

# Ricerca genomica e ICT in Sanità: possibili sinergie

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Genomics of Cancer Unit

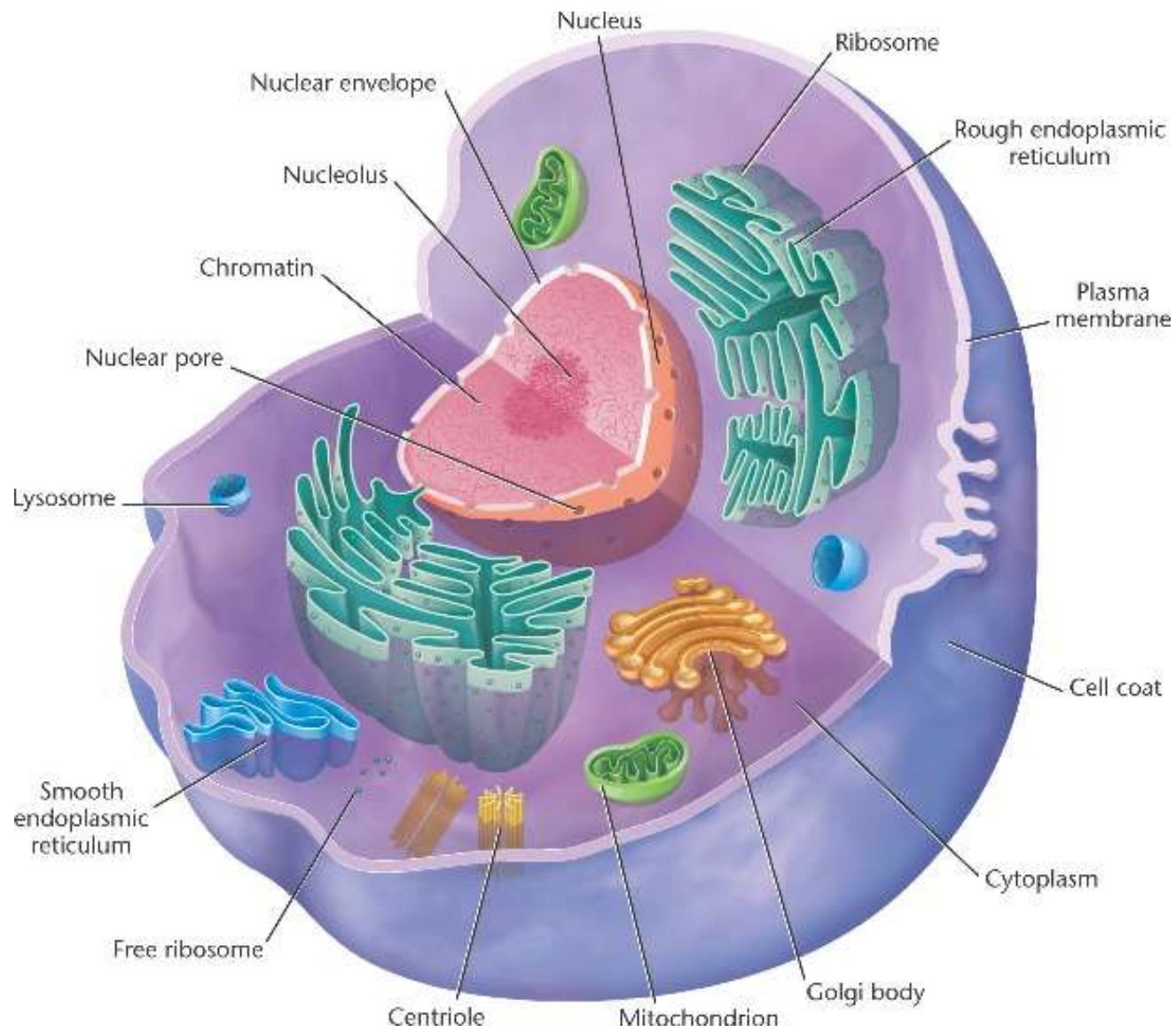
Director, Center for Translational  
Genomics and Bioinformatics

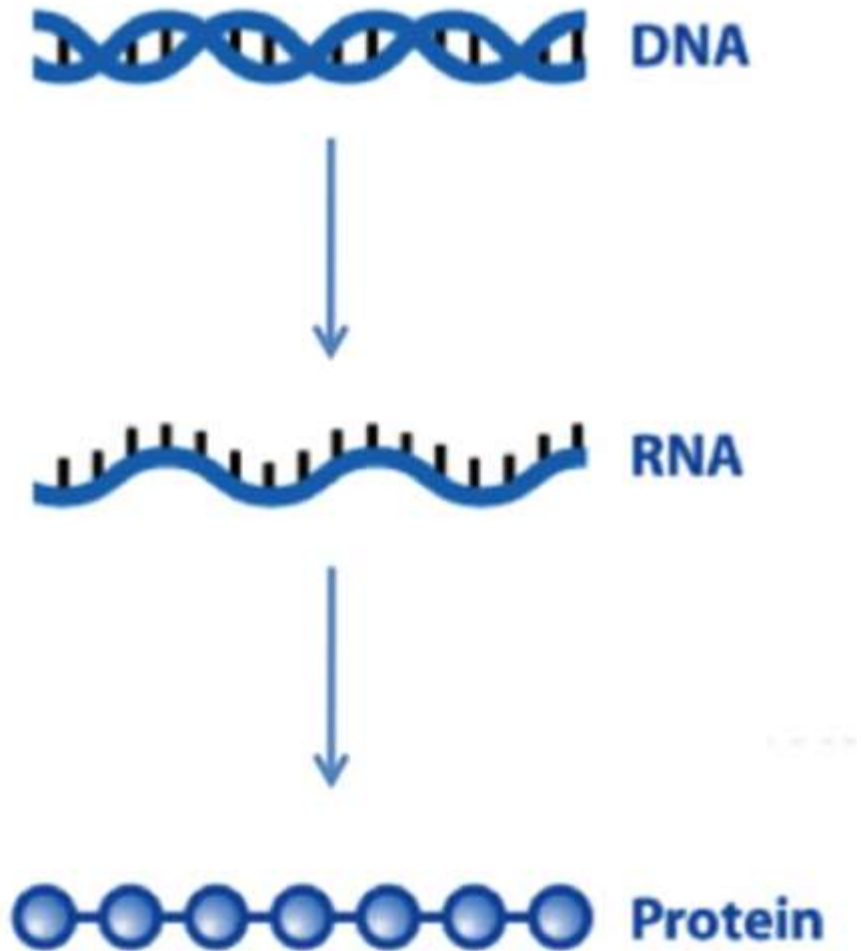
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CONVEGNO ANNUALE AISIS

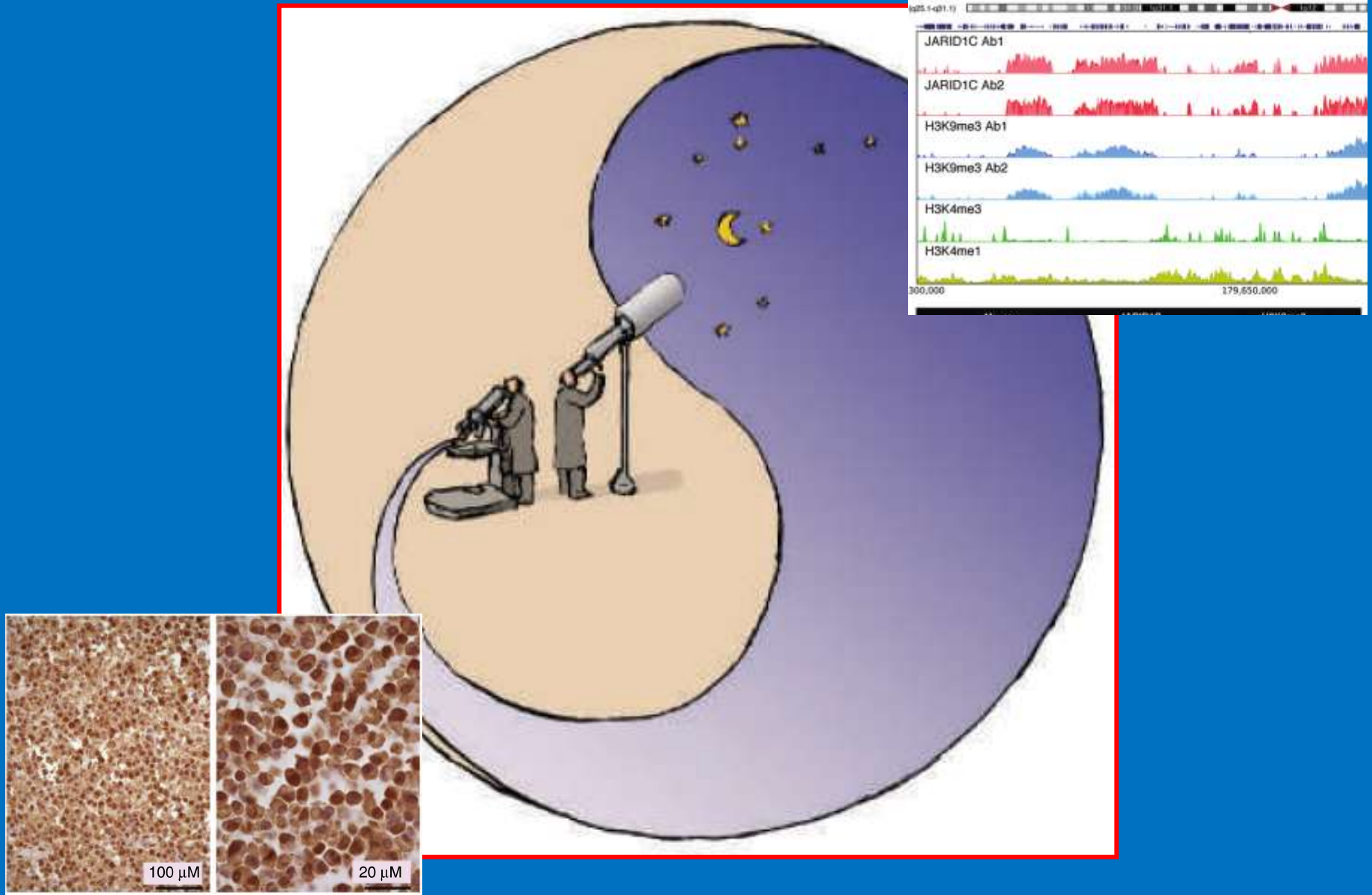
Cagliari

13 Ottobre 2016





*... and metabolites*



Knight, D. (2007). "Kinds of minds." Nature **447**(7141): 149.





