for Health Policy and Clinical Practice, in partnership with one of the world's most respected medical journals, the BMJ, the leading New-York based consumer organisation Consumer Reports, and Bond University. The registration fee is \$475. A call for abstracts ... [Read More....](#)



Allen Frances
Chair DSM IV

What can we do?

1. Raise awareness
2. Inform patients of pros & cons
3. Guidelines for disease definitions
4. Invest in research & policy initiatives
5. ...

Paul_Glasziou@bond.edu.au
www.crebp.net.au

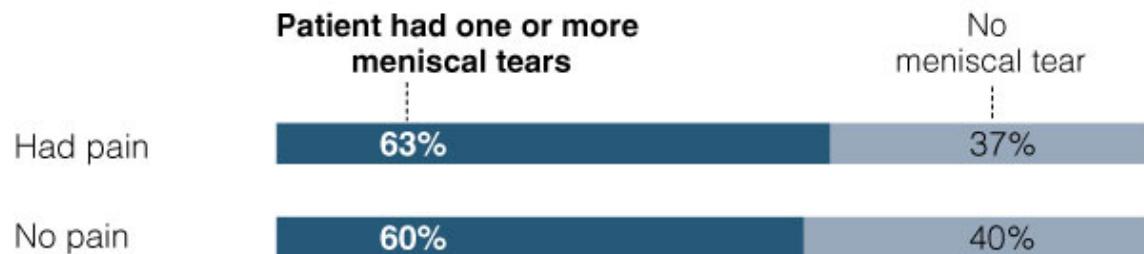


Meniscal tears: MRI vs pain

Before You Go for Surgery...

A study showed that frequent knee pain and stiffness may have nothing to do with the knee injury called a meniscal tear but may be related to arthritis.

PATIENTS WITH EVIDENCE OF OSTEOARTHRITIS

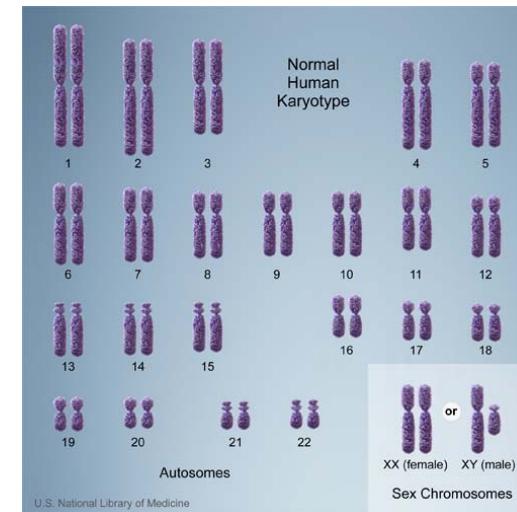


Source: Dr. David T. Felson, Boston University School of Medicine

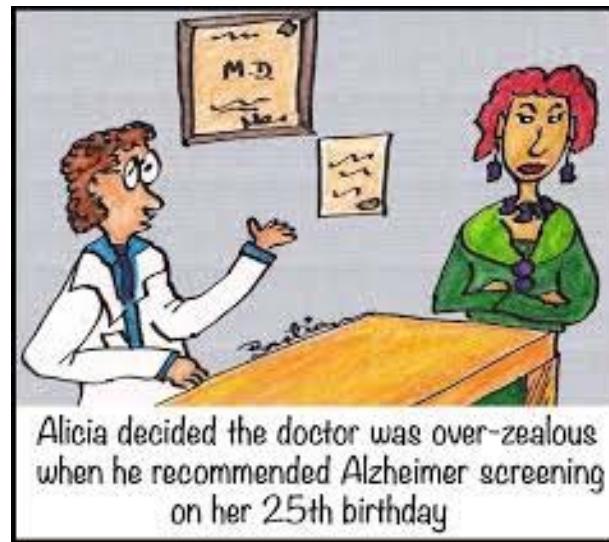
THE NEW YORK TIMES

Beware pseudo-diseases

- 1800's – diseases
 - ▣ Germ theory; endocrine; etc
- 1900's – risk factors
 - ▣ Hypertension; hypercholesterolemia; etc
- 2000's – risk factors for risk factors
 - ▣ Pre-hypertension; pre-diabetes; etc



100% fatality rate!





Knowing they did not need definitive research, an accurate test or effective treatment made developing new conditions for DSM-6 so much easier

Why does Overdiagnosis matter?

