



Qué grandes eventos están
moldeando el futuro de la
medicina de precisión?

IMPRECISION MEDICINE

For every person they do help (blue), the ten highest-grossing drugs in the United States fail to improve the conditions of between 3 and 24 people (red).

1. ABILIFY (aripiprazole)
Schizophrenia



2. NEXIUM (esomeprazole)
Heartburn



3. HUMIRA (adalimumab)
Arthritis



4. CRESTOR (rosuvastatin)
High cholesterol



5. CYMBALTA (duloxetine)
Depression



6. ADVAIR DISKUS (fluticasone propionate)
Asthma



7. ENBREL (etanercept)
Psoriasis



8. REMICADE (infliximab)
Crohn's disease



9. COPAXONE (glatiramer acetate)
Multiple sclerosis



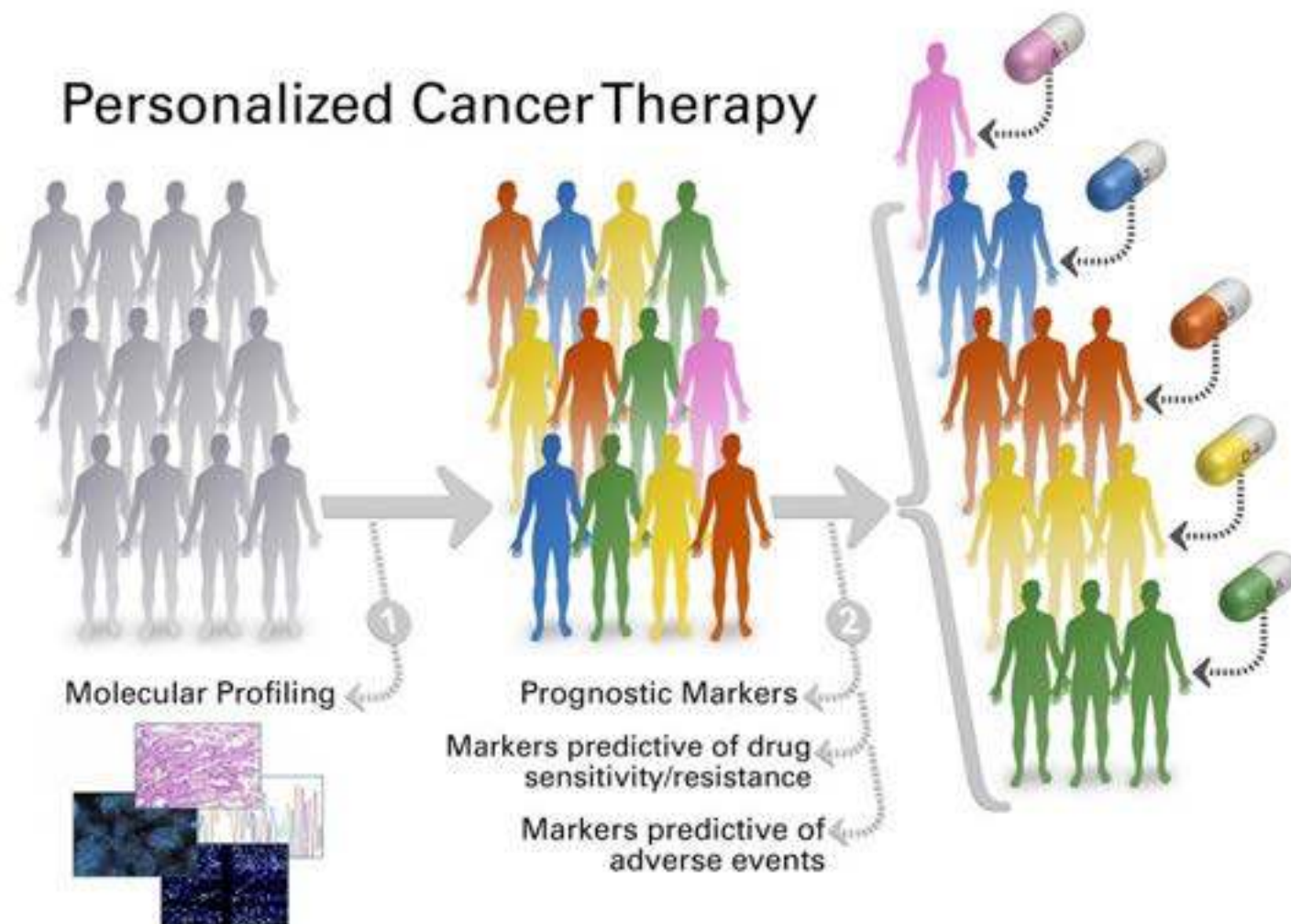
10. NEULASTA (pegfilgrastim)
Neutropenia



Based on published number needed to treat (NNT) figures. For a full list of references, see Supplementary Information at [gs.nature.com/4278](https://doi.org/10.1038/nature04278).