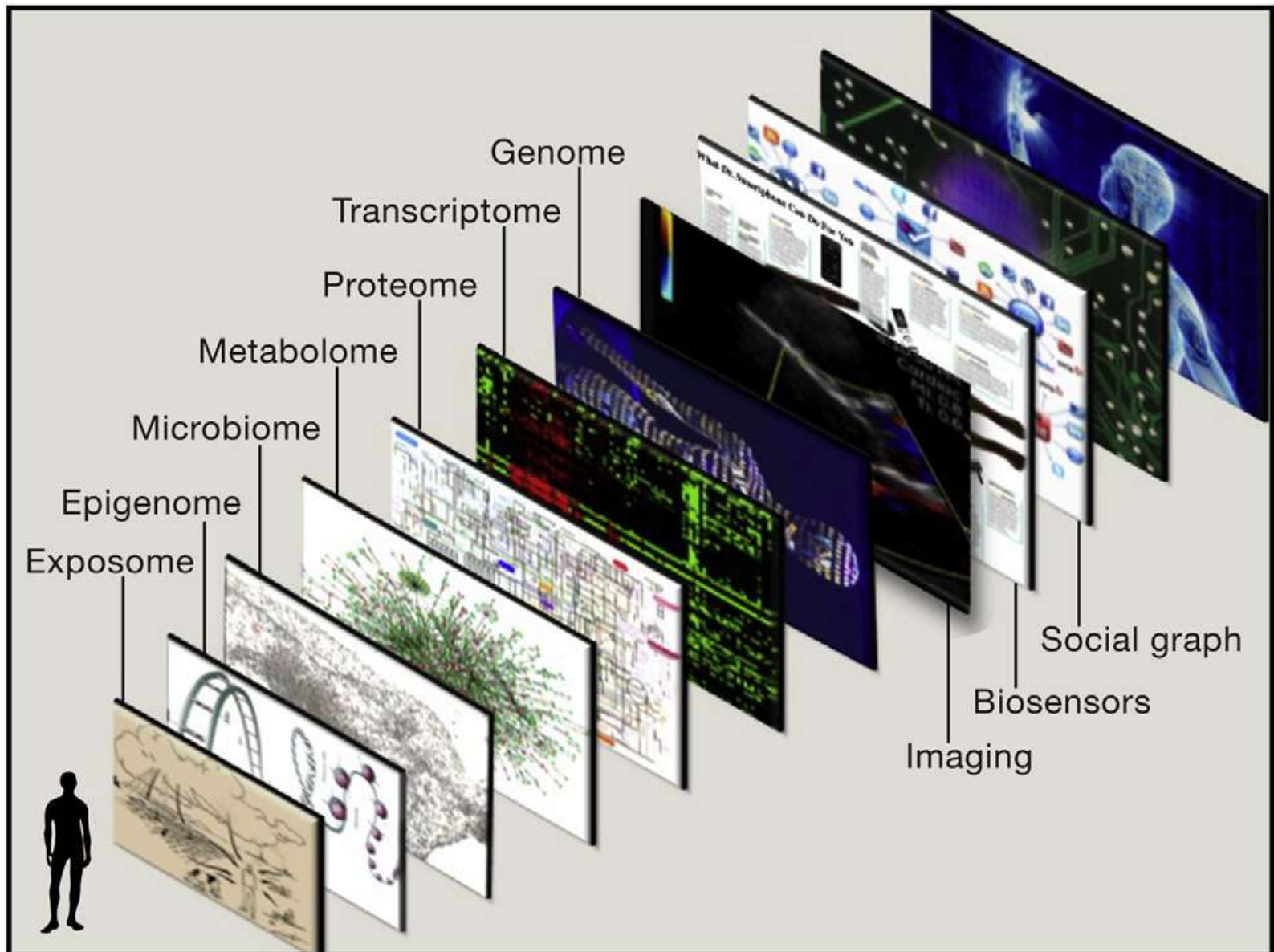


**Health**

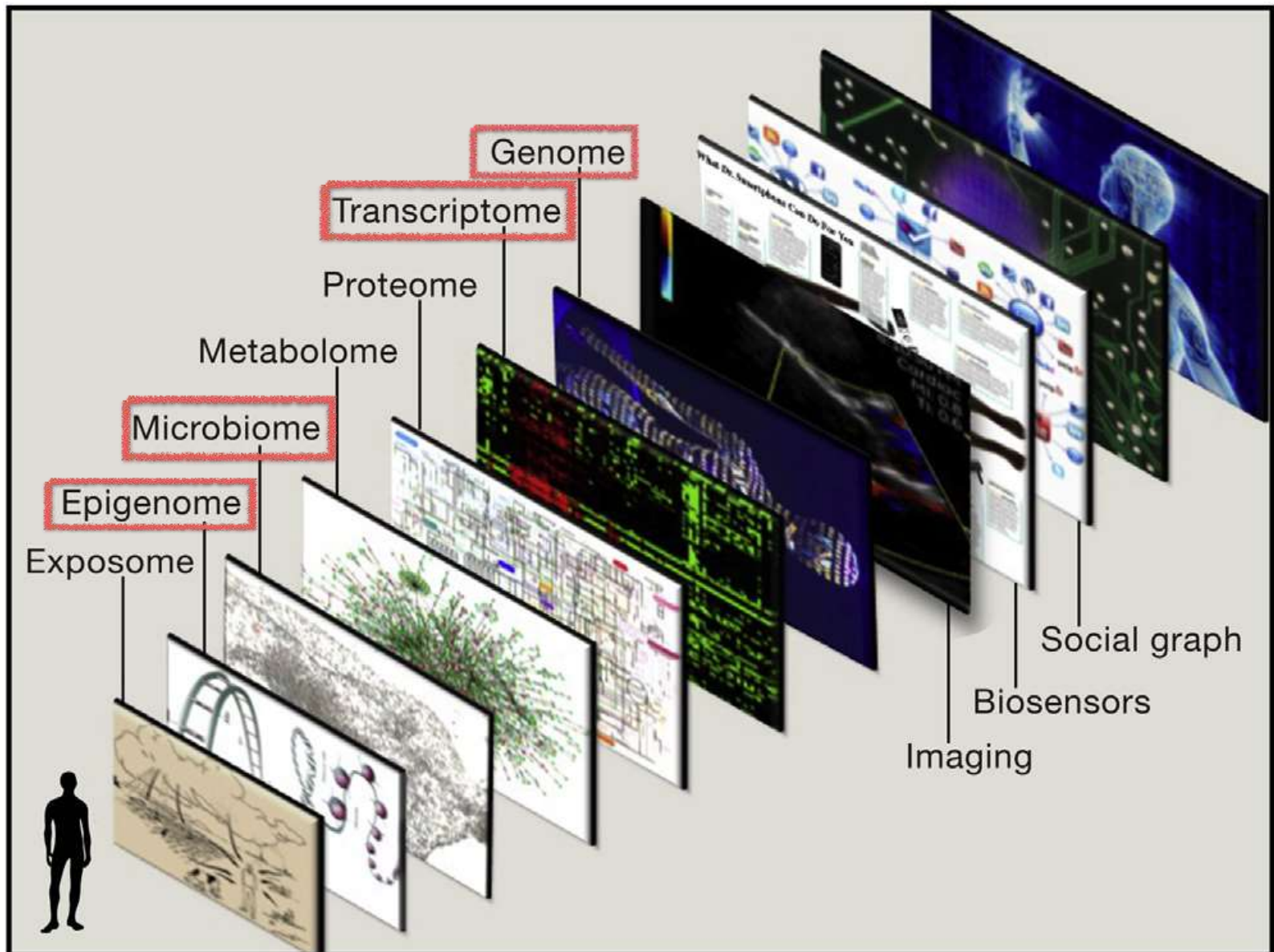
Blood  
Saliva  
Feces  
Hair  
...

**Disease**

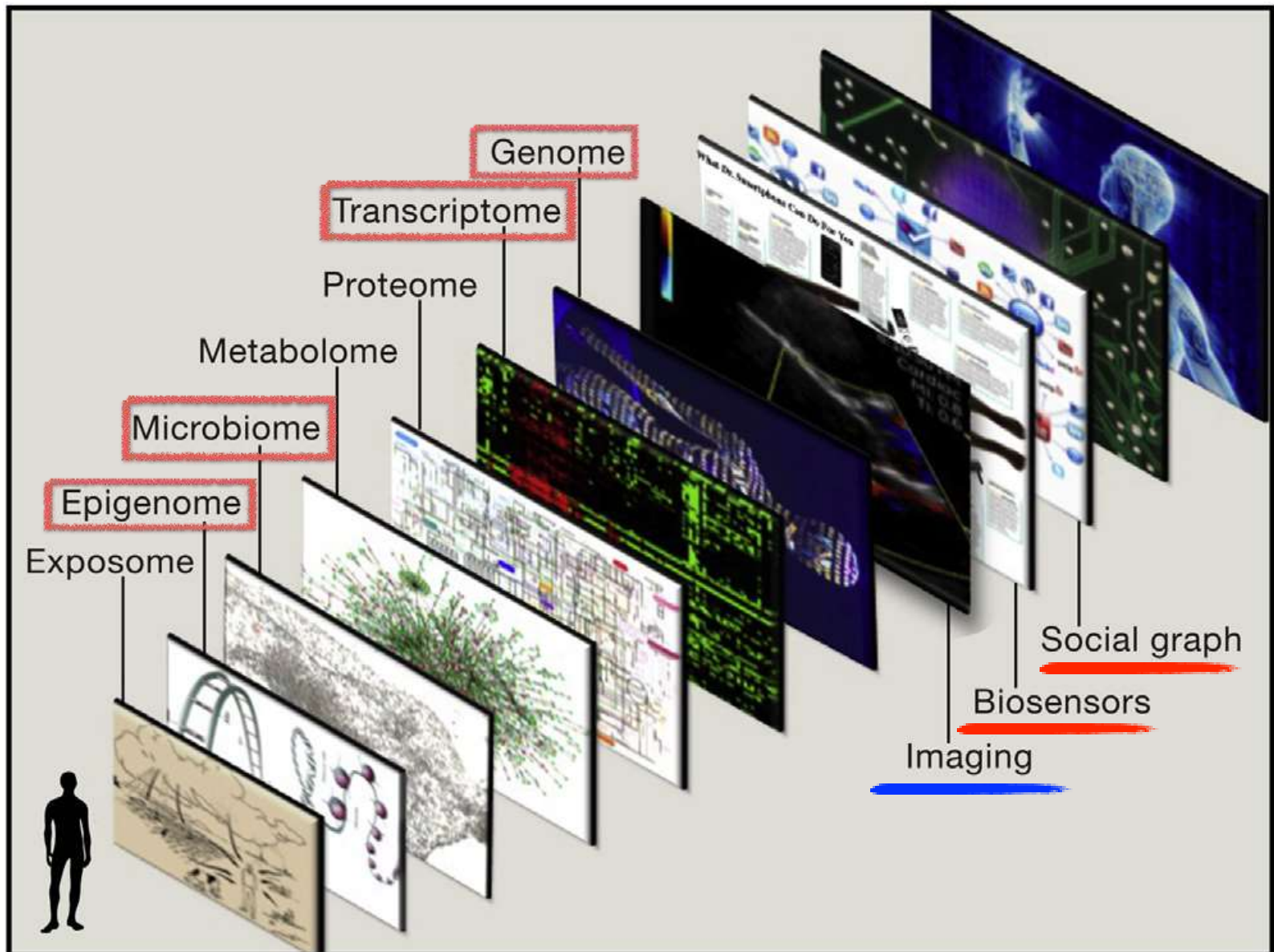
...  
Biopsies  
...