BMJ

BMJ 2013;347:f4247 doi: 10.1136/bmj.f4247 (Published 2 July 2013)

Page 1 of 2

Too much medicine; too little care

Time to wind back the harms of overdiagnosis and overtreatment

Paul Glasziou *professor*¹, Ray Moynihan *senior research fellow*¹, Tessa Richards *analysis editor*², Fiona Godlee *editor in chief*²

¹Bond University, Robina, QLD 4226, Australia; ²BMJ, London WC1H 9JR, UK

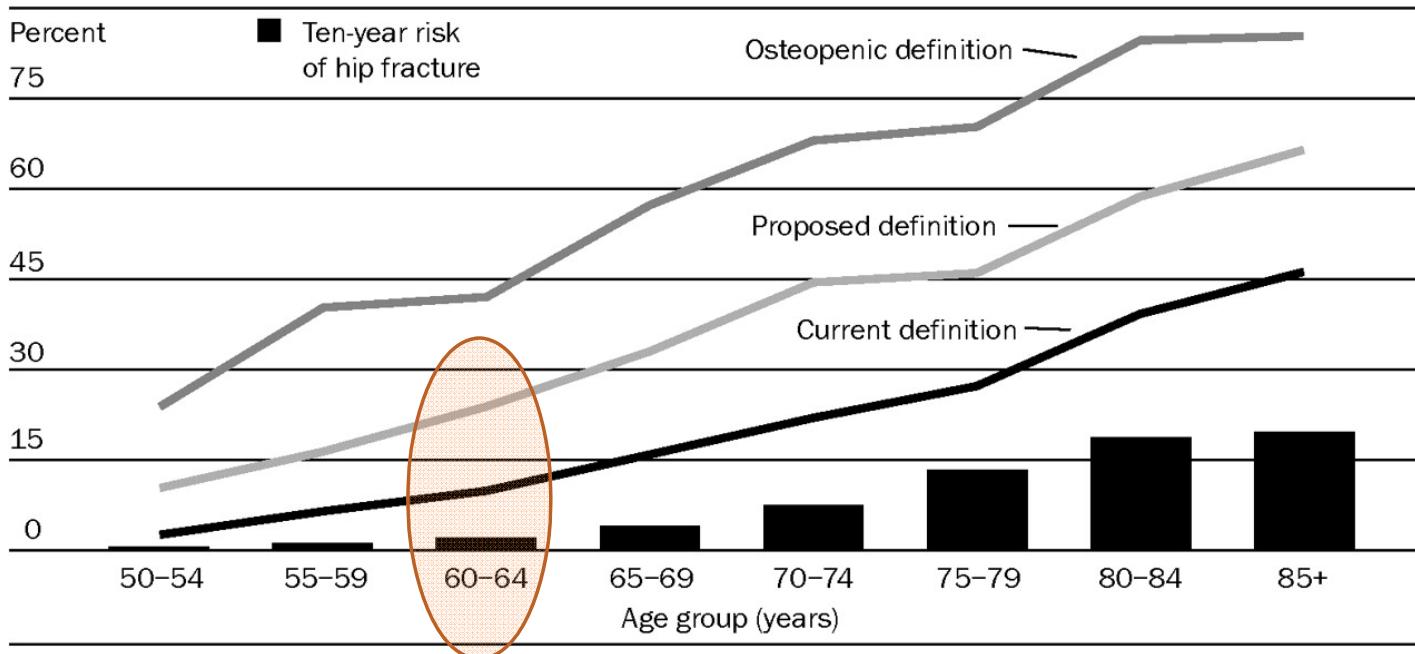
Too much testing of well people and not enough care for the sick worsens health inequalities and drains professionalism, harming both those who need treatment and those who don't.

Margaret McCartney, GP Glasgow,
Author of "The Patient Paradox".

Osteoporosis: Prevalence for changed definition

EXHIBIT 3

Comparison Of Prevalence Of Osteoporosis At Different Disease Definitions, With Ten-Year Risk Of Hip Fracture, For Women Age Fifty And Older, United States, 2000



SOURCE: Authors' analysis of data from the National Health and Nutrition Examination Survey (NHANES III), 1988-94; the 2000 National Inpatient Sample (NIS); and the 2000 U.S. census.

NOTE: For explanation of disease definitions, see text.

