

“I don't know why you say that making a diagnosis is the most important thing a doctor does.

As a general practitioner I hardly ever make a diagnosis.”

General practitioner North London

# Problem Solving

# Decision Making

Malattia

Diagnosi  
(non diagnosi)

# Non diagnosi

Dolore toracico

Dolore addominale non  
determinato

Low back pain

MUPS

Quante diagnosi facciamo (fate)?

Esonerati dall' onere della diagnosi?

# Il Morbo di Schiraldi

0.1%

95%

?

Cos'è la malattia?

Linneo

The diagnoses tabulated in this way are **theoretical abstractions**, but we are inclined to give them a level of credence and reality that tends to exceed that granted to the **patients so labelled**.

In this way, our diagnoses begin to condone structural violence and to excuse social injustice.

Iona Heath president, Royal College of General Practitioners  
BMJ 2012;345:e6595

Every experienced clinician is fully aware that **no two people ever experience the same diagnosed disease in exactly the same way,** and yet the taxonomies of diagnosis and the international classifications that underpin them ignore this underlying truth.

Iona Heath president, Royal College of General Practitioners  
BMJ 2012;345:e6595

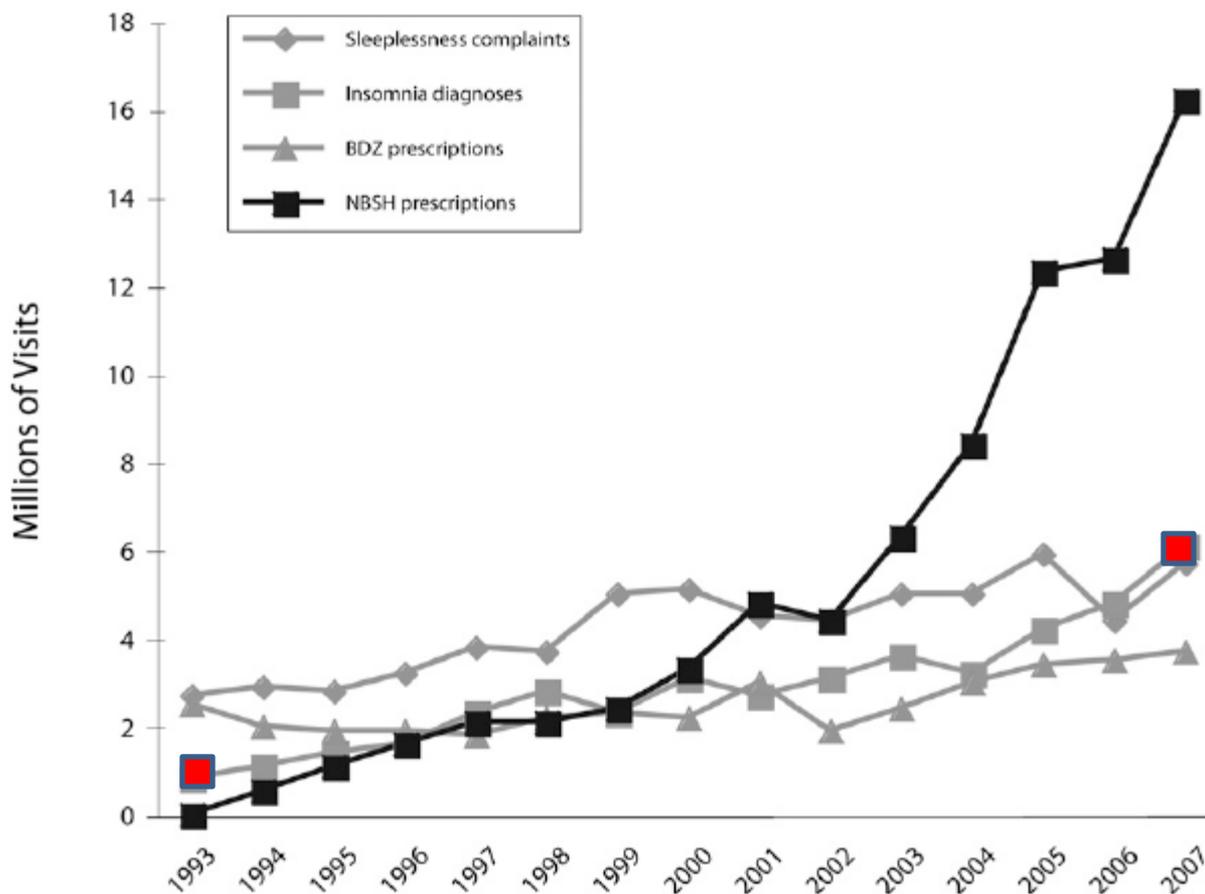
# Medicalising unhappiness

Dowrick C, Frances A BMJ 2013;347:f7140

It allows major depressive disorder to be diagnosed **just two weeks after a bereavement.**

Turning grief into a mental disorder substitutes **a superficial medical ritual** for deep and time honoured cultural ones.