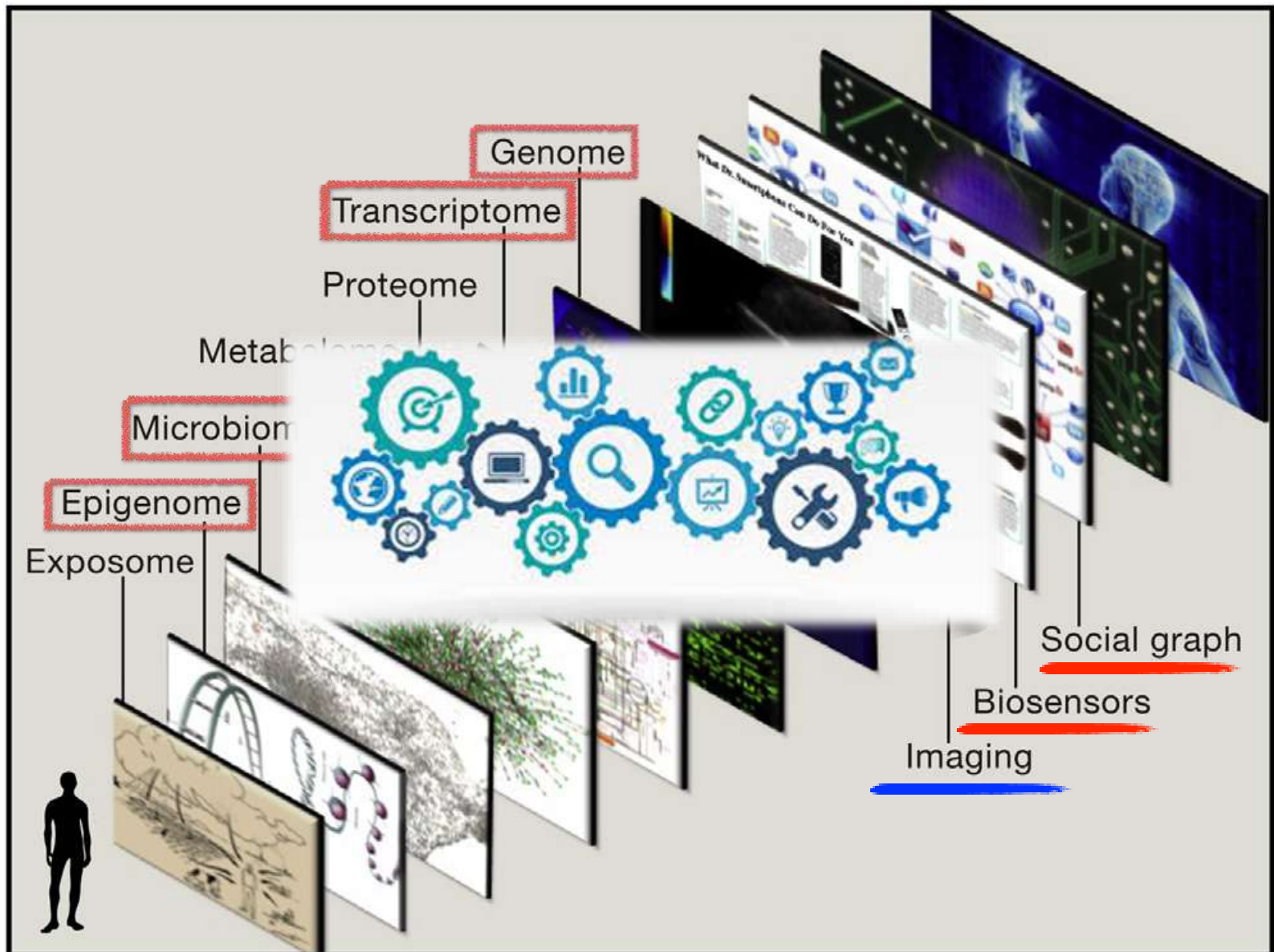


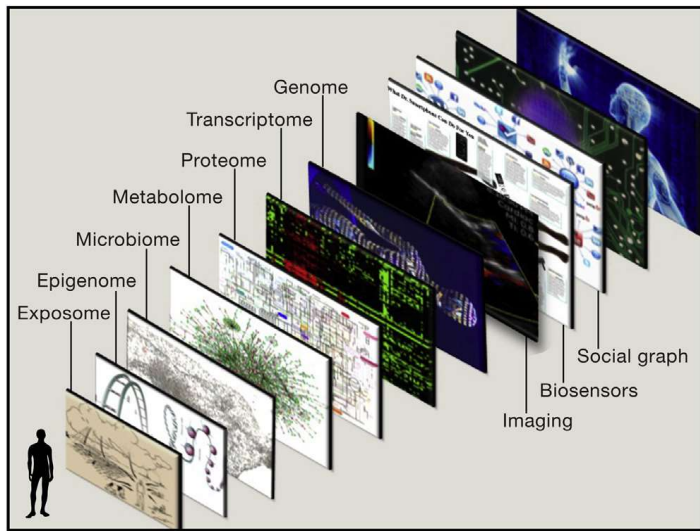








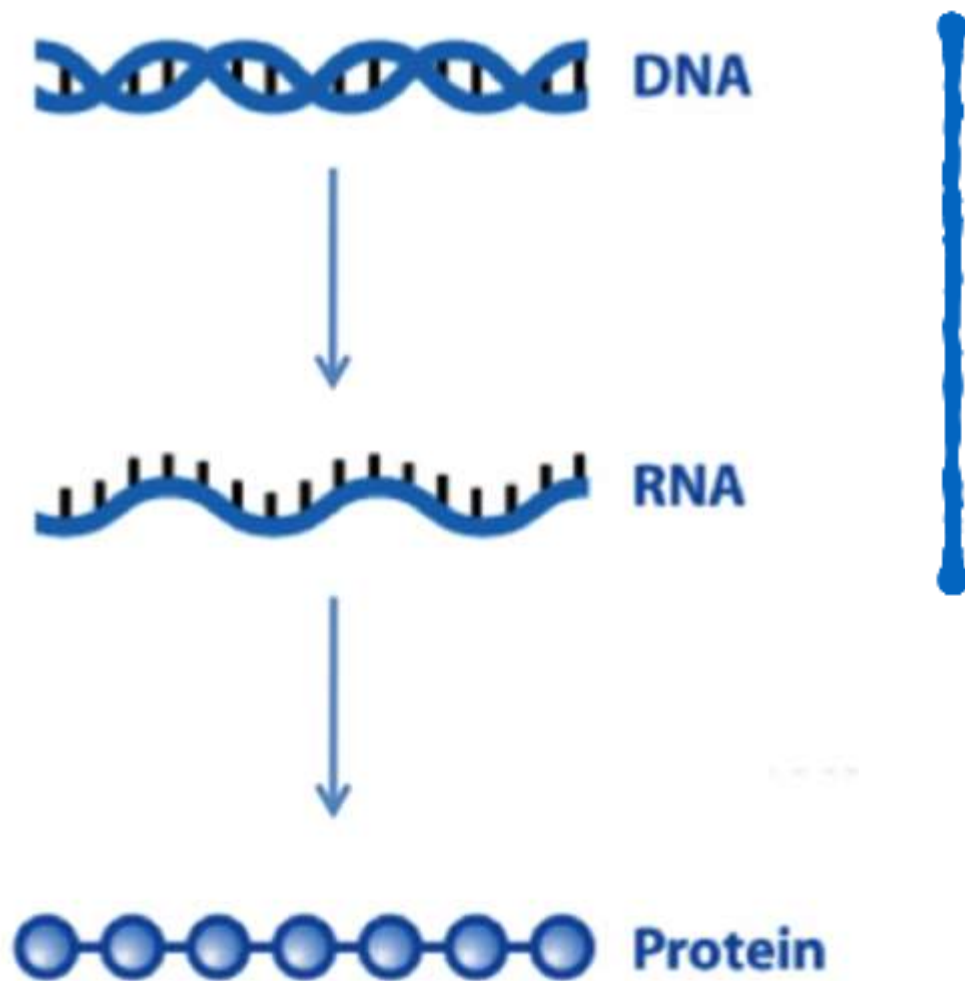








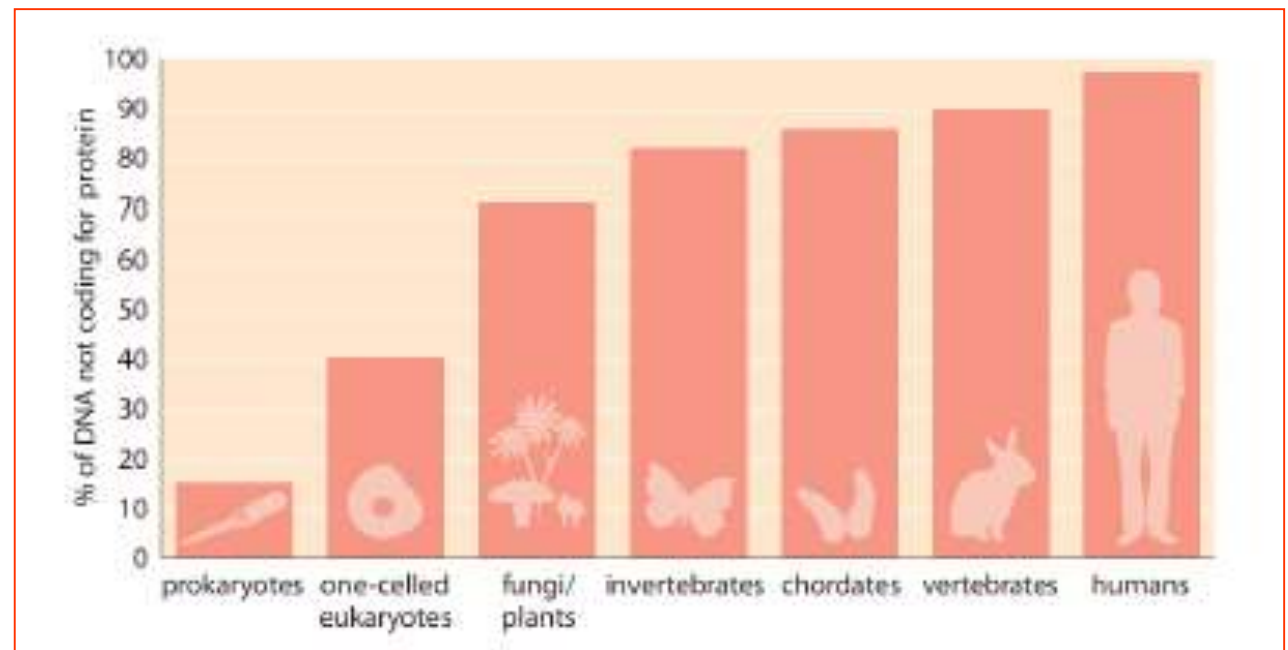




*... and metabolites*

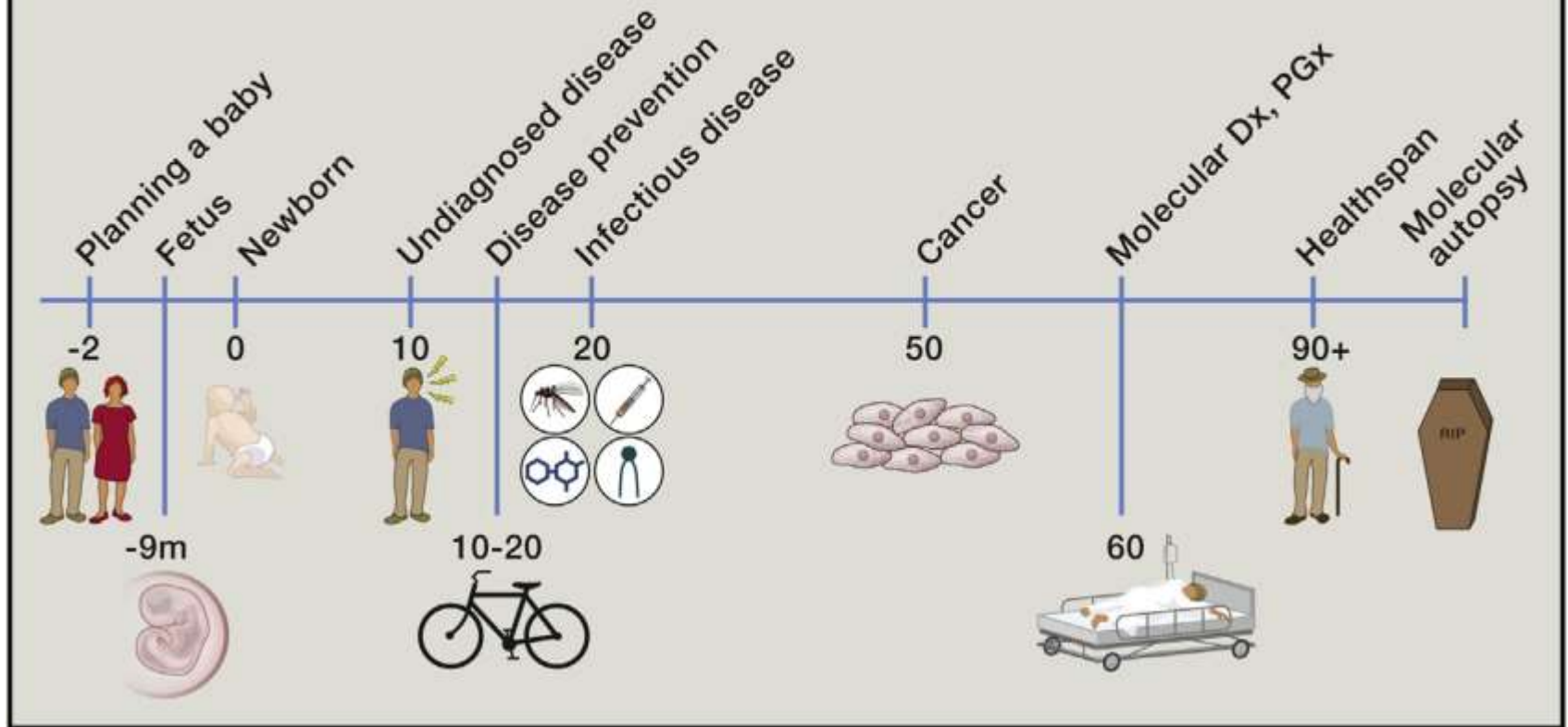
# Quanti geni ci sono nel genoma di...