Precision Medicine

Personalized Cancer Therapy





Next Generation Sequencing

DNA sequencing
is the technology that experienced
the *most dramatic advances*
in the human history

2008 ————— the last ten years ————— 2018

50,000 b

day
1 equipment

0.000016

human genomes
day

100,000,000 USD



18,000,000,000,000 b

day
1 equipment

600

human genomes
day

1,000 USD



illumina®

Projected annual storage in 2025

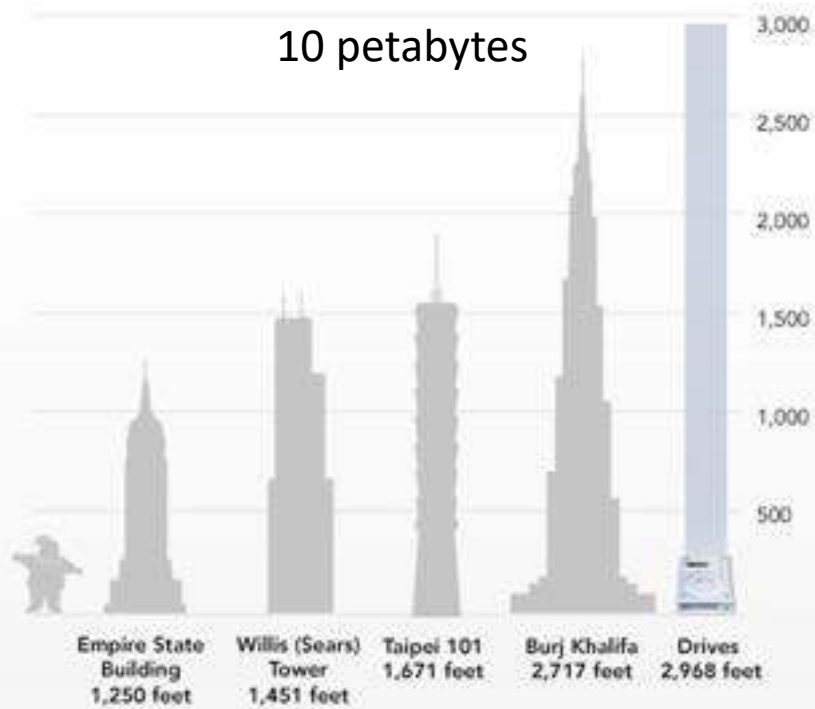
Twitter: 1–17 petabytes per year

Astronomy:
1,000 PB/year

YouTube:
1,000–2,000 PB/year

Genomics:
2,000–40,000
PB/year

BACKBLAZE DRIVES STACKED



