

Qué grandes eventos están
moldeando el futuro de la
medicina y la salud

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

the last ten years

2008

2018

50,000 b

day
1 equipment

18,000,000,000,000 b

day
1 equipment

0.000016

human genomes
day

100,000,000 USD

600

human genomes
day

1,000 USD



illumina®

Projected annual storage in 2025

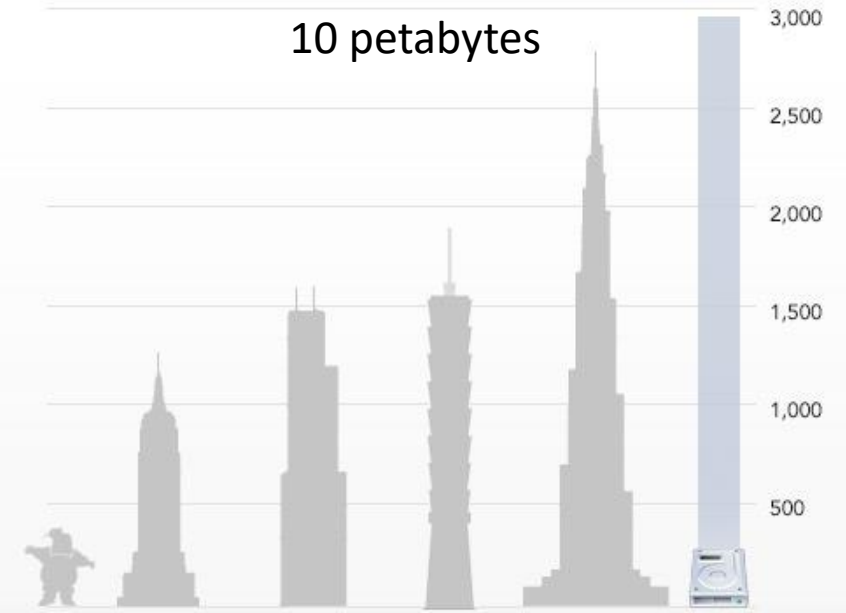
Twitter: 1–17 petabytes per year

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*





THE DNA OF A NATION

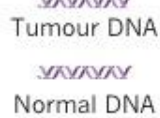
The United Kingdom aims to sequence 100,000 human genomes by 2017. But screening them for disease-causing variants will require innovative software.

~50,000
people with rare
diseases and
their parents



DNA

~25,000
people with
cancer

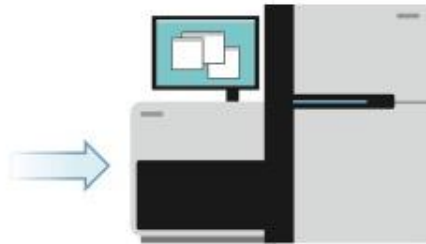


Tumour DNA

Normal DNA

THE CLINICAL GENOME

Genomics England plans to sequence 100,000 genomes by 2017. The genomic data will be crucial for diagnosing and treating disease, but its interpretation will require automated, specialized software.



RECRUITMENT OF 75,000 PEOPLE

The 100,000 Genomes Project is recruiting people with cancer and rare diseases. The genomes of both normal and tumour cells will be sequenced in people with cancer.

NEXT-GENERATION SEQUENCING

The Californian company Illumina will use UK-based high-throughput sequencing machines to produce whole-genome sequences and identify genetic variants.

AUTOMATED INTERPRETATION

Four UK and US companies will use specialized software to automatically analyse the genetic variants that may be linked to disease.

CLINICAL INTERPRETATION

Around 2,000 UK scientists and clinicians will pore over the data to validate or better understand how the variants may cause disease before the information is fed back to patients.