Specie	Numero di geni
E. coli O157:H7 (batterio)	5.416
Drosophila melanogaster (moscerino della frutta)	~17.000

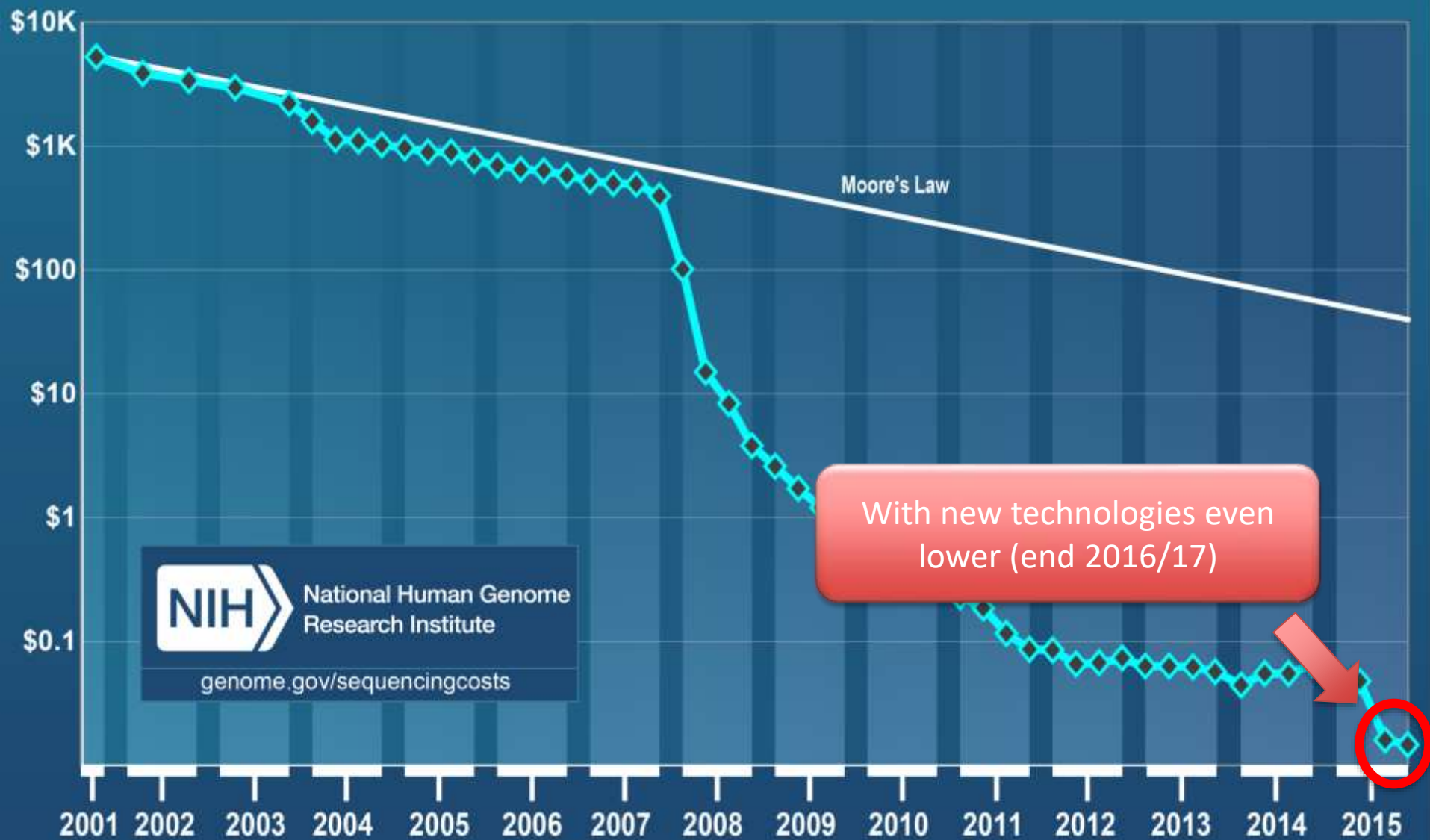


## Individualized genomic medicine

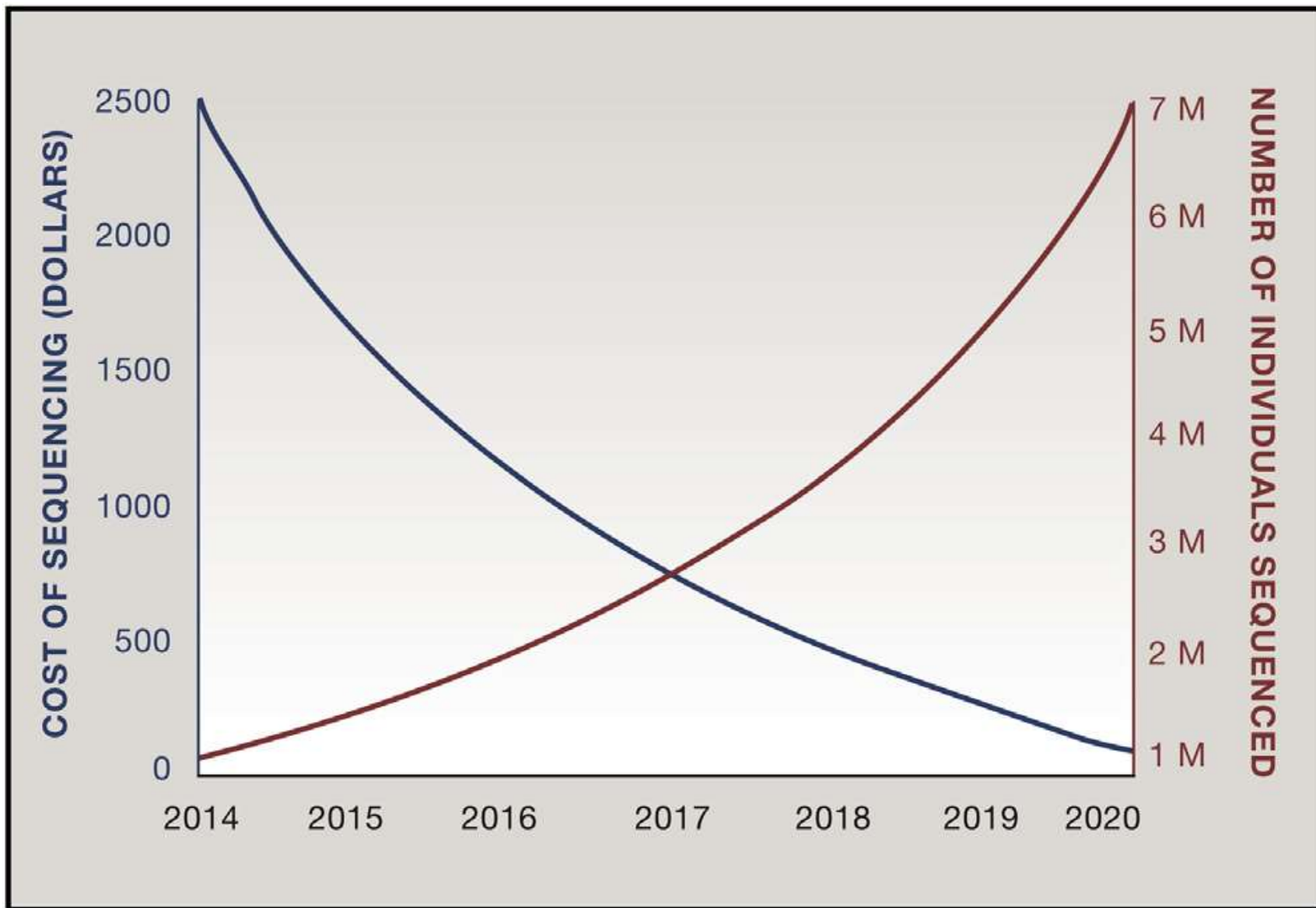
From prewomb to tomb



# Cost per Raw Megabase of DNA Sequence







# Next Generation Sequencing

Enrichment visualization in Genome browsers



Long Reads



Short Reads





BRIEFING ROOM

ISSUES

THE ADMINISTRATION

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## The Precision Medicine Initiative: Data-Driven Treatments as Unique as Your Own Body



Lindsay Holst

January 30, 2015

09:19 AM EDT

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Watch Jo Handelsman, Associate Director for Science in the Office of Science and Technology Policy, explain the Precision Medicine Initiative and its significance.

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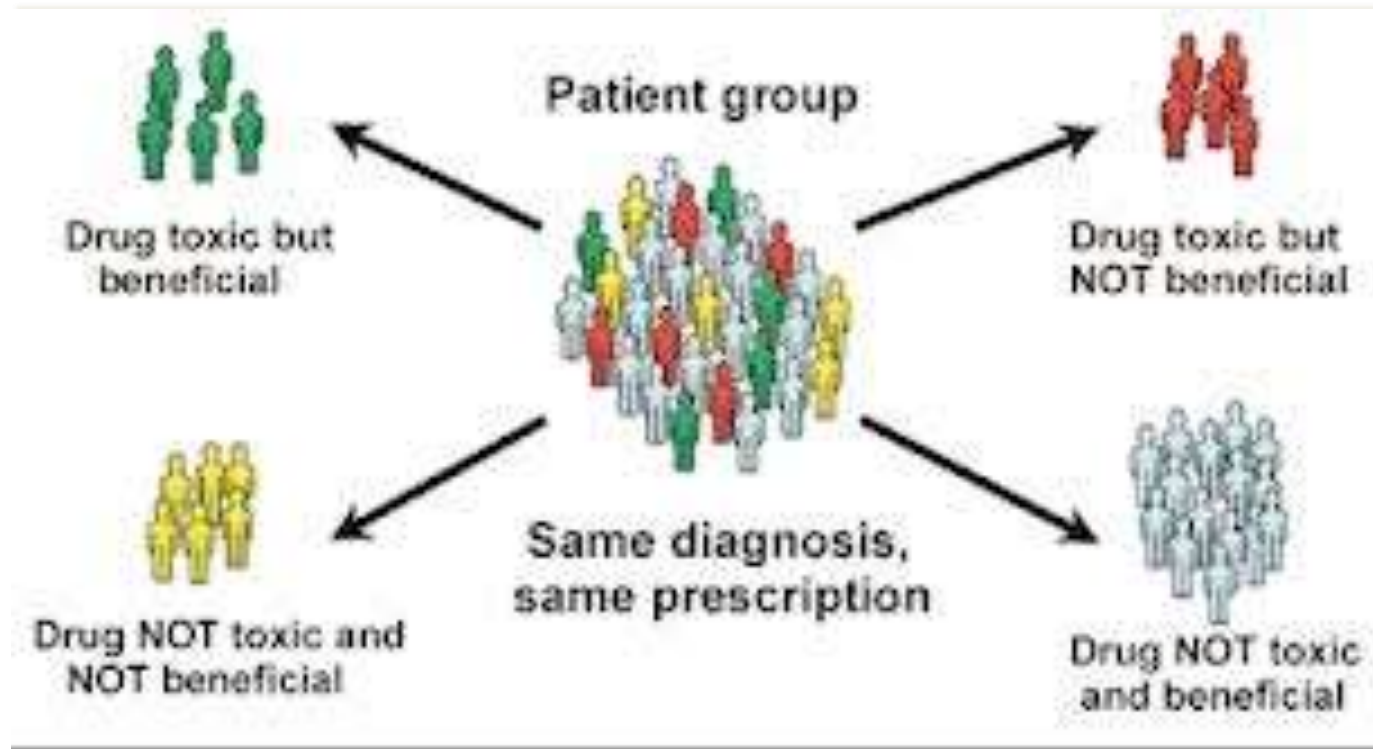
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### PHOTOS OF THE DAY



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# Medicina Personalizzata



“In the future, a family physician might routinely order a whole genome scan, or deploy algorithms that query pharmacogenetic or Mendelian carrier data, or interrogate genomic information.”