M. Brooke Herndon et al. Health Aff 2007;26:1702-1711

HealthAffairs

The many processes of diagnosis

1. Diagnosis uses several processes
 1. Pattern matching
 - Hypothesis & deduction
 - Stepwise refinement
2. “EBM” for diagnosis
 - Using literature for test accuracy
 - sensitivity; specificity
3. ... but there are some short cuts
 - Clinical Prediction Rules

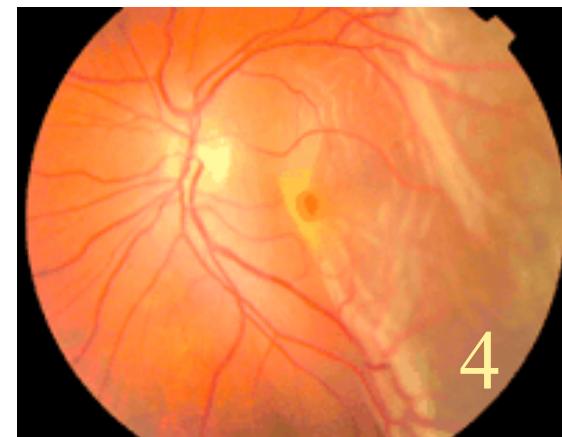


Topic 2: EBM in diagnostics

- Many diagnosis don't have clear “pattern”, e.g, fatigue, dyspepsia, shortness of breath, ...
- Usually no individual feature is 100% sensitivity or specific

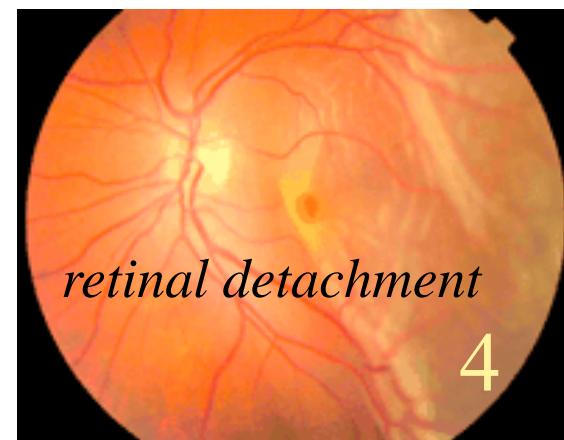
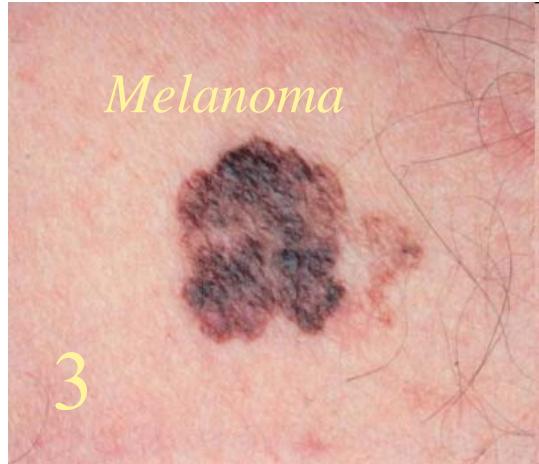
Topic 1: Diagnosis by pattern matching

What is the diagnosis?