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

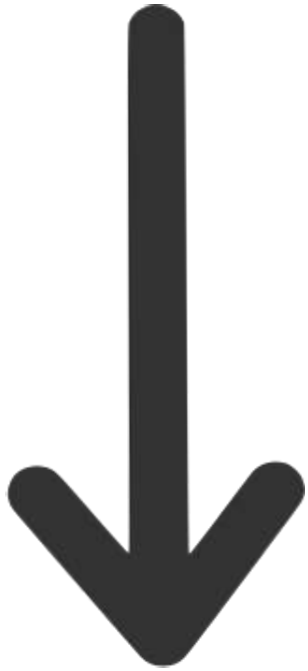
- 1.Visualisation
- 2.Creativity
(find signal in noise)



Population power

HiSeq X Ten

20,000 genomes/year
600 genomes/run



Going backwards

illumina®



MiSeq

3 exomes/run

Individual power

Production power is not a limitation anymore

*Too much information
is good and bad
at the same time*

*Most relevant question in the big data era:
What do we want from the data???*

Genomics route to the clinic

Research

Clinical Diagnostics



Genomics route to the clinic

CAREERS

TRANSITIONS From building houses to building molecules p.13

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

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



DPA_SCHWARTZ/GETTY

Genetic Counsellor
Is the next big thing
in hot professions

GENETICS

Fluent in DNA

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

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



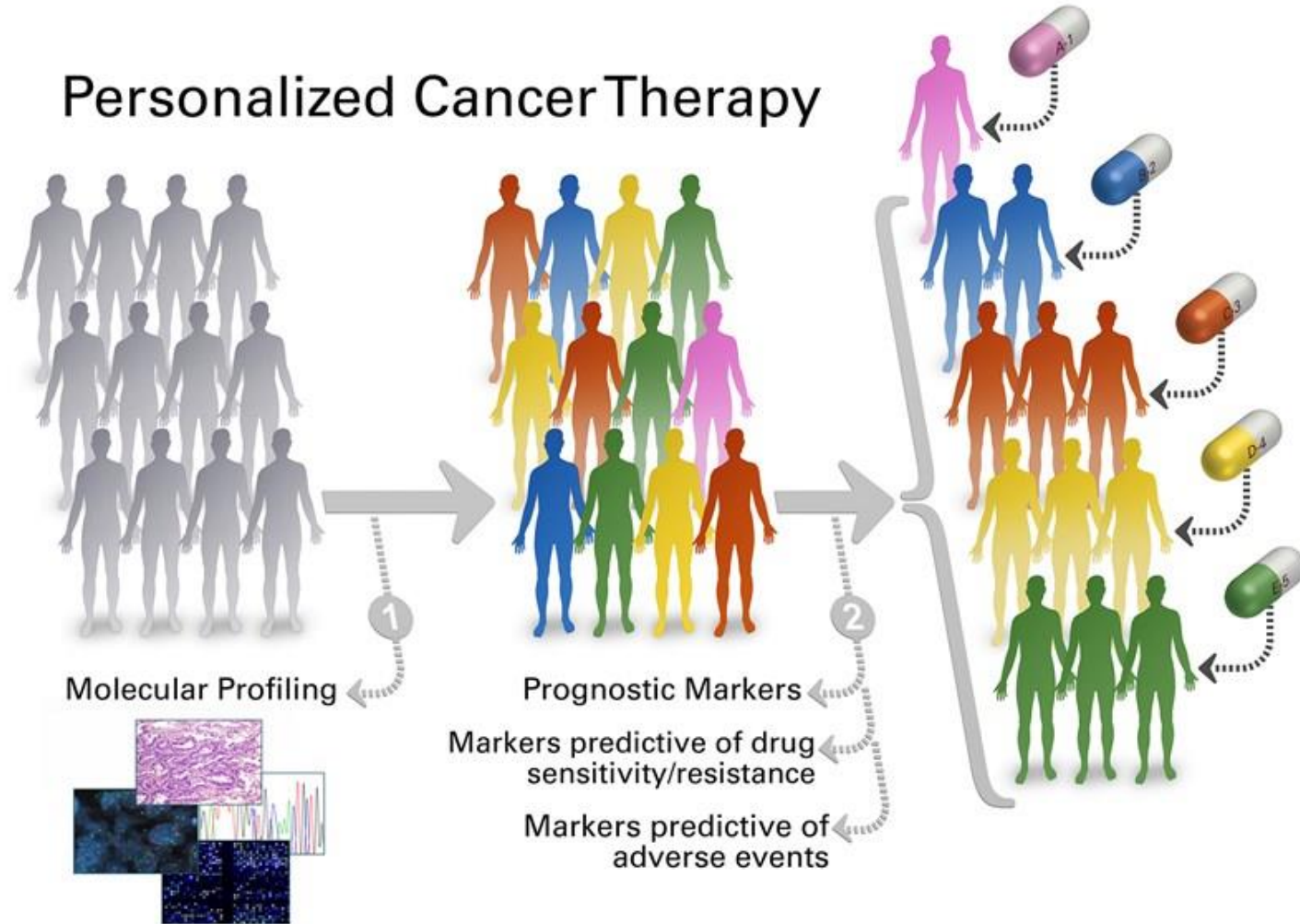
10. NEULASTA (pegfilgrastim)
Neutropenia