Dowrick C, Frances A BMJ 2013;347:f7140



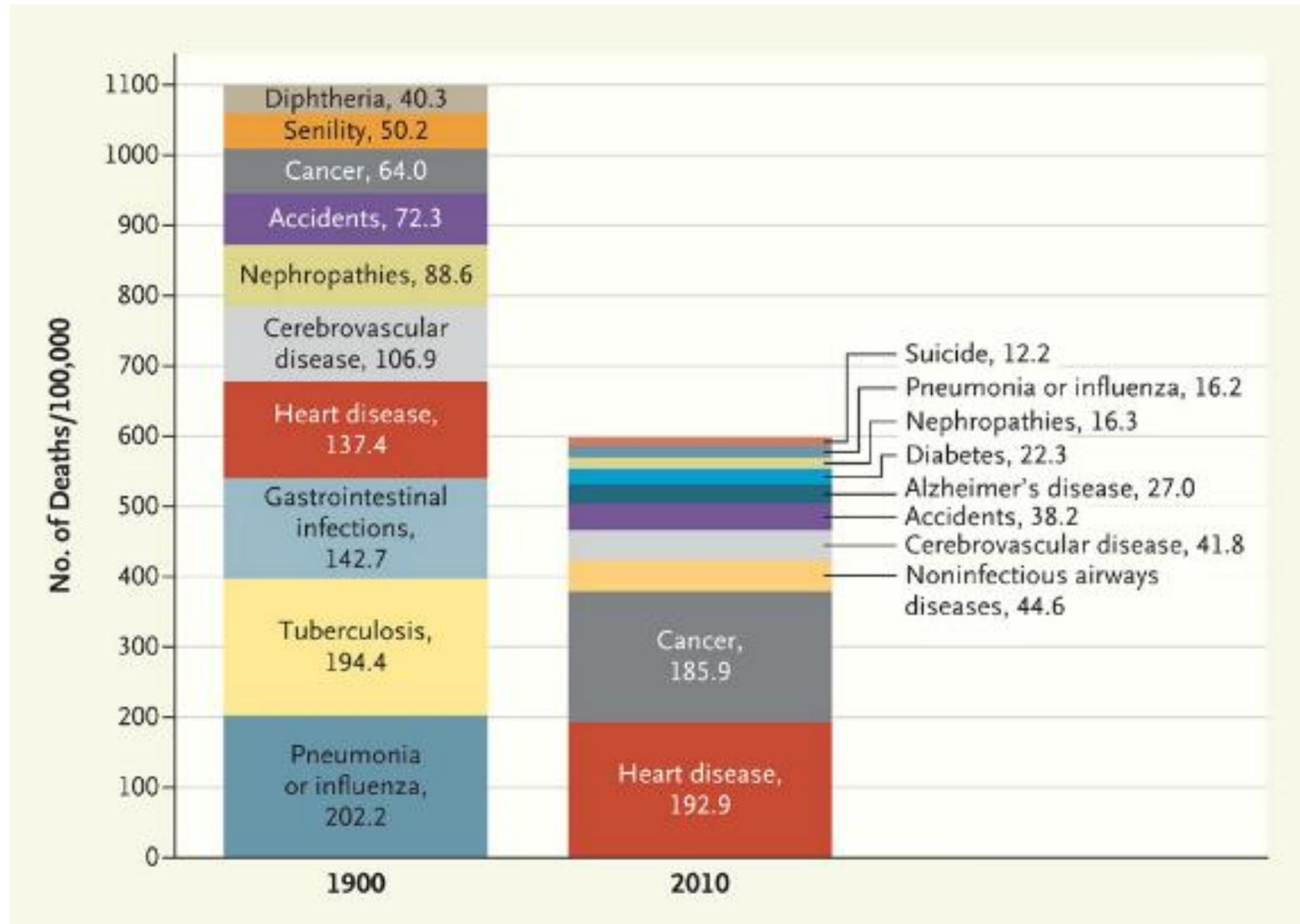
**FIGURE 1—Sleeplessness-related trends of complaint, insomnia diagnosis, benzodiazepine (BDZ) and nonbenzodiazepine sedative hypnotic (NBSH) prescription as a result of physician office visits: United States, 1993–2007.**

Benzodiazepine use is associated with an increased risk of Alzheimer's disease.

Unwarranted long term use of these drugs should be considered as a public health concern.

BMJ 2014;349:g5205

# Il mondo cambia



# Comorbidità

# Fuoco amico

*Critical illness are an intrinsically iatrogenic disorder*

Crit Care 2011 15: 22



Sepsis is not simply the host response to an infection, nor is it the same as sterile inflammation.

Rather, sepsis is **the host's deleterious, non-resolving inflammatory response to infection that leads to organ dysfunction.**

The combination of **genetic factors** and **environmental exposures** provides the crucial mix to produce the biological events that lead to individual diseases and diseases in individuals.

John Bell  
Redefining disease  
Harveian Oration 2010

# Demystifying diagnosis

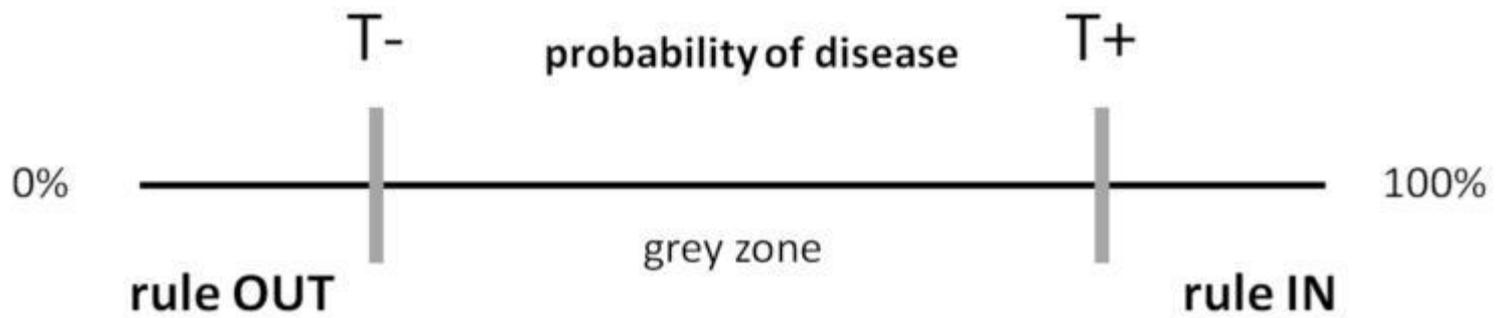
## Against diagnosis

La diagnosi è un'opinione

La revisione di un'opinione

## **Un nodo decisionale.**

Il livello di incertezza al quale è possibile decidere minimizzando le conseguenze prevedibile degli inevitabili errori.



**We believe** in the **objectivity**, **utility**, and **veracity** of test results.

The common problem of overtesting and overreliance on tests is also compounded by an even trickier, and even deeper secret about tests: **we don't understand them.**

David Newman, Hyppocrates' shadow

Faced with uncertainty and the **fear of uncertainty**, clinicians may find themselves ordering batteries of tests, perhaps especially younger clinicians and those working in more litigious environments.

Fiona Goodley BMJ 2014;349:g6665

# disease

present

absent

positive

test

negative

<b>True Positive</b>	<b>False Positive</b>	$\frac{\text{True Positive}}{\text{All Positive}}$
<b>False Negative</b>	<b>True Negative</b>	$\frac{\text{True Negative}}{\text{All Negative}}$
<b>True Positive with disease</b>	<b>True Negative without disease</b>	

Sensitivity

Specificity

**Sn** - positive in disease

**Sn OUT**

**Sp** - negative in health

**Sp IN**

**Sensibilità e specificità sono**  
**caratteristiche del test.**

Ci dicono come funziona il **test** in popolazioni che  
**SAPPIAMO** avere o non avere la malattia.

**Quindi non servono?**

# disease

present

absent

positive

test

negative

<b>True Positive</b>	<b>False Positive</b>	$\frac{\text{True Positive}}{\text{All Positive}}$
<b>False Negative</b>	<b>True Negative</b>	$\frac{\text{True Negative}}{\text{All Negative}}$
<b>True Positive with disease</b>	<b>True Negative without disease</b>	

Positive Predictive Value

Negative Predictive Value

Sensitivity

Specificity

Bayes's theorem of diagnostic probability states that **the predictive value of an abnormal test will vary according to the *probability of disease* in the *population* under study**

**Prevalenza**

# prevalenza

proporzione di una data popolazione in cui  
la malattia è presente

## probabilità **PRE-TEST**

(prevalenza stimata della malattia)

la **prevalenza** della malattia

In **clinica** varia sulla base di

***storia, segni e sintomi***

probabilità **PRE-TEST**

# Quale popolazione?

Un test per HIV ha lo stesso PPV o NPV in un convento di clausura e in un carcere?

