Bio-491
New tools and research strategies in personalized health

Introduction

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Marc Friedli

Thought-Provoking Questions

- What if we could predict your risk of disease years before symptoms appear?
- What if we could tailor treatments based on your DNA to maximize effectiveness and minimize side effects?
- Why do two people with the same diagnosis respond differently to the same treatment?

Program Bio491, 2025

BIO-491 New tools & research strategies in personalized health
Master course
2024-2025

February 20	15.15-16.15	*New tools and research technologies in personalized health: Introduction*	**Marc Friedli**, Scientist, Unit of Precision Medicine, EPFL // Program Manager, Health 2030 (https://health2030.ch)
	16.15-18.00	*Opportunities and challenges of implementing genomic medicine*	**Katrin Männik**, Head of Genomics Strategy, Health 2030 Genome Center, Geneva
February 27	15.15-16.00	*Leveraging Single-Cell and Spatial Data for Personalized Health: A Computational Perspective*	**Raphael Gottardo**, Director, Translational Biomedical Data Science Group @ CHUV & Full Professor in the Faculty of Biology and Medicine @ UNIL
	16.15-17.00	*Digital Epidemiology*	**Marcel Salathé**, Epidemiology Lab, EPFL
	17h15-18h00	*Genomic Medicine*	**Flavia Hodel**, Data scientist, Precision Medicine Unit, CHUV
March 6	15.15-17.00	*Ethical and social issues in personalized health*	**Gaia Barazzetti**, Research Ethics Compliance Officer, Research Office, EPFL
	17h15-19h00	*Highthroughput bioinformatics*	**Ioannis Xenarios,** CHUV-UNIL, Health 2030 Genome Center
March 13	15.15-17.00	*Pharmacogenomics and personalized therapies: the right drug at the right dose for the right patient*	**Caroline Samer**, Pharmacogenomics and Personalised Therapy Unit, HUG & UNIGE
	17.15-19.00	*Leveraging Electronic Health Records, Medical Knowledge, and Information Technologies for Medical Discoveries*	**Mina Bjelogrlic**, Head of Machine Learning, Human-Machine Interfaces in clinical settings Group, UNIGE
March 20	15.15-17.00	*Economical and governance aspects of personalized health*	**Joël Wagner**, Department of Actuarial Science, Faculty of Business and Economics (HEC Lausanne)
	17.15-19.00	*The Role of Organoids in Precision Health*	**Nathalie Brandenberg**, PhD, Co-Founder, SUN bioscience SA and Doppl SA
March 27	15.15-17.00	*Towards personalized treatment strategies in neurological disorders*	**Friedhelm Hummel**, Institute Neuro-X and Defitech Chair of Clinical Neuroengineering, EPFL
April 3	15.15-17.00	*Precision nutrition*	**Murielle Bochud**, Department of Epidemiology and Health System, Unisanté, Lausanne
	17.15-19.00	*Precision medicine in cancer*	**Filipe Martins**, TBC

Date	Schedule	Event
April 10	15.15-16.45	!!! Midterm written exam !!!
April 17		Group Work 1/3 (free schedule)
April 24		*Easter*
May 1		Group Work 2/3 (free schedule)
May 8		Group Work 3/3 (free schedule)
May 15	15.00-19.00	Exam / Group presentation 1/2
May 22	15.00-19.00	Exam / Group presentation 2/2
May 29		*Ascension*

Program Bio491, 2025

Here are a few information about the projects that you will work on:
All the projects will be available on the class' moodle close to the end of lectures schedule

Examples (not actual projects) (The presentations will take place either on **May 15 or May 22**)

- Personalized neurotechnology-based treatment for underdeveloped countries at the example of stroke: a health app-based concept
- Economical and governance aspects of personalized health
- Rapid whole-genome sequencing in hospitalised infants: focus on pediatric oncology
- Personalized Nutrition
- Project in Genomic Medicine
- The Genome of Switzerland: Ethical and Social Challenges
- How to develop precision nutrition based on sustainable diets
- The Matrix
- Build a project around cancer patient care and quality of life.
- Build a therapeutic strategy on a hypothetical patient cancer case.

Program Bio491, 2025

Procedure is as follow:

- Read the projects carefully.
- Choose between the projects and make a selection of three (choice n°1, 2, 3 (1 = best))
- Form a group of 4/5 students (as soon as possible, do not lose any time!!!). Each formed group will have one person send an email to marc.friedli@epfl.ch, cecile.hayward@epfl.ch with the composition of the group and the selected projects listed from 1 to 3.
- We cannot guarantee that your first choice will be attributed to you but we will try. Please also note that all proposed projects will be attributed.
- We are aware that it may be difficult to create groups or find one but we will help you.

As for the presentations:

•Format is very flexible and in a creative and multidisciplinary spirit.

•Projects presentations of **May 15 and May 22** will be in mini-symposium format where groups present and the rest of the class participate by asking questions. This means that each one of you must attend all the presentations, from 15.15 to 19.00.

•**You MUST make contact with the head of the project, and/or TAs for that specific project as soon as project is assigned**, they will guide you and answer your questions. It is expected that you work in close collaboration with them.

•Each presentation will last 40 minutes in total, including questions and discussion. Please prepare a 30-minute presentation and allow 10 minutes for questions/discussion.

The TAs will be at your disposal should you have questions.

🔎 How Does This Course Fit In?

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- This course aims to provide with the knowledge to contribute to the rapidly evolving field of personalized health.

- Overview of the tools, technologies, and challenges shaping personalized health.

- 📅 **Course Roadmap:**
- - Genomics & Big Data
- - Personalized treatment plans & nutrition
- - Digital Health & AI
- - Ethics & Policy
- - Technologies and Applications
- - Economic & Societal Impact

🔎 What do you need to know?

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- Understand the the meaning of personalised healthcare, the revolution it represents in health, what is behind it what are the opportunities and challenges

- Work in groups and a project in a broad manner, in collaboartive and if possible multidiscplinary (group work and presentation) **70% of grade**

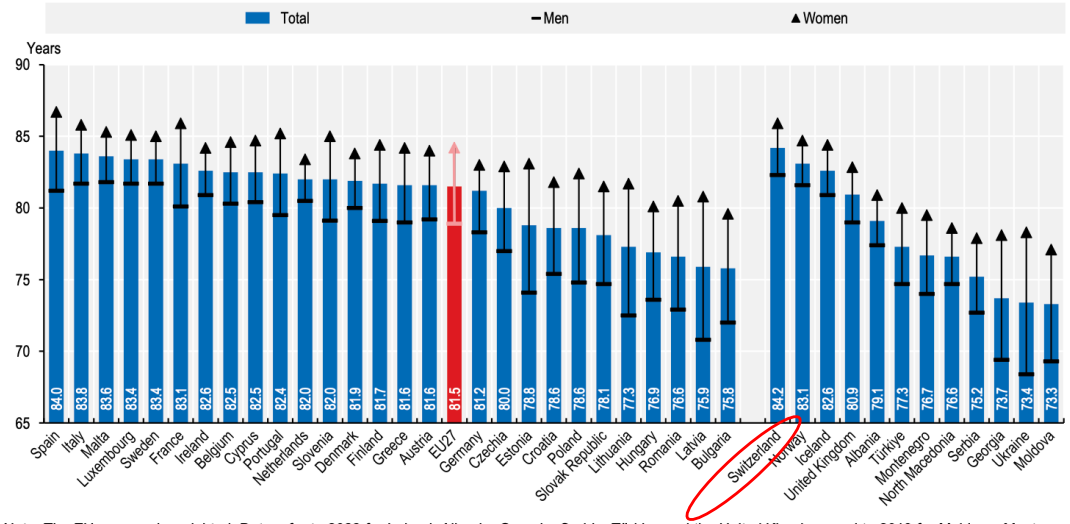
- Understand the general concepts in the individual lectures given. **30% of grade of grade**

Outline

- Health status in Switzerland
- Marching toward Precision medicine
- Definitions of Precision Medicine, Personalized Health, Genomic Medicine
- “Technolomics” and Big Data
- The Human Genome and Genomics
- Clinical applications
- E-Health, M-Health, Electronic patient records, Apps in Health
- AI in Health
- Personalized Health in the international context
- Personalized Health in Switzerland
- Personalized Health Challenges and opportunities

Health status is high in Switzerland

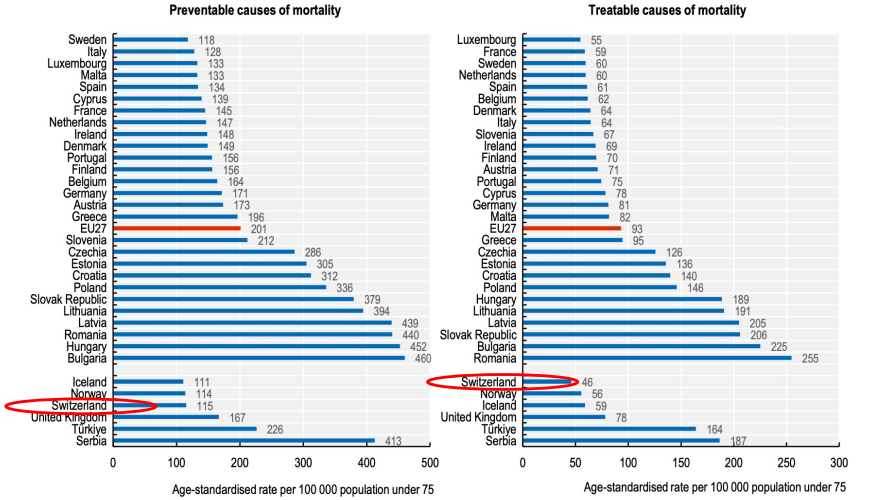
Figure 3.1. Life expectancy at birth, by gender, 2023 (or nearest year)



Note: The EU average is weighted. Data refer to 2022 for Ireland, Albania, Georgia, Serbia, Türkiye and the United Kingdom, and to 2019 for Moldova, Montenegro, North Macedonia and Ukraine.
Source: Eurostat (demo_mlexpec); complemented with ONS for the United Kingdom, TURKSTAT for Türkiye and WHO for Moldova.

USA life expectancy in 2023 in USA: 78.4 years

Figure 6.2. Mortality rates from avoidable causes, 2021



Note: The EU average is weighted. The data for the United Kingdom relate to 2020 (the rates have been calculated by the OECD based on the European population structure).
Source: Eurostat (hlth_cd_apr).

Source OECD

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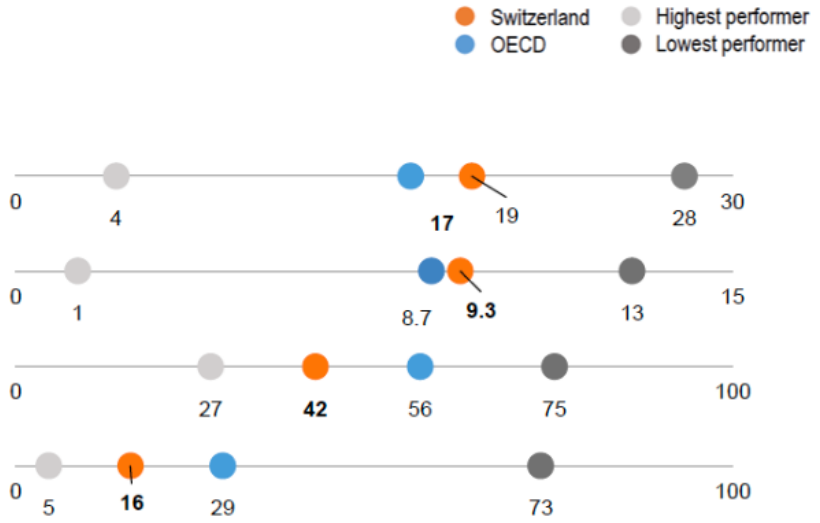
Overweight/Obesity and deaths from particulate matter pollution are lower than OECD average

Smoking (2019 or nearest year)
Daily smokers (% population 15+)

Alcohol (2019 or nearest year)
Litres consumed per capita (population 15+)

Overweight/obese (2019 or nearest year)
Population with BMI>=25 (% population 15+)

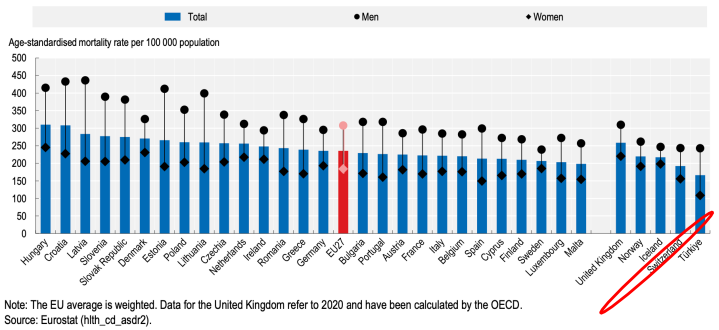
Air pollution (2019 or nearest year)
Deaths due to ambient particulate matter pollution (per 100 000 population)



Source OECD

Cancer survival

Figure 3.9. Cancer mortality in EU countries by gender, 2021 (or nearest year)

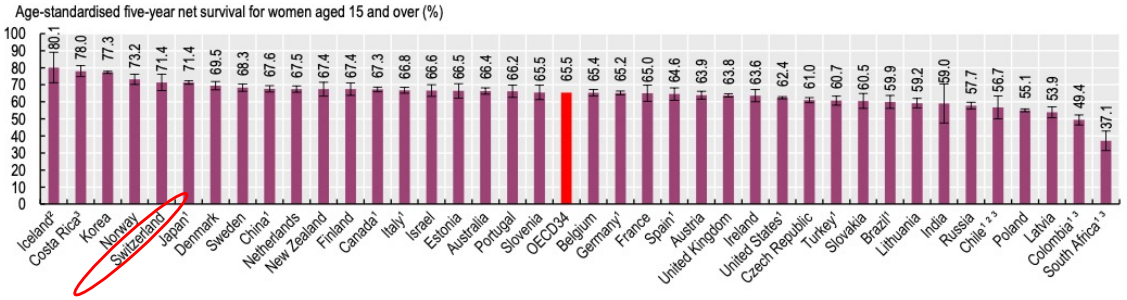


Note: The EU average is weighted. Data for the United Kingdom refer to 2020 and have been calculated by the OECD.
Source: Eurostat (hlth_cd_asdr2).