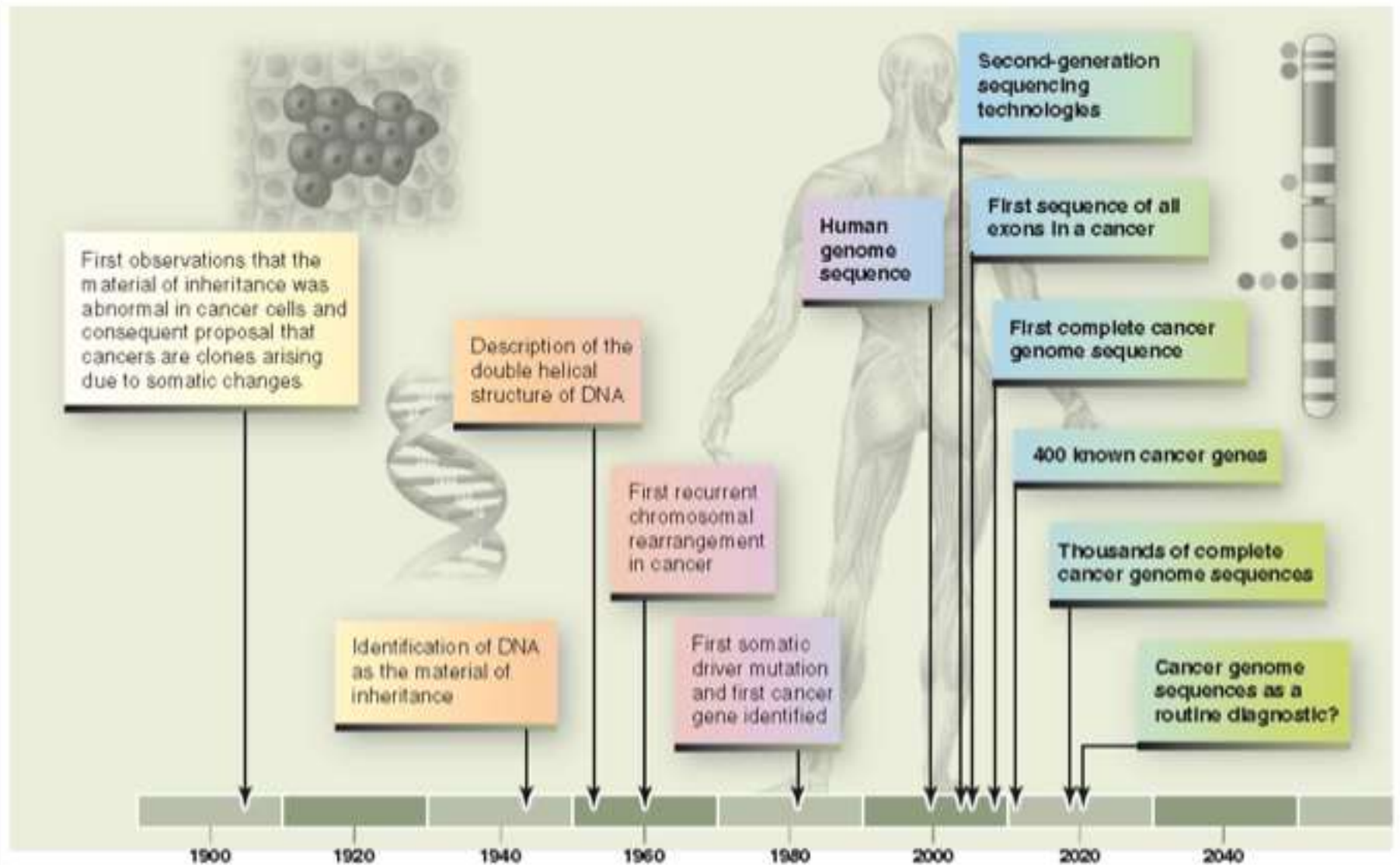


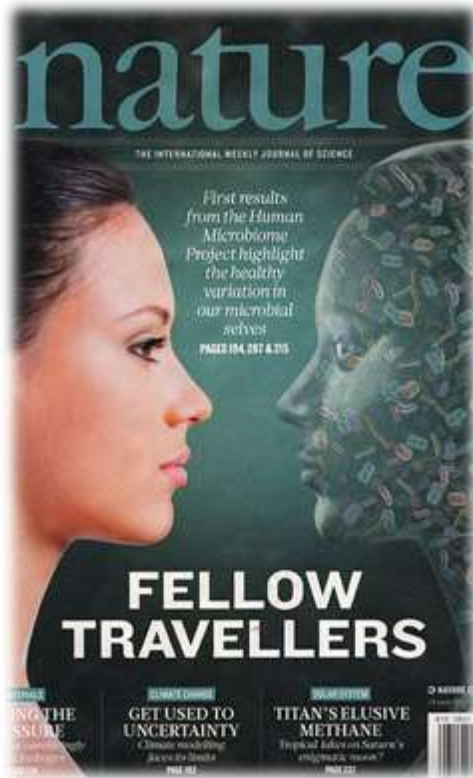






\* Doctor in this chart refers to your regular doctor or a medical geneticist.

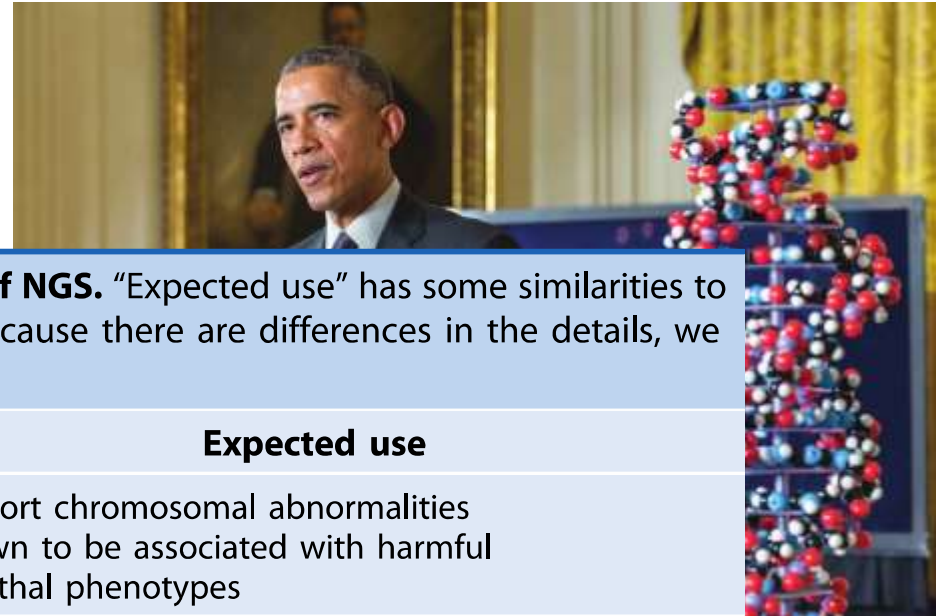
† May be a single meeting or multiple meetings depending upon your questions and concerns.







	Stage in life	Human developmental stage	Microbiome composition	Biologic functions of the microbiome
	Fetus	Developing	Minimal	?
<b>Birth</b>				
	Child	Developing	Developing	Choreography of development
	Adult	Reproduction	Mature	Homeostasis, host fitness
	Elderly person	Aging	Senescent	Neutral or negative fitness



discusses the Precision

**Table 1. Range of currently envisioned uses of NGS.** “Expected use” has some similarities to the FDA regulatory term “intended use,” but because there are differences in the details, we simplify here to “expected use.”

Test	Expected use
Noninvasive fetal testing	To report chromosomal abnormalities known to be associated with harmful or lethal phenotypes
Targeted gene panels to test a patient’s tumor	To report any somatic mutations in known cancer driver genes
Carrier tests for individuals of reproductive age	To report whether an individual carries known or new pathogenic mutations in known heritable disease genes
Whole-genome sequencing tests for children with congenital diseases or conditions	To aid in discovery and mechanistic elucidation of likely causal mutations
Whole-genome sequencing tests for patients with end-stage cancers	To detect neoantigens that can be used for immune-therapy applications
Whole-genome sequencing tests for individuals to calculate risk of common disease	To detect variants associated with risk for a wide range of phenotypes, including rare diseases, common diseases, and drug responses





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Research



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best advocate.

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ETHAN HAWKE

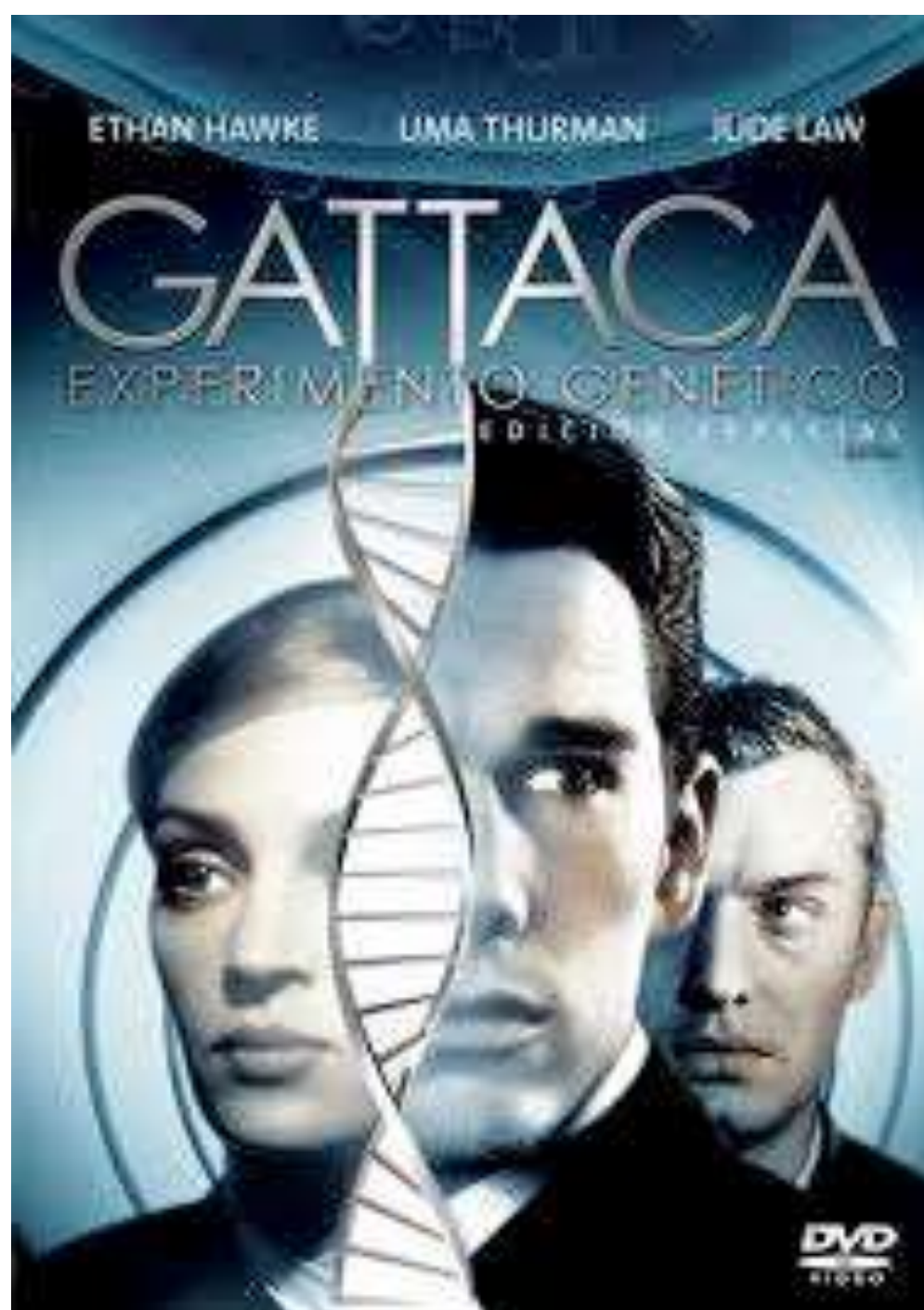
LIMA THURMAN

JUDE LAW

# GATTACA

EXPERIMENTO GENÉTICO

EDITION SPECIALE



DVD  
VIDEO





**First research applications approved**

## News



### UK Biobank welcomes imaging funding

Proposals could see 100,000 participants scanned



### Science Minister visits UK Biobank

Minister hears how the project is building for the future



### UK Biobank opens for research



### Access Procedures

Scientists – find out how to use the resource



### UK Biobank in the news

Launch generates lots of interest in pioneering study



### Long-term follow-up of health

How far have we got?

