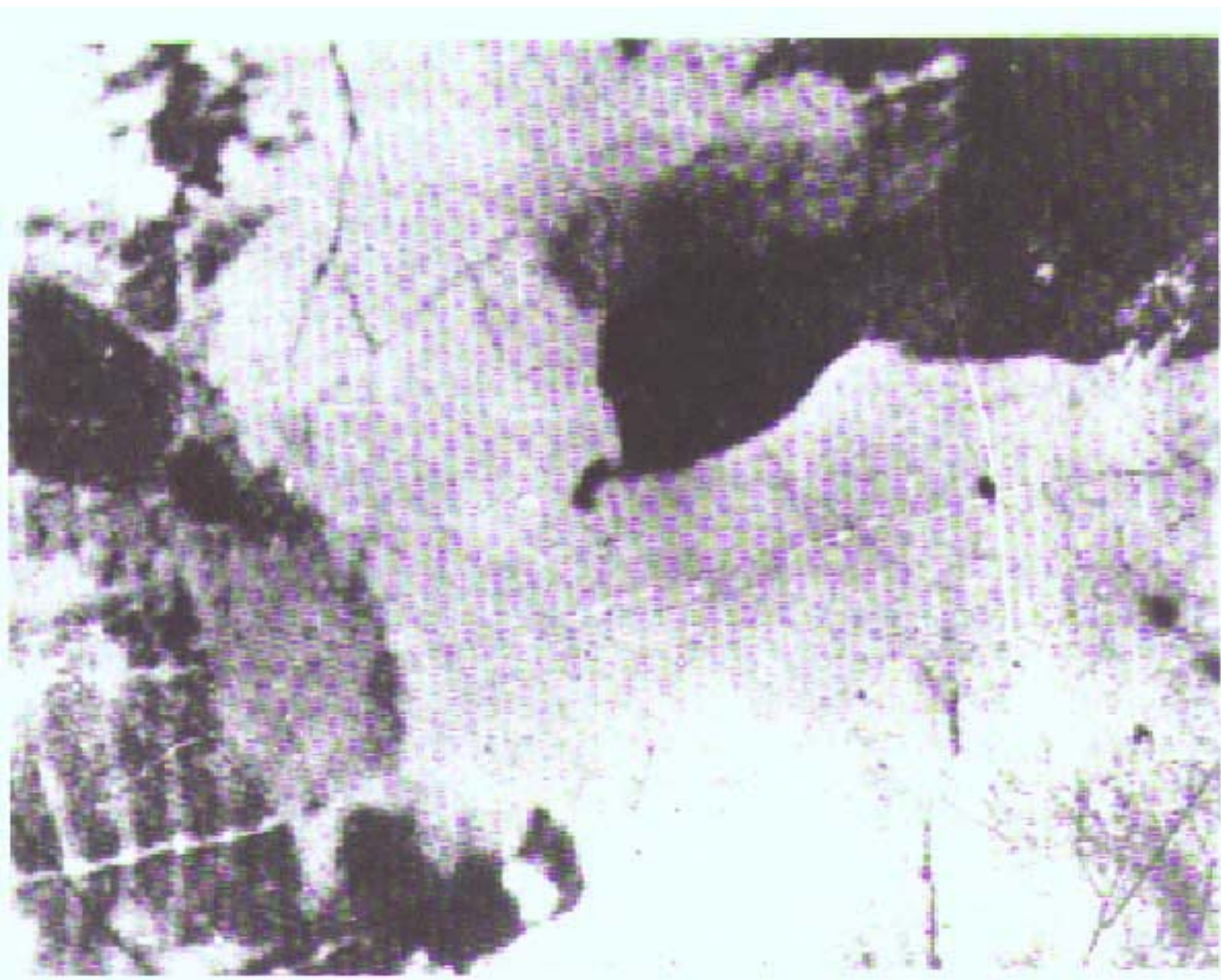


# Diagnosis: an impossible but essential task?

Professor Paul Glasziou  
Bond University, Australia  
[Paul\\_Glasziou@bond.edu.au](mailto:Paul_Glasziou@bond.edu.au)

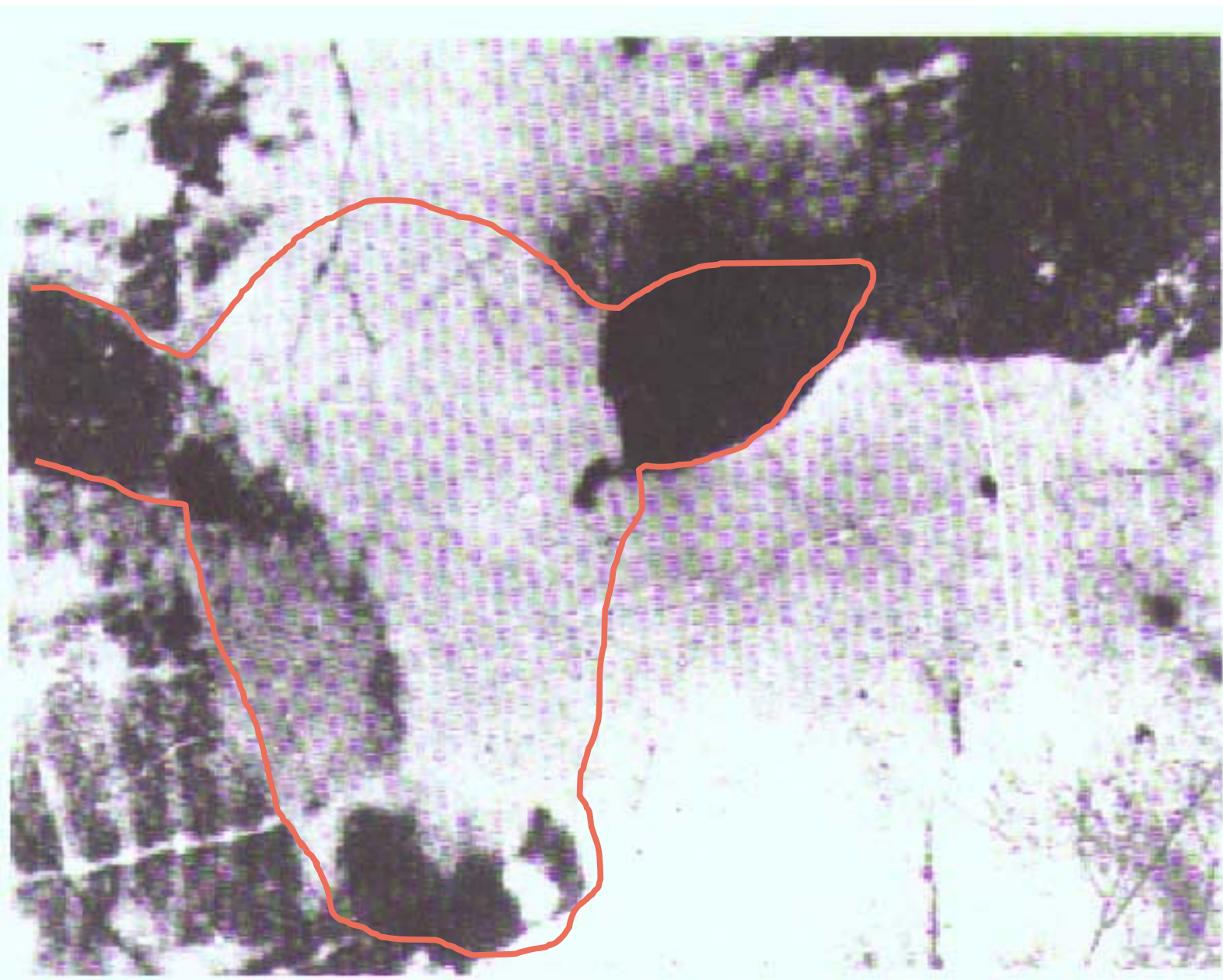


WONCA Copenhagen 2016



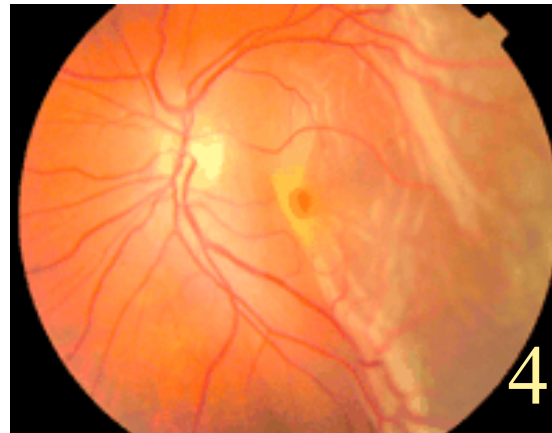
What is this?





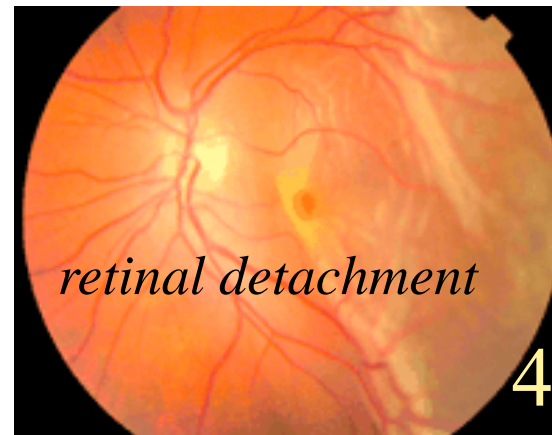
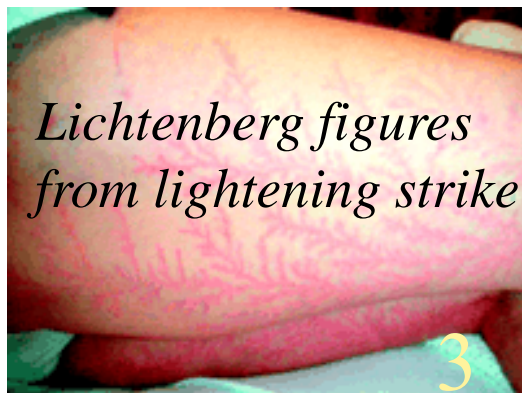
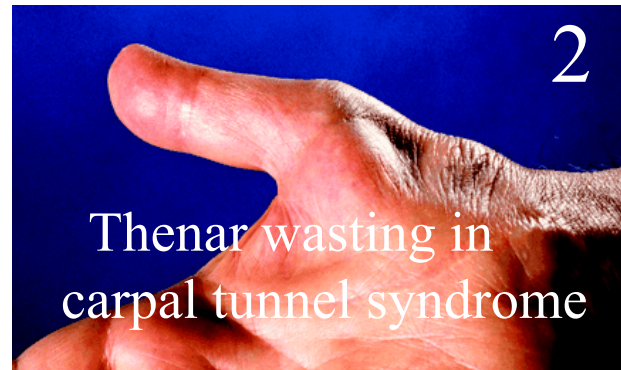
What is this?