* 6,195 drives x 5.75 inches of drive height = 35,621 inches or 2,968 feet

*Biological interpretation
(social, legal issues)
Lagging Behind*



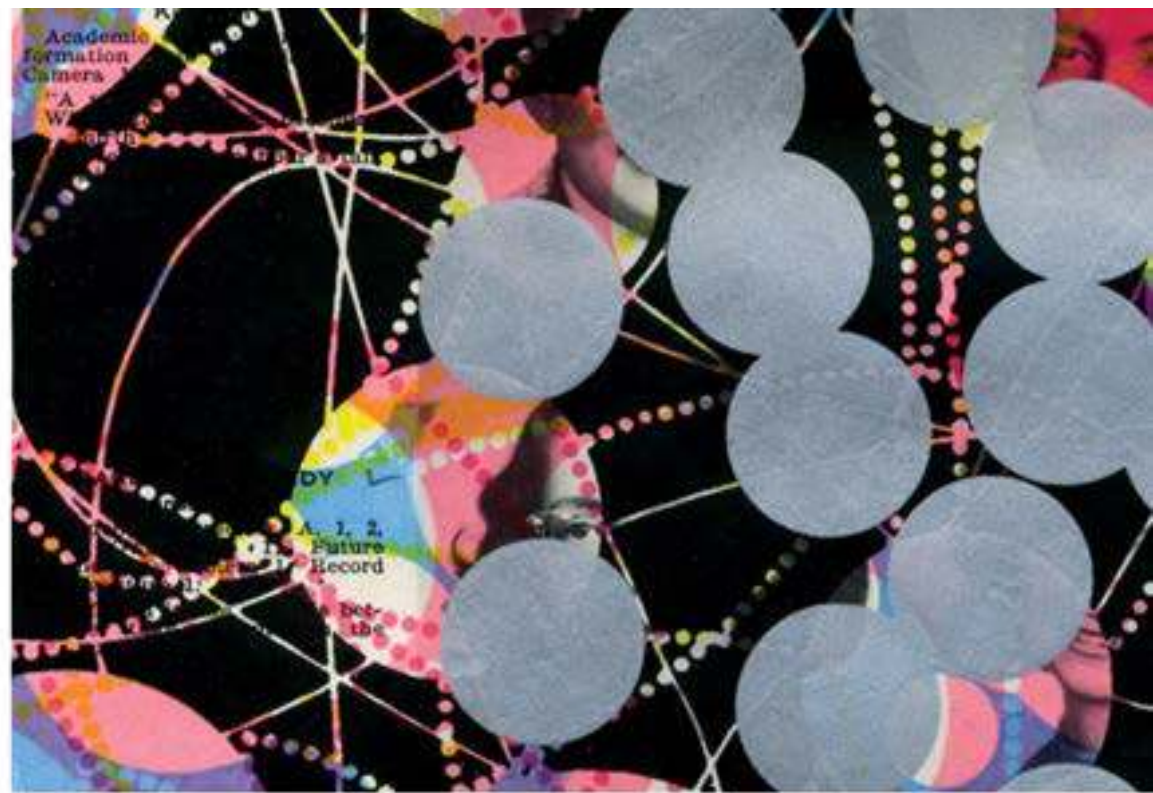
*Technology
at the forefront*





Harvard
Business
Review

El valor de los datos



DATA

Data Scientist: The Sexiest Job of the 21st Century

by Thomas H. Davenport and D.J. Patil

FROM THE OCTOBER 2012 ISSUE

So you wanna be a data scientist? A guide to 2015's hottest profession

Science

- 1.Math
- 2.Machine learning

Data

- 1.Statistics
- 2.R programming

Art

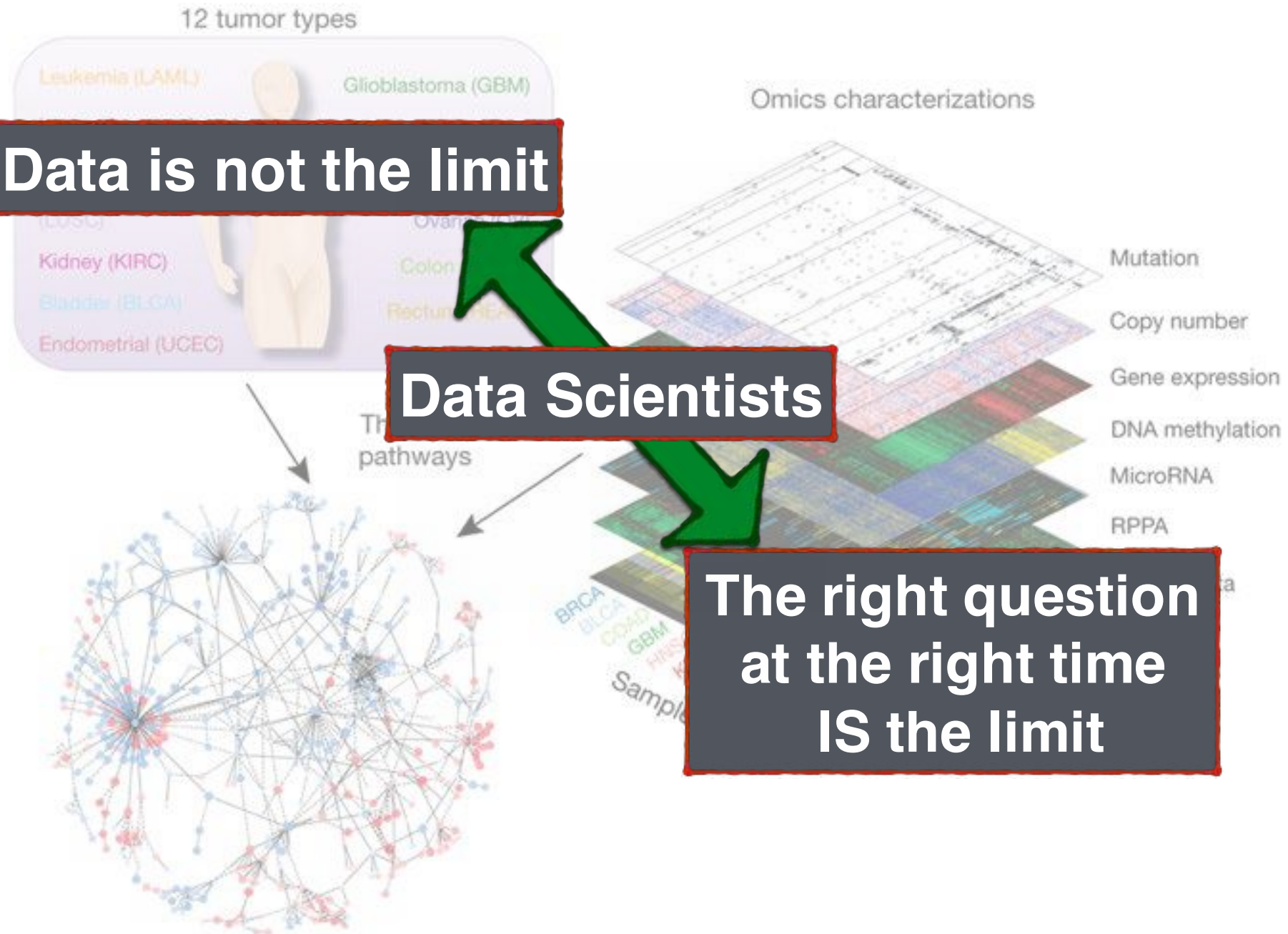
- 1.Visualisation
- 2.Creativity
(find signal in noise)