## Scientists



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[UK Biobank opens for](#) 29/06/2012

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## Latest Quotes

*"We're testing our physical activity monitor - so if you receive an invite asking if you'd wear one, please do try it out."*

Sir Rory Collins, UK Biobank Principal Investigator





The UK Biobank stores millions of samples from UK citizens.

- Sangue, saliva, urina
- Giornata tipica:
  - > 700-1000 volontari
  - > 4-5 tecnici, 12 robot
- In 10 anni...
- Alzheimer's (9.000 individui)
- Tumore alla mammella (10.000)
- Malattia cardiovascolare (28.000)



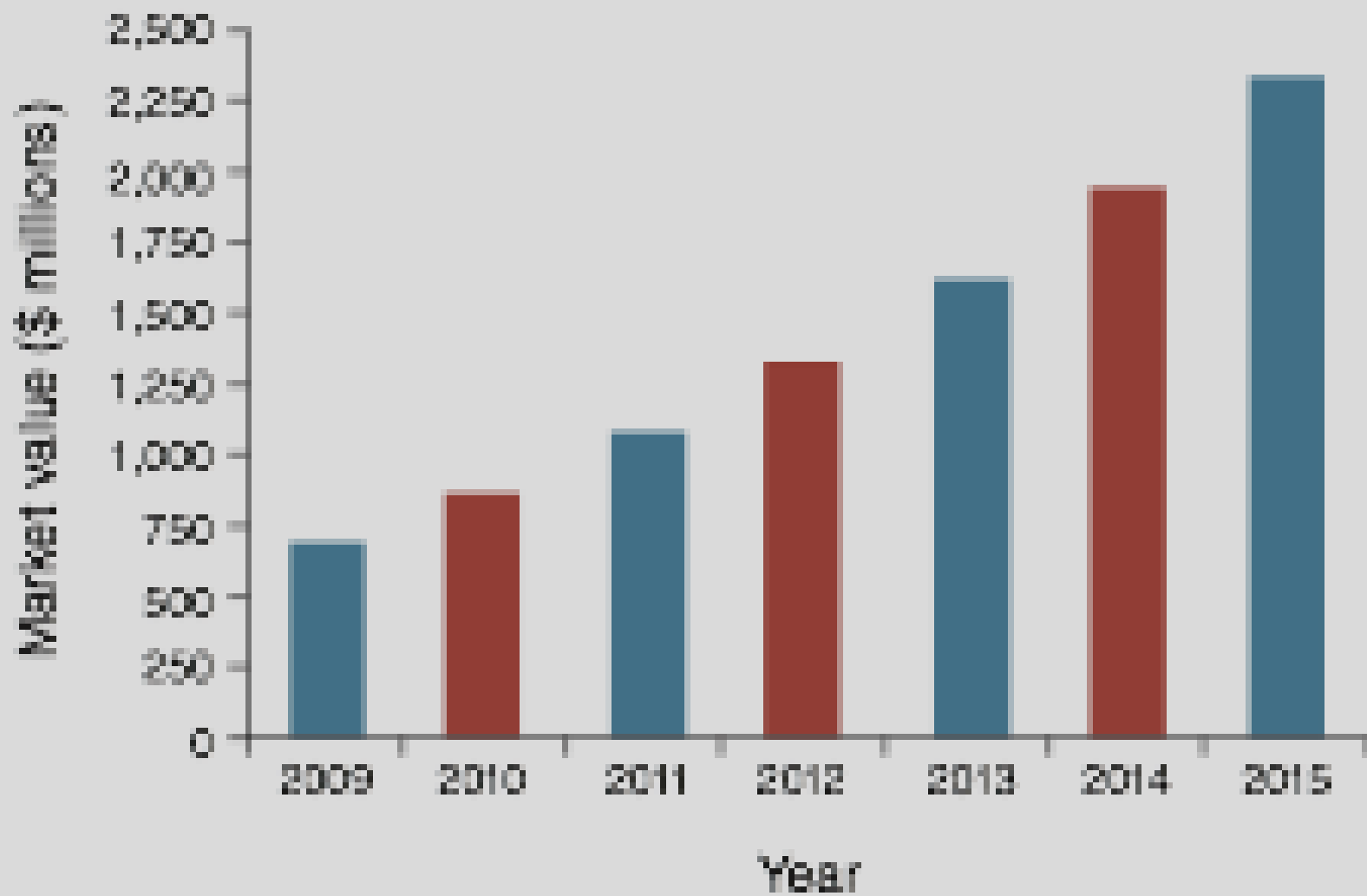
# Qualche dato...

- > 300.000 milioni campioni tissutali mantenuti in banche tissutali USA (circa 1999)
- Aumentano ad una frequenza di più di 20 milioni all'anno



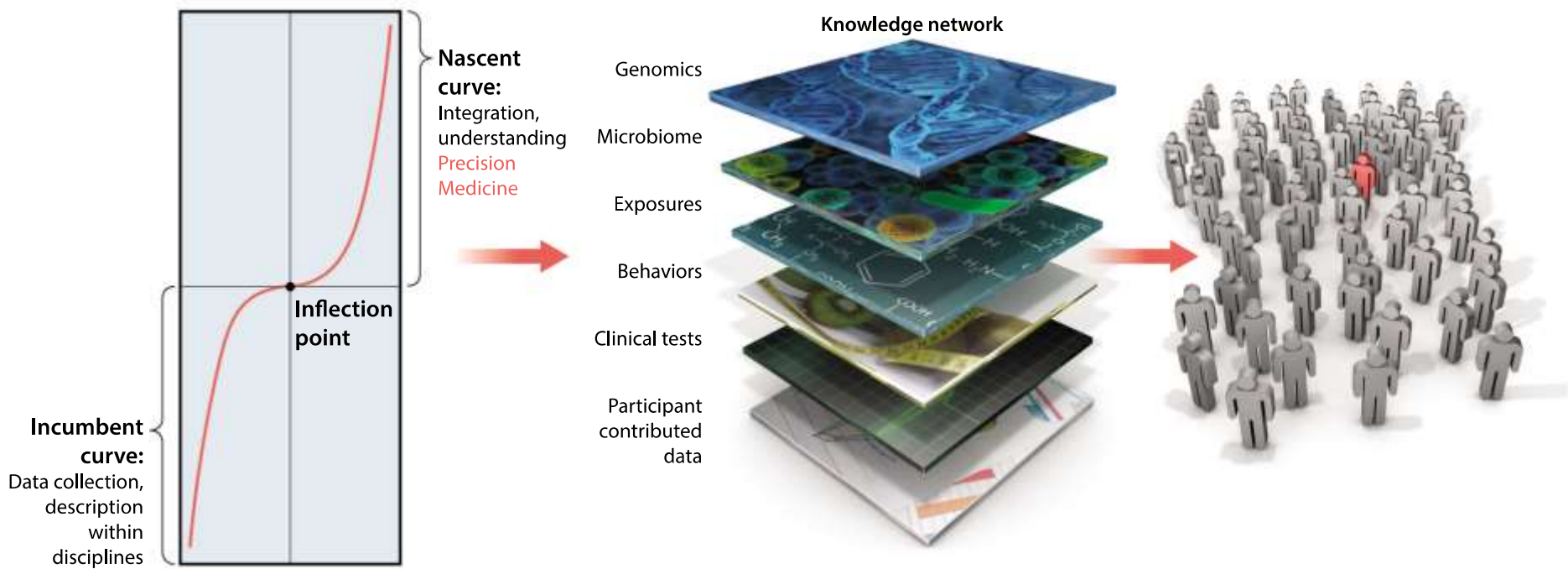
# Caratteristiche...

- Campioni (DNA, RNA, cellule, tessuti, tumori, sangue ...)
- Condizioni (specifiche patologie, tumori...)
- Anatomia (occhi, ossa, cartilagine...)
- “dati” (analisi molecolari, microarrays...)