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



# Diagnosis by visual pattern matching

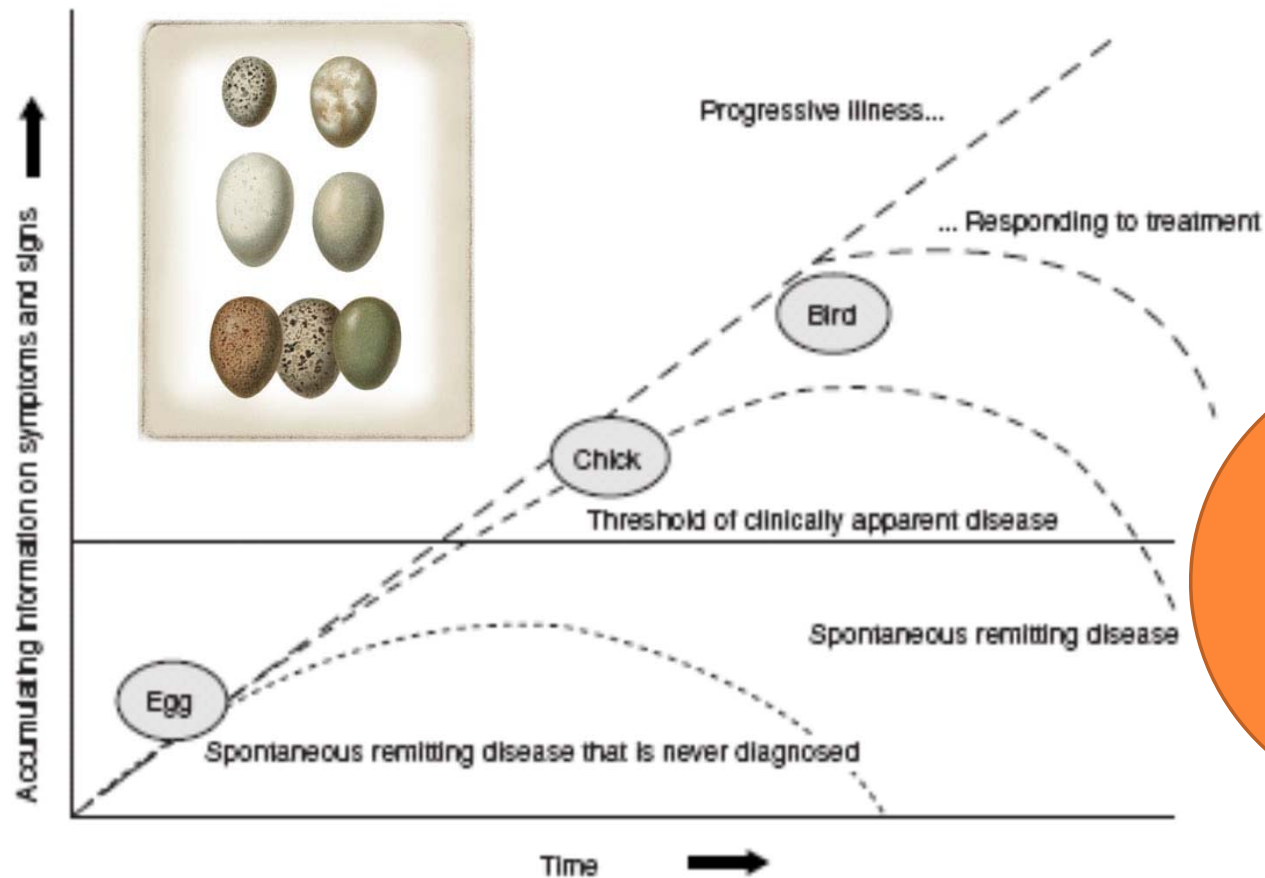
quick – but requires experience of pattern





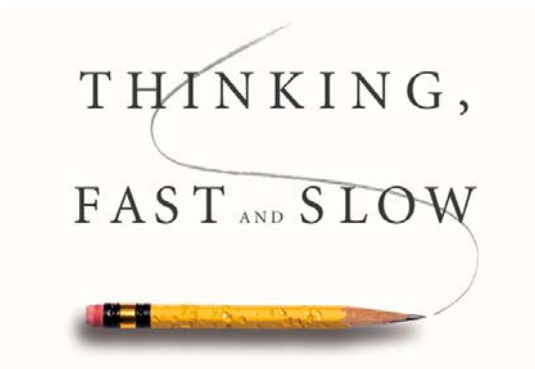
# Patterns change with time

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



Diagnosis not  
essential for  
spontaneously  
remitting  
diseases

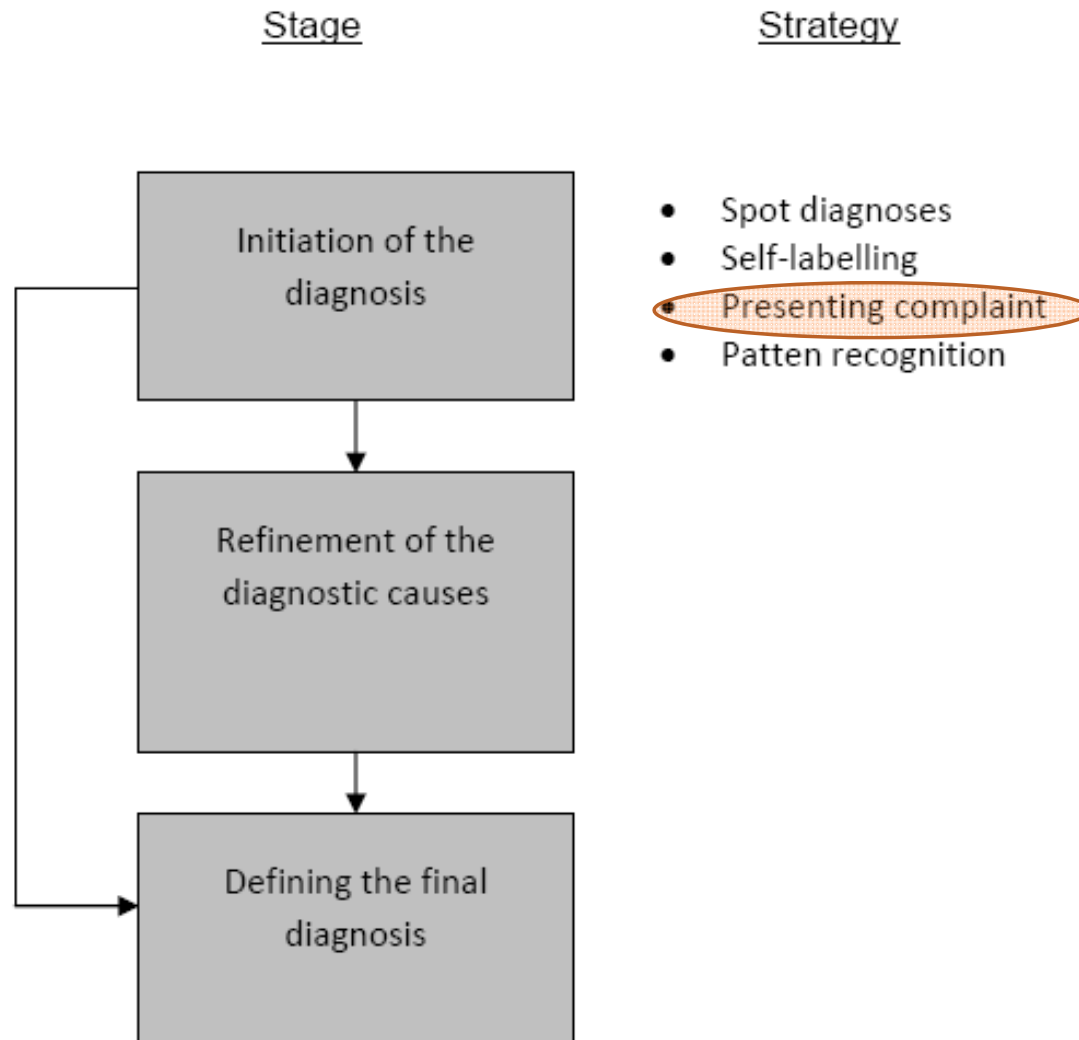
# Types of Diagnostic Thinking processes