Una popolazione di un individuo

Tra due specchi

L'insostenibile leggerezza dell' ST

# Likelihood ratio

$$LR_{+} = \frac{\text{Sens}}{1 - \text{Spec}}$$

$$LR_{-} = \frac{1 - \text{Sens}}{\text{Spec}}$$

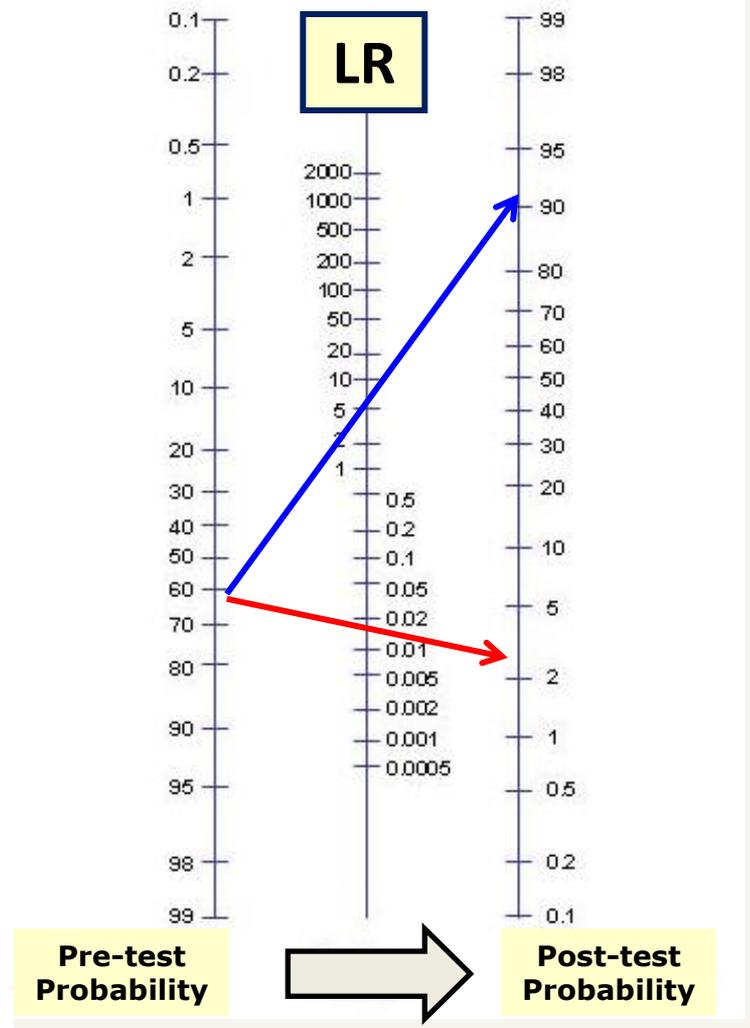
$$LR_{+} = \frac{VP}{FP}$$

$$LR_{-} = \frac{FN}{VN}$$

<b>LR+</b>		<b>LR-</b>
>10	diagnostico	<0.1
5-10	forte	0.1-0.2
3-5	moderato	0.3-0.5
1.1-3	debole	0.5-0.9
<b>1</b>	inutile	<b>1</b>

LR indica di quanto un segno clinico o un test diagnostico **alzano** o **abbassano** la probabilità pretest della malattia (quella malattia).

## Revisione della probabilità

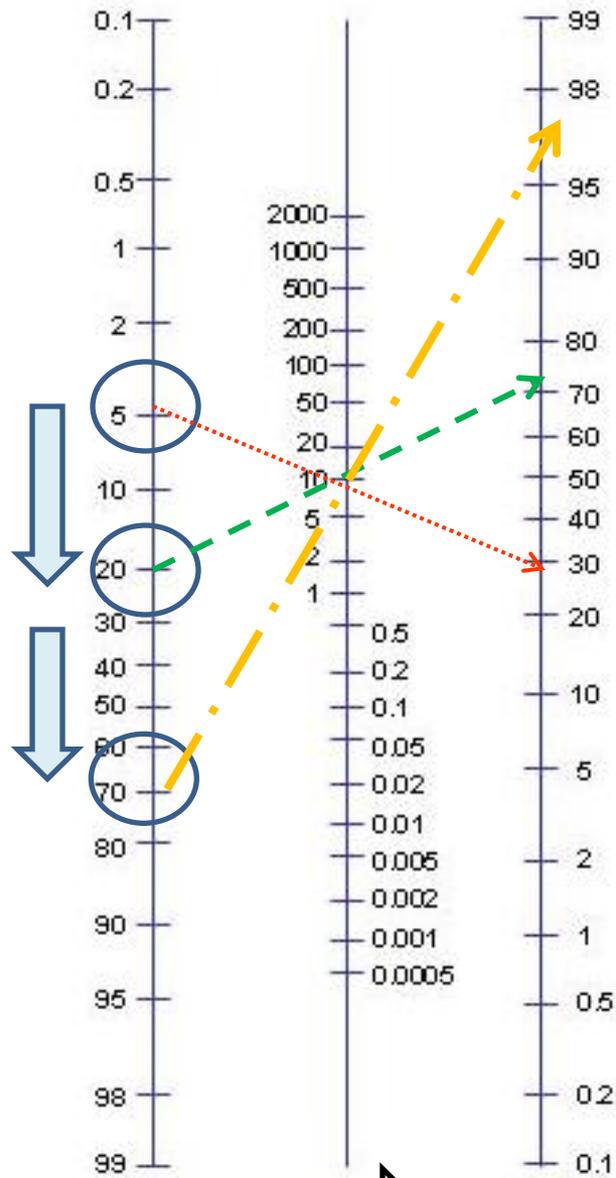


# Bayes ha vinto la guerra

By updating our initial belief about something with objective new informations, we get a new and improved belief. This is an elegant statement about learning from experience.

(S.Bertsch McGrayne)

Se oggi c'è il sole.

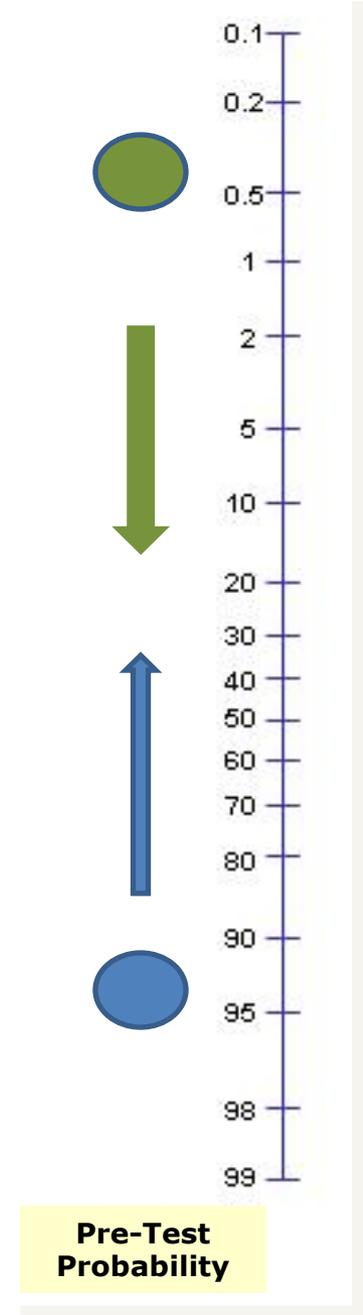


**Pre-Test Probability**

**Post-test Probability**

# Come arrivare ad una ragionevole probabilità a priori (pretest)?

- Impressione soggettiva
- Scores clinici
- Anamnesi
- Esame obiettivo



“To be a good diagnostician, a physician needs to acquire a ***large set of labels for disease***, each of which binds an idea of the illness and its symptoms, possible antecedents and causes, possible developments and consequences, and possible intervention to in part cure or mitigate the illness.