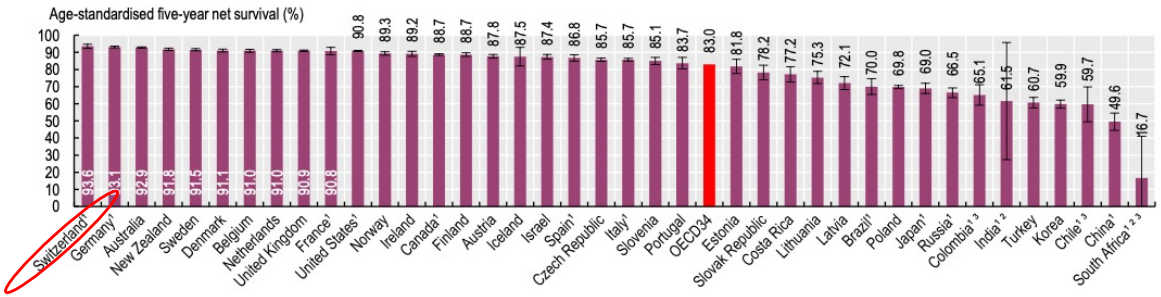
Cancer Mortality

five-year net 2010-2014

Uterine cervix



Melanoma



Source: OECD London School of Hygiene and Tropical medicine

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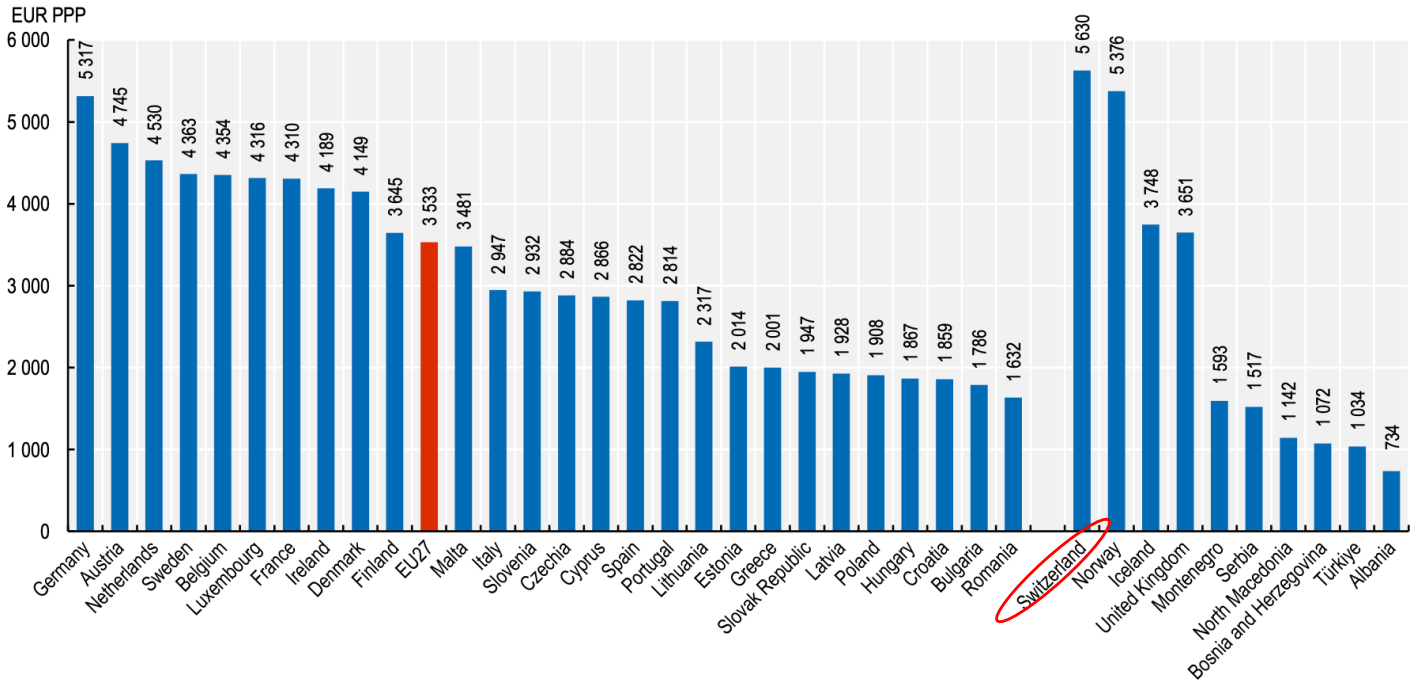
Cost of health in Switzerland is amongst the highest

Figure 5.1. Health expenditure per capita, 2022 (or nearest year)



Note: The EU average is weighted (based on OECD calculations).
Source: OECD Health Statistics 2024; Eurostat (hlth_sha11_hf); WHO Global Health Expenditure Database.

Source OECD

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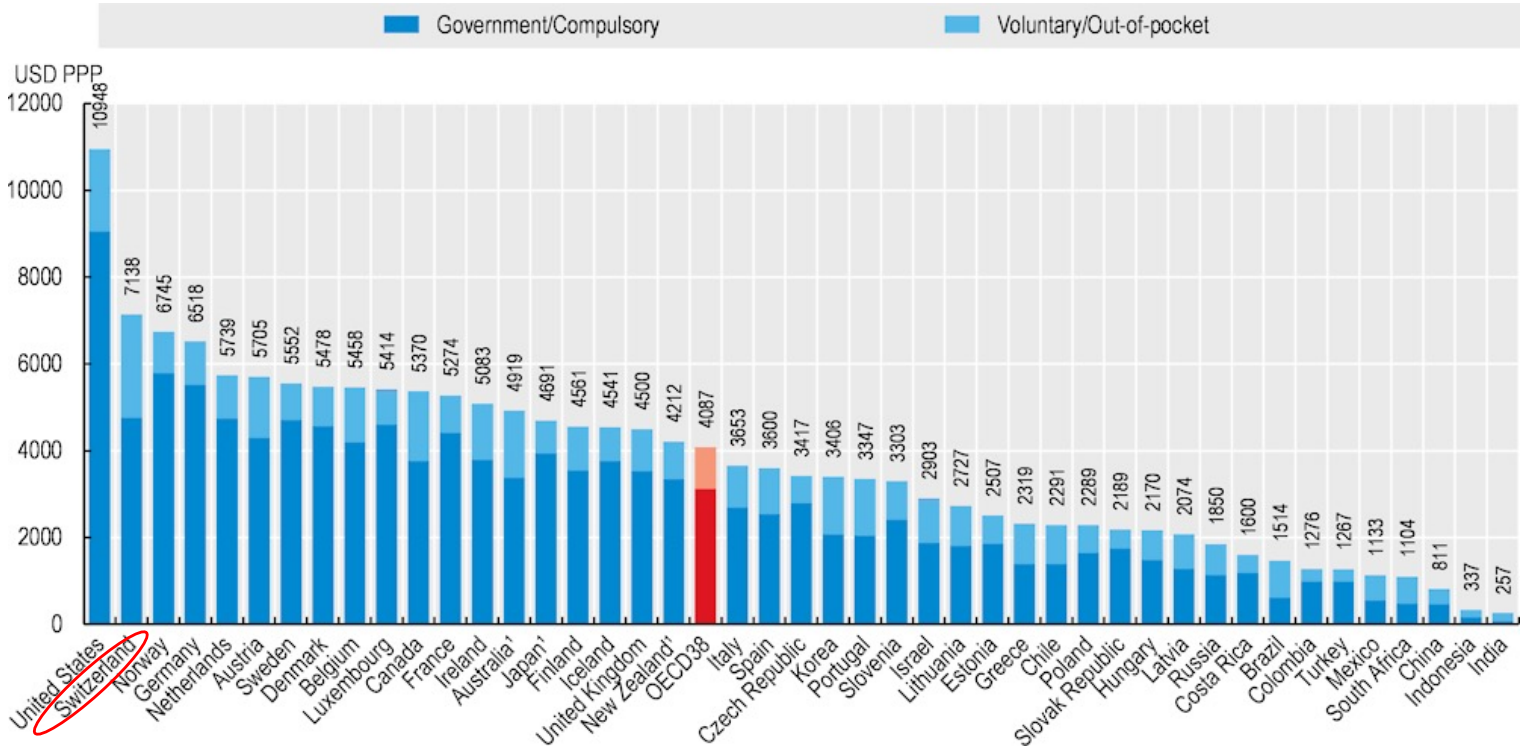
Health expenditure per capita, 2021



Source OECD

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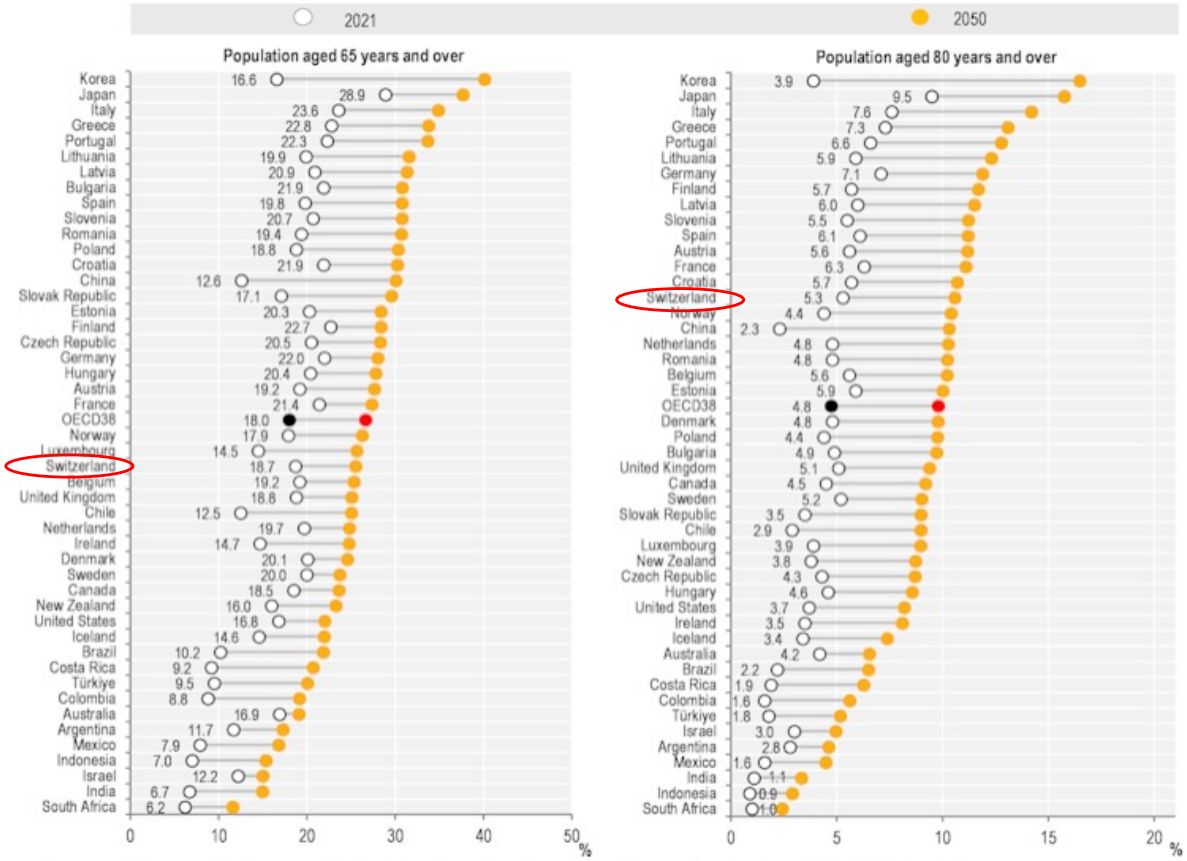
Society is aging

Figure 10.1. **Share of the population aged 65 and over and 80 and over, 2021 and 2050**



Sources: OECD Health Statistics 2023, OECD Historical Population Data and Projections (1950-2060) database.