- 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**

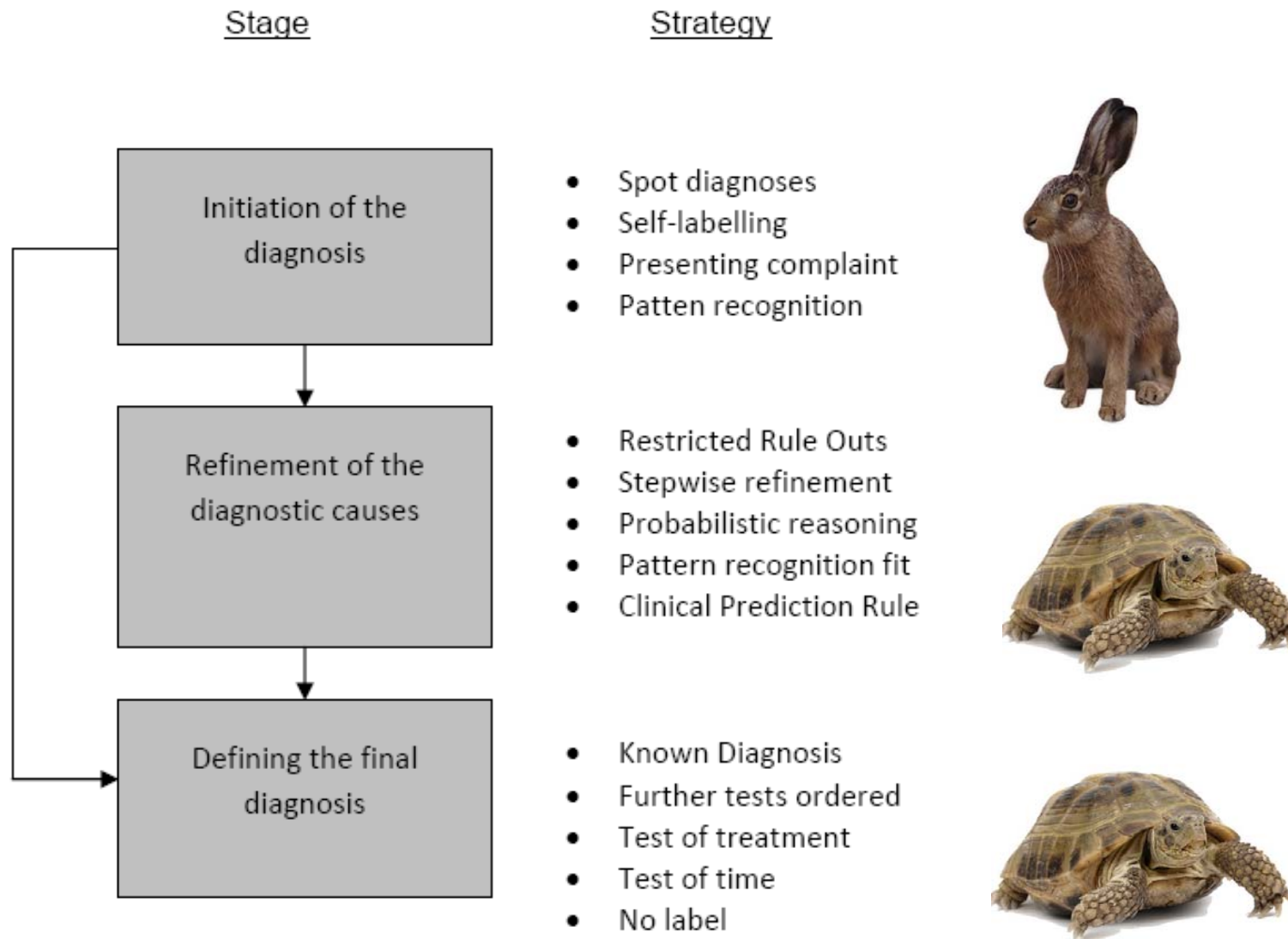


Patient 2: Few specific features,

- 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  
 $\kappa = 0.75$  between observers A and B and  
 $\kappa = 0.54$  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  $\leq 90$  g/L

Pallor	Hemoglobin $\leq 90$ g/L (n)	Hemoglobin $\geq 91$ 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

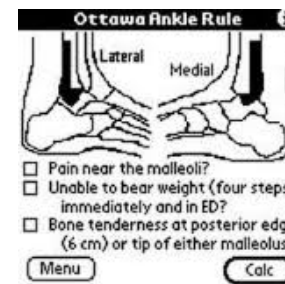
- 0–10 Scale

**Apgar Scoring System**

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

- Ottawa Rules

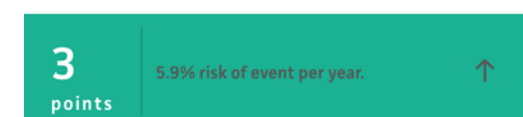
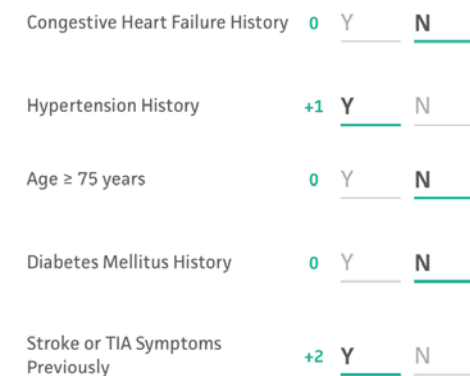
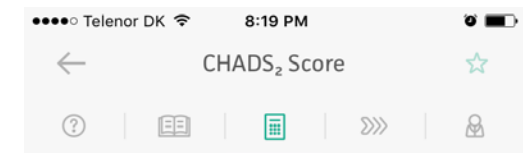
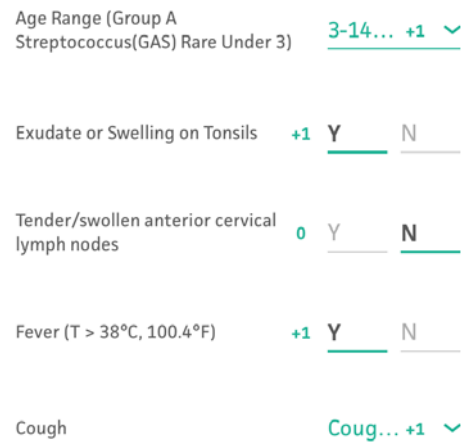
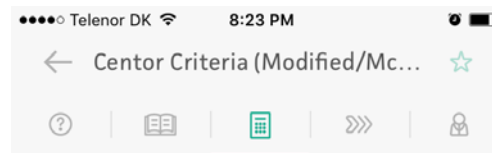
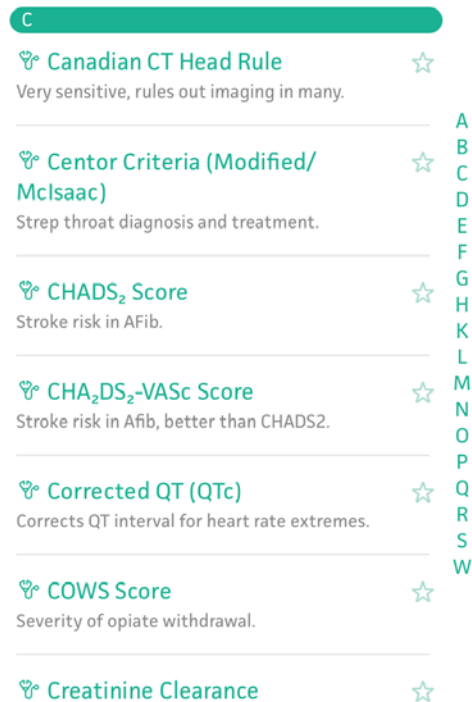
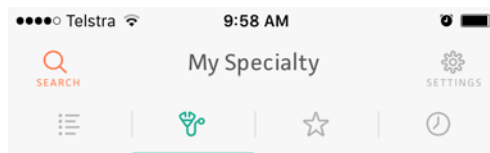
- Ankle
- Knee
- Cervical spine



- Well's DVT & PE rules

# Clinical Prediction Rules as 'instant' experience

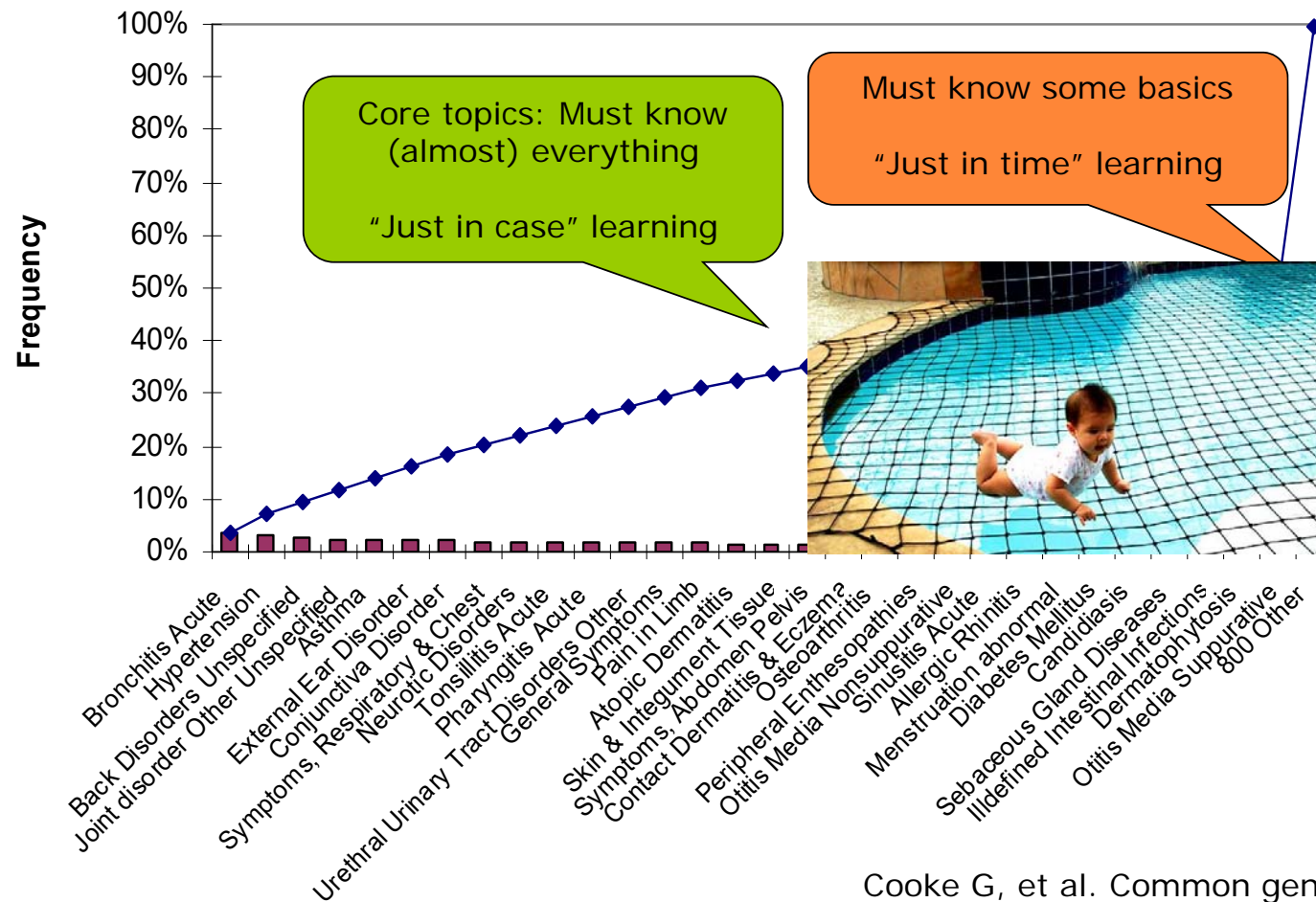
MDCalc – free App, 170 prediction rules; 53 for GPs





# Half GP consultations are for 30 conditions

## Other half are for over 800 conditions



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

# 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

**13,000 Aussie males don't know they have Klinefelter's Syndrome and remain untreated**  
**Are you one of them?**

The physical signs of Klinefelter's Syndrome are:

- Under-functioning testicles that are hard and abnormally small - peanut size.
- Infertility (recent technological advances can assist KS males to father a biological child)
- reduced life-span if untreated.
- Increased risk of both diabetes and heart disease.
- Rudimentary breast development, fatty hips and thighs, poor muscle strength and mass.
- Psychological issues including depression.
- KS boys often have learning, speech, physical and attitude problems.