Kahaneman

Learning medicine consists in part of learning the language of medicine.”

Kahaneman

Pattern recognition

Conoscere per riconoscere.

Bisogna vedere malati.

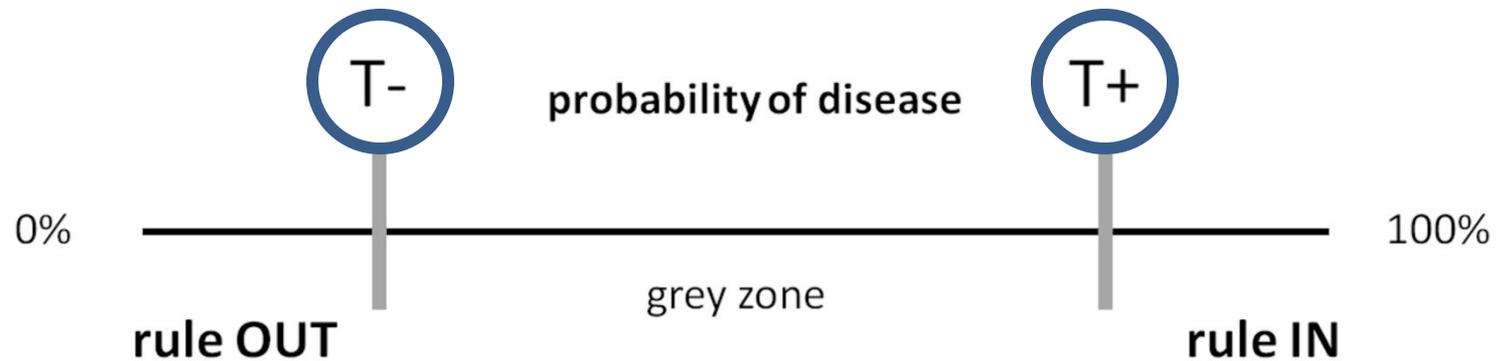
**Bisogna studiare.**

*Diagnostic accuracy does not depend as much on strategy as **on mastery of contents.***