Based on published number needed to treat (NNT) figures. For a full list of references, see Supplementary Information at go.nature.com/4dr78f.

Precision Medicine

Personalized Cancer Therapy



DEEP MEDICINE

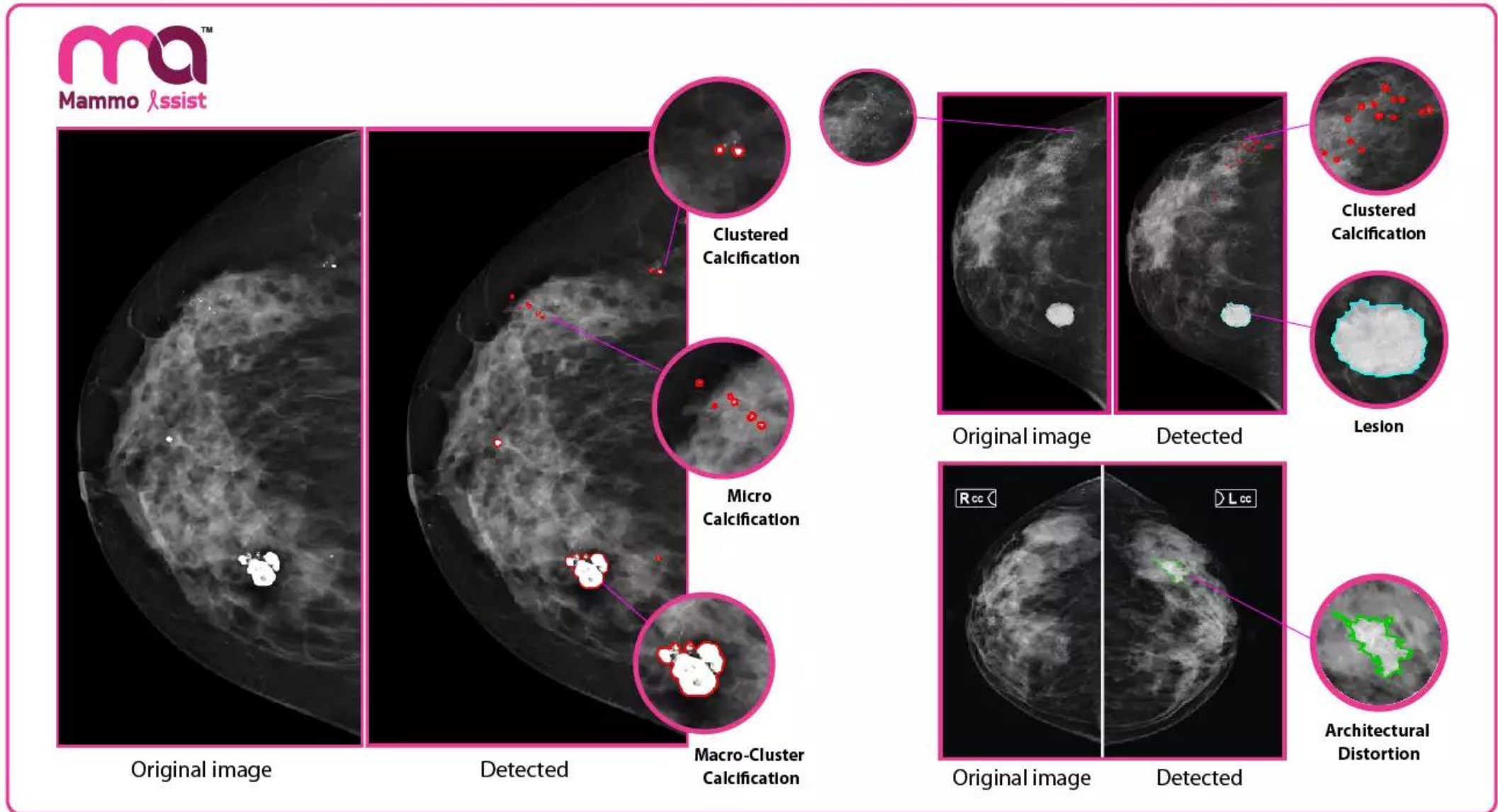
HOW ARTIFICIAL
INTELLIGENCE
CAN MAKE
HEALTHCARE
HUMAN AGAIN

ERIC TOPOL

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



AI: Deep learning in image classification in Early Breast Cancer



AFib - Genética

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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

Apple Watch.
Helping your patients
identify early warning
signs.