Big Data in genomics

Data is not the limit

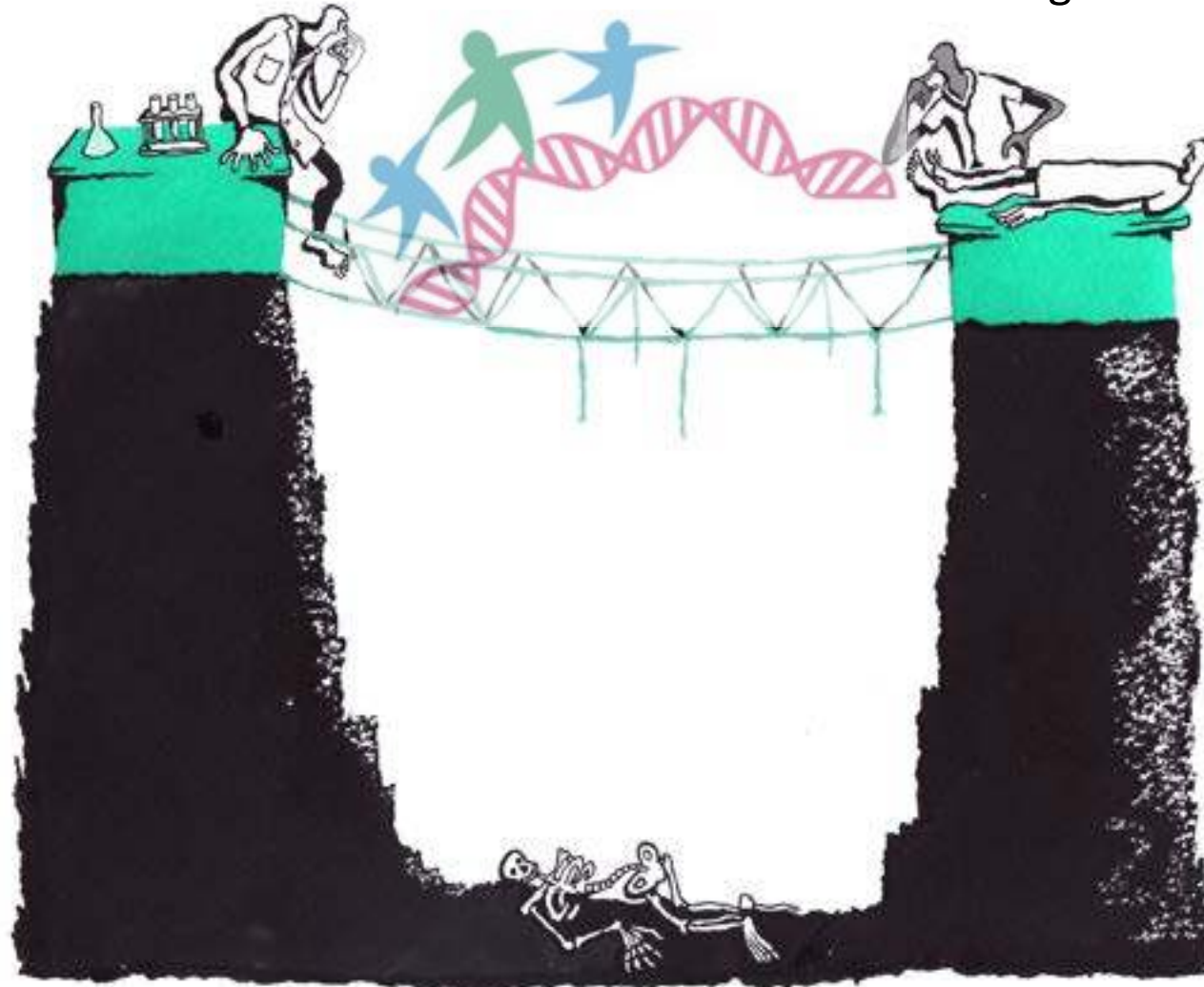
Data Scientists

**The right question
at the right time
IS the limit**



Research

Clinical Diagnostics/Therapies



La traducción al paciente

CAREERS

TRANSITIONS From building houses to building molecules **p.153**

FUTURE PLANS Three steps to prepare for the next five years go.nature.com/bpdlrc

NATUREJOBS For the latest career listings and advice www.naturejobs.com

DRA_SCHWARTZ/GETTY



GENETICS

Fluent in DNA

As genomics migrates to the clinic, job options are emerging for genetic counsellors to explain the meaning in mutations.

Genetic Counsellor
Is the next big thing
in hot professions

04

Inteligencia Artificial (Aumentada)