J.Kassirer

# Melioidosi

# Varcare quella **soglia**...



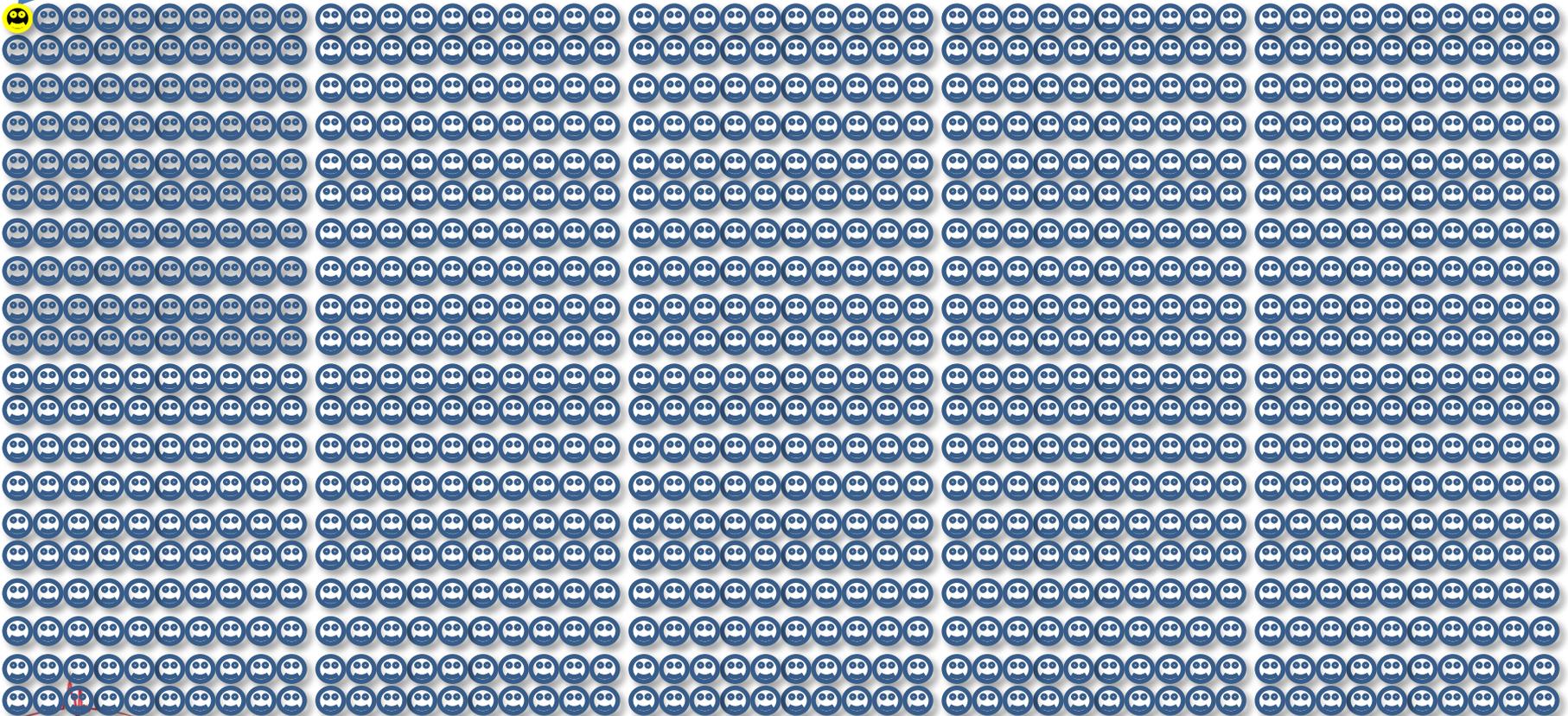
0.1%

95%

2%

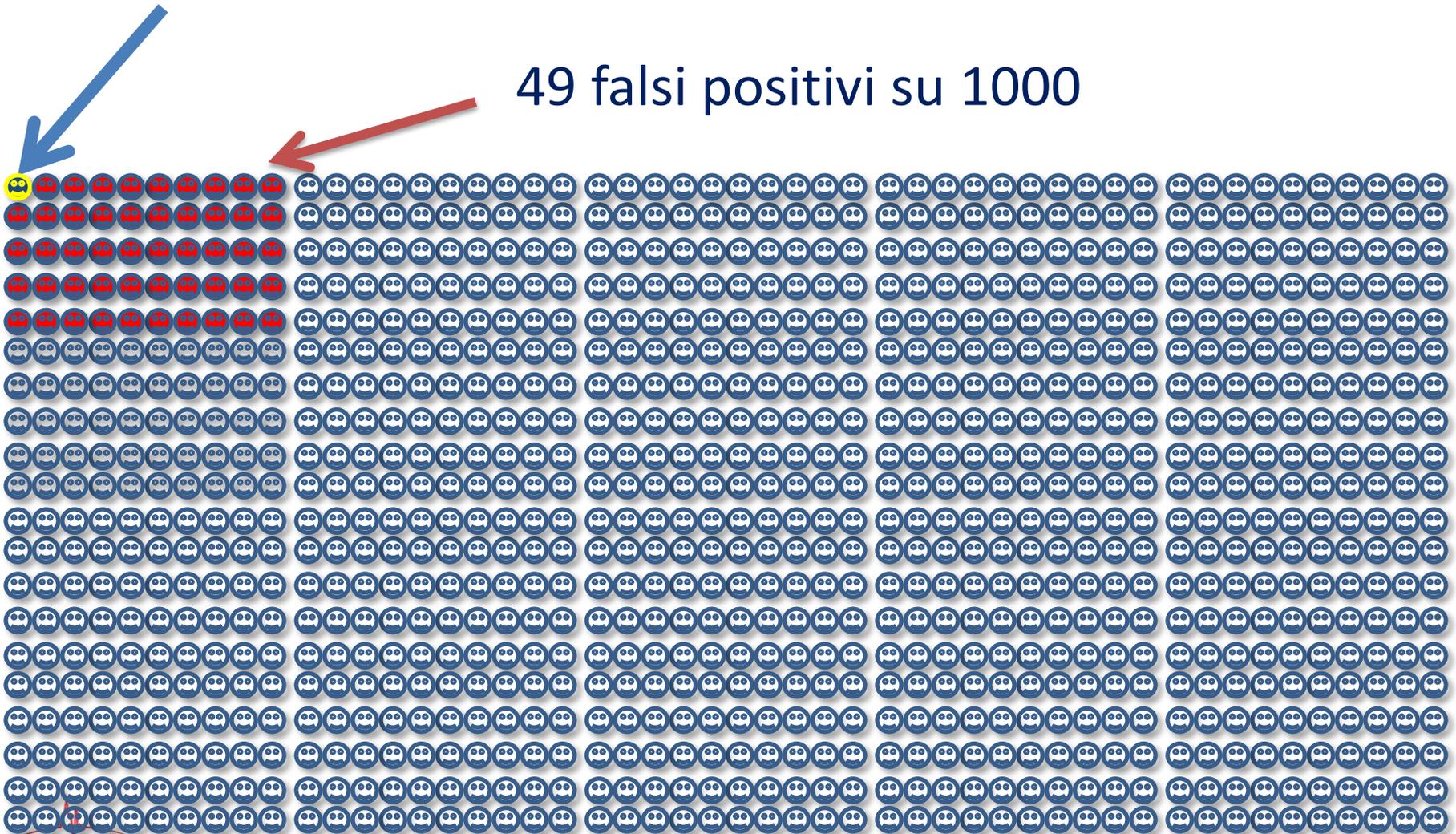
Poveri Bayes e Laplace

1 malato su 1000 = 0,1%



1 malato su 1000 = 0,1%

49 falsi positivi su 1000



**With a very low prevalence** even a very sensitive test might produce an **unacceptably high number of false positives.**

This is the great problem with **screening** that great hits the public imagination, sometimes even **against evident demonstrations of uselessness.**

Minchia, Intern Emerg Med. 2012 Oct;7 Suppl 3:S173-9.

Siamo esseri umani.

Ragioniamo (o dovremmo ragionare) come  
esseri umani.

Decidiamo in condizioni di incertezza.

Siamo pigri.

# *Dual Process Theory*

Intuitivo



Analitico

Prendere le scorciatoie.

Energy saving brains.

Heuristics.

Strategie cognitive istintive.

Funzionano, e piuttosto bene. Ma non sempre.

# Distorsioni del giudizio

## Pregiudizi

# Tra Bayes e bias

Availability (memorability)

Representativeness

Base rate neglect

Anchoring

Confirmatory bias

Overconfidence

# Il contesto

# Demonstrable biases in the medical setting

- Gender
- Education
- Race
- Ethnicity
- Obesity
- Psychiatric illness
- Age
- Socioeconomic status
- Sexual orientation
- Substance abuse disorders
- Chronic and complex illness

I will not permit considerations of  
***age, disease or disability, creed, ethnic origin,  
gender, nationality, race, sexual orientation,  
or social standing***

to intervene between my duty and my patient.

*WMA Declaration of Geneva 2006*

# Intolerance of error and culture of blame drive medical excess

Hoffman JR BMJ 2014;349:g5702 (14 oct )

The medical culture of **shame** and **blame**, which can lead practitioners to deny and hide errors, has also fed the **pretension that modern medicine is based on perfected science**, which in turn implies that any error, and indeed any adverse outcome, represents unacceptable failure.

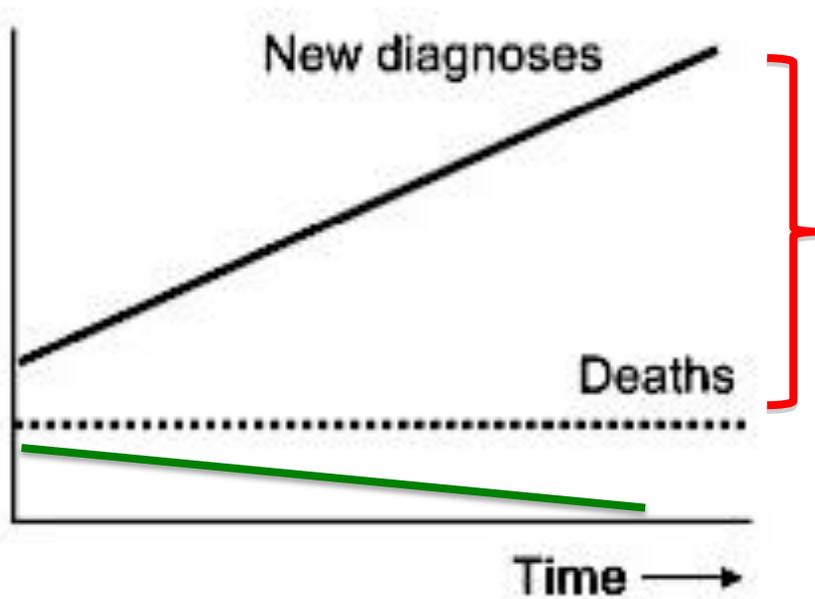
This pretension has also been sold to the public.

Hoffman JR BMJ 2014;349:g5702

Crediamo nelle narrazioni

A zero tolerance for error and uncertainty drives the culture of overdiagnosis and overtreatment.