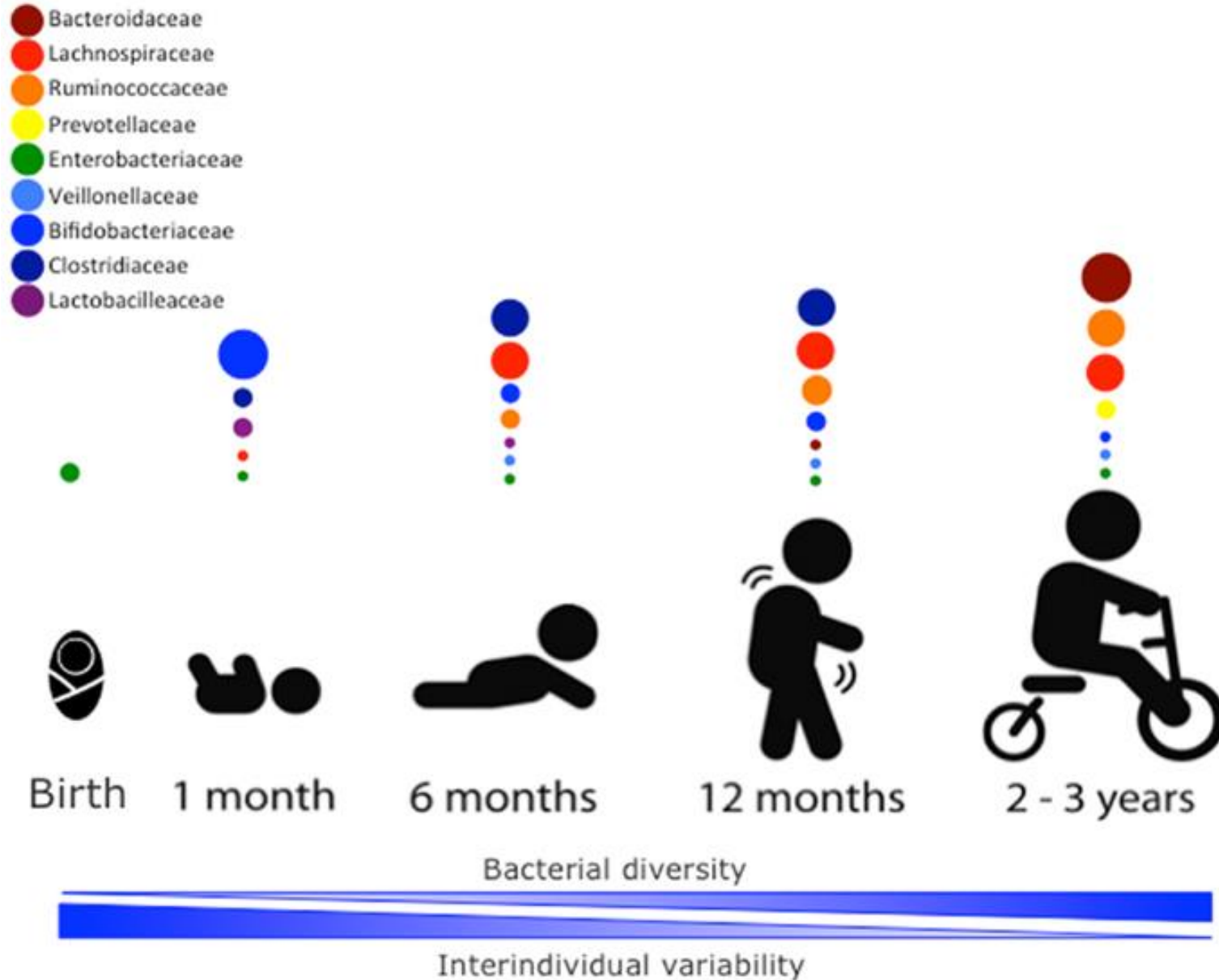


10%

Human

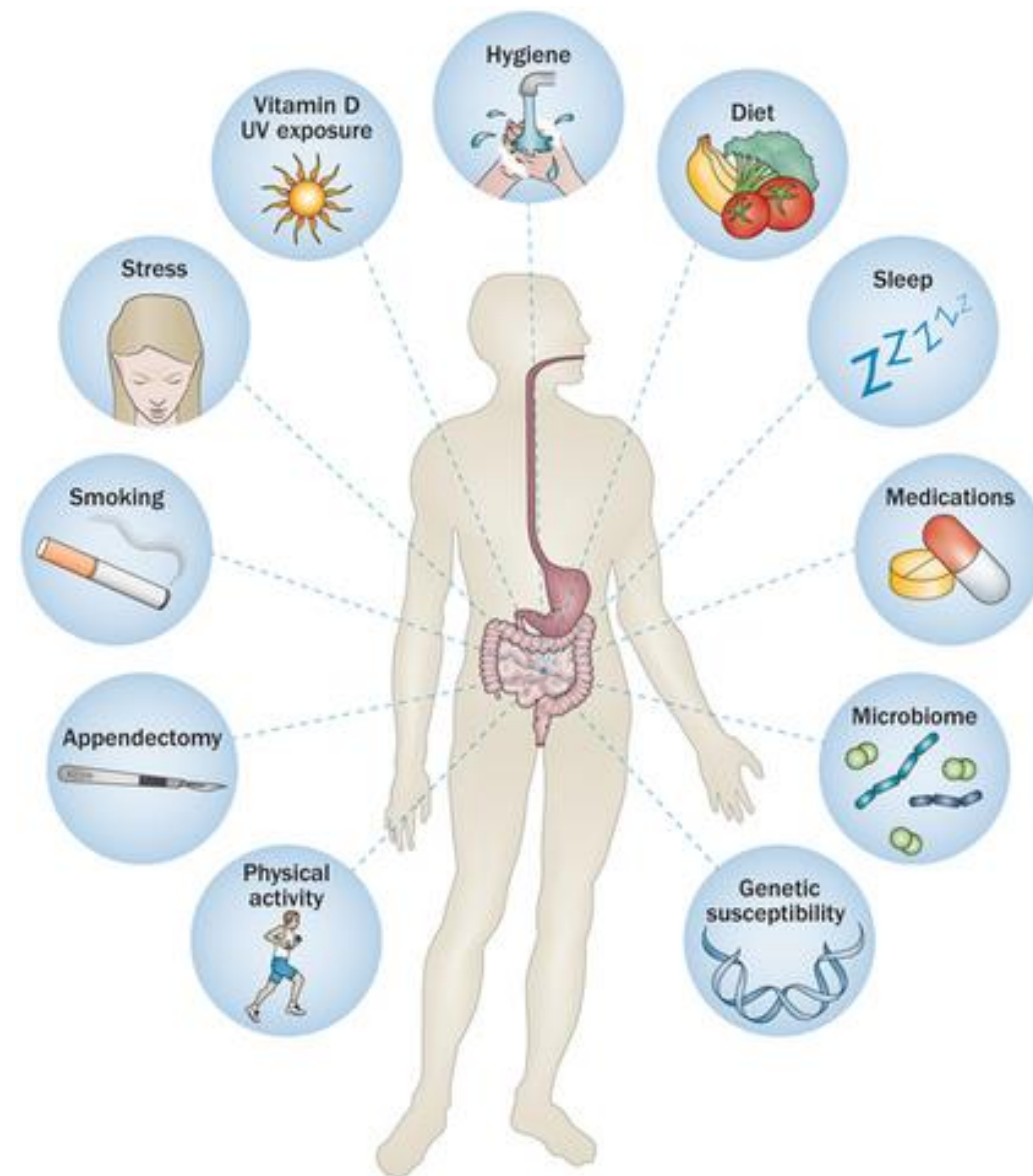


Evolución del Microbioma



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^{1,4}, Niv Zmora^{5,6,7}, Meirav Pevsner-Fischer⁵, David Israeli⁸, Noa Kosower^{1,2}, Gal