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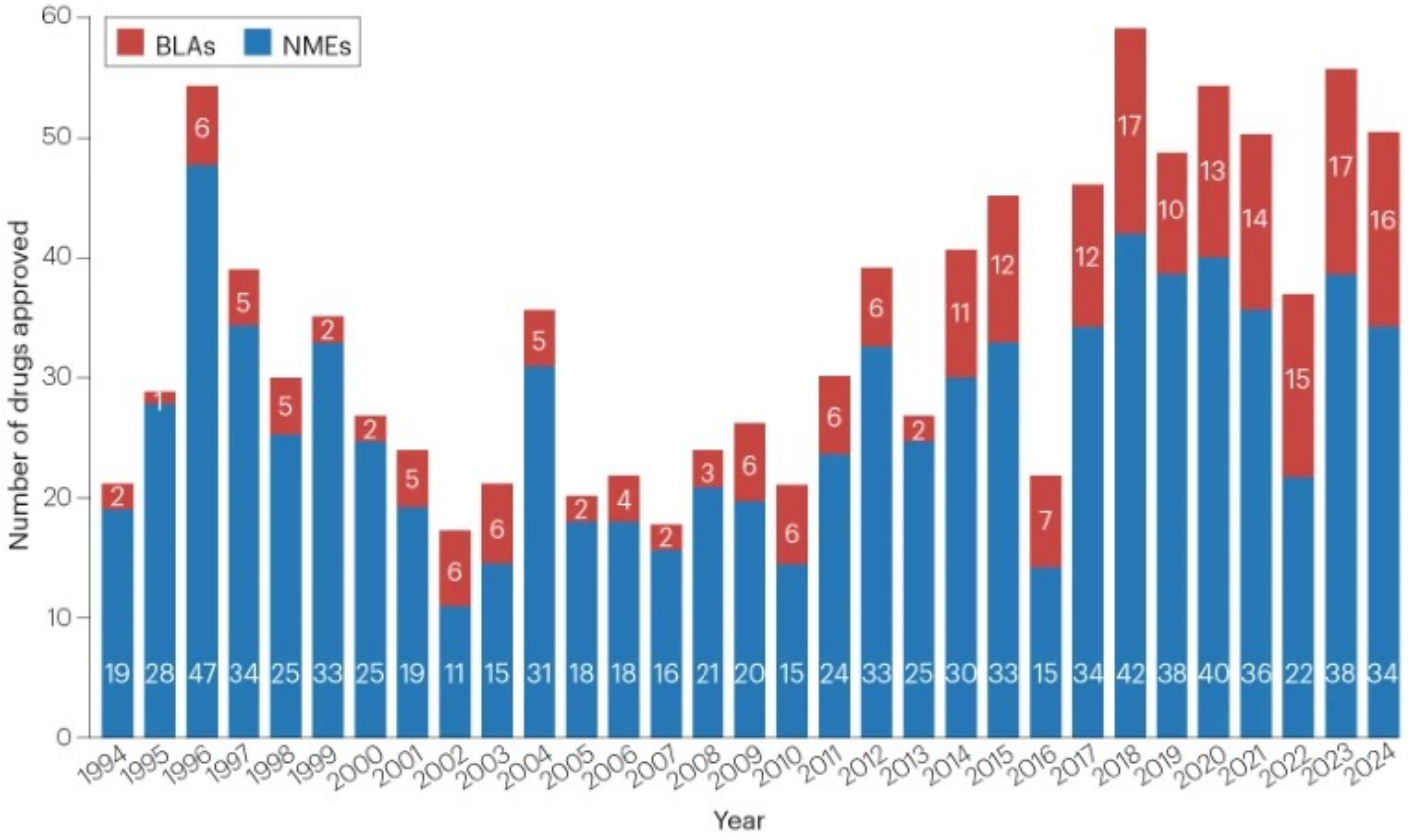
New drugs - few new molecules



new molecular entities (NMEs)
biologics license applications (BLAs)

Nature reviews drug discovery

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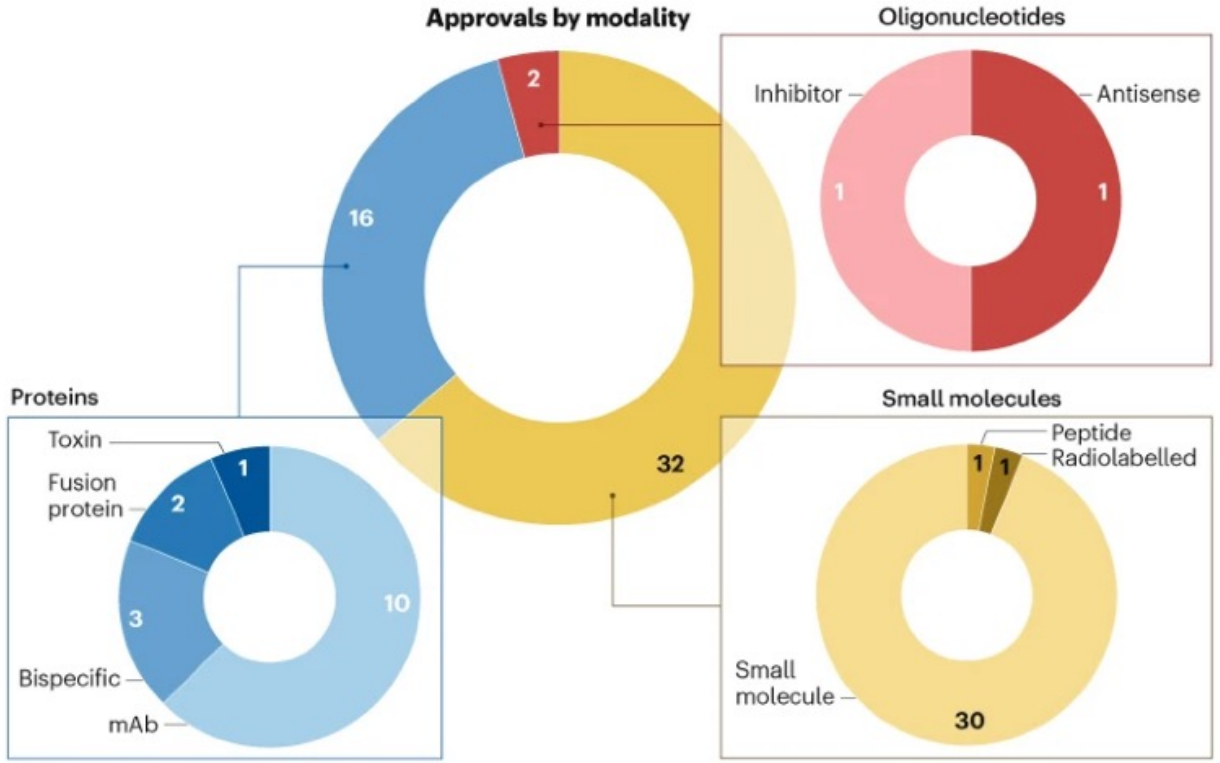
Approvals by modality



biologics license applications (BLAs)

new molecular entities (NMEs)

Nature reviews drug discovery

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📌 What is Precision Medicine/Personalized Health?

- Precision Medicine (PM) is about customizing disease prevention, diagnosis, and treatment for individuals based on:

- ✓ Genetics
- ✓ Environment
- ✓ Lifestyle

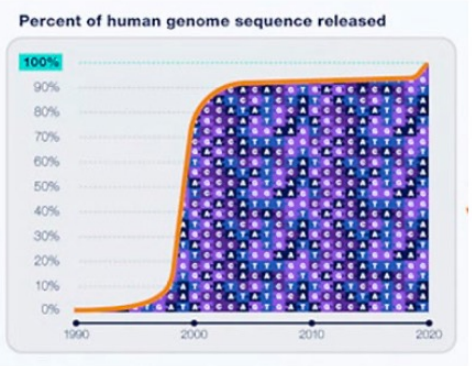
Definitions

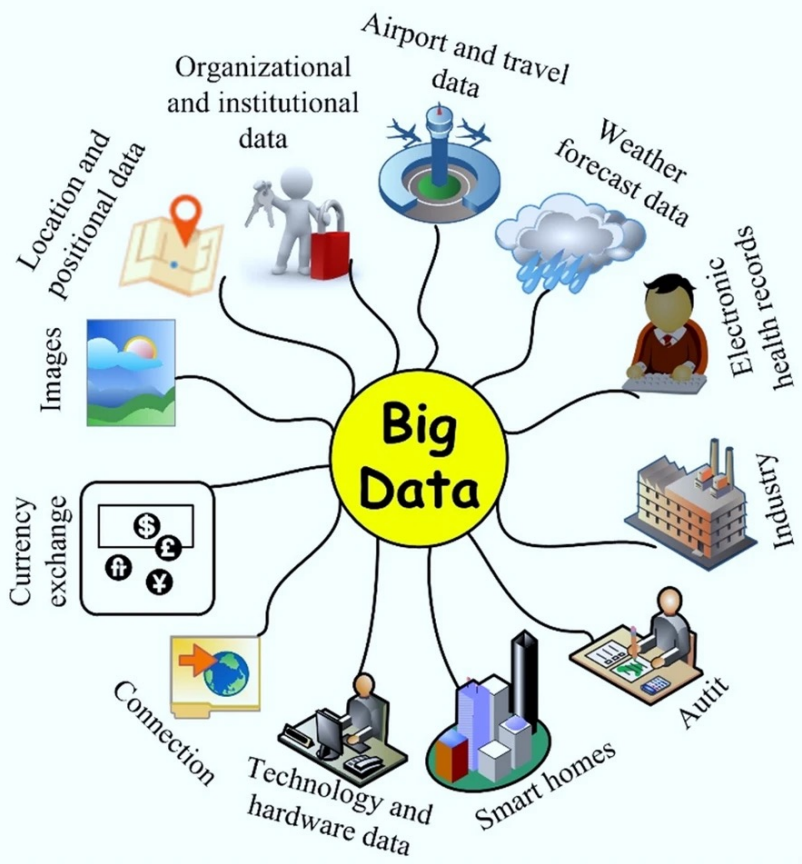
1.Precision Medicine: This approach is about tailoring disease treatment and prevention to individuals by considering their genetic variability, environment. The goal of precision medicine is to determine which treatment and prevention strategies will work best for specific groups of people, moving away from the one-size-fits-all approach of traditional medicine. It aims to provide the right treatments to the right patients at the right time, utilizing advances in technology to make medical care more precise for prevention, diagnosis, and treatment of diseases.

2.Personalized Health (or Health Care): While often used interchangeably with precision medicine, personalized health casts a wider net. It encompasses not just the customization of medical treatment based on genetic or biomolecular understanding but also integrates a broader spectrum of patient-specific factors — including lifestyle, environmental factors, and personal preferences — into care planning and decision-making. Personalized health aims for a holistic approach to health and wellness, emphasizing prevention and individualized care plans.

3.Genomic Medicine: This is a subset of precision medicine that specifically uses genetic information from an individual's genome to guide the diagnosis, treatment, and prevention of disease. Genomic medicine focuses on how an individual's genetic makeup affects their response to medication or their risk of developing certain diseases. This field relies heavily on technologies like Next Generation Sequencing (NGS) to identify genetic variants that may influence health and disease outcomes.

Stars aligned at the dawn of the 21st century

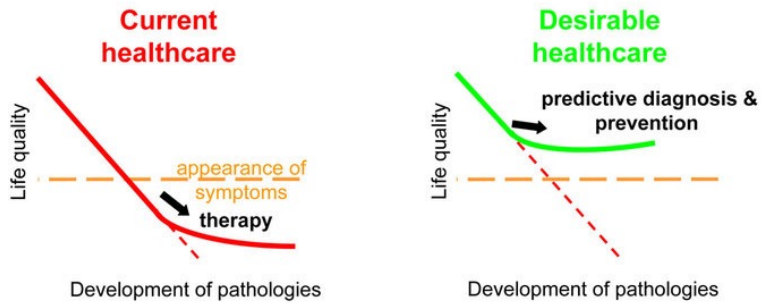




Marching towards precision medicine

Exploit new technologies emerging from biomedical research("technolomics"), bioengineering (wearable devices) and data science in order to:

- Shift emphasis from reaction to prevention
- Predict susceptibility to disease
- Improve disease detection
- Preempt disease progression
- Customize disease-prevention strategies
- Prescribe more effective drugs
- Avoid prescribing drugs with predictable side effects
- Reduce the time, cost and failure rates of pharmaceutical clinical trials
- Eliminate trial-and-error inefficiencies that inflate health care costs and undermine patient care



Why is Precision Medicine Important?

- 🚑 Traditional medicine treats diseases reactively—after symptoms appear.

- 🧬 Precision medicine shifts healthcare to prevention, using genomics, big data, and AI to anticipate health risks and optimize treatments.