When diagnosed, treatment provides KS males with vastly improved mental and physical health and lifestyle opportunities.  
**Please consult your doctor.**

**check your balls .com.au**

**Self examination is the first step to your wellbeing.**

**LAWLEY** [www.lawleypharm.com.au](http://www.lawleypharm.com.au)  
A Lawley men's health initiative.



# Syndrome Without A Name



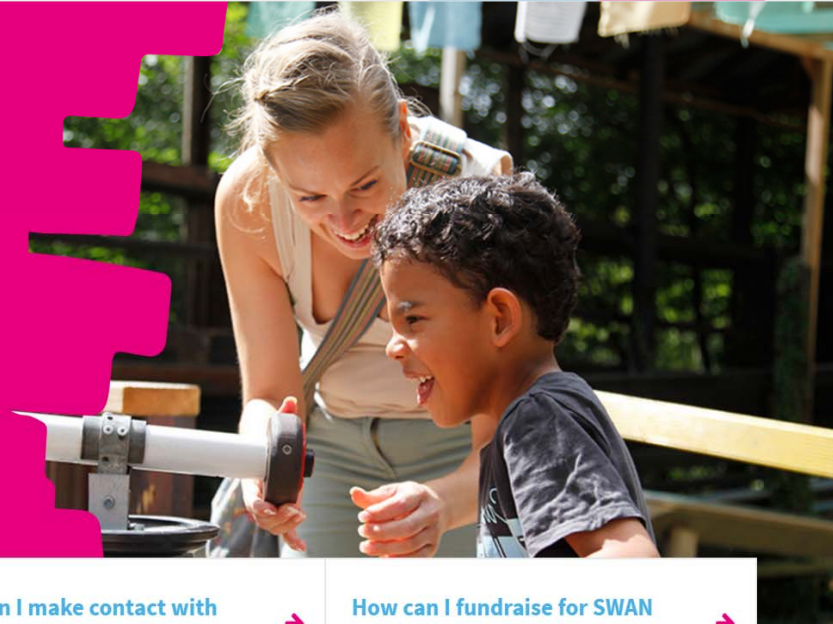
[Support & Information](#) [Get Involved](#) [News & Events](#) [About us](#) [Contact Us](#)

[Donate](#)

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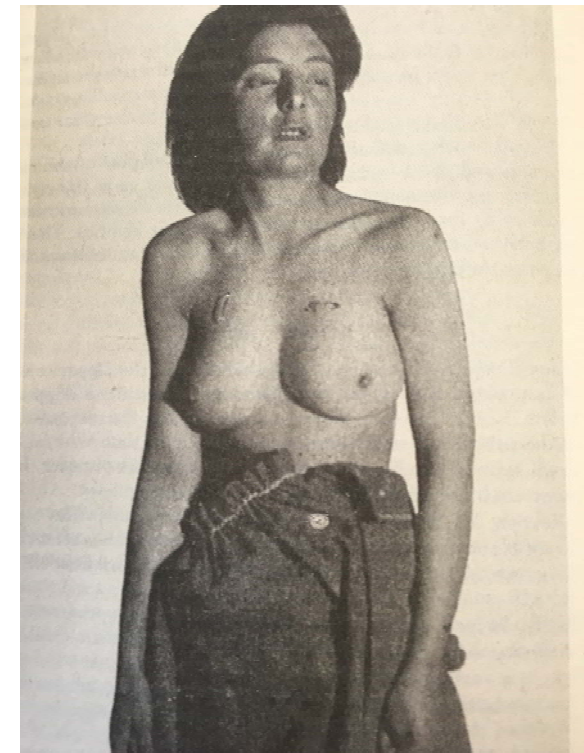
**We support families  
and those affected  
by a syndrome  
without a name**



[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
- 1/2 consults involve 30 conditions



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## Diagnosis in general practice

### Latest from The BMJ

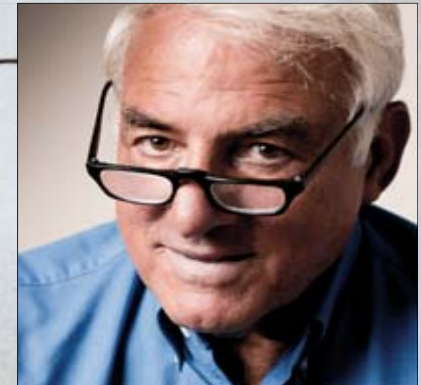
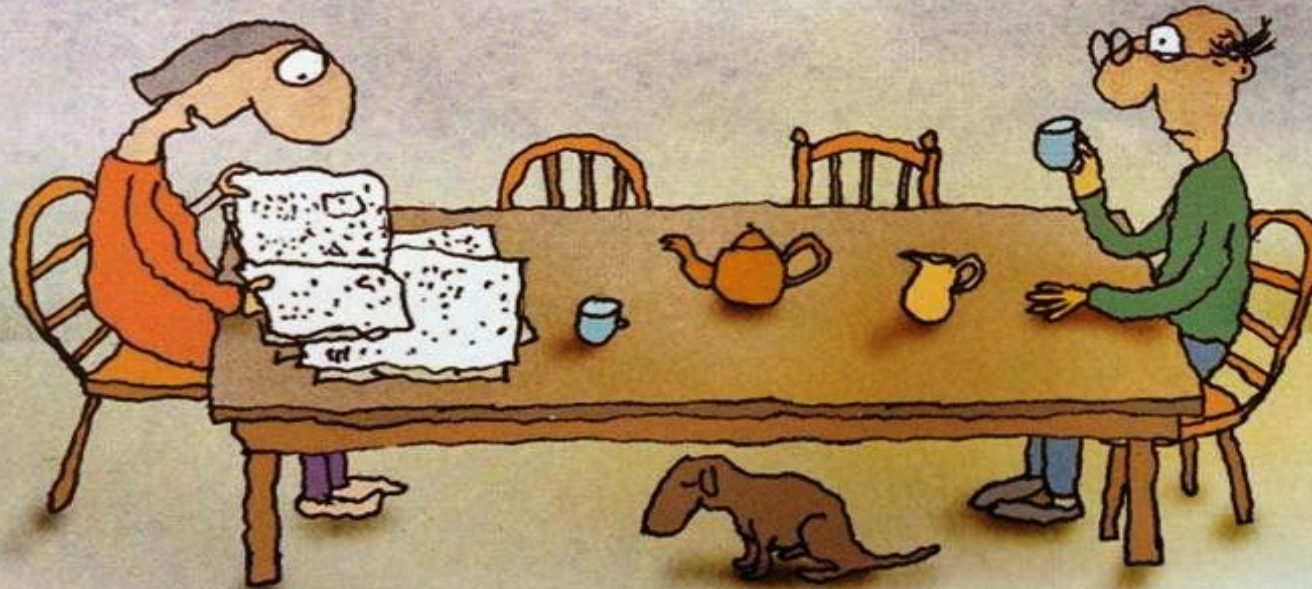
Practice <a href="#">Clinical prediction rules</a> Published 07 August 2009	Practice <a href="#">Acute diarrhoea in adults</a> Published 15 June 2009
Practice <a href="#">Test of time</a> Published 15 June 2009	Practice <a href="#">Diagnosis using “test of treatment”</a> Published 24 April 2009
Practice <a href="#">Chronic cough in adults</a> Published 24 April 2009	Practice <a href="#">Diagnostic strategies used in primary care</a> Published 20 April 2009

« first < previous 1 2 3 next > last »

[www.bmj.com/specialties/diagnosis-general-practice](http://www.bmj.com/specialties/diagnosis-general-practice)

# Overdiagnosis

oh darling... what a pity...  
I think your interesting personality  
has just been classified as  
a personality disorder.



Allen Frances  
Chair DSM IV

**saving**

**nor•mal** (nôr/n

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.**

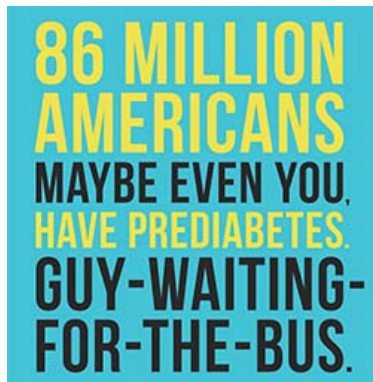
'Chair of the DSM-IV Task Force



# Three types of Overdiagnosis



Early detection key to treating thyroid cancer, experts say



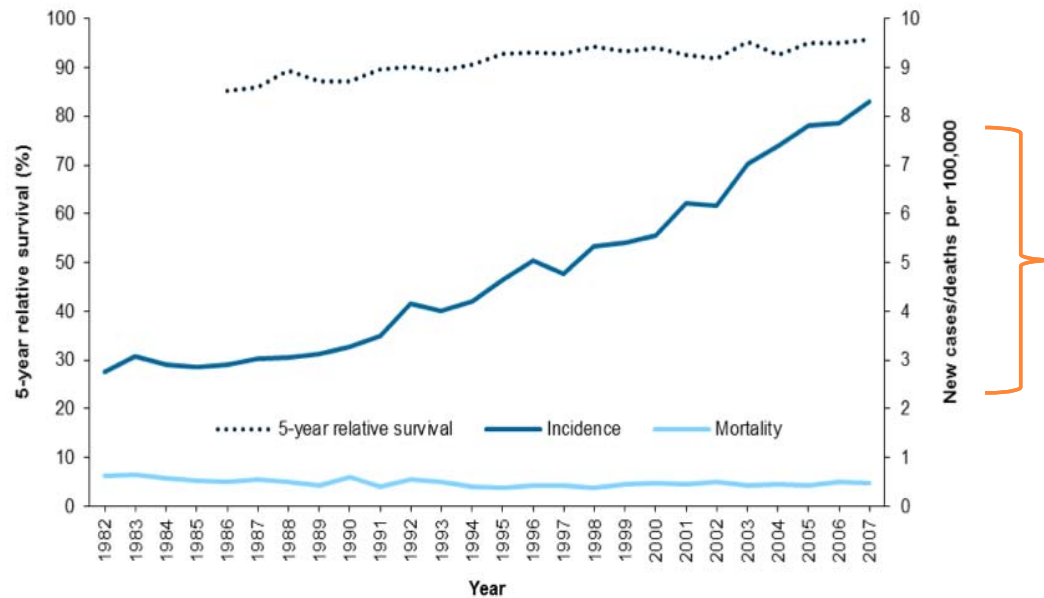
1. Overdetection

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.