Hoffman JR BMJ 2014;349:g5702



overdiagnosis



# Harming the healthy



# Overdiagnosis

```
graph TD; A([Overdiagnosis]) --> B[Tests<br/>Treatments<br/>Harms]; A --> C[Medicalisation]; A --> D[Diverting resources];
```

Tests  
Treatments  
Harms

Medicalisation

Diverting  
resources

Overdiagnosis

Overtreatment

Conflitti di interesse

Illusioni di beneficio

# Drivers of overdiagnosis

- Technological changes detecting ever **smaller** “**abnormalities**”
- Commercial and professional **vested interests**
- Conflicted panels produce **expanded disease definitions** and writing **guidelines**

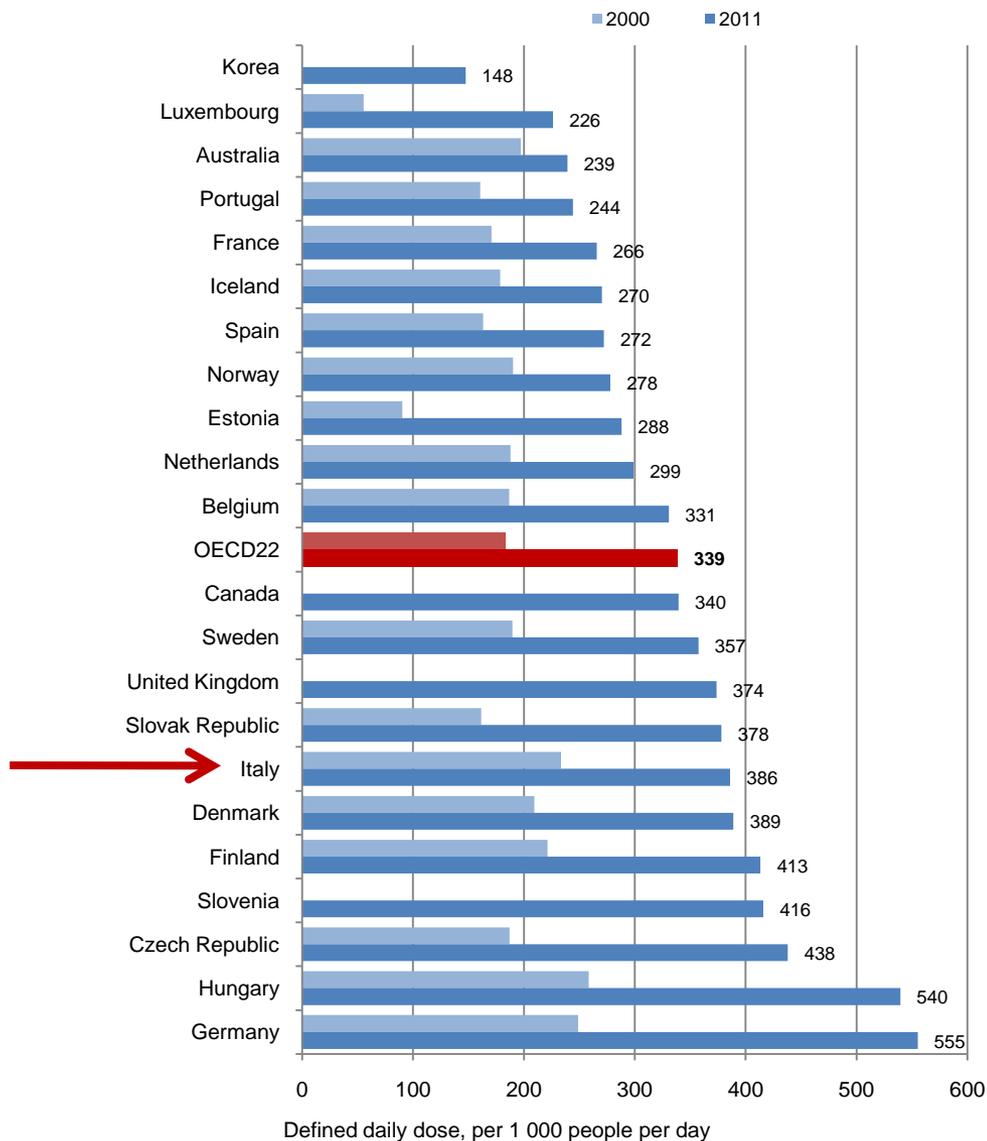
# *Different ways to expand disease definitions*

Creating new categories of **pre-disease**

**Lowering** diagnostic **thresholds**

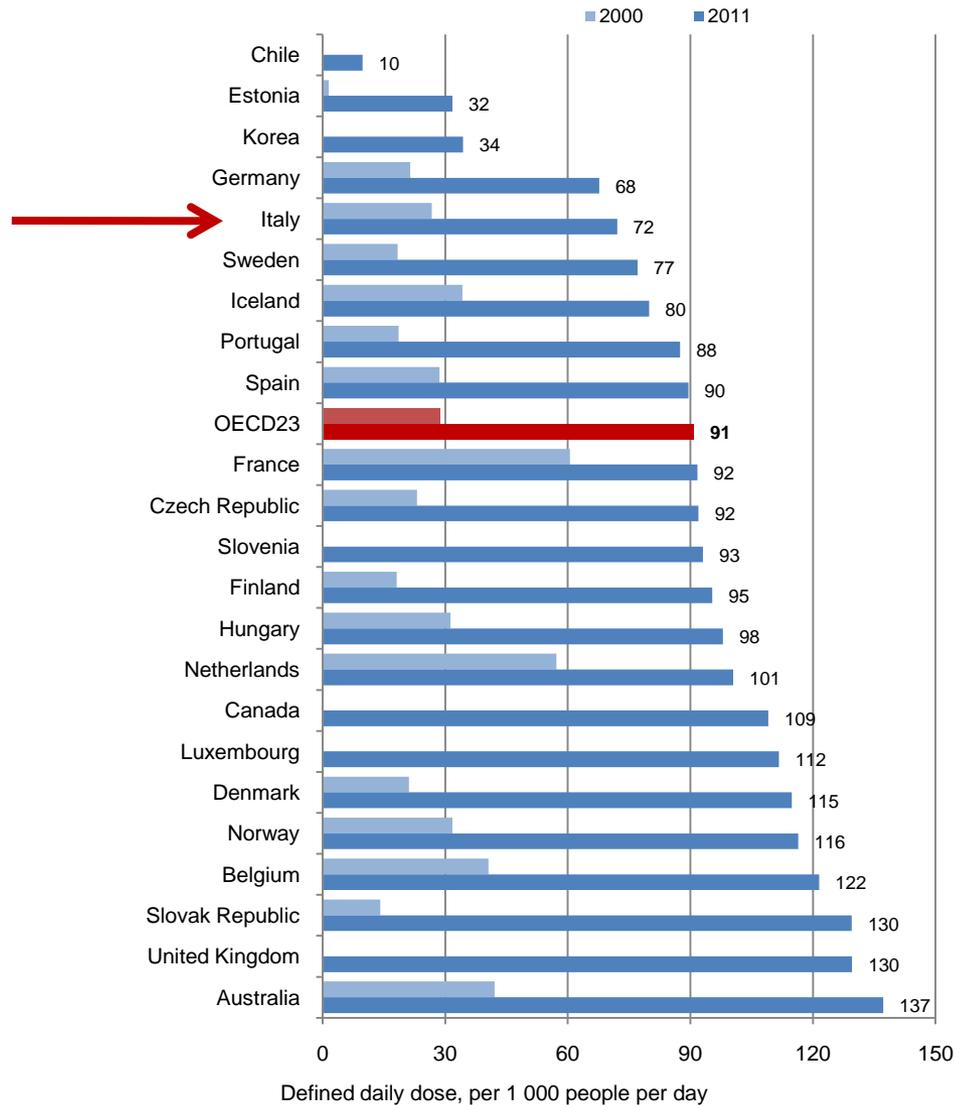
**Earlier diagnosis**, different diagnostic method