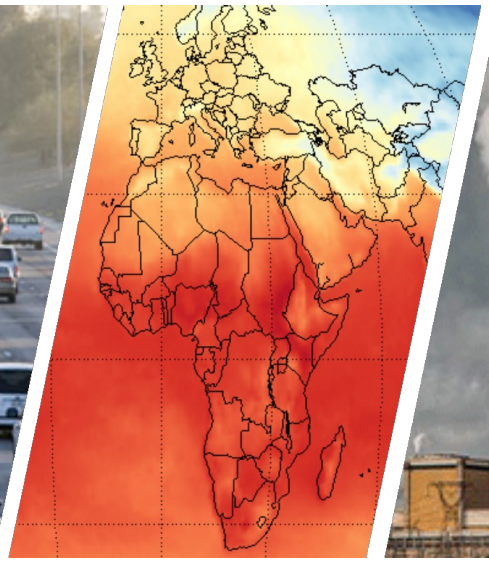
A major milestone in precision medicine: 2001 the complete sequence of the human genome

2001 2007 2023 …

Big data: all the things that surround you

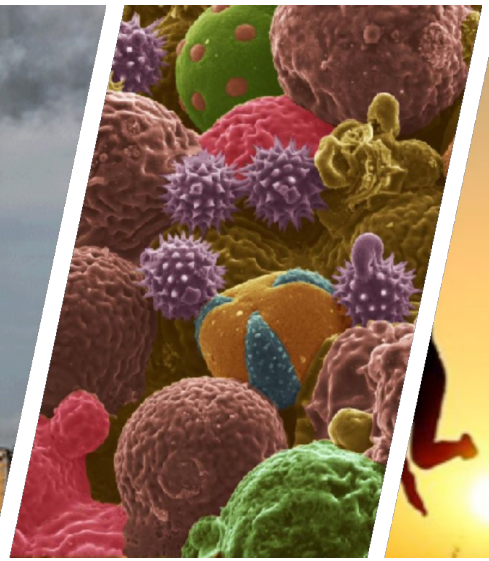
Traffic

Climate



Pollution

Allergens



Populations

Big data: all the things that you do

Food

Exercise

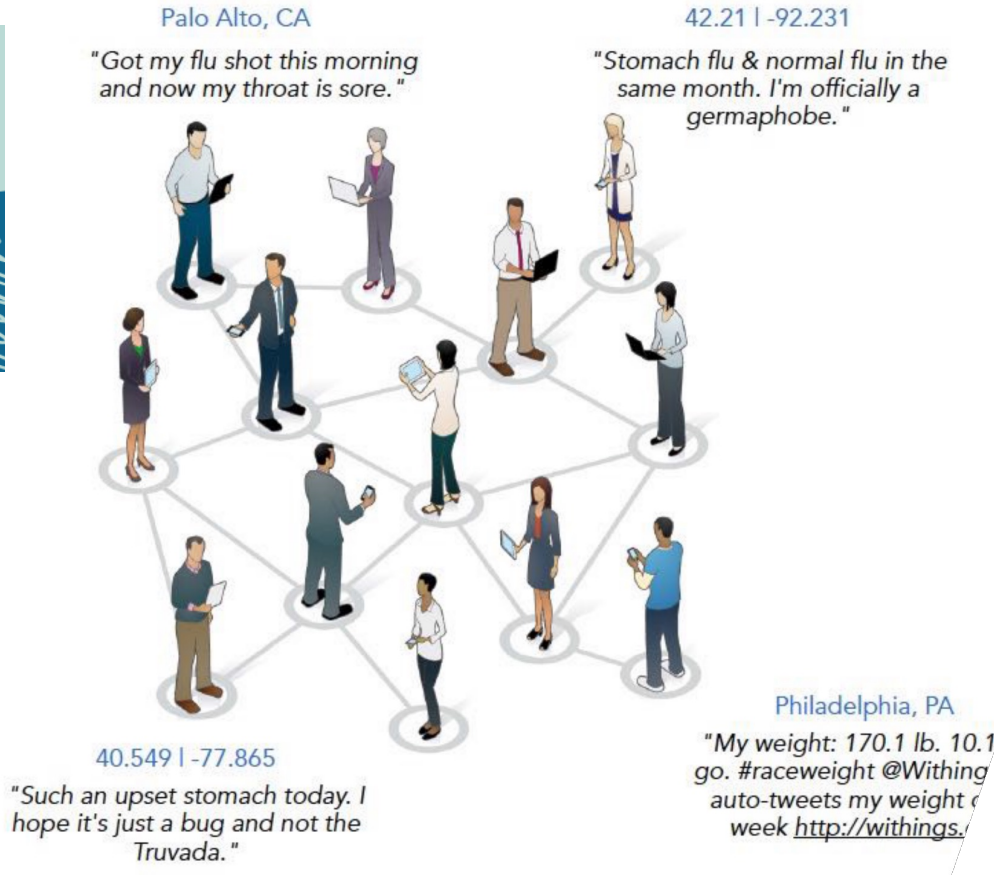
Travels

Social interactions

Vital parameters

Big data: everything you share

- Text
- Images
- GPS data
- Biological data
- Sounds
- Videos



Big data: all the things you are

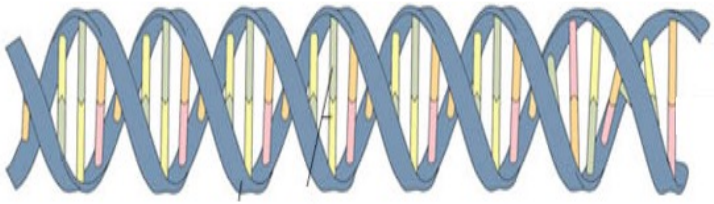
~40’000 billion ($4x10^{13}$) human cells

x25 →

~ 1 million billion (10^{15}) bacteria

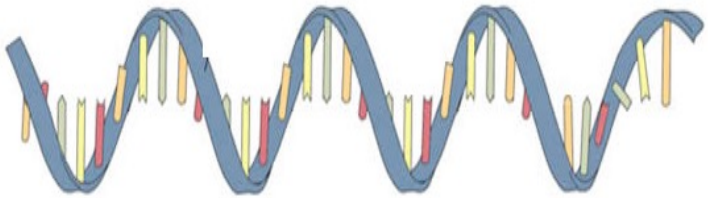
The central dogma

DNA ATGC



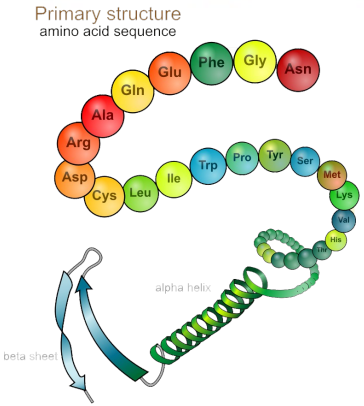
transcription

RNA AUGC

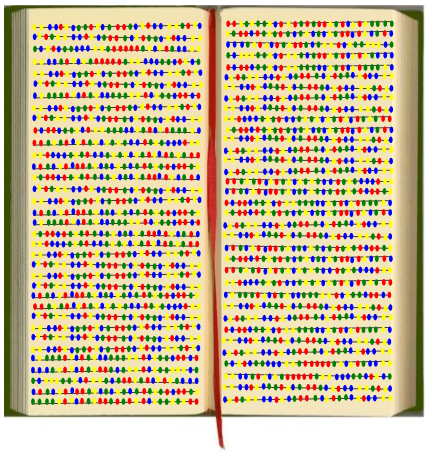


translation

Proteins 20 amino acids



My Genome



Only about 4Gb

A book of 2x3 billion characters (G, A, T, C)

~1000 “The Lord of the Rings”

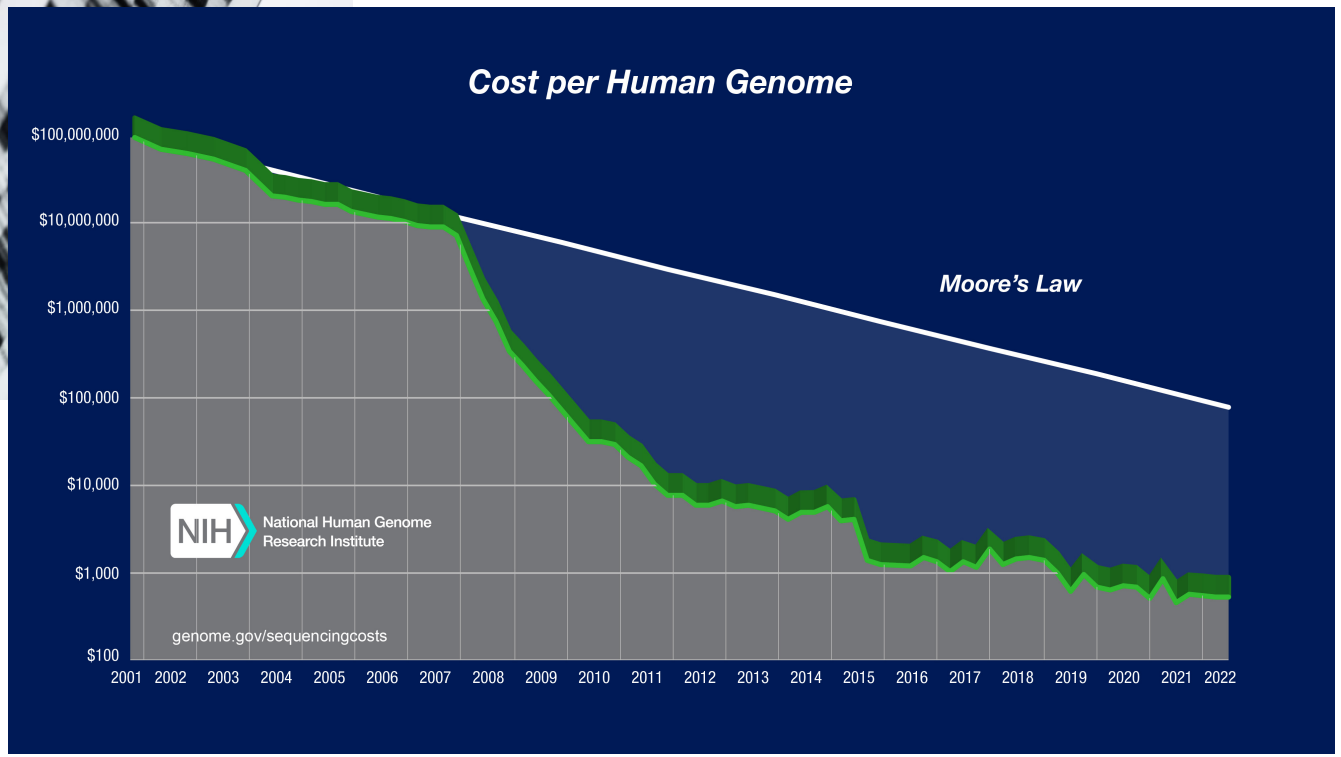
99.9% identical betwenn to human beings

99% identical to the chimpanzee genome

Genomics: the versatile pillar of precision medicine

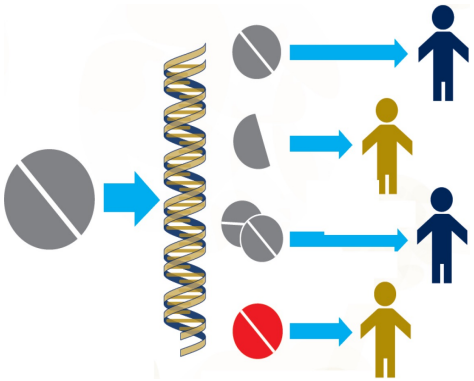
Genome | Epigenome | Transcriptome | Metagenome

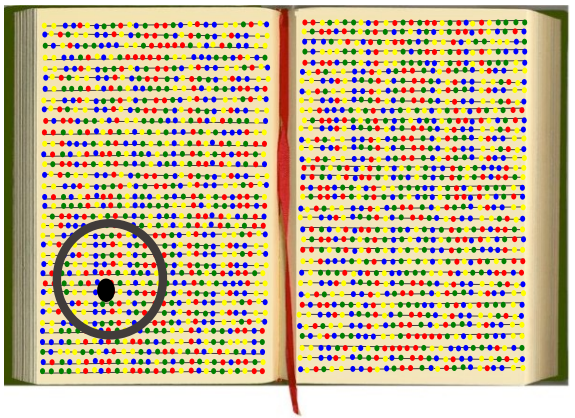
Genomics: now a cheap technolgy



Genomics: What for?

- Monogenic disorders
- Pharmacogenetics





- Simple or more complex risk factors

Genomics: What for?