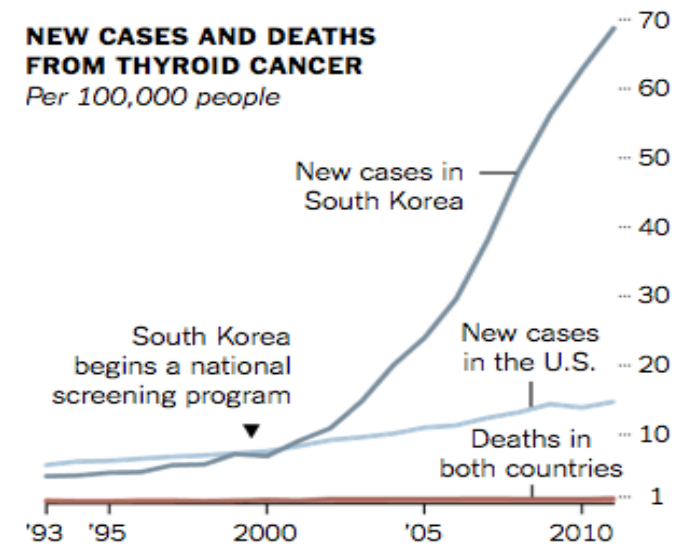
Figure 4.73: Yearly trends in incidence, mortality and 5-year relative survival of thyroid cancer, 1982 to 2007

## 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



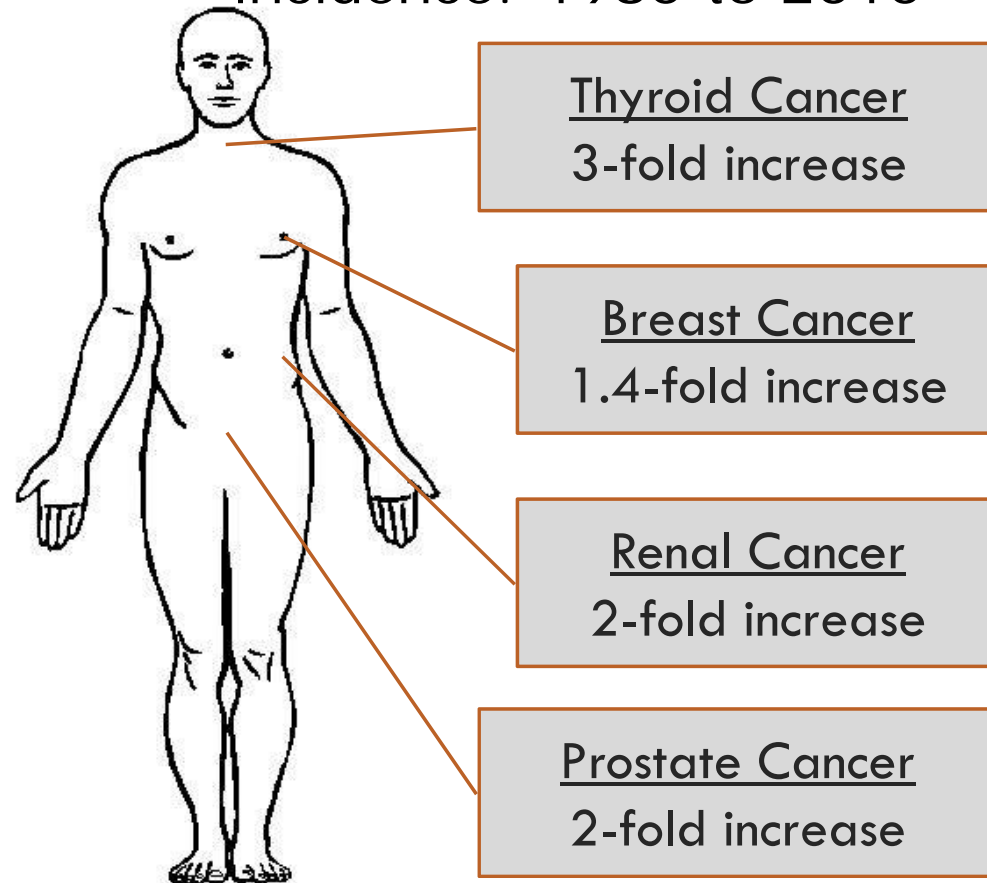
Sources: New England Journal of Medicine; National Cancer Institute

By The New York Times

# Overdetection

## CANCER

Increase in adult cancer incidence: 1980 to 2010



# Overdetection

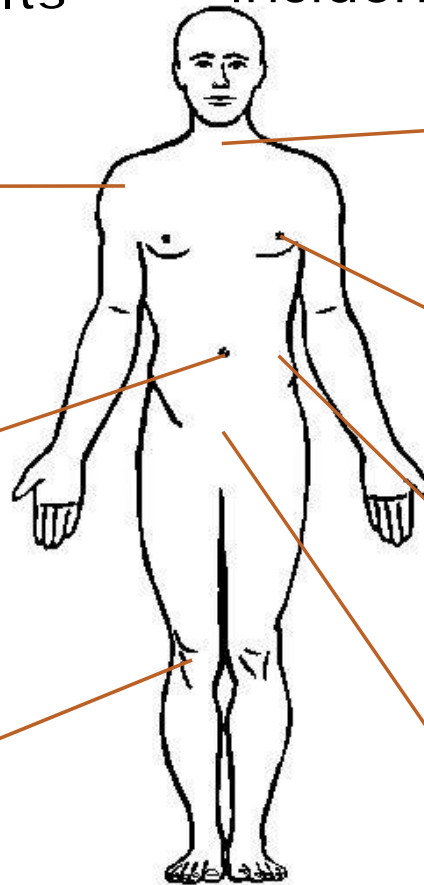
## MUSCULOSKELETAL

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

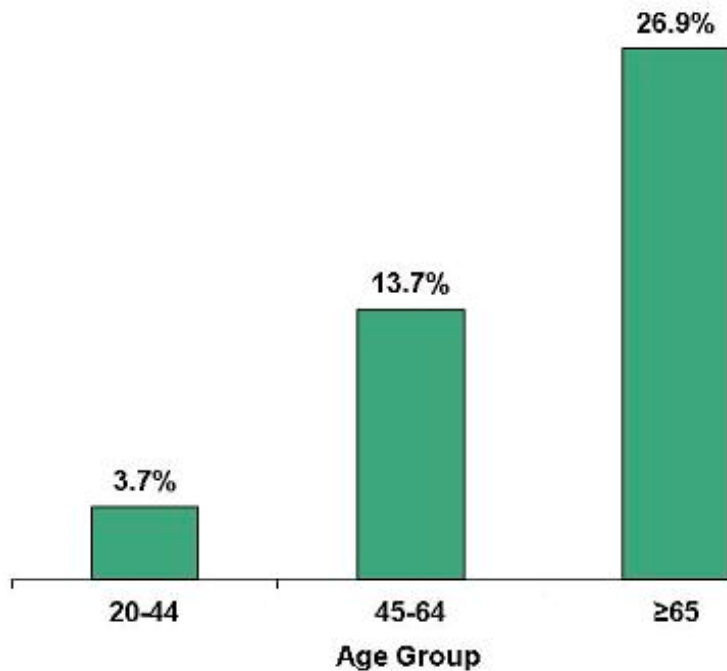
Renal Cancer  
2-fold increase

Prostate Cancer  
2-fold increase



## B. Expanding definitions of Diabetes

US Prevalence by age

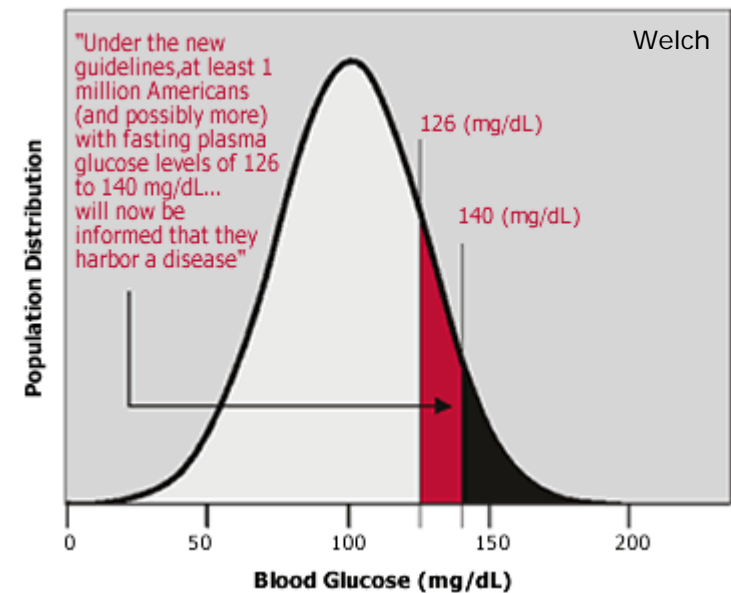


**86 MILLION  
AMERICANS  
MAYBE EVEN YOU,  
HAVE PREDIABETES.  
GUY-WAITING-  
FOR-THE-BUS.**

[DoIHavePrediabetes.org](http://DoIHavePrediabetes.org)



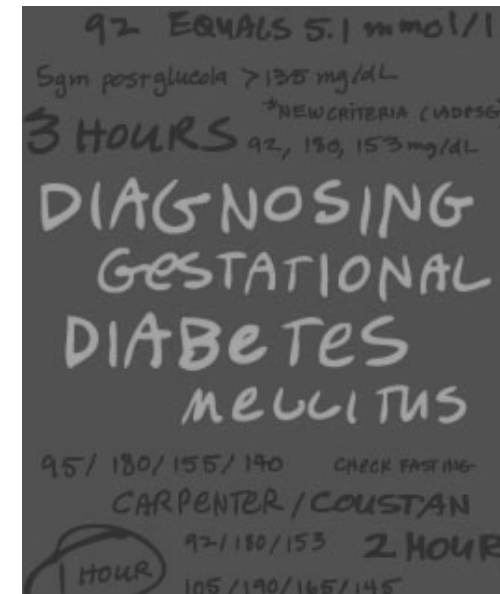
Change of Definition



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<sup>1\*</sup>, Georga P. E. Cooke<sup>1</sup>, Jenny A. Doust<sup>1</sup>, Lisa Bero<sup>2</sup>, Suzanne Hill<sup>3</sup>, Paul P. Glasziou<sup>1</sup>

<sup>1</sup> Bond University, Robina, Australia, <sup>2</sup> University of California, San Francisco, San Francisco, California, United States of America, <sup>3</sup> 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](http://mghocd.org/bddbp)

\*Content is being used for illustrative purposes only and any person depicted in the content is a model.

The advertisement features a black and white close-up of a man's face on the right side, looking directly at the camera with a serious expression. The text is on the left, with a QR code and the MGH logo below the main text.

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.

**F**inally! After 8 years of intensive research, this men's health 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 flab, 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.

**No Two People Are The Same, No Two Testosterone Treatments Should Be Either**

Unlike other men's health solutions, this breakthrough is tailor-made for your body. It is based on your blood test results and your hormone levels.

Until recently, it has only been available to private patients. Now, to combat the silent epidemic haunting Australian men, it is being released publicly for the first time.

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

Australian Male Hormone Clinic. "I have put together a team of leading medical doctors, hormone specialists, research scientists and nursing support staff to create this breakthrough service for Australian men."

"It is helping men to feel like men again and be their best. So we are offering a limited number of free assessments today."

**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.

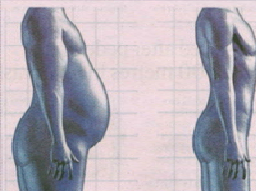
The Australian Male Hormone Clinic's groundbreaking treatment on the other hand stimulates natural testosterone production.