DEEP MEDICINE

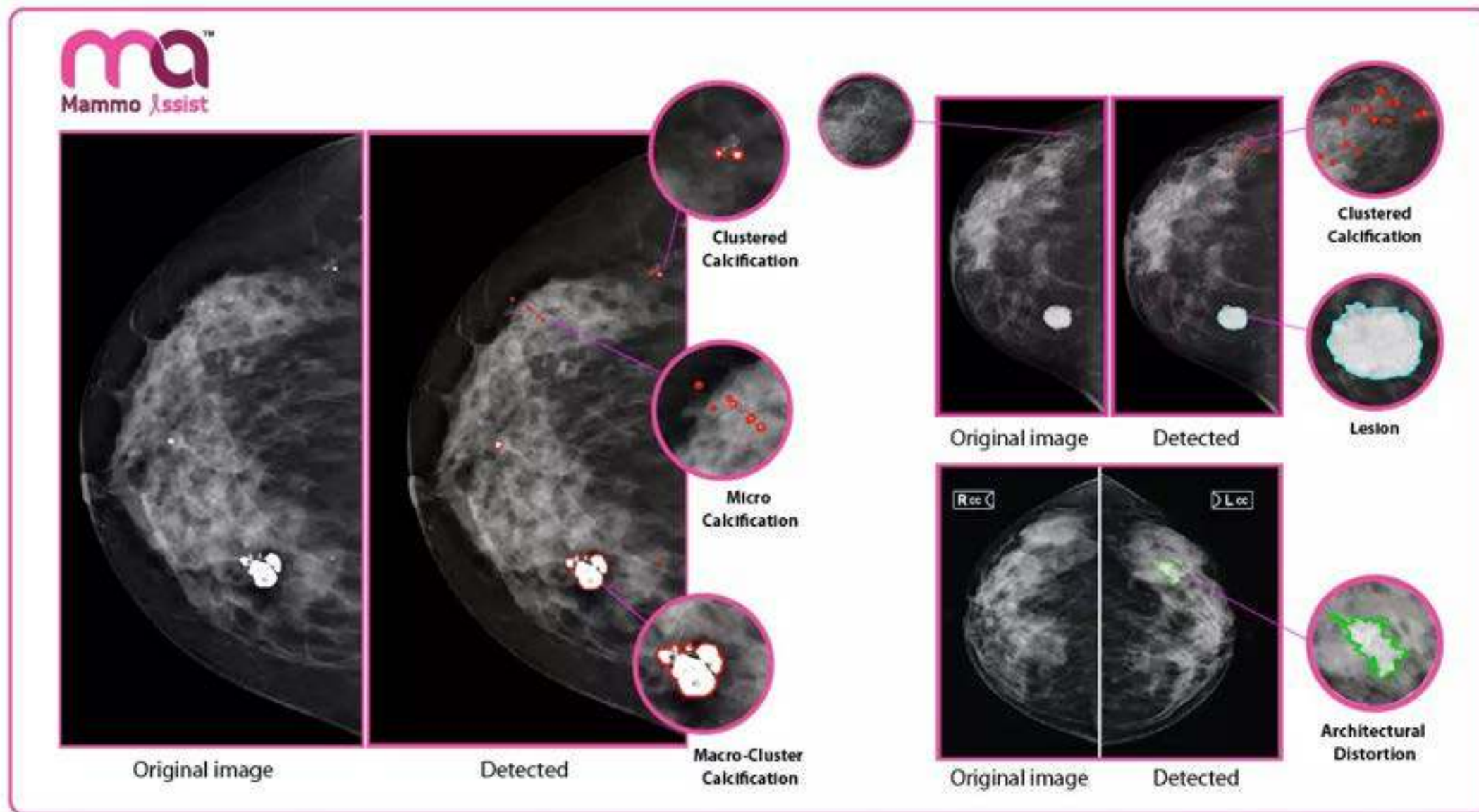
HOW ARTIFICIAL
INTELLIGENCE
CAN MAKE
HEALTHCARE
HUMAN AGAIN

ERIC TOPOL

With a foreword by
ABRAHAM VERGHESE,
author of Cutting for Stone



AI: Deep learning in image classification in Early Breast Cancer



Fibrilación Atrial Familiar

info@heritas.com.ar

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cardio
HERITAS CARDIOMIOPATÍAS HEREDITARIAS

Plat. Tecnológicas

Asesoría genética

Servicios

Novedades

Acerca de Heritas

Contacto



SÍNDROME DE QT LARGO

SÍNDROME DE BRUGADA

SÍNDROME DE QT CORTO

TAQUICARDIA VENTRICULAR

CATECOLAMINÉRGICA POLIMÓRFICA

FIBRILACIÓN ATRIAL FAMILIAR

La fibrilación atrial (FA) familiar es una manifestación asociada a diversos fenotipos electrofisiológicos y/o inclusive cardiopatías estructurales, en los que el desarrollo de FA puede presentarse en forma primaria.

GENES COMPROBADOS

GJA5	KCNQ1	SCN5A	
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GENES EMERGENTES

ABCC9	DSC2	EMD	HCN4
JPH2	KCNA5	KCND3	KCNE1
KCNE2	KCNE3	KCNJ2	KCNJ8
LMNA	MYH6	NKX2-5	NPPA
SCN3B	SCN4B		

AFib – AI machine learning





Dispositivos móviles

Editorial

The Lancet

NEJM

THE NEW ENGLAND JOURNAL OF MEDICINE
REVIEW ARTICLE
FRONTIERS IN MEDICINE
Mobile Devices and Health
Ida Sim, M.D., Ph.D.
MOBILE HEALTH — THE APPLICATION OF SENSORS, MOBILE APPS

JAMA

VIEWPOINT
**Personal Health Records
More Promising in the Smartphone Era?**
Christian Dameff, MD
As health care delivery organizations shift from implementation of electronic health records to optimization, reported in

Wearable technology and lifestyle management:
the fight against obesity and diabetes

In January, The Lancet published two Commission reports on the current lack of conclusive clinical evidence showing the efficacy of the technologies for promoting behavior



JAMA

VIEWPOINT
**Wearable Devices for Cardiac Rhythm Diagnosis
and Management**

James E. Ip, MD
Division of Cardiology,
Department of
Medicine, Cornell

With the increasing use of direct-to-consumer medical devices, it is paramount for clinicians to recognize the potential utilization for patient management. The