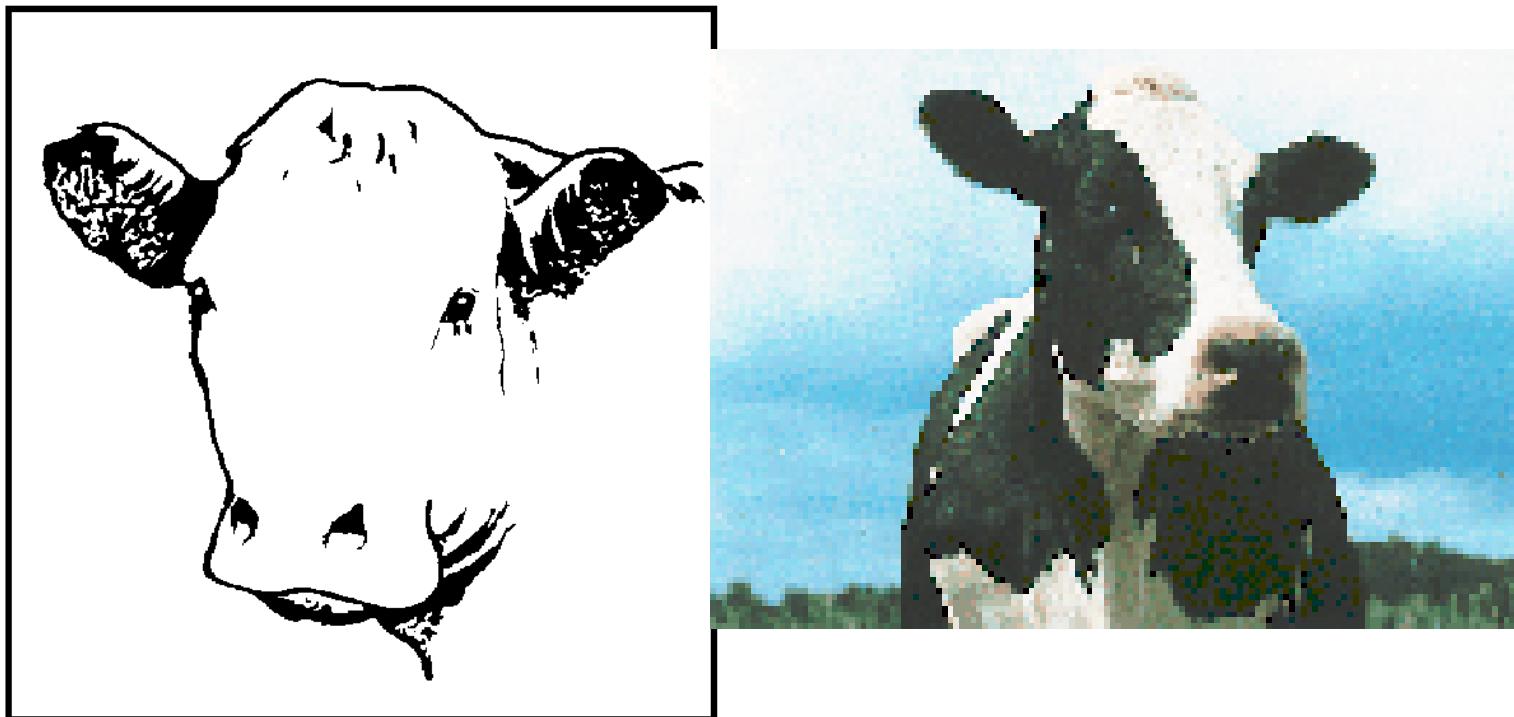


Topic 1: Diagnosis by pattern matching

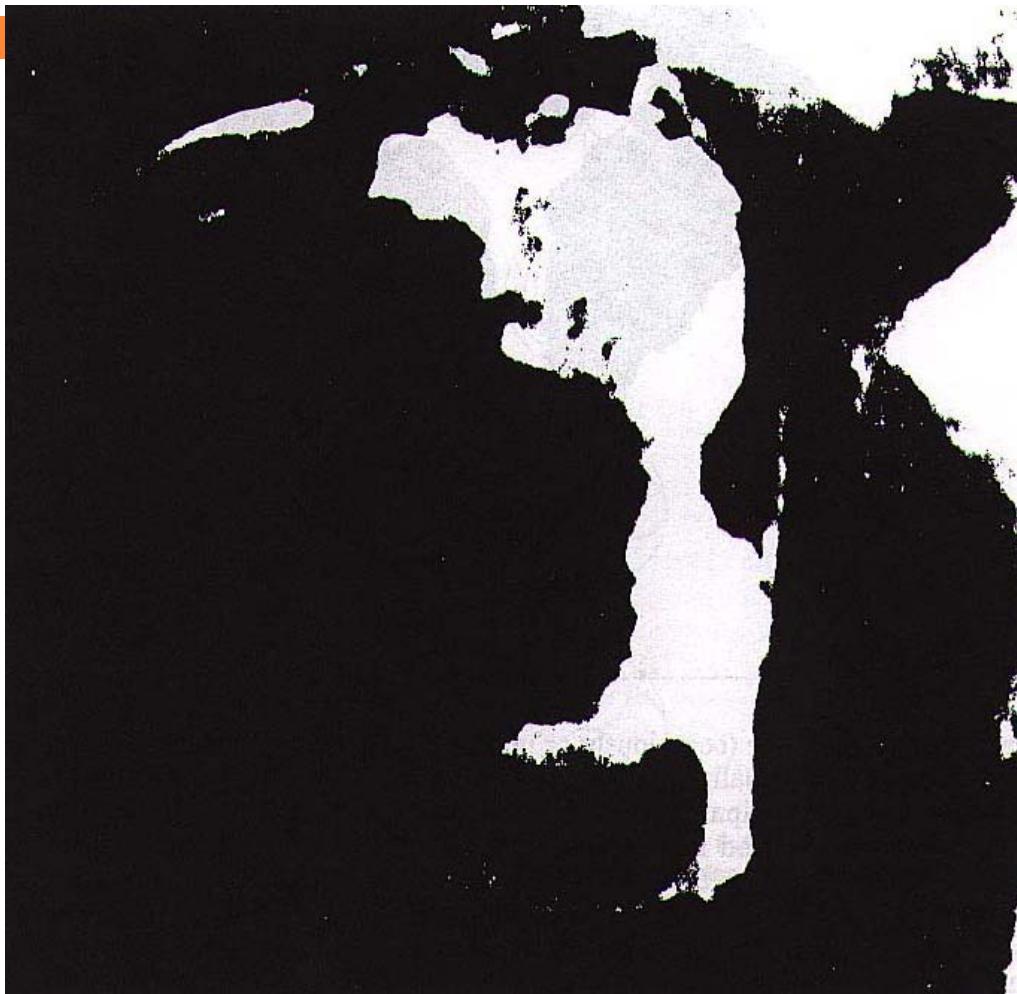
What is the diagnosis?