"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 jaw dropping.

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.

### 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 flab vanishes. Patient has more energy, more stamina and is more attractive to opposite sex.

**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 to bed 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 dud. 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

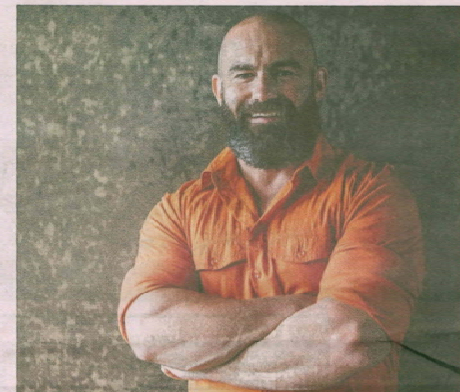
nothing to do with his personality, bank balance or looks. It's because his body is releasing alpha testosterone hormones.

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

Great results. Clinically supported data. Natural treatment.

### SAY GOODBYE TO LOW T TODAY!

- Increase muscle size
- Wake up in the morning bursting with powerful energy
- Stay razor sharp and "focused" all day long
- Feel happier, younger, more alive
- Satisfy sexual partners
- Command more respect
- Burn off pudgy flab
- Restore masculinity and strength



### 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.

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 see 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.



Be your best  
amhc.com.au

Overdiagnosis is hidden in  
the garden of diagnosis



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

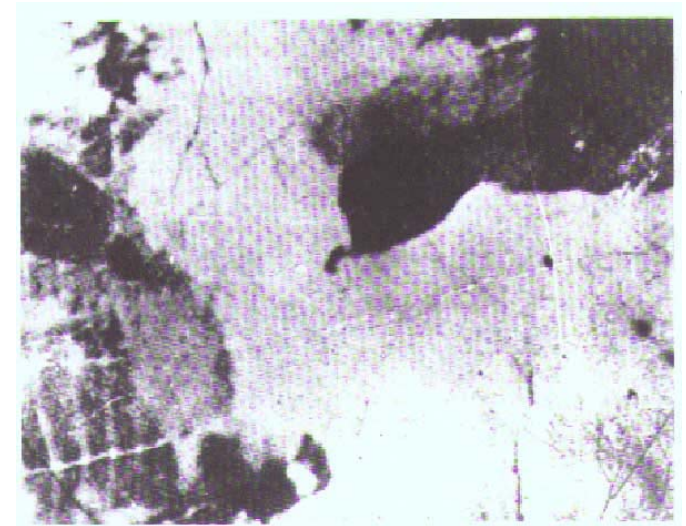
# Diagnosis: impossible but essential task

## 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.bmj.com/specialties/diagnosis-general-practice](http://www.bmj.com/specialties/diagnosis-general-practice)

[www.preventingoverdiagnosis.net](http://www.preventingoverdiagnosis.net)



What is this?

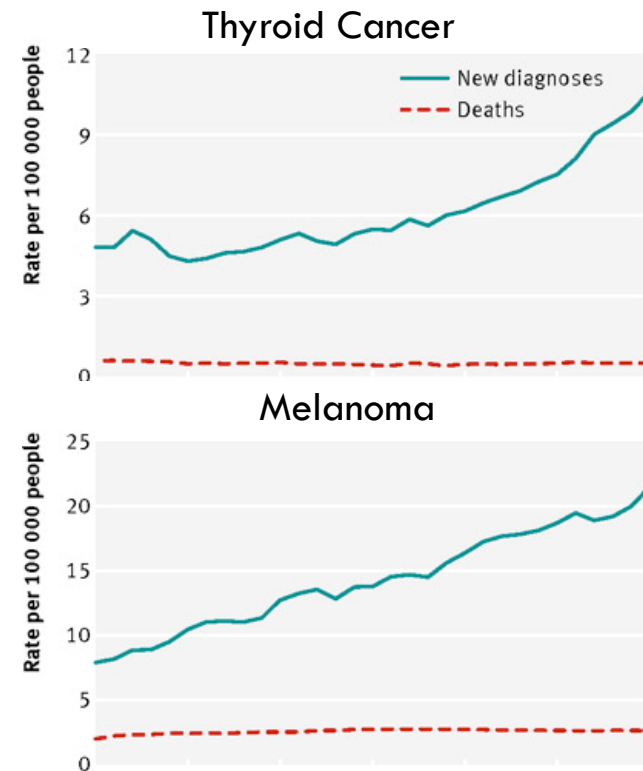
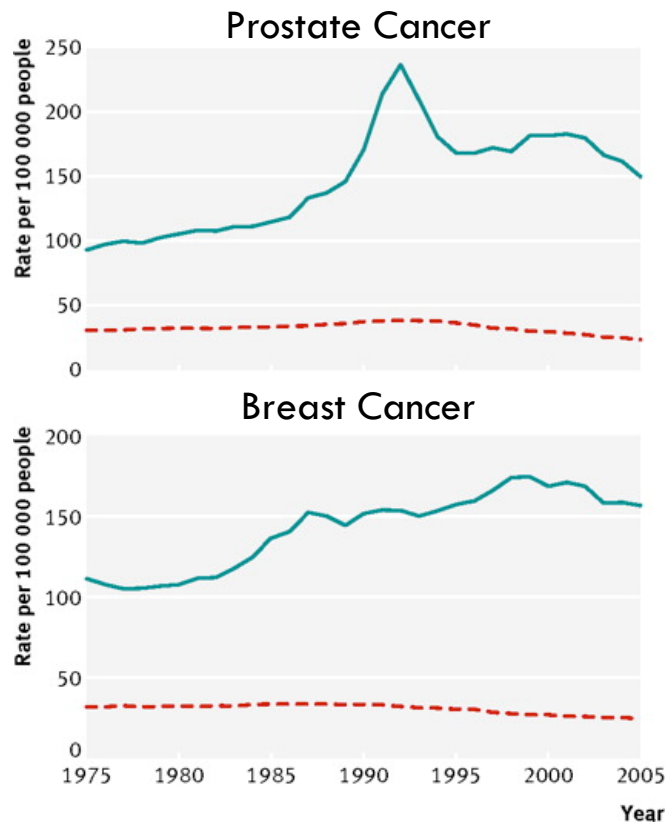






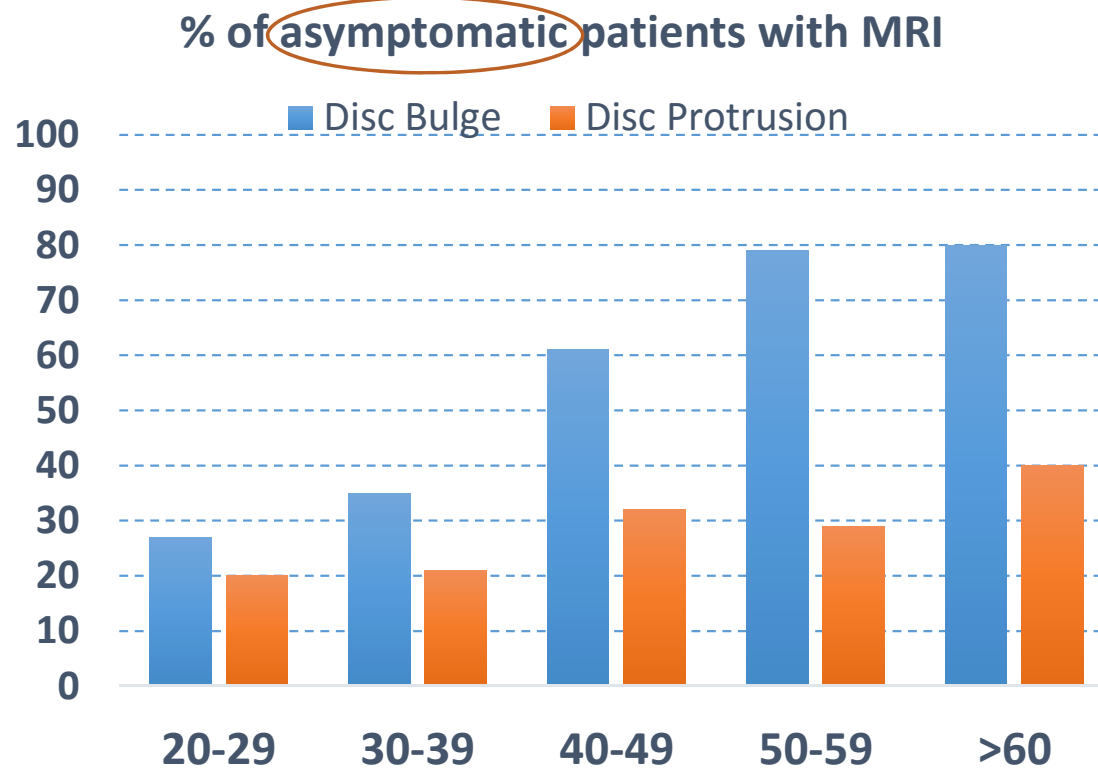
## A. Overdetection: 4 cancers

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*

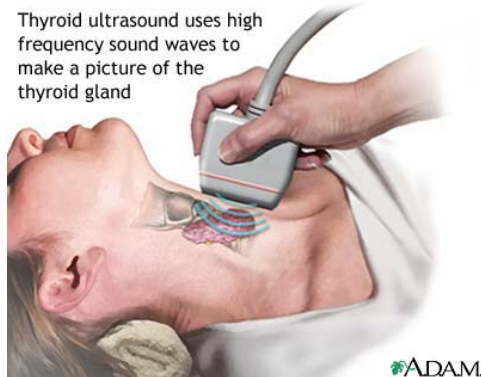
For most, treatment is not needed or is harmful.