Combining data from PPG sensors with accelerometers can help identify some arrhythmic cardiac detection at rest may indicate

www.nature.com/npjdigitalmedicine

Opinion

NATURE

npj | Digital Medicine

PERSPECTIVE OPEN
**Best practices for analyzing large-scale health data from
wearables and smartphone apps**
Jennifer L. Hicks¹, Tim Althoff², Rok Sosic³, Peter Kuhar⁴, Bojan Bosnjakovic⁵, Abby C. King^{6,7}, Jure Leskovec^{8,9} and Scott E. Delp¹⁰

Smartphone apps and wearable devices for tracking physical activity and other health behaviors have become popular in recent years and provide a largely untapped source of data about health behaviors in the free living environment. The data are large in



JS | BIOLOGY

RESEARCH ARTICLE
**Digital Health: Tracking Physiomes and
Activity Using Wearable Biosensors Reveals
Useful Health-Related Information**
Xiao Li^{1,2}, Jessilyn Dunn^{1,2,3}, Denis Salins^{1,2,3}, Gao Zhou¹, Wenyu Zhou¹, Sophia Miryam Schüssler-Florenza Rose^{2,4}, Dalia Perelman⁵, Elizabeth Colbert², Ryan Runge¹, Shannon Rego², Ria Sonecha¹, Somalee Datta¹, Tracey McLaughlin⁶, Michael P. Snyder¹