We learn basic patterns



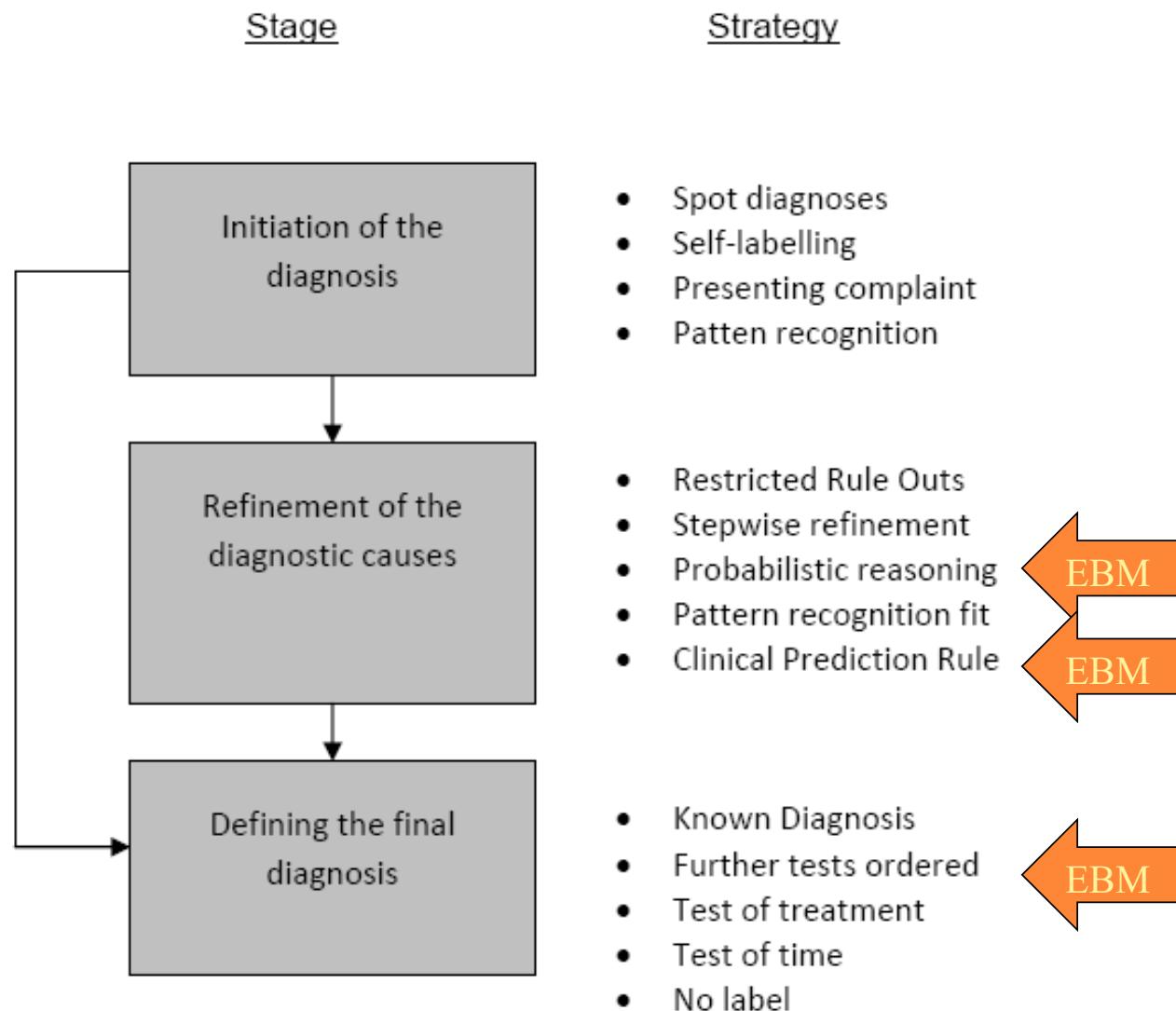
Then see them in new situations.