Malka^{1,2}, Bat Chen Wolf^{1,2}, Tali Avnit-Sagi^{1,2}, Maya Lotan-Pompan^{1,2}, Adina Weinberger^{1,2}, Zamir Halpern^{7,9}, Shaf Carmi¹⁰, Jingyuan Fu^{3,11}, Cisca Wijmenga^{3,12}, Alexandra Zhernakova³, Eran Elinav^{5,§} & Eran Segal^{1,2,§}

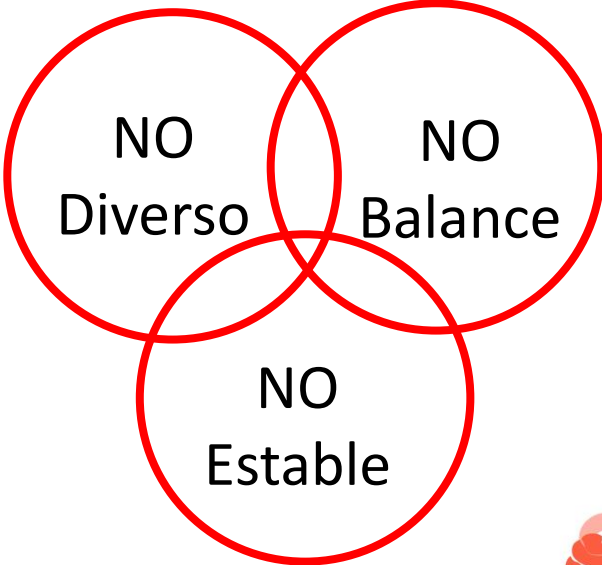
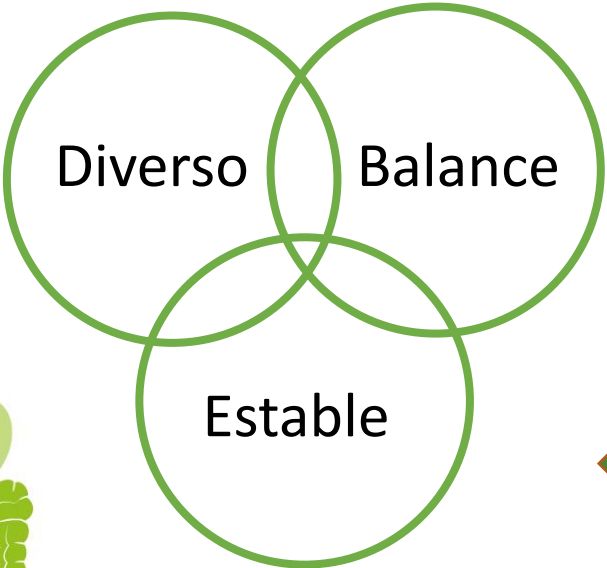


Salud y Microbioma

+Saludable

Medio ambiente:
*Dieta, estilo de vida,
Salud mental, Antibióticos*

+Propenso a enfermedades



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