Stratification of cancer therapy

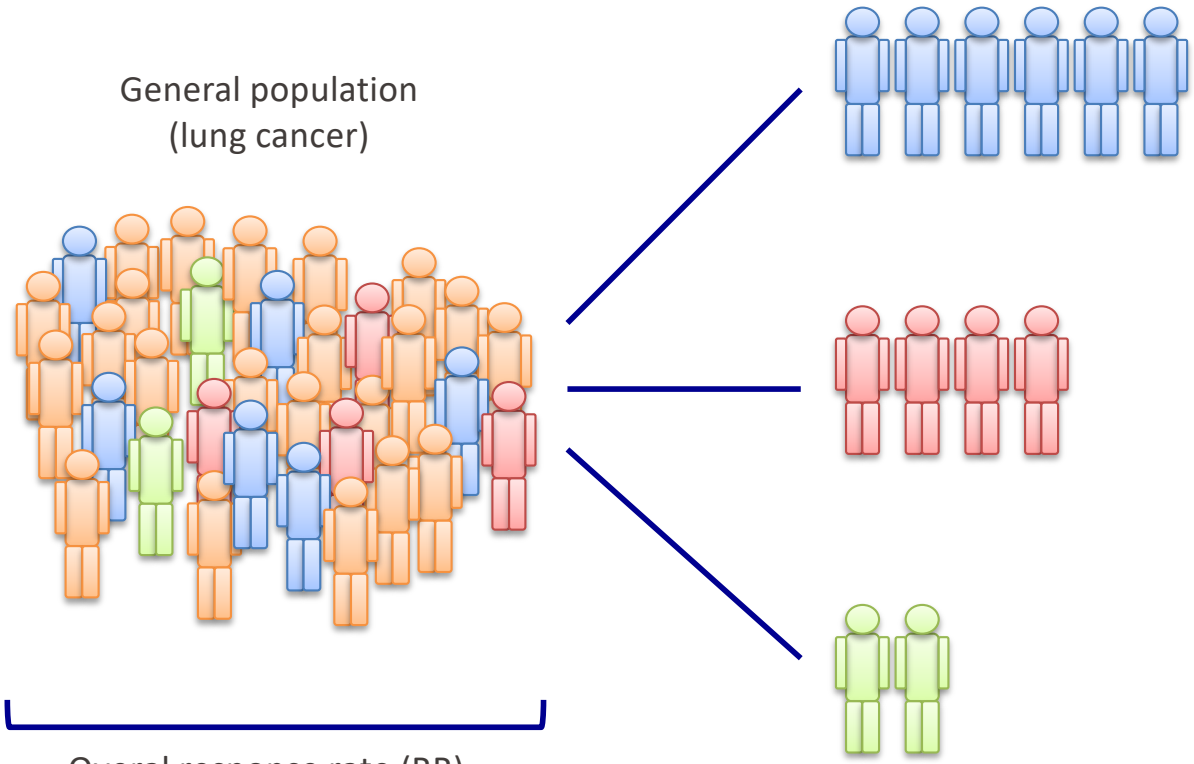
Treatment 1

Treatment 2

Treatment 3

Genomics for stratified cancer therapy

Stratification of cancer therapy



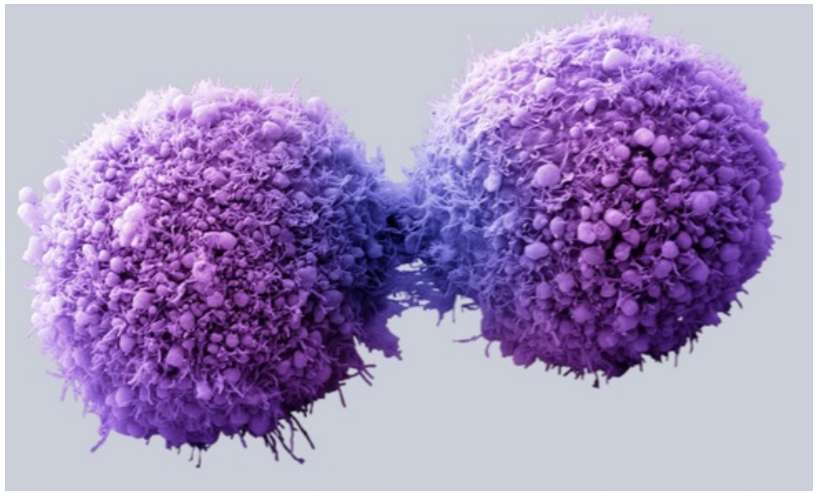
EGFR mutation (15%)
70% response rate to Erlotinib

ALK rearrangement (4%)
80% response rate to Crizotinib

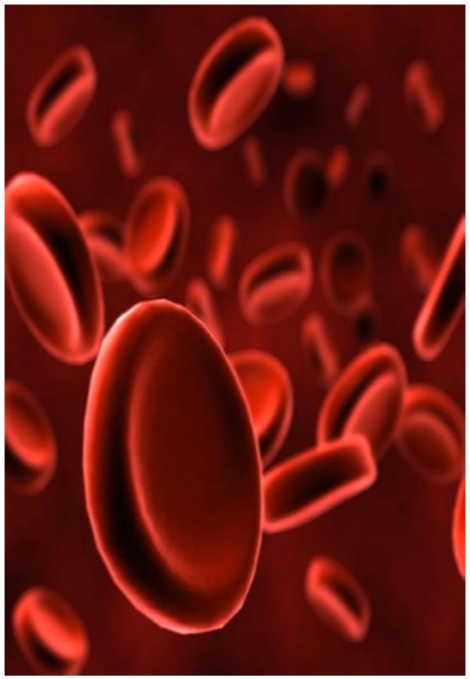
$V^{600}E$ BRAF mutation (2%)
40% response rate to Vemurafenib

Genomics and cancer: other applications

- Identification of genetic risk factors
- Staging of primary tumours
- Characterization of metastases
- Discovery of tumour antigens
- Monitoring of treatments



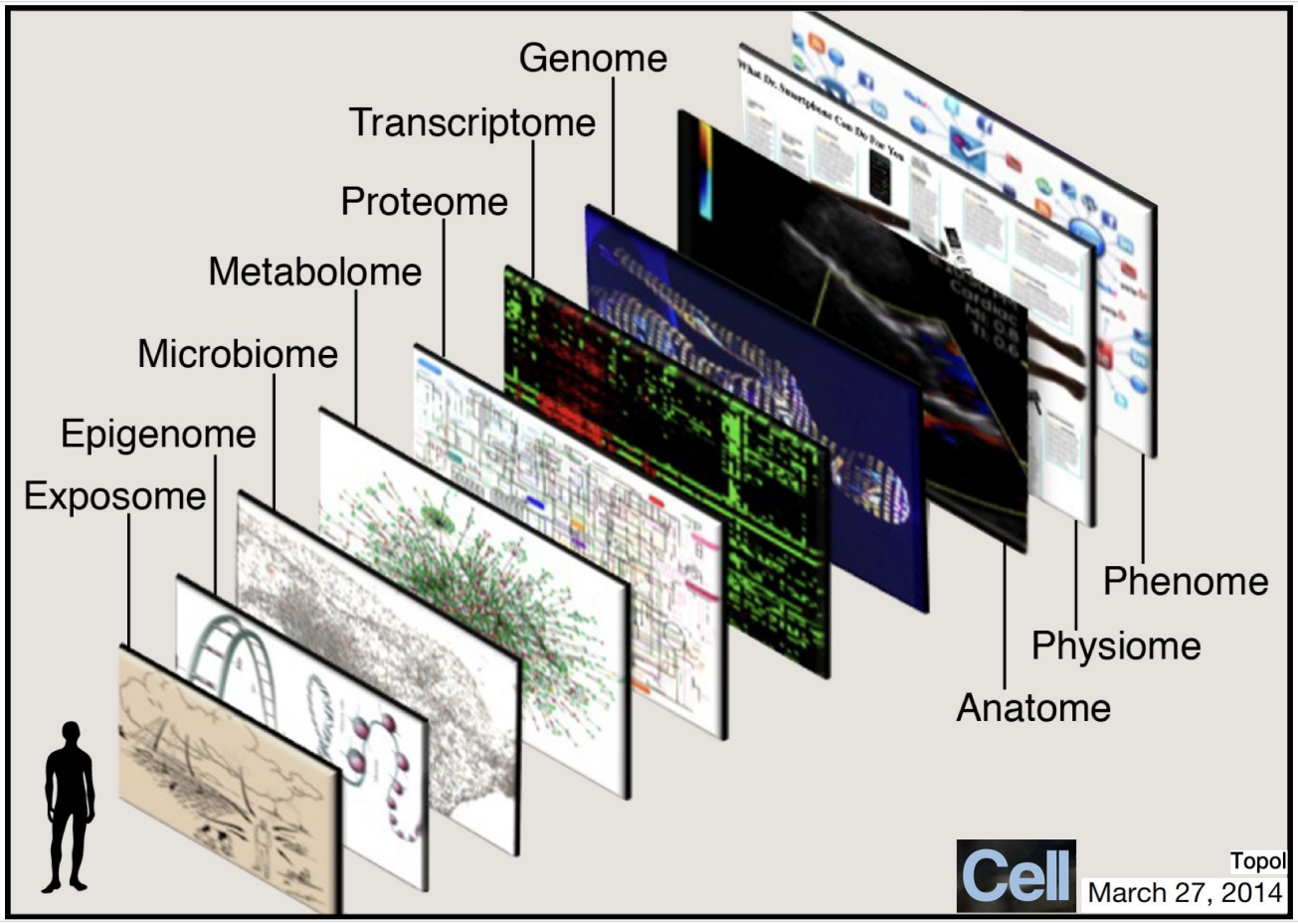
Other “omics”



Proteomics

Metabolomics

“Personomics”



Topol, E. Cell 2014 Mar 27;157(1):241-53.

E-health, m-health, electronic patient record, etc.

More than 300,000 health-related apps are available through main providers
Examples:
- Apps used for diagnostic purposes (e.g. heart rhythm analysis)
- Apps that operate a medical device (e.g. volume adjustment for a hearing aid)
- Apps that are used for specific and individual evaluation of patient data and provide therapeutic suggestions (e.g. contraception calendar with individual display)
- Apps that calculate a medication dosage (e.g. suggestions for corrective insulin)

Potential issues: usefulness, cybersecurity, data protection, interoperability, etc.

National surveillance: www.e-health-suisse.ch

Electronic patient record is progressively introduced in Switzerland:

- compulsory for hospitals, nursing homes and newly established medical practices since beginning of 2022
- still optional for other private practitioners, pharmacists and home care services

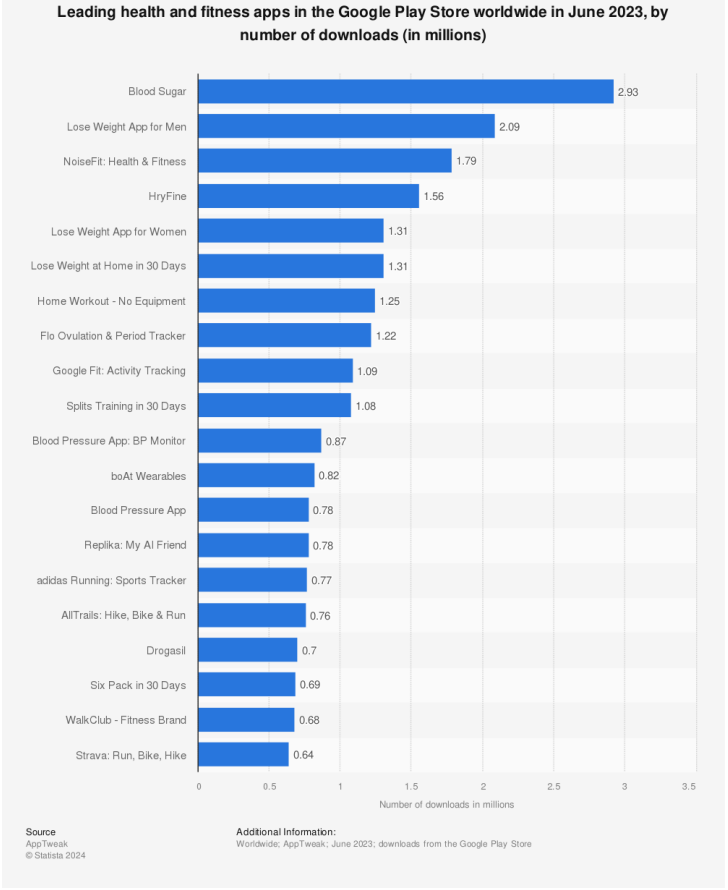
The apps of health

Cell Metabolism
Clinical and Translational Report

A Smartphone App Reveals Erratic Diurnal Eating Patterns in Humans that Can Be Modulated for Health Benefits

Shubhroz Gill[1] and Satchidananda Panda[1,*]
[1]Regulatory Biology Laboratory, The Salk Institute for Biological Studies, La Jolla, CA 92037, USA
*Correspondence: satchin@salk.edu
http://dx.doi.org/10.1016/j.cmet.2015.09.005

STRAVA



AI's Role in Precision Medicine

- - AI enables fast and accurate analysis of vast medical datasets
- - Helps in disease diagnosis, treatment recommendations, and personalized therapies
- - Large Language Models (LLMs) like GPT-4 or Med-PaLM 2 assist with medical question answering
- - AI can complement clinical decision-making but still requires validation and oversight

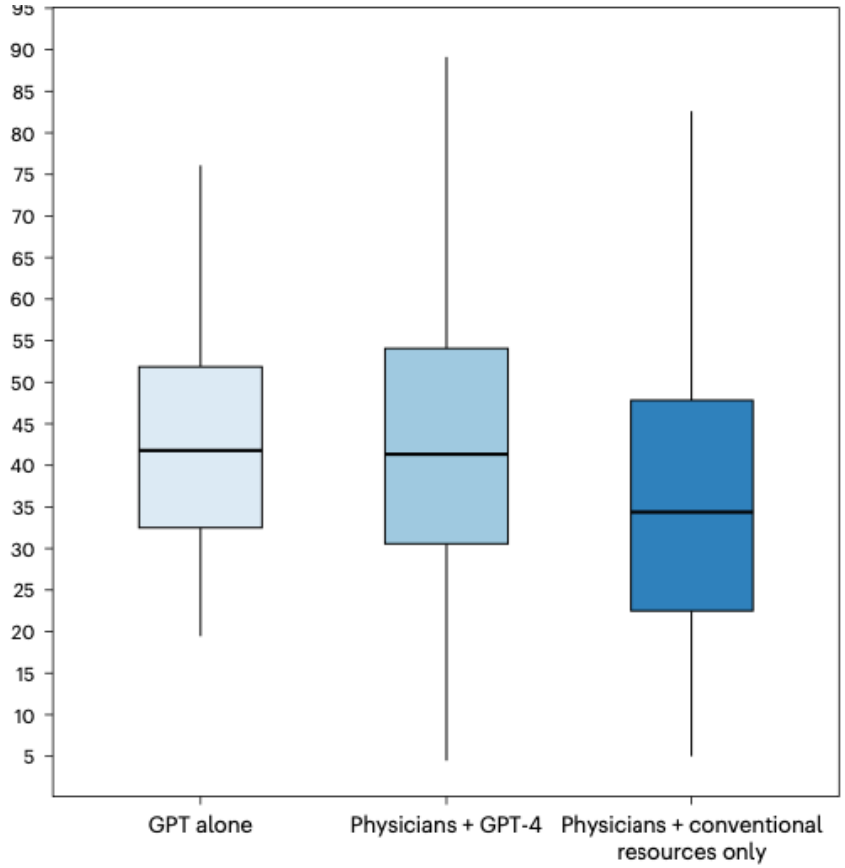
🤖 Will AI Replace Doctors?