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



**PROVEN DIAGNOSTICS**  
PROVEN DIAGNOSTICS  
212 Broadhead Road  
Bethlehem, PA 18017  
Lab Administration  
212 Broadhead Road  
Bethlehem, PA 18017

NAME	PATIENT, TEST	OFFICE USE ONLY	PHONE	DOB	AGE	SEX
W0001001	W0001001	12/15/1970		01/01/1970	39 Yrs	M
W0001001	W0001001	06/29/2009 08:00	06/29/2009 08:05	ORDERING DR	DOC, TEST	
Test Description		Result	Abnormal	Reference Range	Units	Lab
BASIC METAB PANEL		Results: 06/29/2009 08:18 Status: F				
BUN	16			6-20	mg/dL	P1
CREATININE	0.9			0.7-1.3	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	250		H	70-120	mg/dL	P1
RESULTS RECHECKED						
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

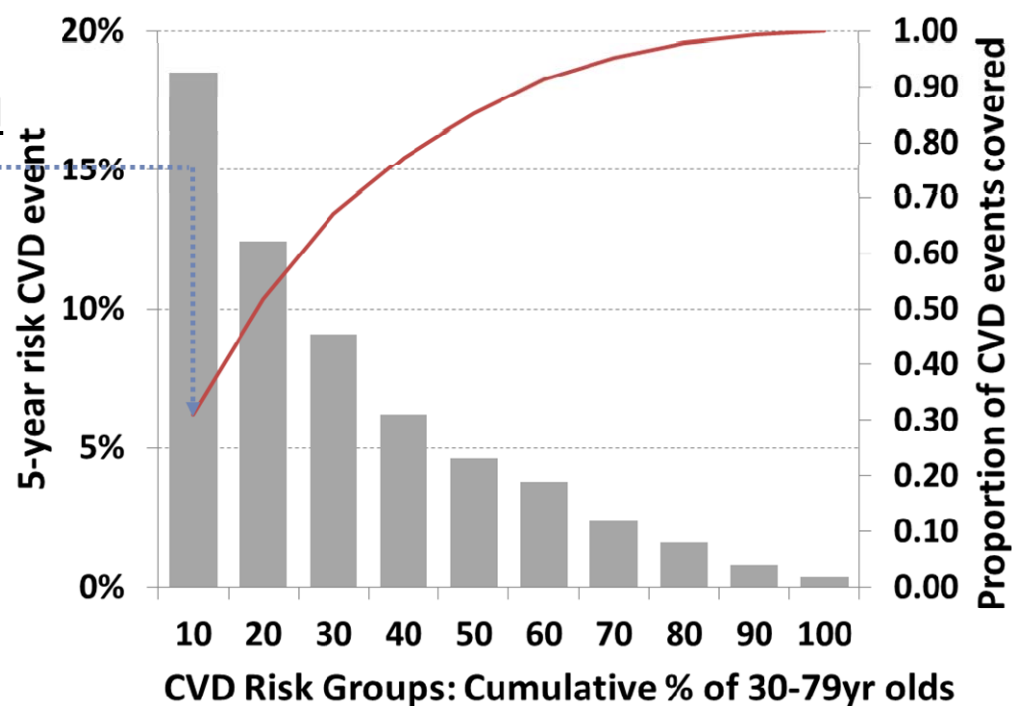
Performing Laboratory Information:  
PL - PERFORMED AT PROVEN DIAGNOSTICS LABORATORY 212 BROADHEAD RD BETHLEHEM PA 18017

# 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

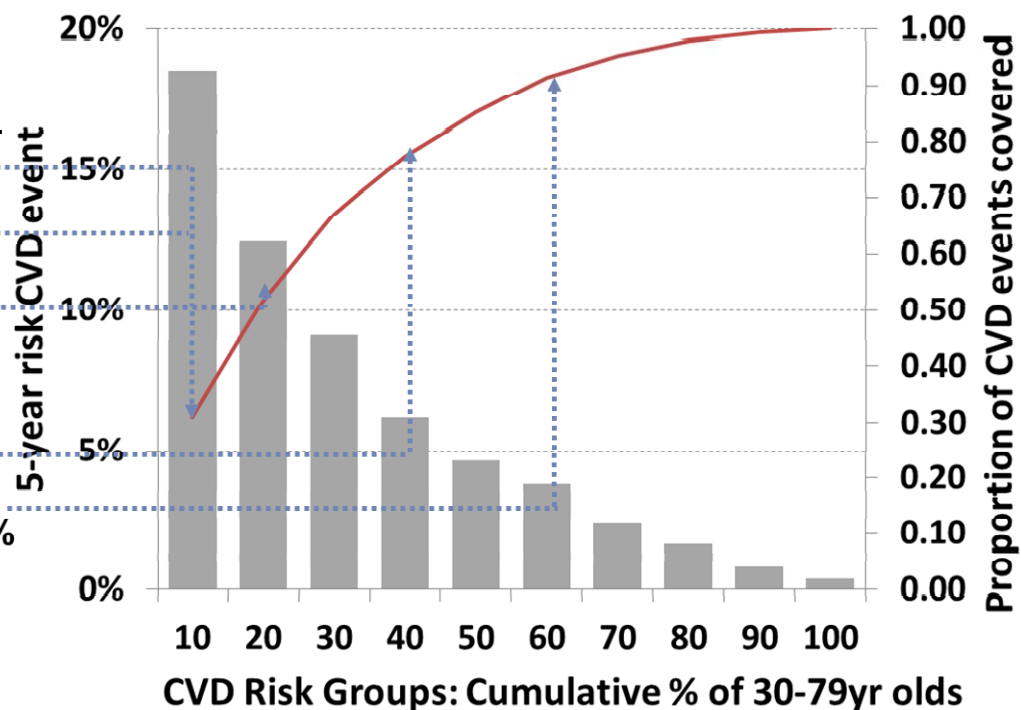
1990's New Zealand 15%

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-yr)



# 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

### Sackett's "Normal"s

Table 5.1 Six definitions of normal

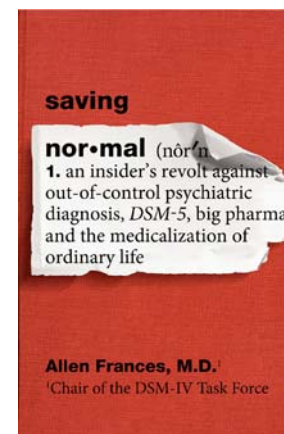
1. *Gaussian*: the mean  $\pm 2$  standard deviations (SD) – this one assumes a normal distribution for all tests and results in all "abnormalities" having the same frequency.
2. *Percentile*: within the range, say of 5–95% – has the same basic defect as the Gaussian definition. Implies a specificity 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



# 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



Conference partners



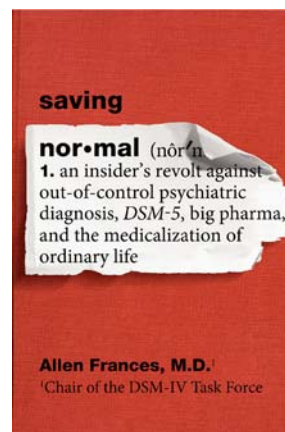
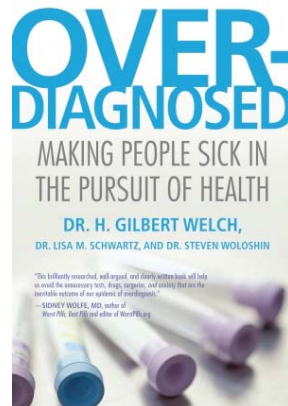
2013 Dartmouth  
2014 Oxford  
2015 Washington

Home About Readings Conference Blog Contact



LESS IS MORE  
- = + MEDICINE

HOME THE IDEA TOOLS BLOG [FRANÇAIS] ABOUT



(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

PLOS MEDICINE

## 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<sup>1\*</sup>, Georga P. E. Cooke<sup>1</sup>, Jenny A. Doust<sup>1</sup>, Lisa Bero<sup>2</sup>, Suzanne Hill<sup>3</sup>, Paul P. Glasziou<sup>1</sup>

<sup>1</sup> Bond University, Robina, Australia, <sup>2</sup> University of California, San Francisco, San Francisco, California, United States of America, <sup>3</sup> 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.”



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**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](mailto:Paul_Glasziou@bond.edu.au)  
[www.crebp.net.au](http://www.crebp.net.au)





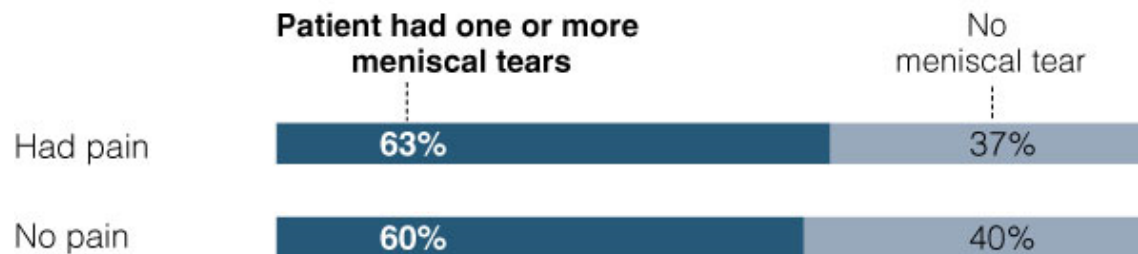


# Meniscal tears: MRI vs pain

## 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

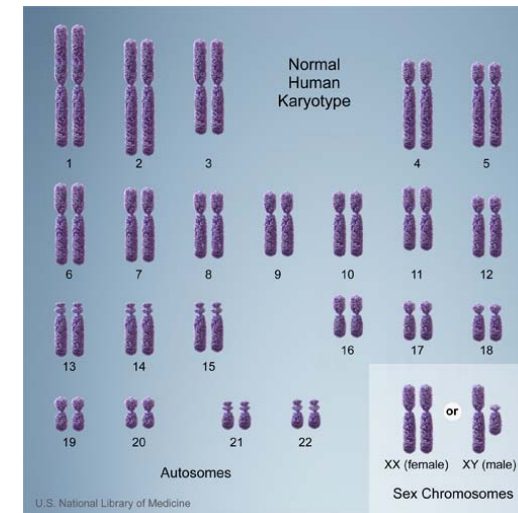


Source: Dr. David T. Felson, Boston University School of Medicine

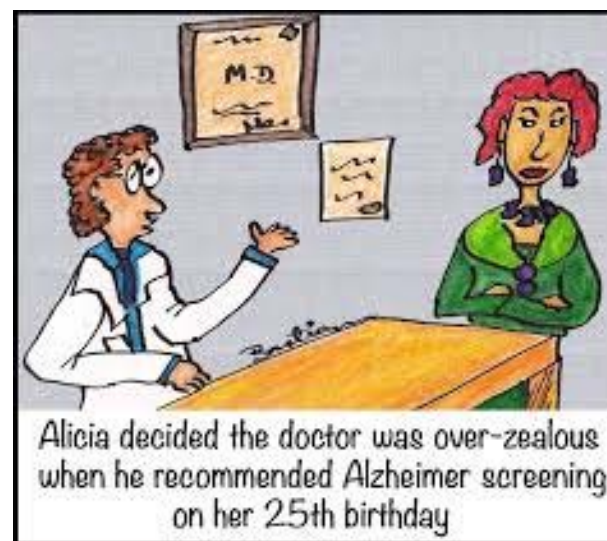
THE NEW YORK TIMES

# 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

Statistically-funny.blogspot.com



# 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*<sup>1</sup>, Ray Moynihan *senior research fellow*<sup>1</sup>, Tessa Richards *analysis editor*<sup>2</sup>,  
Fiona Godlee *editor in chief*<sup>2</sup>