Diagnosis and level of experience with a presentation

1. No experience: use first principles / processes
Vascular, **I**nfection/**I**nflammatory, **N**eoplastic, **D**egenerative,
Intoxication/**I**atrogenic, **C**ongenital, **A**llergic, **T**raumatic, ...
2. Some experience: known differential diagnosis
What is common/important?
How to rule these in/out?
3. Great Experience: Pattern Matching
(shortcut? Clinical prediction rule)

Diagnosis: Partly (learned) intuition, partly analysis



3. We can't know all diseases

Could this be PFAPA?

- Recurrent sore throat,
sore mouth, fever
(every 4-5 weeks)



Patient -> Paediatrician -> ENT - GP

WIKIPEDIA The Free Encyclopedia

pfapa wiki

Article Discussion Read Edit View history Search

Periodic fever, aphthous stomatitis, pharyngitis and adenitis

From Wikipedia, the free encyclopedia

Periodic fever, aphthous stomatitis, pharyngitis and adenitis or periodic fever aphthous pharyngitis and cervical adenopathy (PFAPA) syndrome is a medical condition, typically starting in young children, in which high fever occurs periodically at intervals of about 3–5 weeks, frequently accompanied by aphthous ulcers, pharyngitis and/or adenitis. The syndrome was described in 1987 and named two years later.^{[1][2][3]}

Contents [hide]

- 1 Signs and symptoms
- 2 Cause
- 3 Treatment
- 4 Prognosis
- 5 References

Signs and symptoms



Classification and external resources

DiseasesDB 33682

30 most common consults = 48%

Table 2. The 30 most frequently managed problems

Rank	Problem type	Percent of problems (n=305 738)	Rate per 100 encounters (n=194 100)	Rank	Problem type	Percent of problems (n=305 738)	Rate per 100 encounters (n=194 100)
1	Hypertension	5.7	9.0	16	Test results	1.2	1.8
2	Immunisation/vaccination: all	4.2	6.7	17	Urinary tract infection	1.1	1.8
3	Acute upper respiratory tract infection	3.3	5.1	18	Dermatitis, contact/allergic	1.0	1.6
4	Depression	2.9	4.6	19	Pregnancy	1.0	1.5
5	Diabetes: non-gestational	2.3	3.7	20	Sleep disturbance	1.0	1.5
6	Lipid disorders	2.1	3.4	21	Sinusitis acute/chronic	0.9	1.4
7	General check-up	1.9	3.0	22	Gastroenteritis	0.8	1.3
8	Osteoarthritis	1.7	2.7	23	Vitamin/nutritional deficiency	0.8	1.3
9	Back complaint	1.7	2.6	24	Malignant neoplasm of skin	0.8	1.3
10	Prescription	1.6	2.5	25	Abnormal test results	0.8	1.2
11	Oesophagus disease	1.6	2.4	26	Atrial fibrillation/flutter	0.8	1.2
12	Female genital check-up	1.5	2.4	27	Oral contraception	0.8	1.2
13	Acute bronchitis/bronchiolitis	1.5	2.3	28	Solar keratosis/sunburn	0.8	1.2
14	Asthma	1.3	2.1	29	Ischaemic heart disease	0.7	1.1
15	Anxiety	1.2	2.0	30	Viral disease, not otherwise specified	0.7	1.1
	Cumulative total top 15	34.6%	—		Cumulative total top 30	47.8%	—

Cooke G, et al. Common general practice presentations, AFP, 2013