- ❌ No, LLMs cannot replace medical doctors, but they will assist them.

- ✅ **What AI Can Do Well:**
- - Medical Knowledge Retrieval & Decision Support
- - Automating Documentation & Patient Education
- - Drug Discovery & Genomic Analysis

- ❌ **What AI Cannot Do:**
- - Perform Physical Exams & Clinical Judgment
- - Handle Unstructured, Rare Cases
- - Provide Personalized Human Interaction

Will it be safe for doctors to NOT use AI?



nature medicine

Article

https://doi.org/10.1038/s41591-024-03456-y

GPT-4 assistance for improvement of physician performance on patient care tasks: a randomized controlled trial

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Check for updates

A list of authors and their affiliations appears at the end of the paper

While large language models (LLMs) have shown promise in diagnostic reasoning, their impact on management reasoning, which involves balancing treatment decisions and testing strategies while managing risk, is unknown. This prospective, randomized, controlled trial assessed whether LLM assistance improves physician performance on open-ended management reasoning tasks compared to conventional resources. From November 2023 to April 2024, 92 practicing physicians were randomized to use either GPT-4 plus conventional resources or conventional resources alone to answer five expert-developed clinical vignettes in a simulated setting. All cases were based on real, de-identified patient encounters, with information revealed sequentially to mirror the nature of clinical environments. The primary outcome was the difference in total score between groups on expert-developed scoring rubrics. Secondary outcomes included domain-specific scores and time spent per case. Physicians using the LLM scored significantly higher compared to those using conventional resources (mean difference = 6.5%, 95% confidence interval (CI) = 2.7 to 10.2, $P < 0.001$). LLM users spent more time per case (mean difference = 119.3 s, 95% CI = 17.4 to 221.2, $P = 0.02$). There was no significant difference between LLM-augmented physicians and LLM alone (−0.9%, 95% CI = −9.0 to 7.2, $P = 0.8$). LLM assistance can improve physician management reasoning in complex clinical vignettes compared to conventional resources and should be validated in real clinical practice. ClinicalTrials.gov registration: NCT06208423.

Large language models (LLMs) show considerable abilities in diagnostic reasoning, outperforming previous artificial intelligence (AI) models and human physicians in their ability to construct helpful differential diagnoses, explain reasoning and collect historical information from standardized patients[1–3]. LLMs have not yet been shown to perform similarly in management reasoning, which encompasses decision-making around treatment, testing, patient preferences, social determinants of health and cost-conscious care, all while managing risk[4–8].

While there is overlap, clinical reasoning is often considered to include both diagnostic and management reasoning. The study of diagnostic reasoning has a century-long history with many metacognitive frameworks and assessment methods, while management reasoning processes are a comparatively recent area of study[9–11]. Current frameworks in management reasoning include context-dependent concepts, such as shared decision-making, dynamic relationships and competing priorities between medical systems and individuals, the physician–patient relationship and time constraints inherent in modern clinical encounters[9,12–14]. Unlike diagnostic reasoning, which can be thought of as a classification task with often a single right answer, management reasoning may have no right answers and involves weighing trade-offs between inherently risky courses of action; even inaction through 'watchful waiting' is a deliberate choice with potential risks

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The Future of Medical AI

- Multimodal AI: Integrating genomics, imaging, and clinical text
- Specialty-Specific LLMs: Fine-tuned models for cardiology, oncology, etc.
- Clinical Integration: Embedding AI into electronic health records (EHRs)

🏥 Future of AI + Human Collaboration

- 🤖 AI as an Assistant: Helps analyze data, suggest treatments, and detect errors
- 👨‍⚕️ Doctors as Decision-Makers: Use AI insights but apply ethics & clinical judgment
- 🔬 AI Augmenting Specialties: Useful in radiology, pathology, genomics, and documentation
- ⚖️ Ethical, regulation & Safety Concerns:
  - Bias in AI Models
  - Hallucinations & Errors
  - Patient Trust & Data Privacy

conflAnce: AI-Powered Medical Chatbot by HUG

Enhancing Access to Reliable Medical Information

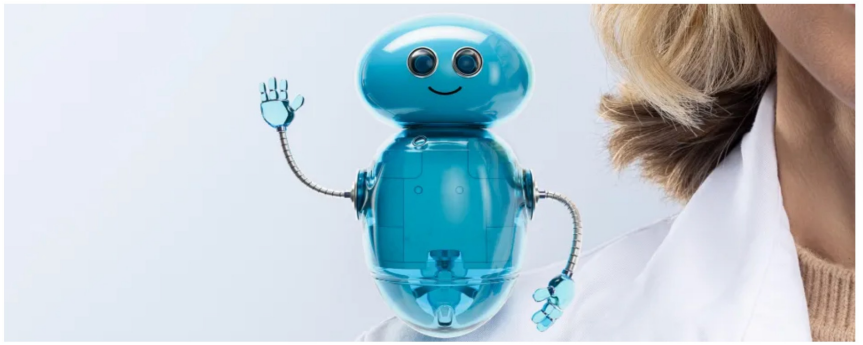
- **First AI-powered general medicine chatbot in Switzerland**
- Provides **reliable, verified medical information** based on the HUG Primary Care Service (SMPR)
- Available **24/7** for both patients and doctors
- Answers questions on **30+ chronic conditions** (e.g., anemia, depression, hay fever)
- Uses **Large Language Model (LLM) technology**, ensuring **data privacy & anonymity**
- **Not a diagnostic tool** but supports informed discussions with doctors

- **Addresses physician shortages & long wait times**
- Helps patients find **trustworthy medical information** instead of unreliable online sources
- **Supports ongoing patient-doctor communication** between consultations
- Enhances accessibility for **multilingual users**
- Improves **health literacy** by making complex medical content more understandable

Hôpitaux Universitaires Genève

COMMUNIQUÉ DE PRESSE

« conflAnce », le chatbot d'information médicale générale, une première suisse



Lundi 10 février 2025

chatbot conflAnce

Meanwhile in the USA

Healthy Technology Act of 2025: AI Prescribing Bill Introduced in Congress

The year is 2030, you have been in the doctor's office for two hours, answering what seems like hundreds of questions on their augmented reality computer. You are excited about meeting your new doctor. Finally, the office staff robot announces, "the computer will see you now." Your initial reaction is, "What!?!" The private equity firm that owns the practice has apparently just bought the recently-released HAL 35 computers to replace your retiring physician.

How did we get here? In a move that could reshape the landscape of medical practice, Representative David Schweikert has introduced the Healthy Technology Act of 2025 in the United States House of Representatives. This legislation aims to amend the Federal Food, Drug, and Cosmetic Act to allow artificial intelligence (AI) and machine learning technologies to qualify as practitioners eligible to prescribe drugs, under certain conditions.

Key Provisions

H.R. 238, initially introduced on January 7, 2025, proposes that AI systems could be authorized to prescribe medications if: (1) the AI is approved, cleared, or authorized by the Food and Drug Administration (FDA) and (2) the state in which the AI operates authorizes its use for prescribing medication.

This bill represents a significant step towards integrating AI into core medical practices, potentially transforming how medications are prescribed and managed.

Previous Congressional Efforts

Importantly, this is not the first time that Congress has considered the role of AI in healthcare. In fact, Representative Schweikert introduced similar legislation in the 118th Congress and it died in committee with no discussion.

Arguments in Favor

- reduce medication errors and improve patient safety,
- enhance efficiency in healthcare delivery,
- provide more personalized treatment plans based on vast data analysis
- alleviate physician burnout by automating routine tasks.

Arguments Against

- loss of human judgment in medical decision-making
- data privacy and security risks
- increased potential for fraud with manipulated

AI writing scripts

- possibility of AI perpetuating or exacerbating existing biases in healthcare, and liability issues in cases of AI-related errors.

Personalized health: the international context

Programs in precision medicine / personalized health launched in the UK, USA, Denmark, etc…

A new field, with many trials and errors, a good sharing spirit in the public sector and a strong interest in the private sector (e.g. GAFAM et al.)

Switzerland has **strengths**:

size, federal organization, intra-national differences, quality of medical care, excellence of research, track record with cohorts, support of population to biomedicine, wealth, etc.

Switzerland has **weaknesses**:

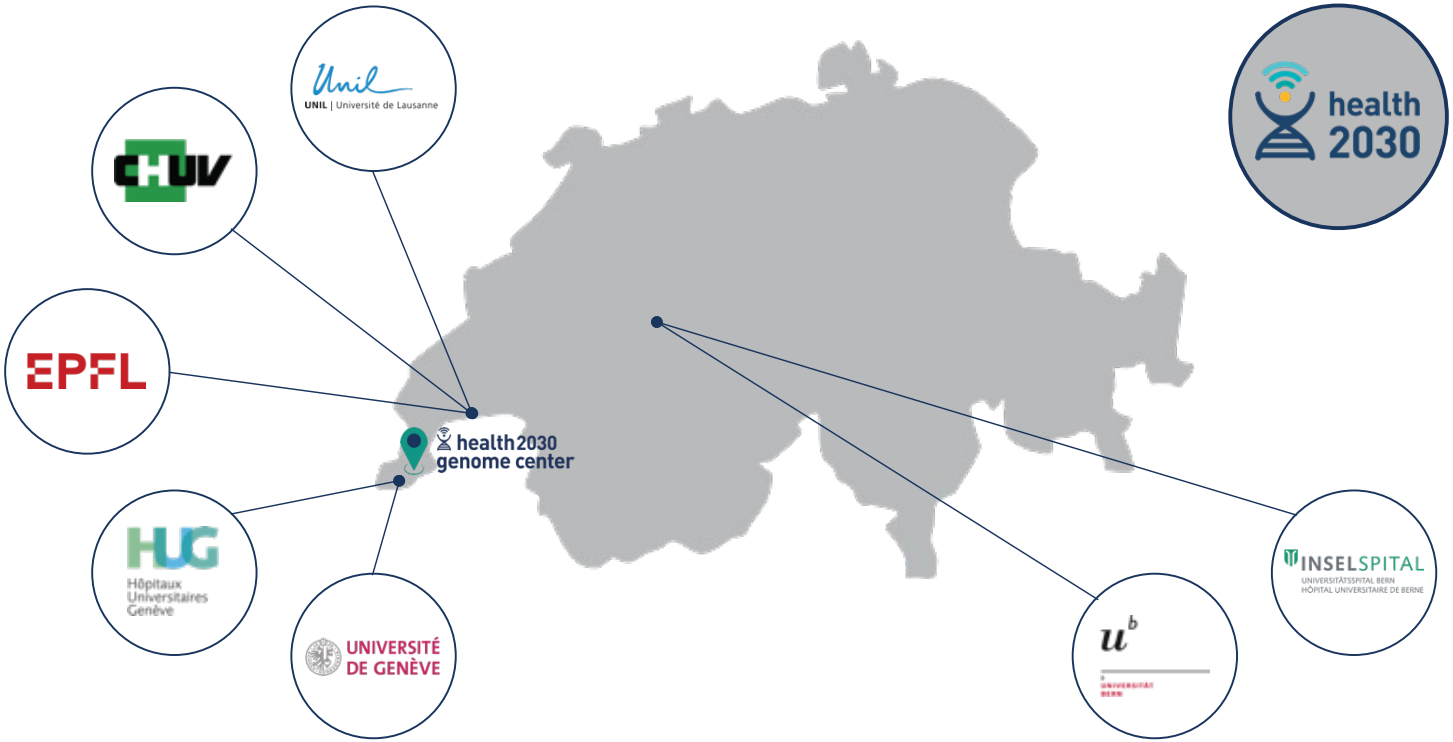
size, federal organization, intra-national differences, splitting of health and research/innovation/education between different federal and between different federal and cantonal departments, late- joining tendency, indigent state of digitalization

Personalized health: Swiss national programs 2017-2024

SPHN
SERI, SNF, ASSM
120 Million for hospitals and universitites

PHRT
ETH domain
100 Million for hospitals and universitites

Personalized health: Western Switzerland initiative



- A multi-institutional initiative to exploit the potential of new technologies “Omics and Big data” for the field of health