2019-2020

10%
Human



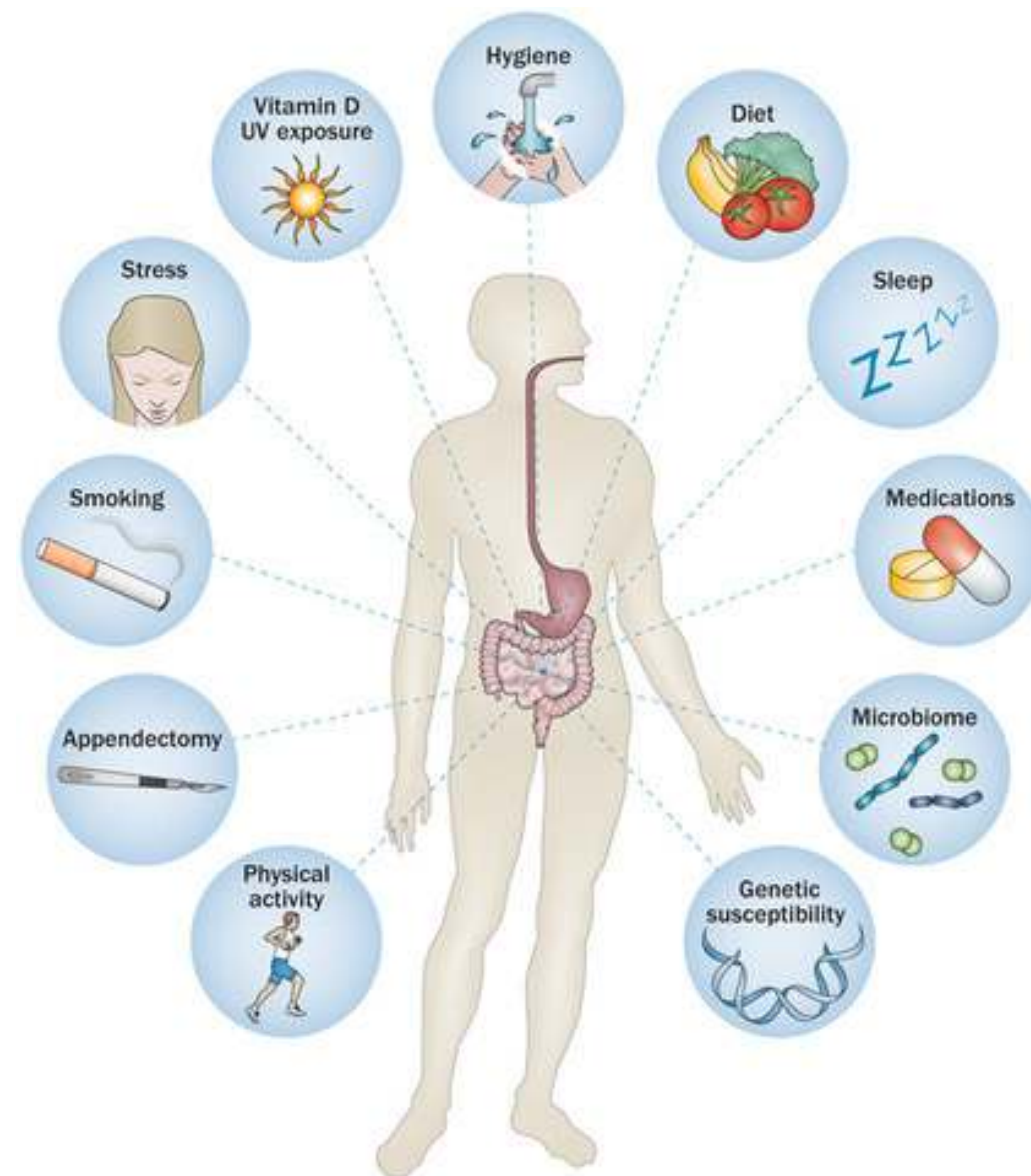
ARTICLE

nature
International journal of science

doi:10.1038/nature25973

Environment dominates over host genetics in shaping human gut microbiota

Daphna Rothschild^{1,2*}, Omer Weissbrod^{1,2*}, Elad Barkan^{1,2*}, Alexander Kurilshikov³, Tal Korem^{1,2}, David Zeevi^{1,2}, Paul I. Costea^{1,2}, Anastasia Godneva^{1,2}, Iris N. Kalka^{1,2}, Noam Bar^{1,2}, Smadar Shilo^{1,2}, Dar Lador^{1,2}, Arnau Vich Vila^{3,4}, Niv Zmora^{5,6,7}, Meirav Pevsner-Fischer⁵, David Israeli⁸, Noa Kosower^{1,2}, Gal Malka^{1,2}, Bat Chen Wolf^{1,2}, Tali Avnir-Sagi^{1,2}, Maya Lotan-Pompan^{1,2}, Adina Weinberger^{1,2}, Zamir Halpern^{7,9}, Shai Carmi¹⁰, Jingyuan Fu^{3,11}, Cisca Wijmenga^{3,12}, Alexandra Zhernakova³, Eran Elinav⁵ & Eran Segal^{1,2}§

