FUTURE PERSPECTIVES

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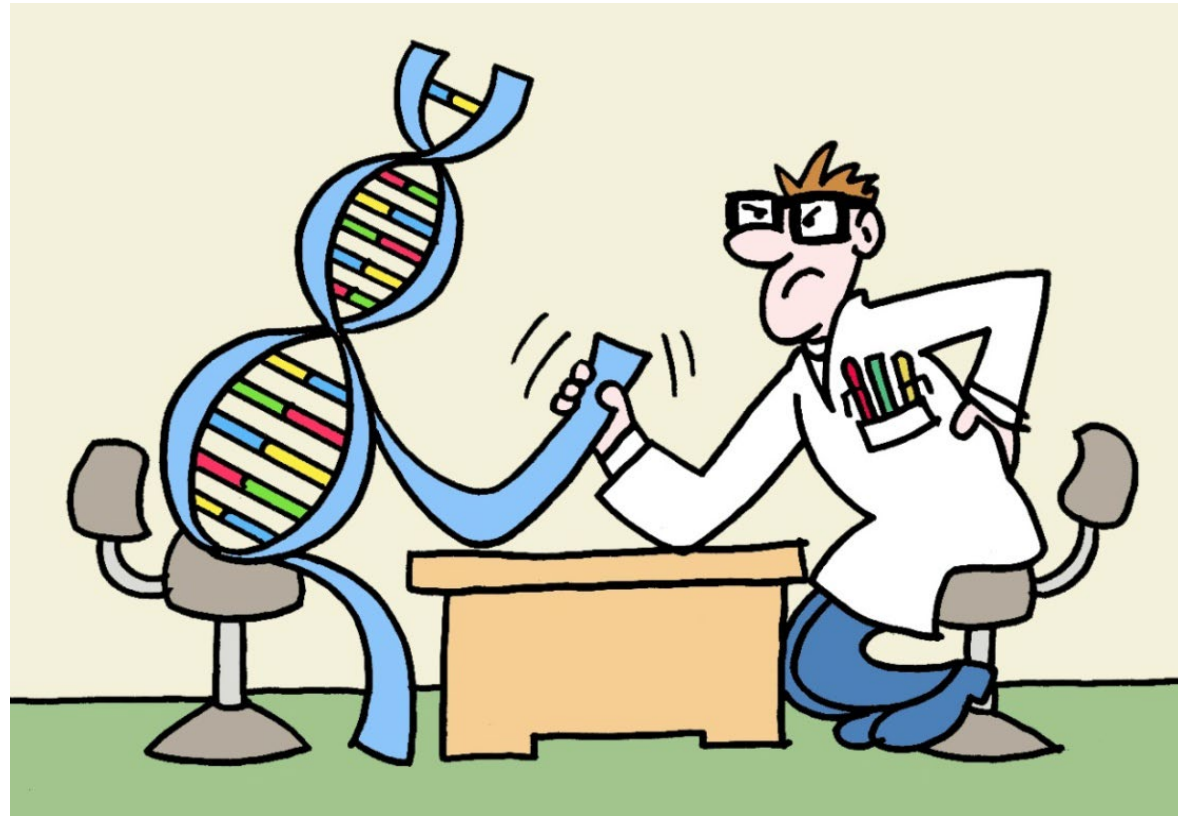
Universitair Ziekenhuis Brussel