Swiss genomic medicine: the Health 2030 Genome Center

health2030 genome center

A Hub for developing genomic medicine in Switzerland

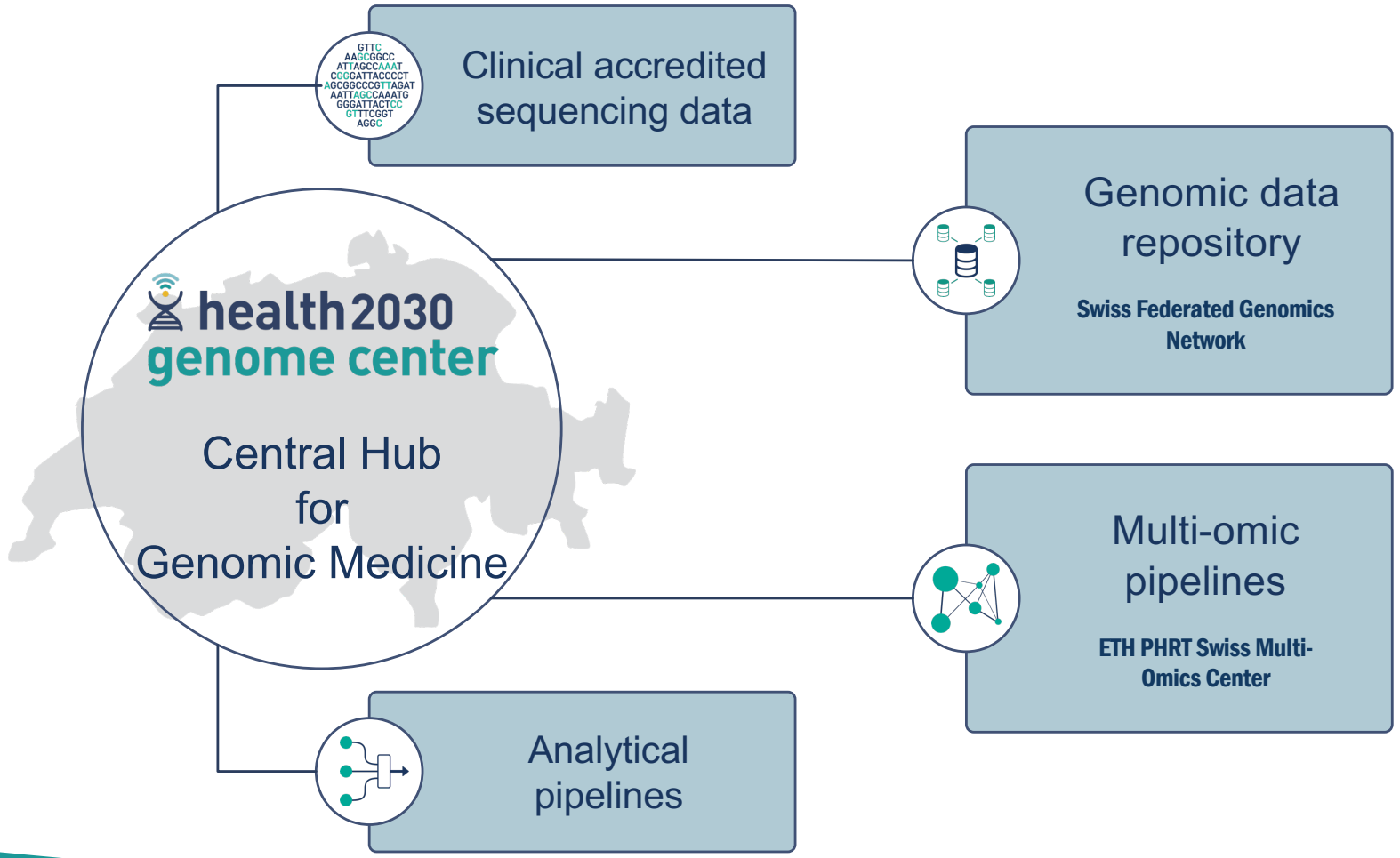
Health 2030 Initiative >> Health 2030 Genome Center

- The goal of Health 2030 Initiative is to explore and exploit the potential of new technologies in the fields of health and personalized medicine and has the partners: EPFL, HUG, UNIGE, CHUV, UNIL, InselSpital and UNIBE

>>*https://health2030.ch/*

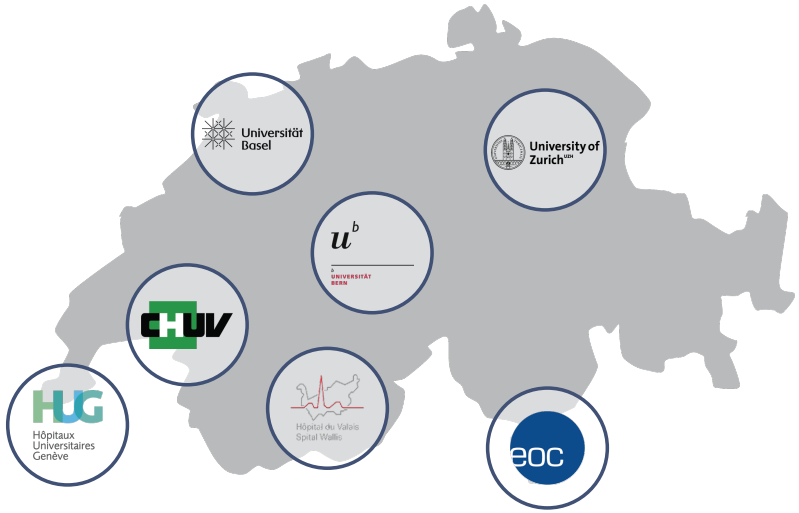
- The Health 2030 Genome Center is the **genomic medicine arm** of the Health 2030 initiative, but serves the **entire** Swiss community

>> *https://www.health2030genome.ch/*

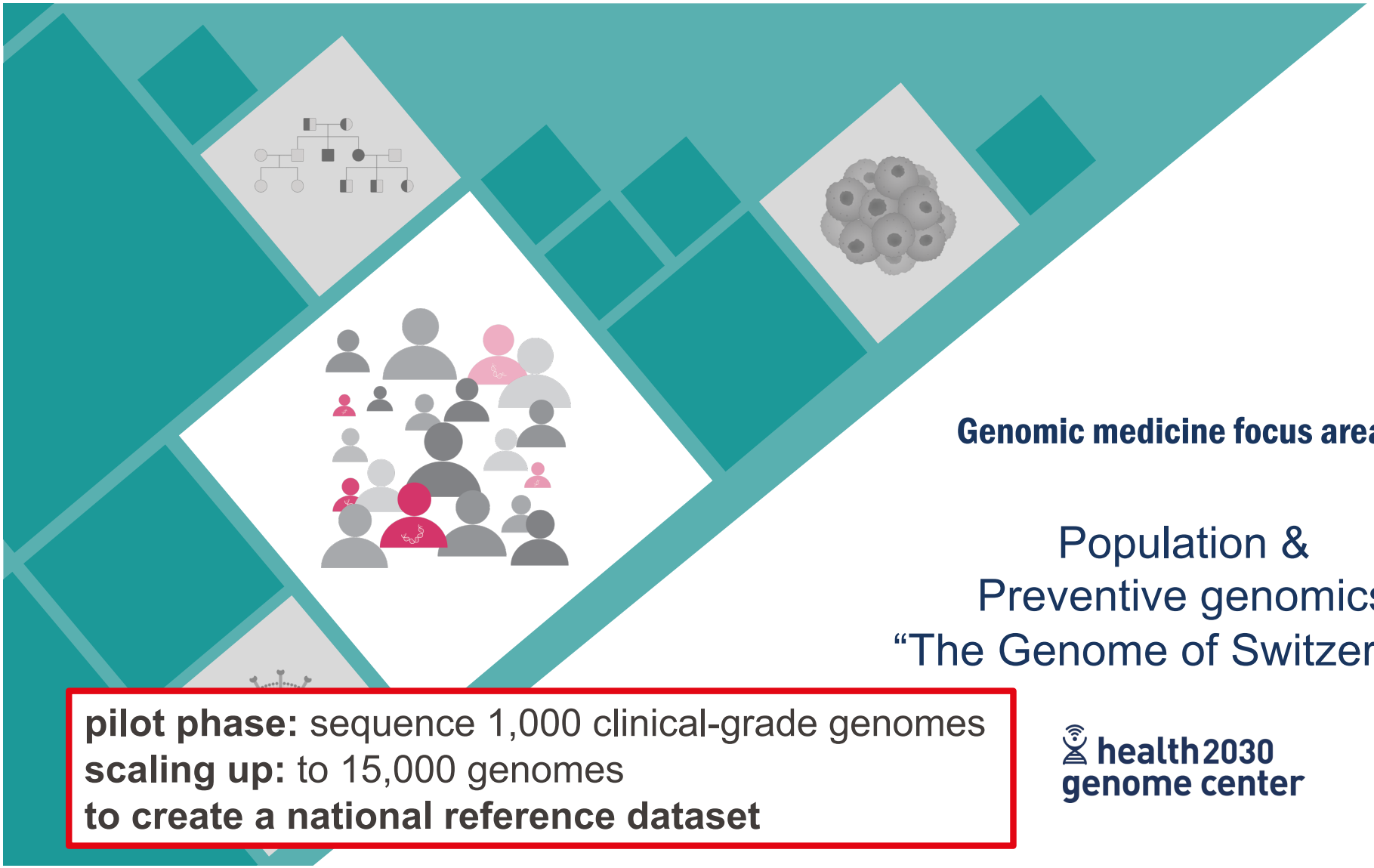

Clinical accredited sequencing data
Genomic data repository
Swiss Federated Genomics Network
health2030 genome center
Central Hub for Genomic Medicine
Multi-omic pipelines
ETH PHRT Swiss Multi-Omics Center
Analytical pipelines

Swiss National SARS-CoV-2 Surveillance Program

- Phase 1: March 2021-March 2022
  Roughly 32% of nation-wide samples sequenced in the Genome Center
- Phase 2: April 2022-December 2022
  Roughly 44% of nation-wide samples sequenced in the Genome Center
- Phase 3: January 2023–December 2023
  100% nation-wide samples sequenced in the Genome Center
- Phase 4: January 2024–
  100% nation-wide samples sequenced in the Genome Center



health2030 genome center



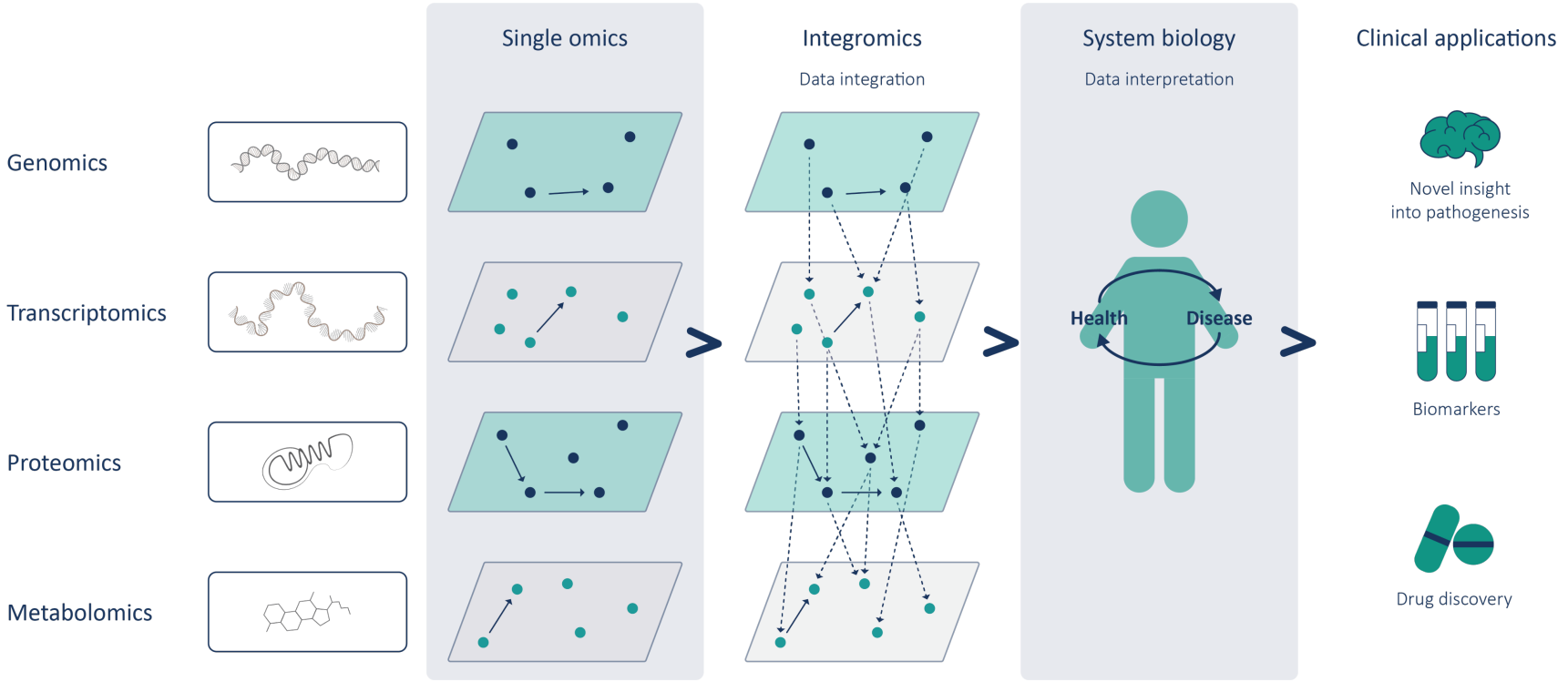
Genomic medicine focus areas:

Population &
Preventive genomics
“The Genome of Switzerland”

pilot phase: sequence 1,000 clinical-grade genomes
scaling up: to 15,000 genomes
to create a national reference dataset

health2030
genome center

Personalized Health: Swiss Multiomic Center: Integration of Swiss-scale clinical and molecular data



health2030 genome center

ETH BOARD

Strategic Focus Area Personalized Health and Related Technologies

Adapted from DOI: 10.1038/s41582-020-0350-6

Meanwhile, across the world

- United Kingdom:

As of 30 November 2023, the UK Biobank has unveiled data from whole genome sequencing of its half a million participants.