# Hypertension drugs consumption, 2000 and 2011 (or nearest year)



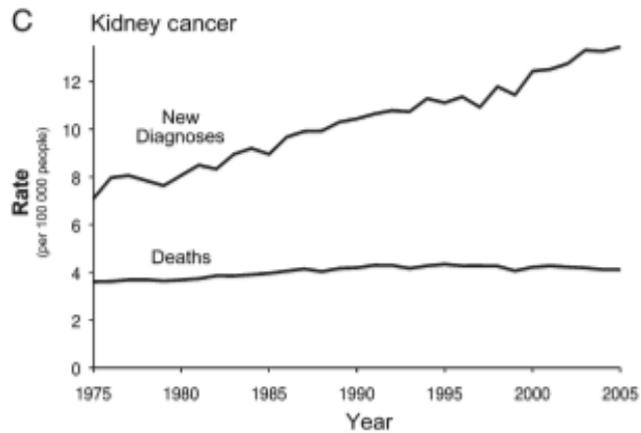
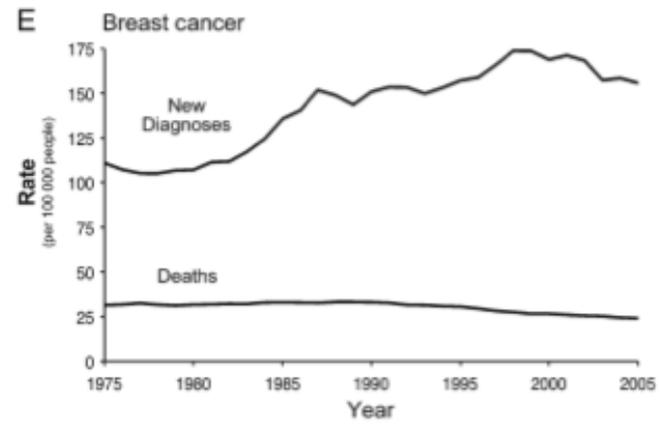
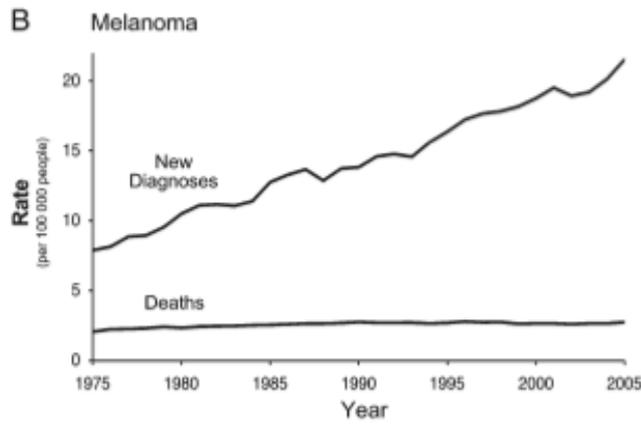
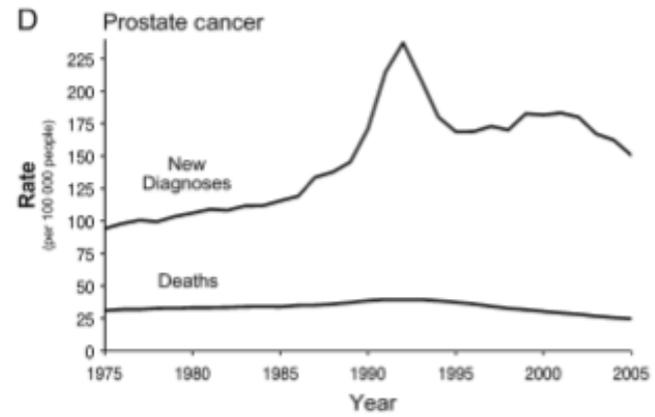
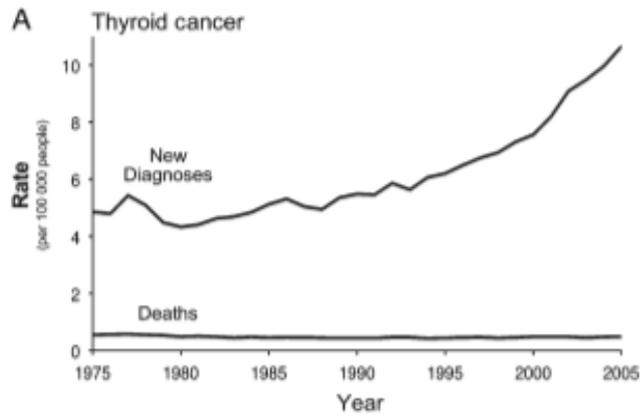
From: Health at a Glance: Europe 2012