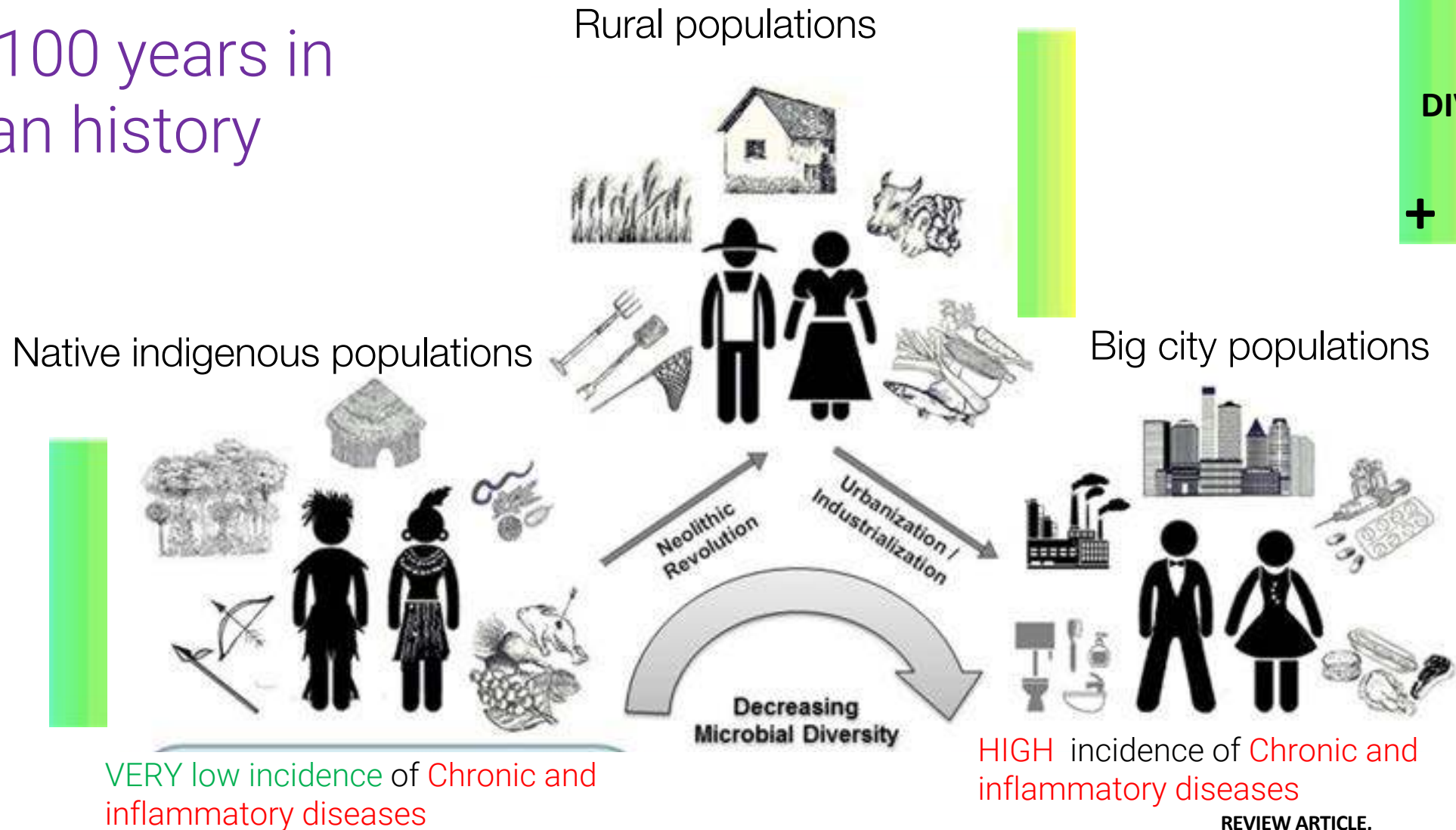


Microbioma Humano y salud



- ❑ Obesidad
- ❑ Intolerancia Alimentos
- ❑ Diabetes
- ❑ Autoinmunidad
- ❑ Cancer colon
- ❑ Homeostasis riñon
- ❑ Enfermedad periodontal
- ❑ Esofagitis
- ❑ Gastritis
- ❑ Gastroenteritis
- ❑ Colon Irritable
- ❑ Enfermedad de Crohn
- ❑ Artritis Reumatoidea
- ❑ Parkinson
- ❑ Enfermedades psiquiatricas
- ❑ Autismo
- ❑ Schizofrenia
- ❑ Infertilidad

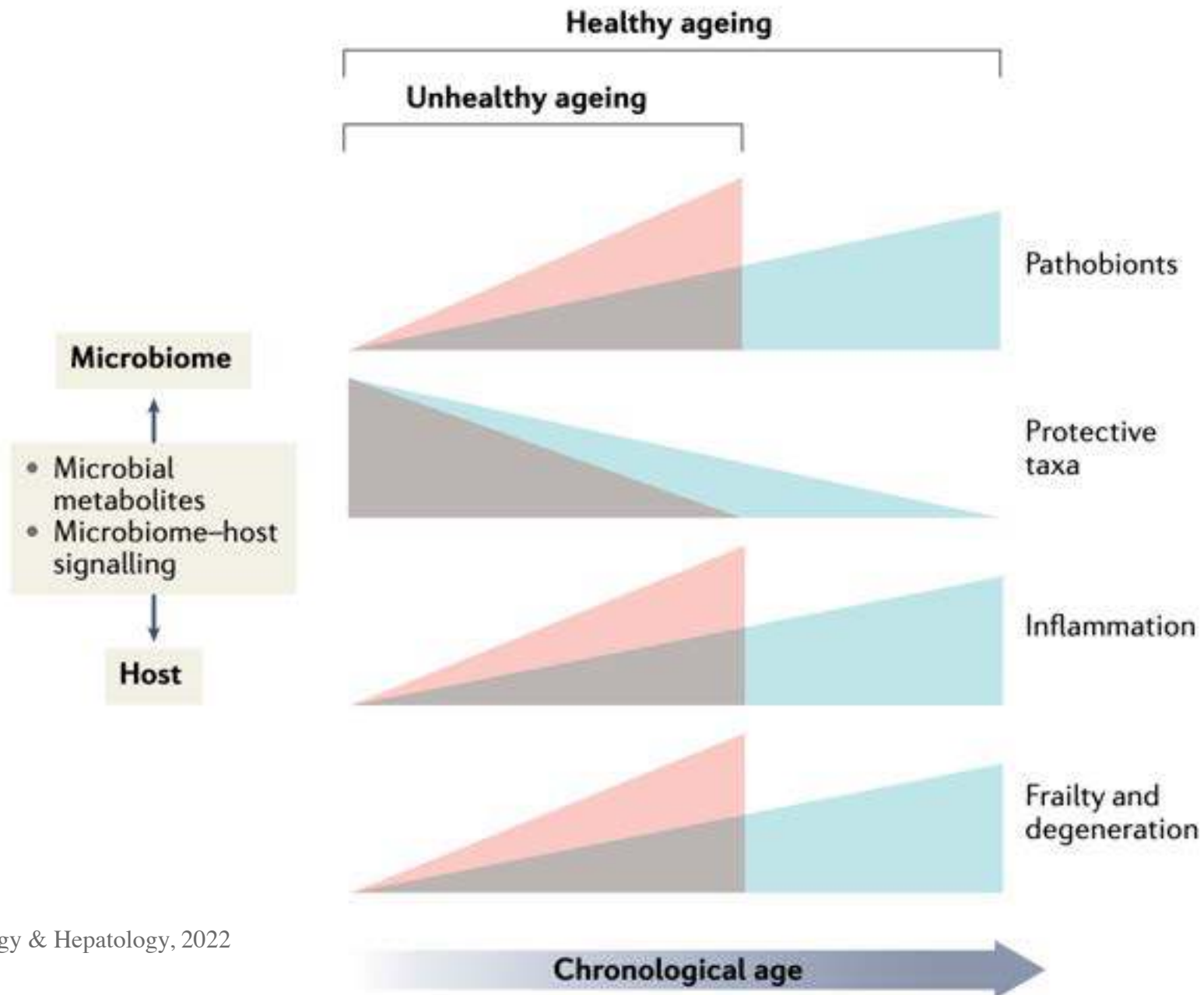
Industrialization effect- Last 100 years in human history



REVIEW ARTICLE,
Gupta VK, Paul S and Dutta C (2017)
Front. Microbiol., 23 June 2017

The gut microbiome as a modulator of healthy ageing

Srinivasan Chakraborty^{1,2}, Pooja Choudhary^{1,2} and Paul W. O'Toole^{1,2*}





Next Generation Sequencing
El valor de los datos
La traducción al paciente
Inteligencia Artificial
Dispositivos móviles
Microbioma Humano