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

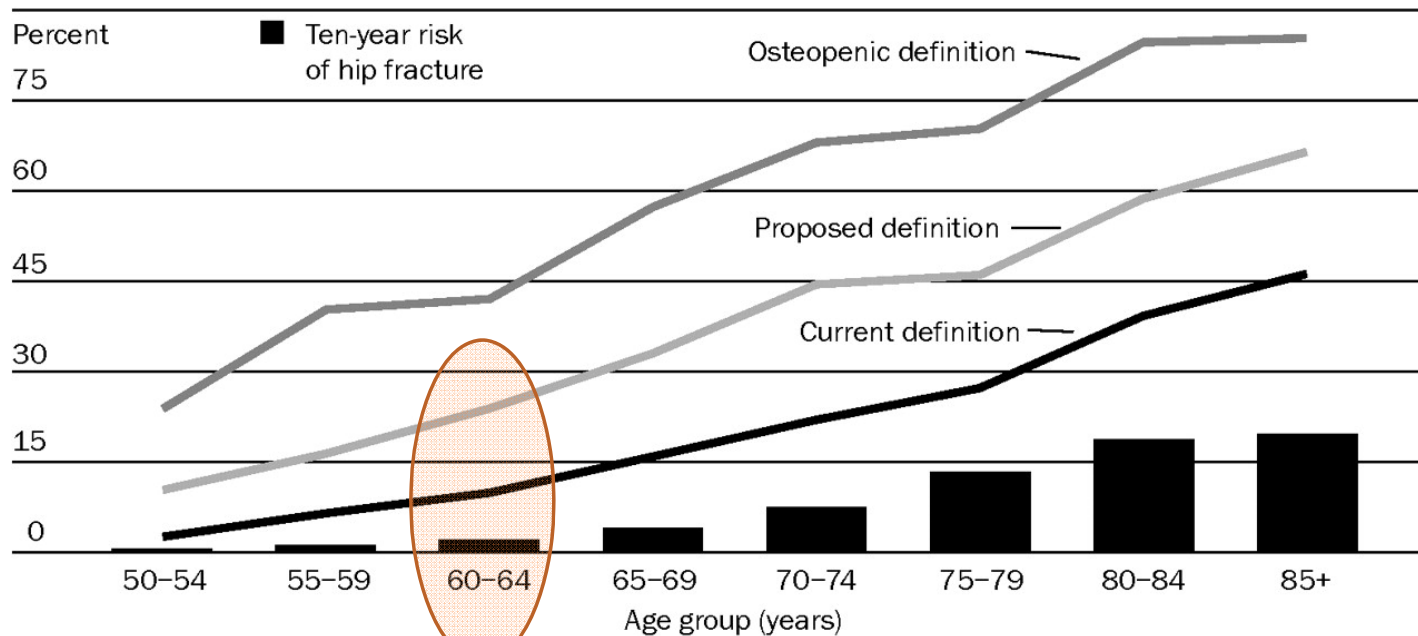
Too much testing of well people and not enough care for the sick worsens health inequalities and drains professionalism, harming both those who need treatment and those who don't."

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

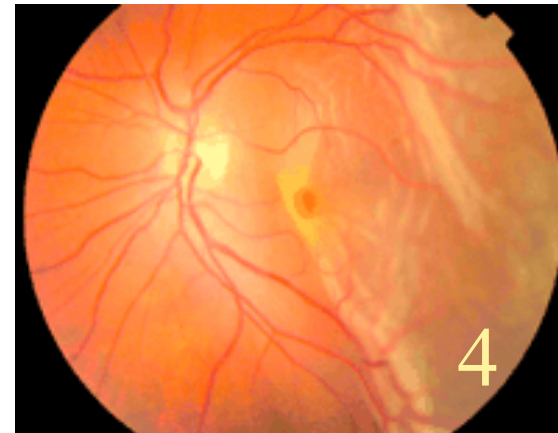
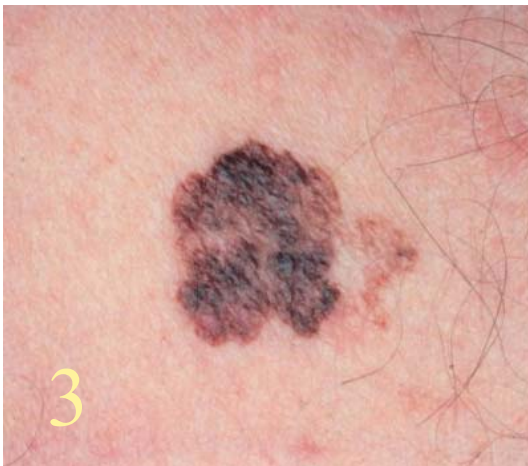


## Topic 2: EBM in diagnostics

- 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

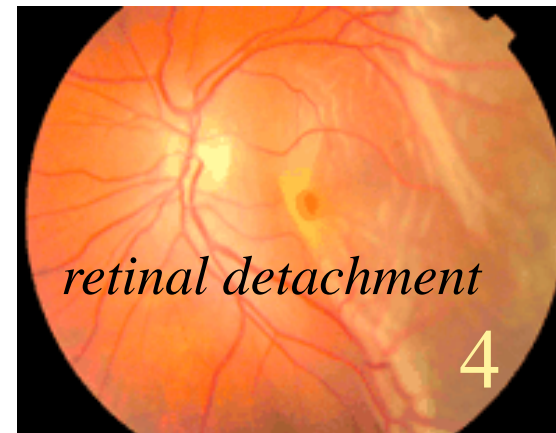
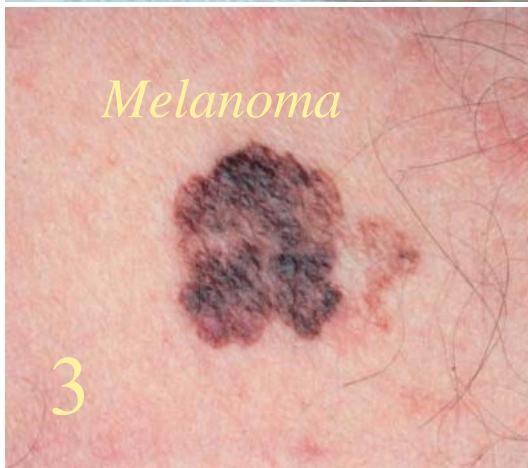
## What is the diagnosis?





# Topic 1: Diagnosis by pattern matching

## What is the diagnosis?



# We learn basic patterns



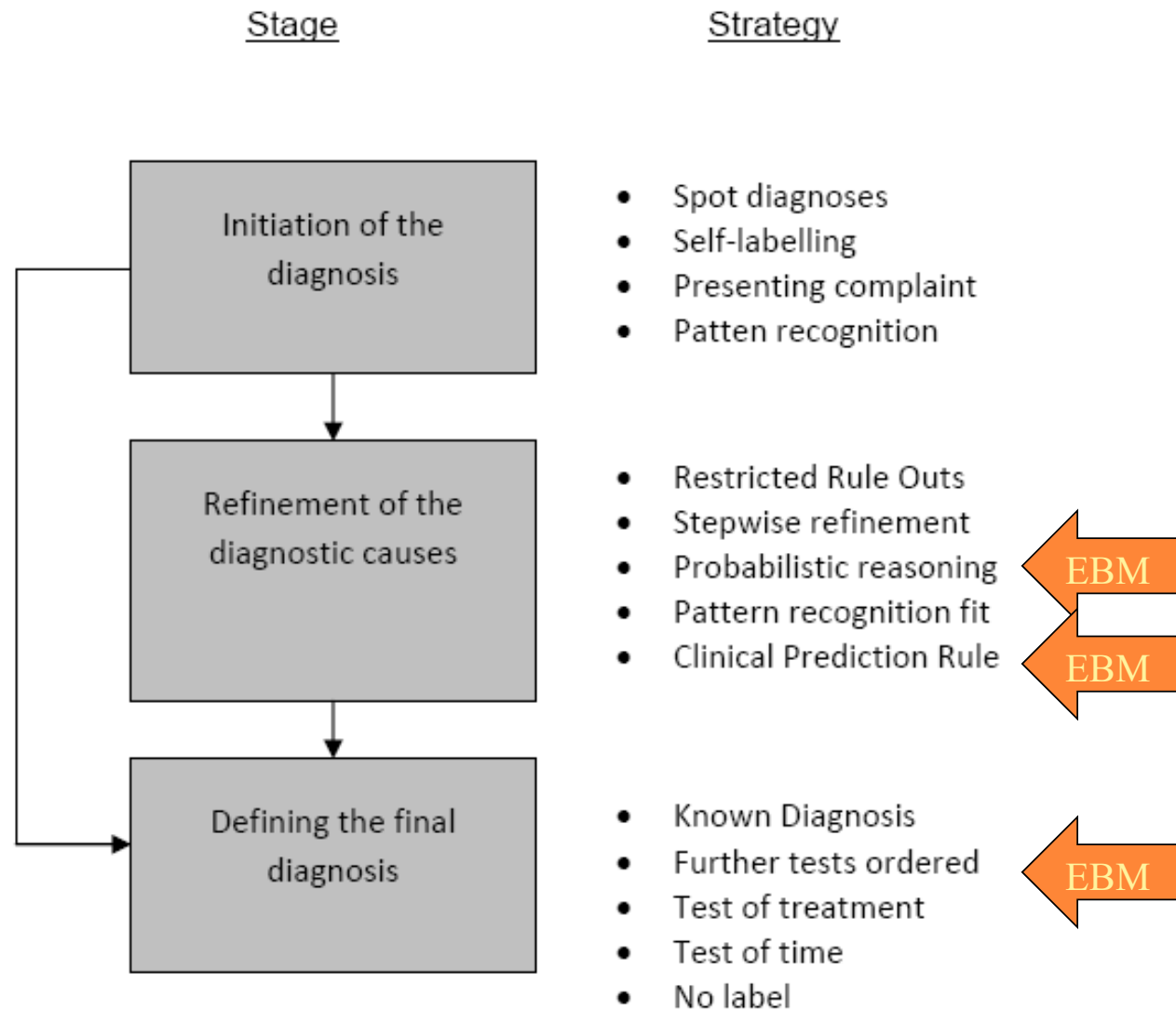
Then see them in new situations.



# Diagnosis and level of experience with a presentation

1. No experience: use first principles / processes  
Vascular, Infection/Inflammatory, Neoplastic, Degenerative,  
Intoxication/Iatrogenic, Congenital, Allergic, Traumatic, ...
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



Heneghan, BMJ 2008

### 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



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The Free Encyclopedia

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pfapa wiki

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## 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.<sup>[1][2][3]</sup>

Contents [hide]



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

**Periodic fever, aphthous stomatitis, pharyngitis and adenitis**

Classification and external resources

DiseasesDB 33682

### Signs and symptoms



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6-140.

# 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