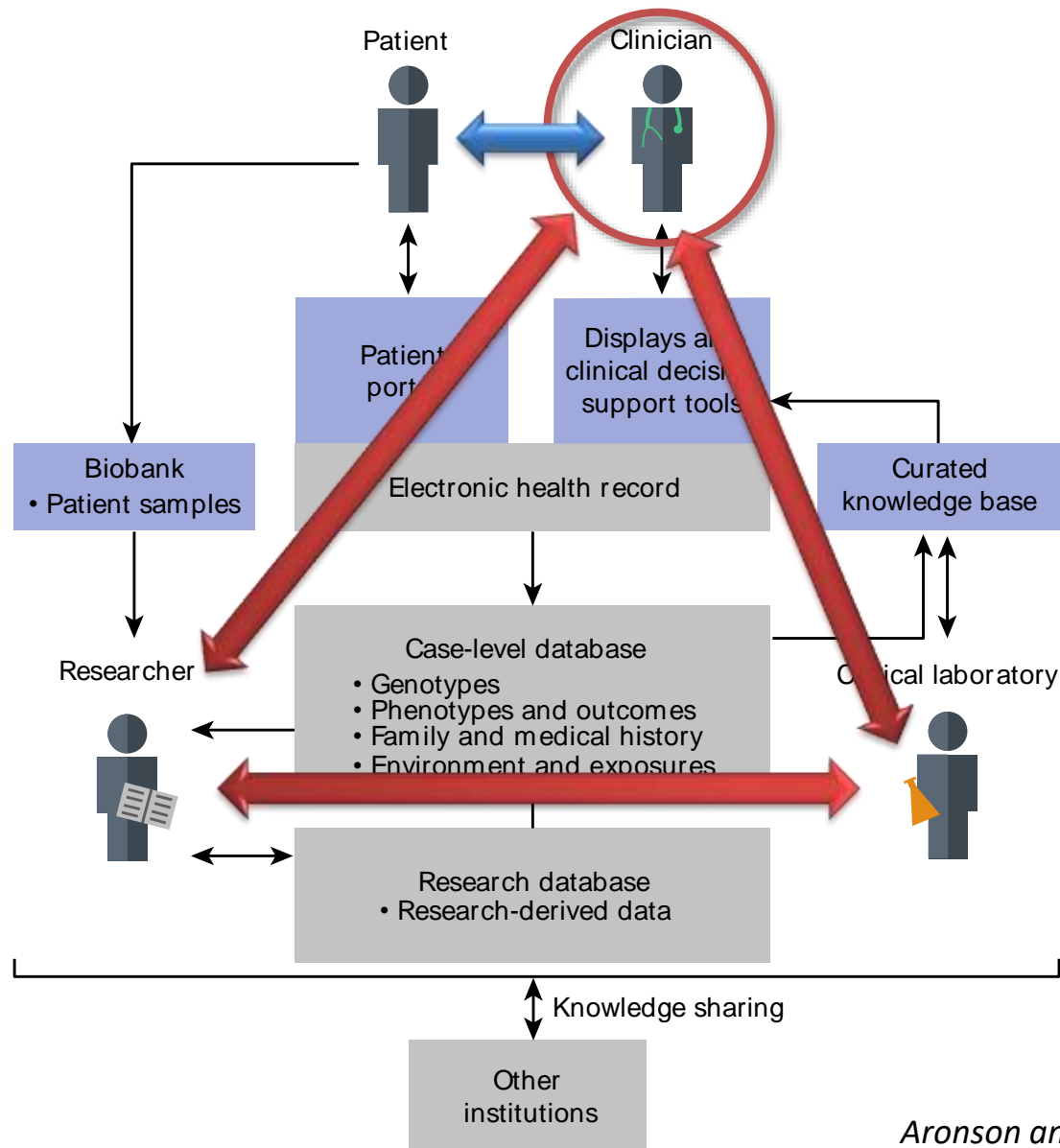


# Precision medicine: Beyond the inflection point

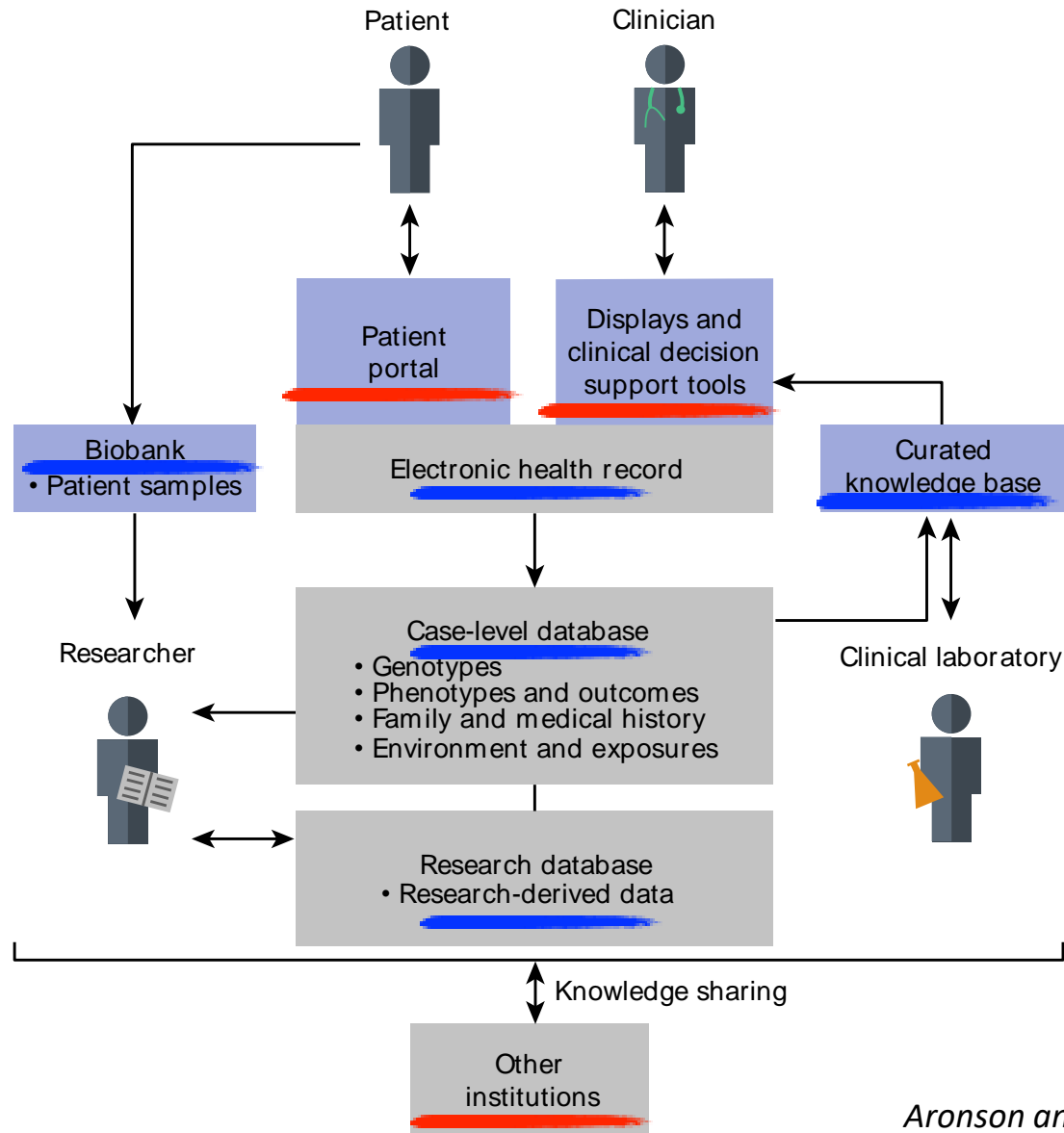




# The precision-medicine ecosystem a new role for the clinician



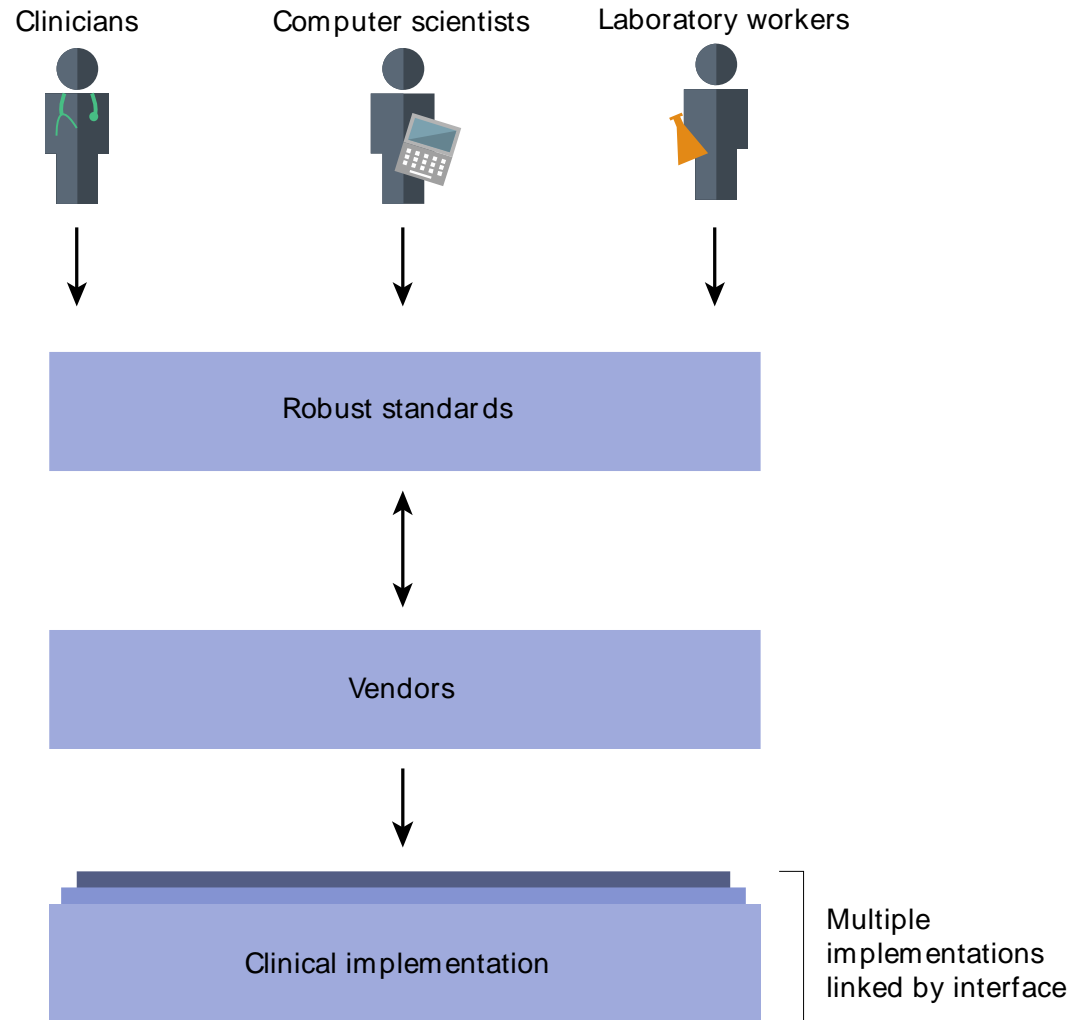
# The precision-medicine ecosystem a new role for ITC