- United States:

'All of Us' Research Program (NIH) aims for 1 million genomes.

- Japan:

Development of the JG1 (Japanese reference genome)

- United Arab Emirates:

The Emirati Genome Programme, using AI for genomic data.
350’000 human genomes


->Personalized and preventive healthcare.
->Personalized genetic research
->Enhancing clinical analysis for rare diseases.

Personalized health: economic opportunites

Acceleration of discovery of new biomarkers and therapeutic targets

Reduction of health cost by disease prevention and treatment stratification

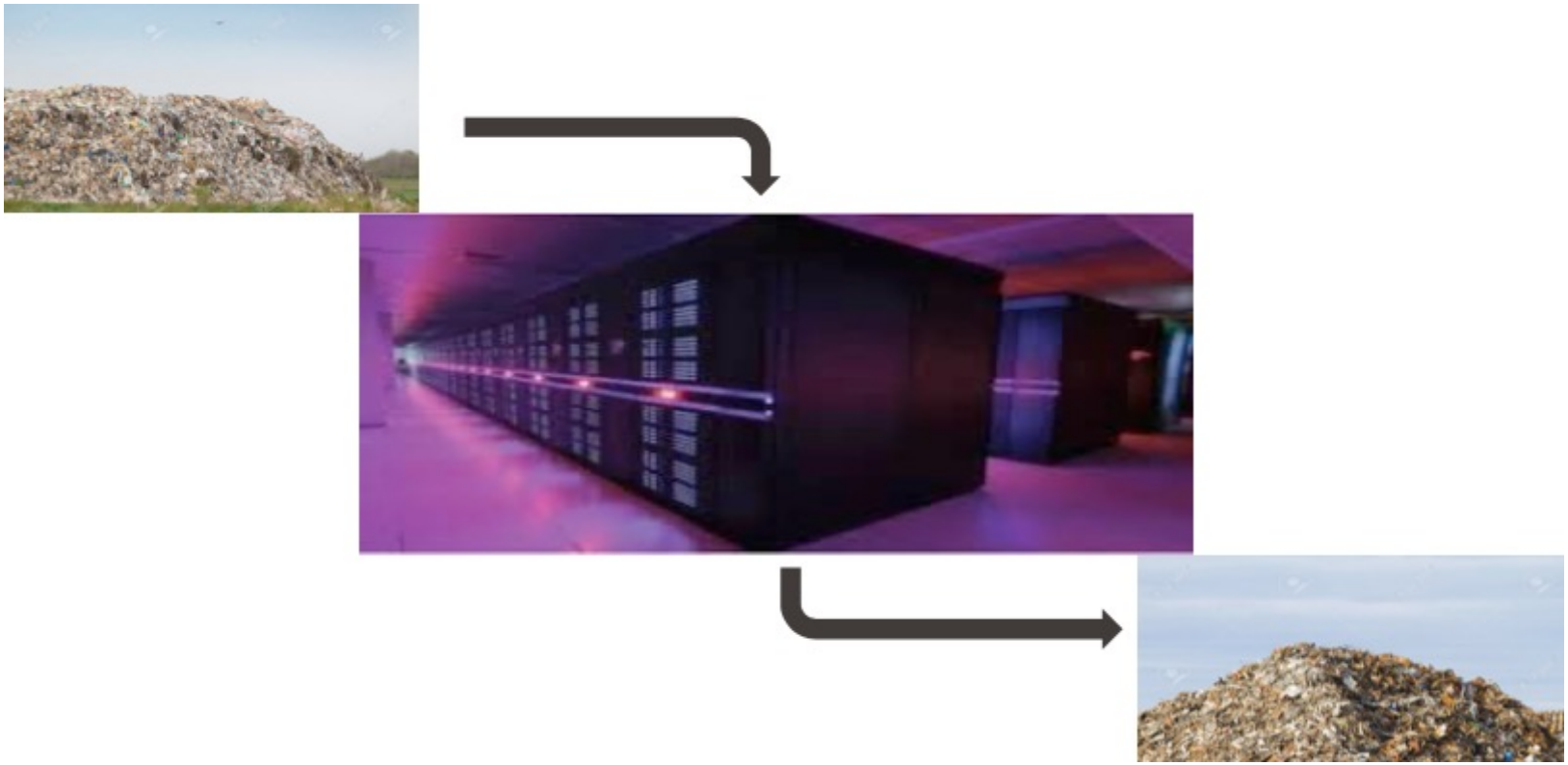
Development of a new industry based on:

- “wellness”
- Impact assessment
- Digitalization, storage and interpretation of health-related data.
- “Theranostics”
- Biosensors and other techniques of personal or environmental monitoring

Marc Friedli

Personalized health: technlogical challenges

Quality of data ("Rubbish in, rubbish out")



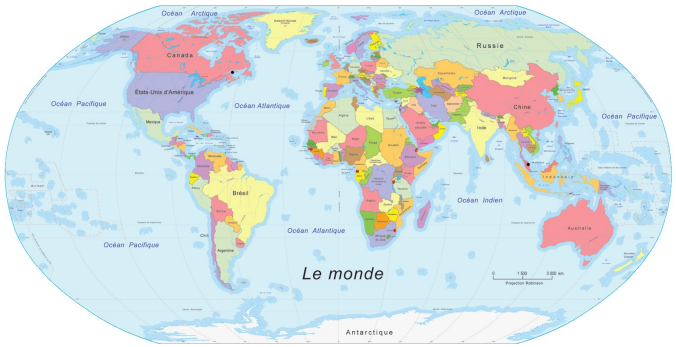
Semantics

Marc Friedli

Systems interoperabity

FAIR principles

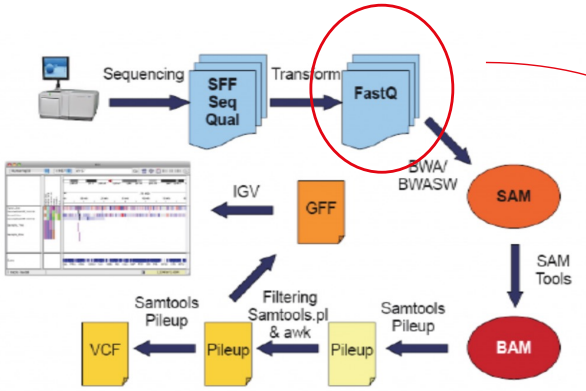
- Findable
- Accessible
- Interoperable
- Reusable



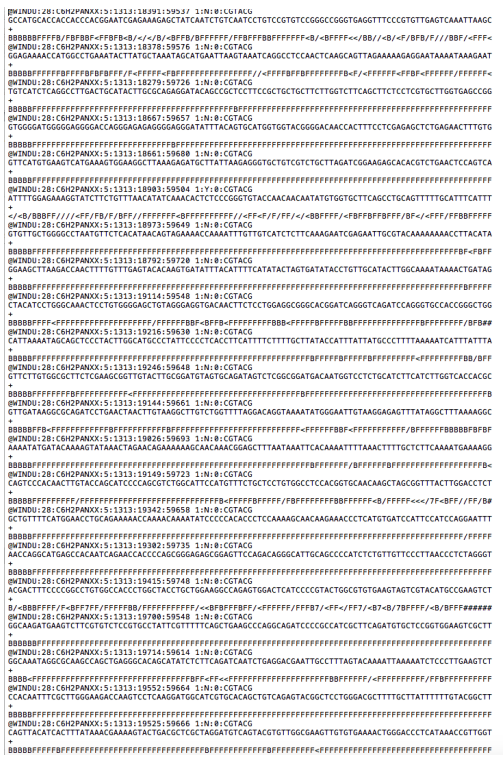
Marc Friedli

Storage, encryption, analysis, restitution of “smartified” data

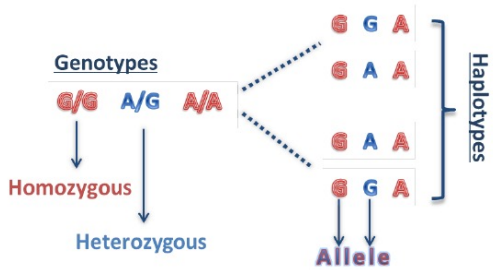
Sequencing data

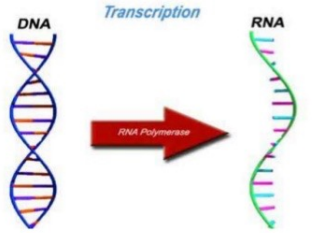


FastQ



200 million lines





DNA Transcription

Marc Friedli

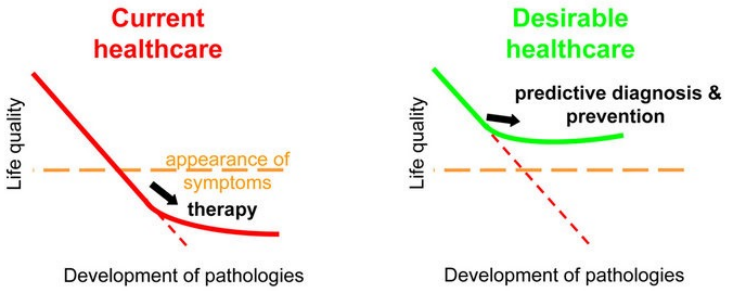
Appropriate use of biobanking

Biobanks are expensive

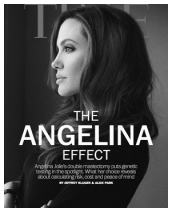
->ensure samples stored can be reused

Personalized Health: societal challenges

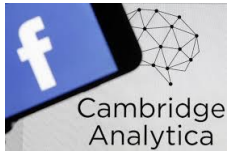
- Restructure health management, as one moves from disease treatment to **prevention**



- Avoid that our health care be controlled by large IT & internet companies (GAFAM & al.) and the governments that are behind them



- Discuss of **incidental genetic findings**, whether for individual or for relatives
- Protect privacy but **promote collectivization of data**
- Consolidate **anti-discrimination** laws



Personalized Health: a holistic endeavour

Communication

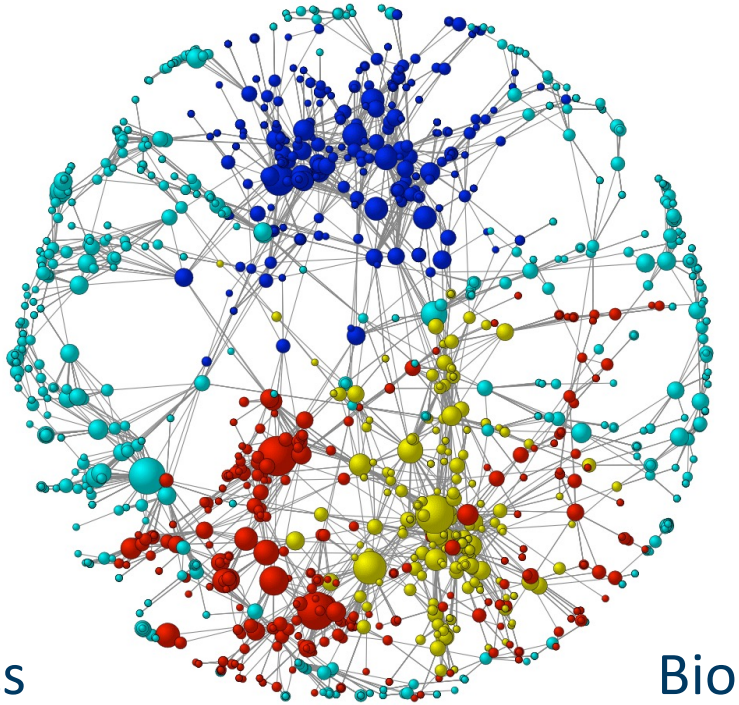
Education

E-health – M-health

Genomics

Other
“technolomics”

Big Data



Digital epidemiology

Digitalized
medical record

Bioinformatics

Biobanks

Ethics

Sociology

Law

Psychology

Health economics

Marc Friedli


Thank you
DNA is not our destiny; it's just a very useful tool
Ewan Birney
EPFL
health2030
marc.friedli@epfl.ch