## Anticholesterols consumption, 2000 and 2011 (or nearest year)

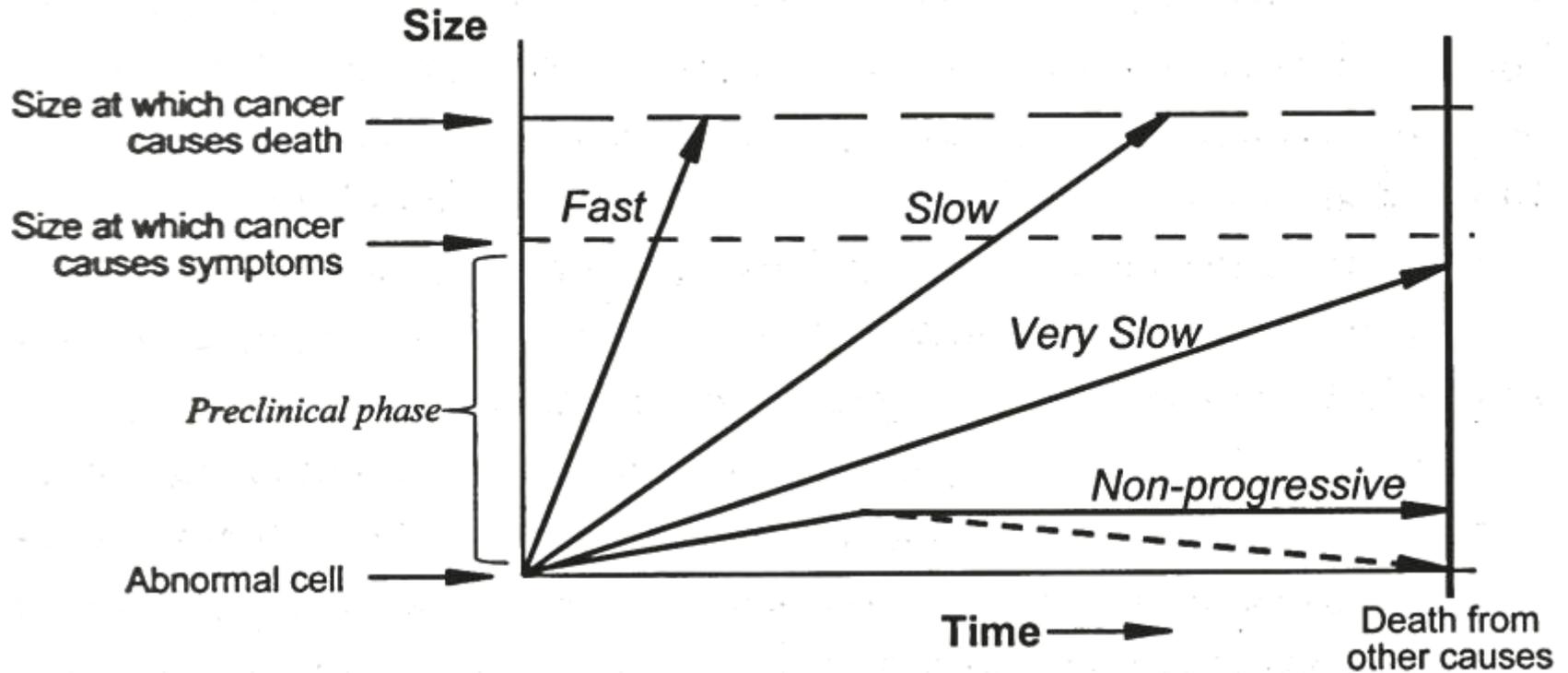


From:  
Health at a Glance: Europe 2012

Exaggeration of benefits  
and suppression of harms



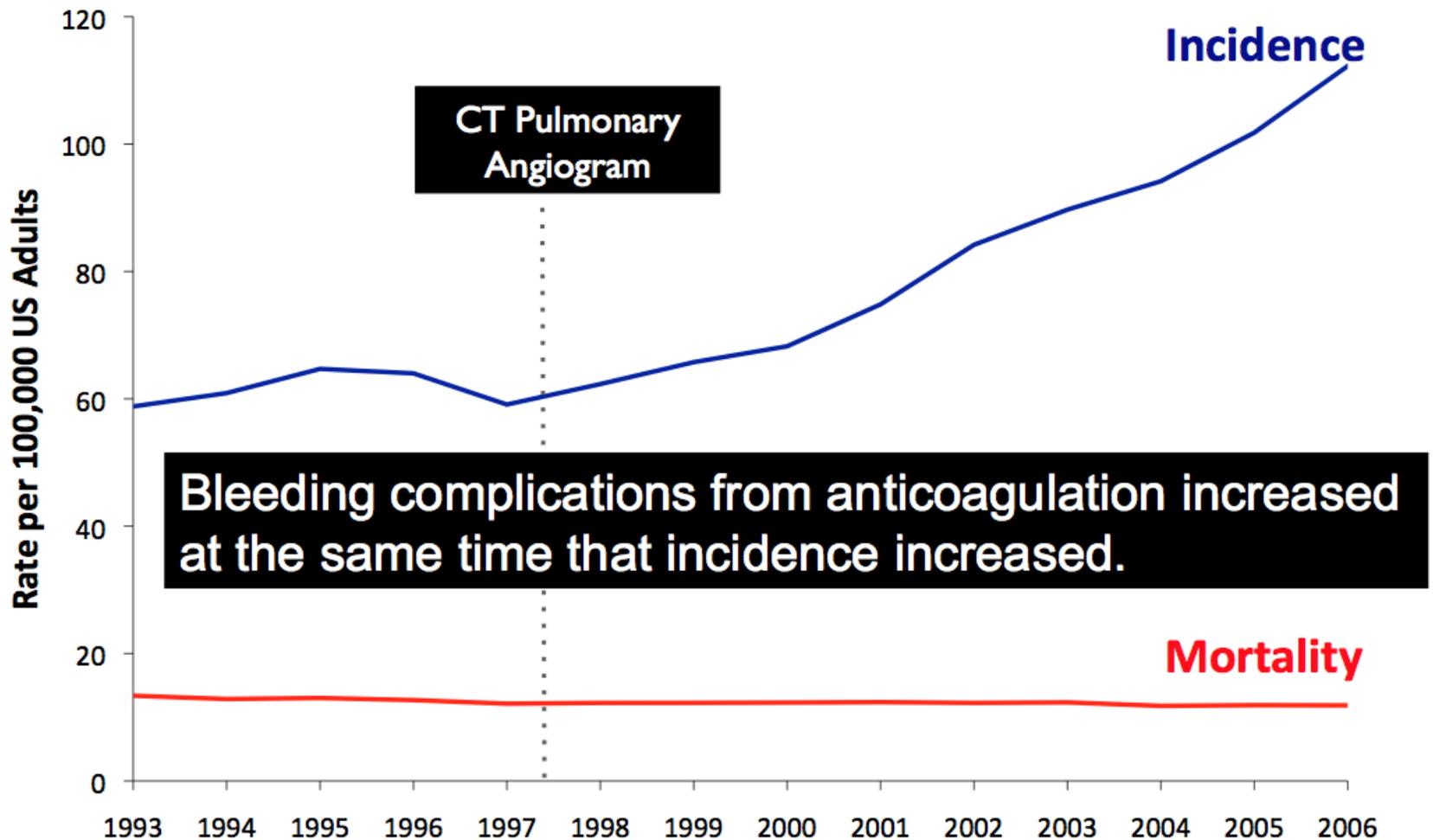
# The Heterogeneity of Cancer Progression



J Natl Cancer Inst 2010;102:605-613

# Diagnosis and treatment of pulmonary embolism: a metaphor for medicine in the evidence-based medicine era.

Prasad V Arch Intern Med. 2012; 172(12):955-958.



Weiner, Arch Intern Med 2012;307(22):2400-240

# La diagnosi come strumento

"C'e` da congratularsi con lui per essere morto di una malattia tanto rara, ma più ancora per avere trovato medici capaci di riconoscerla e darle il suo vero nome."

*Jan Potockj,  
Manoscritto trovato a Saragozza*

Clinicians and patients need to acknowledge the limits of medicine and embrace the uncertainty that lies beyond.

Only because we do not understand everything and because we cannot control the future is it possible to live.

Iona Heath, 2014