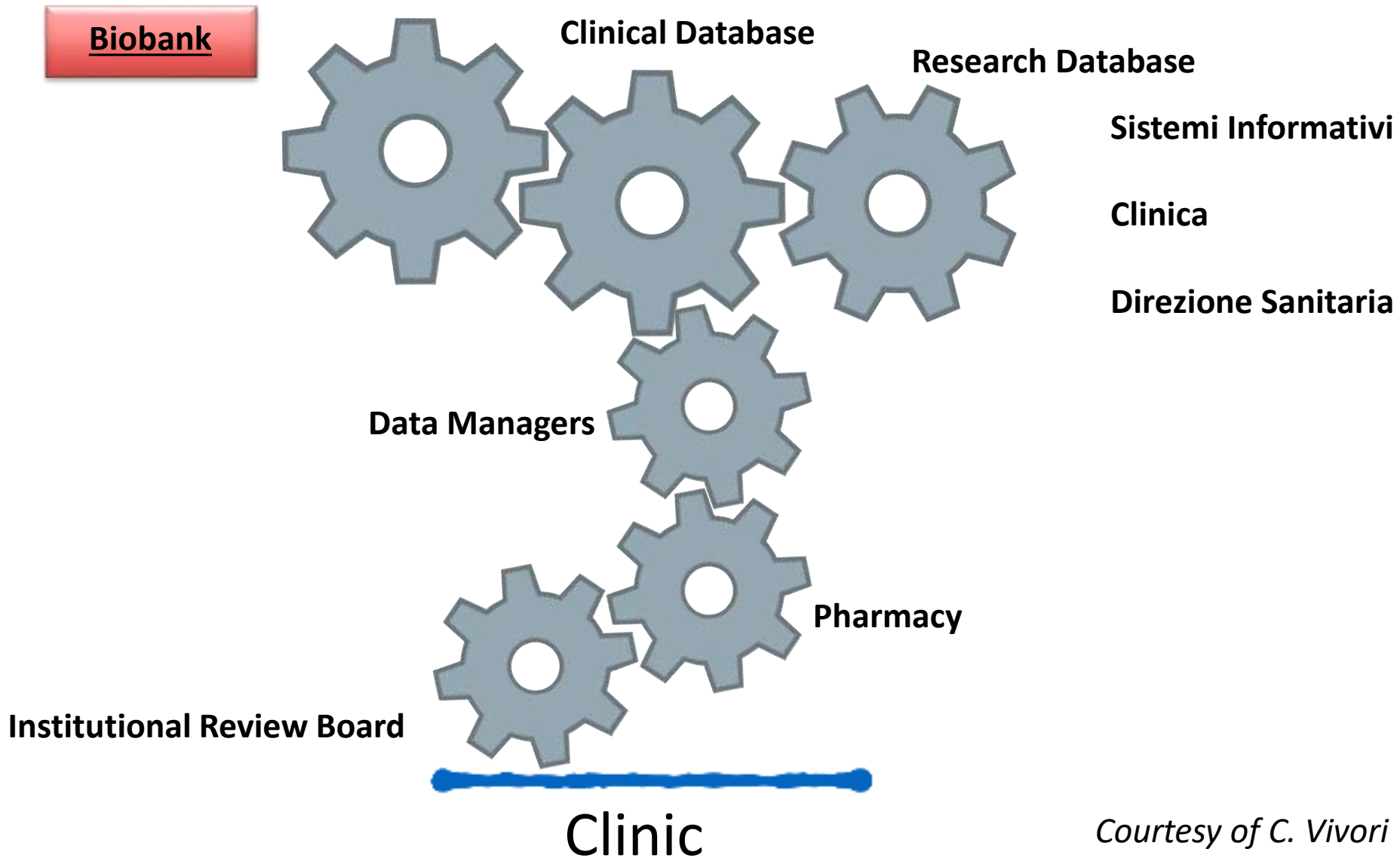
# The precision-medicine ecosystem

## a new role for ITC

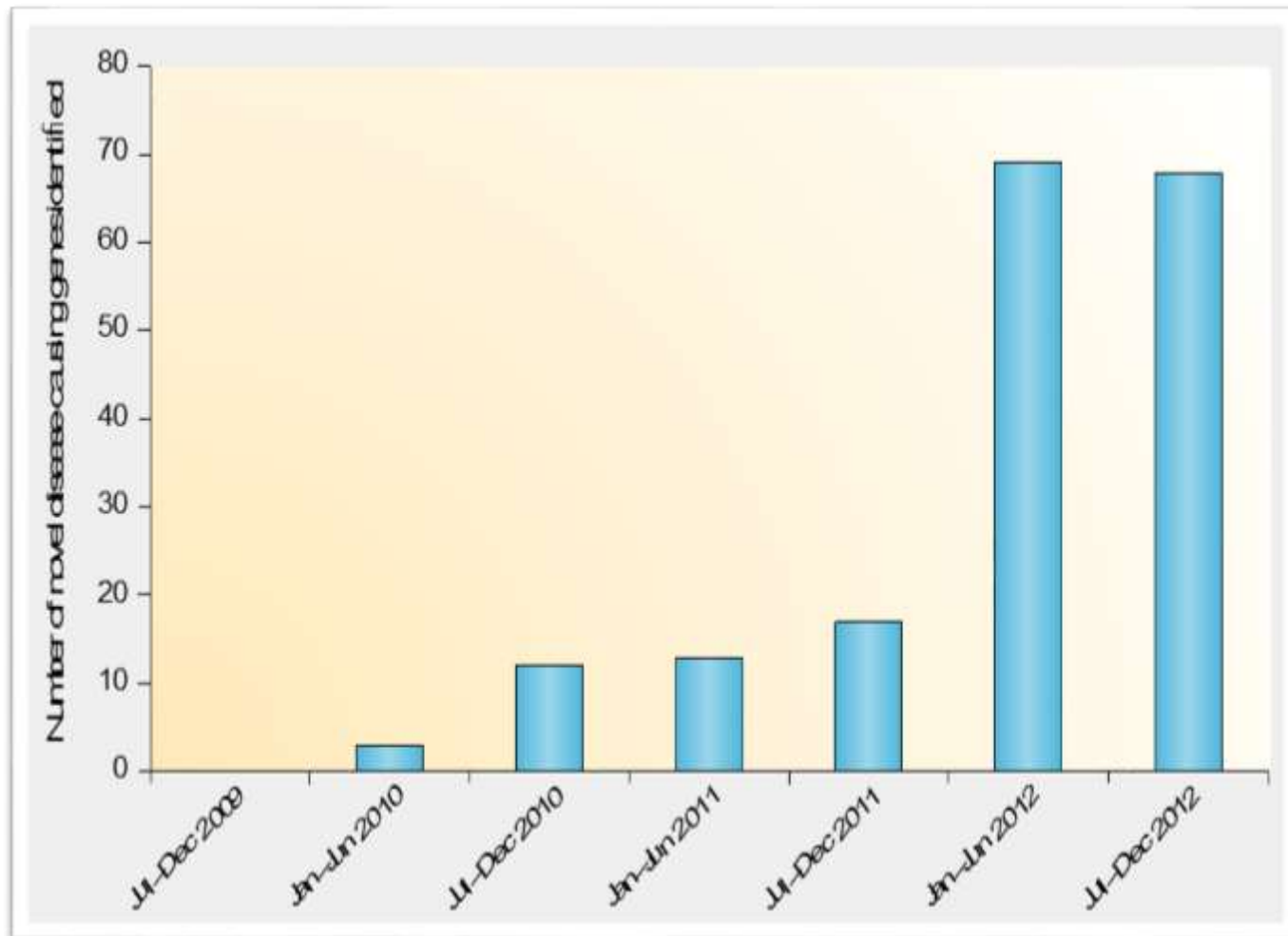




# Infrastructure



*Courtesy of C. Vivori*



## SOUNDING BOARD

**Precision Medicine — Personalized, Problematic, and Promising**

J. Larry Jameson, M.D., Ph.D., and Dan L. Longo, M.D.

Discovery

Assay Design

Diagnostic Tool

Clinic

Heterogeneous disease

New diagnostic test

Refined disease classification,  
according to prognostic  
implications

Subtype A

Subtype B

Subtype C

New targeted treatment

Clinical research  
Outcomes  
Safety  
Cost

Clinical guidelines

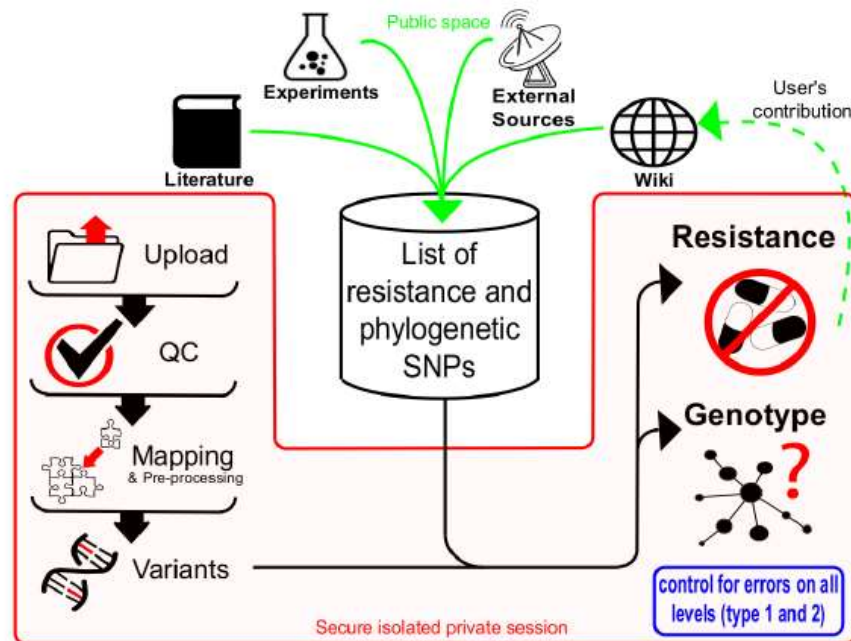
Clinical implementation

Adoption by  
physicians and  
health systemsAdoption by  
payersAdoption by  
patients





# Genomics of Infectious Diseases

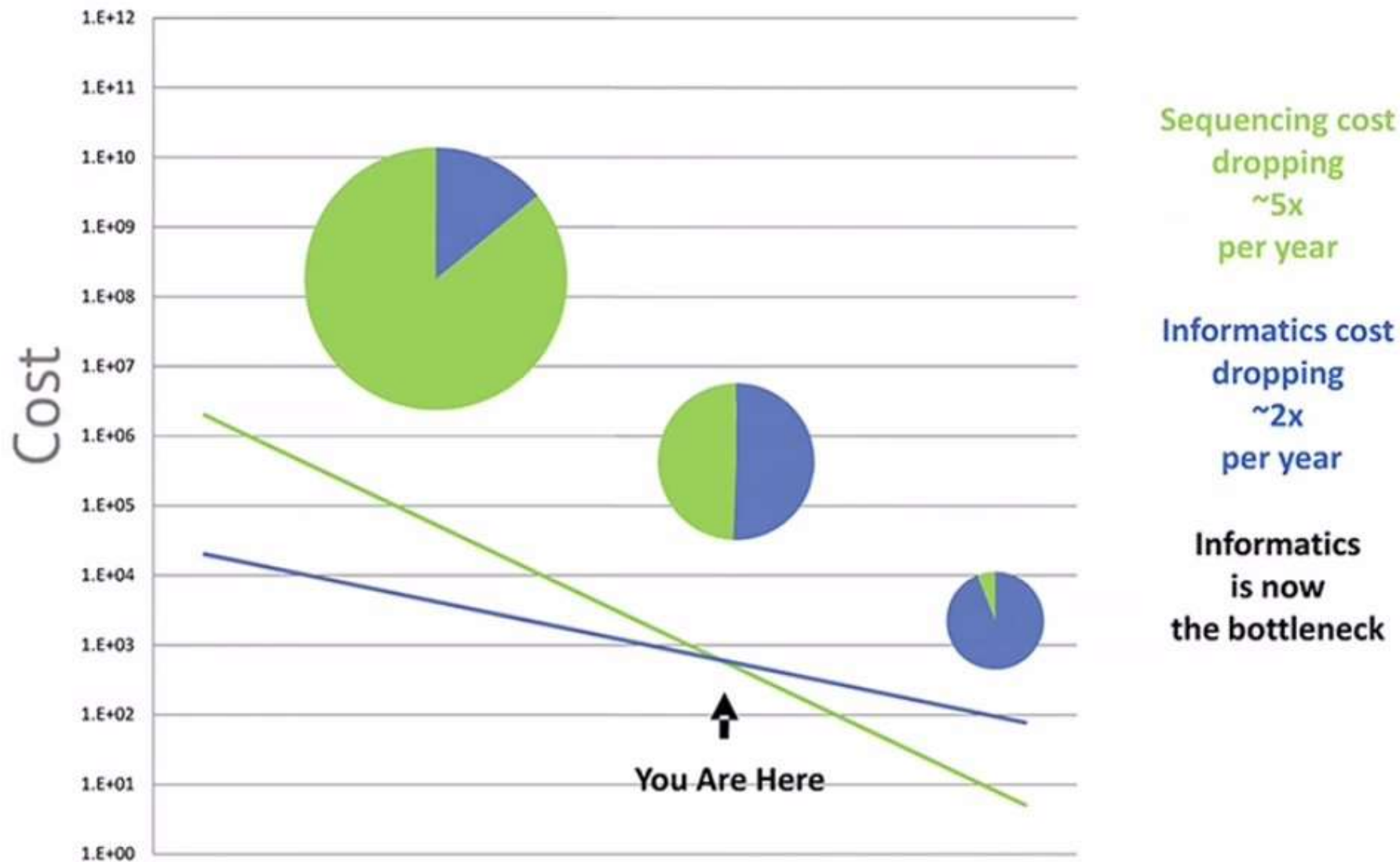


## EMERGING BACTERIAL PATHOGENS UNIT

TB Supranational Reference Laboratory  
WHO Collaborating Centre in TB Laboratory Strengthening (ITA-98)

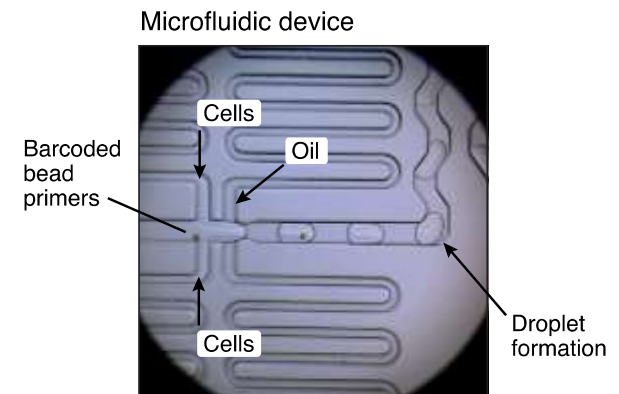
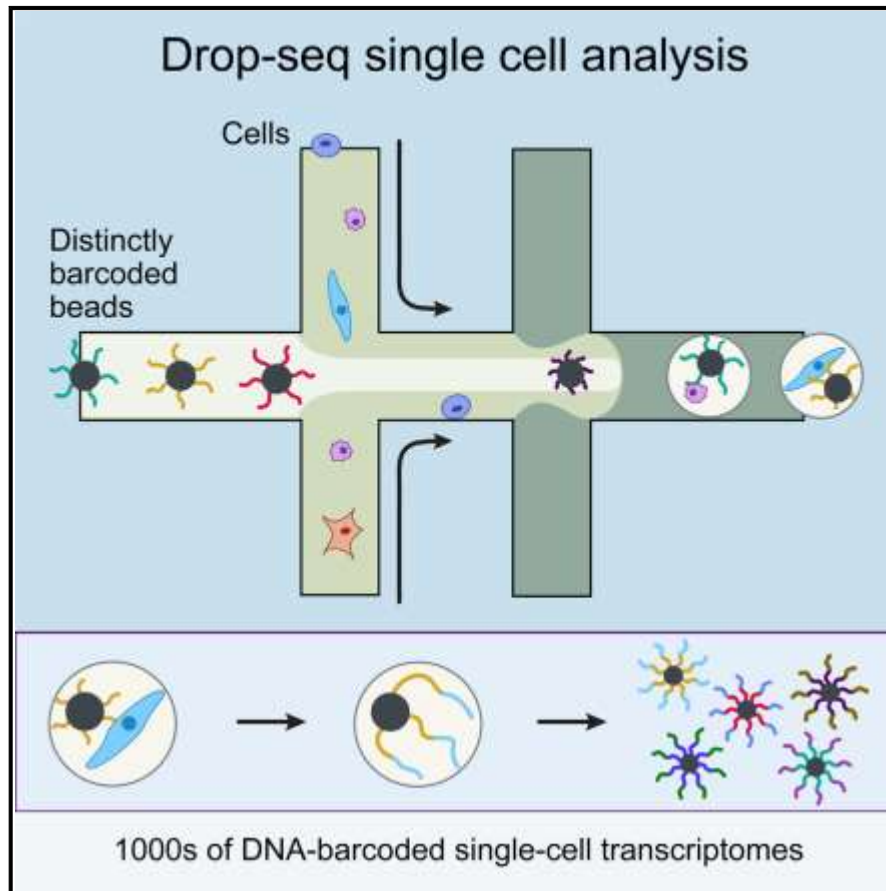


# DNA Sequencing Economics



# Highly Parallel Genome-wide Expression Profiling of Individual Cells Using Nanoliter Droplets

## Single-cell genomics



Our soul is cast into a body, where it finds number, time,  
dimension. Thereupon it reasons, and calls this nature necessity,  
and can believe nothing else

Blaise Pascal