CMG
UZ Brussel

40 jaar Centrum Medische Genetica UZ Brussel

VUB

DOCTORS VERSUS GENOMES

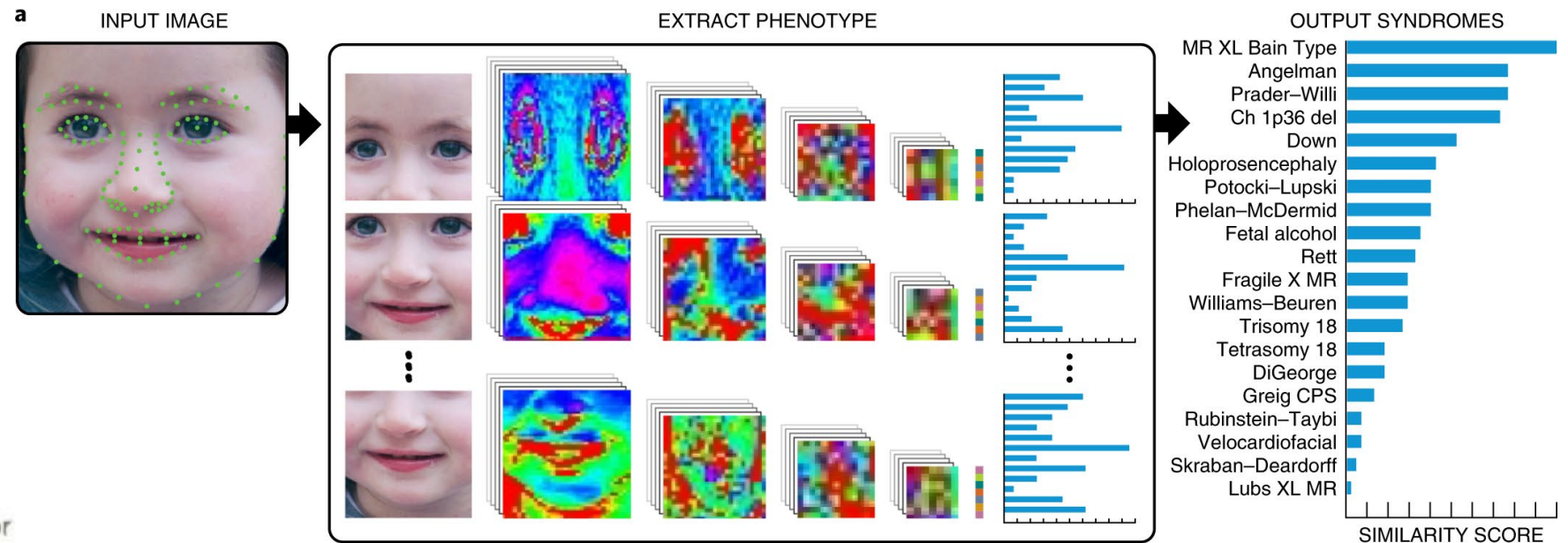in rare possibly genetic diseases

ARTIFICIAL INTELLIGENCE

Machine learning

Diagnose cancer from a liquid biopsy

Predict how cancer will progress

Score genomic variants

Face to gene
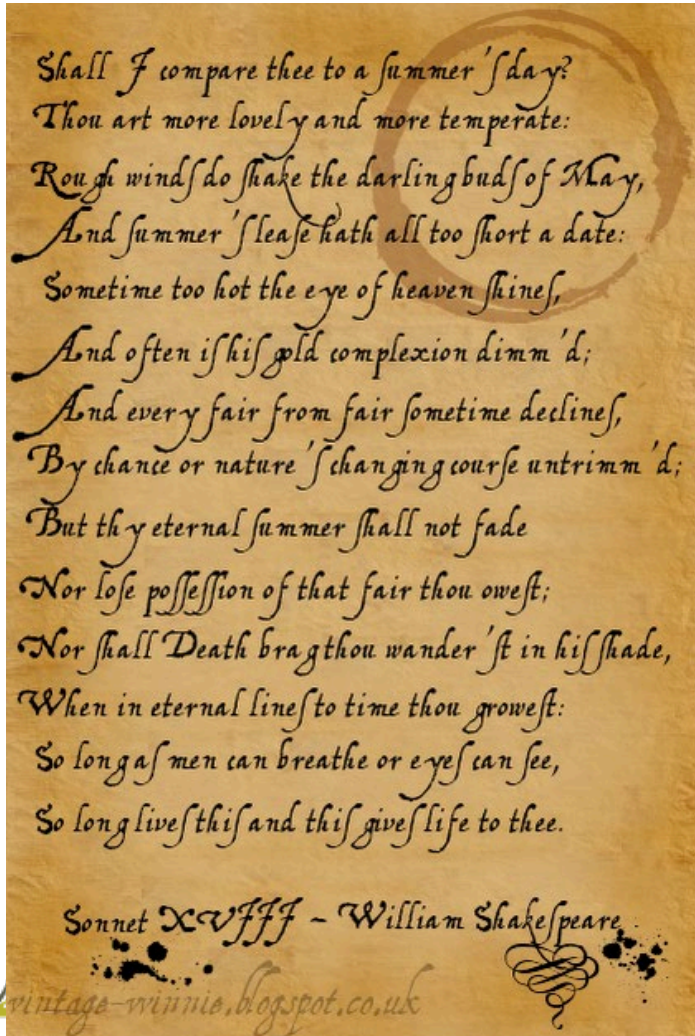
THE FUTURE OF DNA SEQUENCING

Green E, Rubin E, Olsen MV. Nature, 12 October 2017

"Surprises are a certainty: trends in DNA sequencing will be driven by killer applications, not by killer technologies."

FROM TEXT TO DNA CODES

TEXT TO BINARY CODE
Binary ones and zeroes represent the ASCII code for part of Shakespeare's *Sonnet 18*.

...1000100001010101110011110000001001100010001...

...Thou art more lovely and more...

BINARY TO TRIPLET CODE
The binary file is mathematically converted into 'trits': the zeroes, ones and twos of a three-digit code.
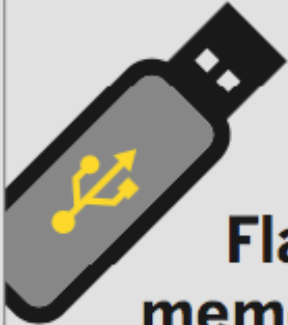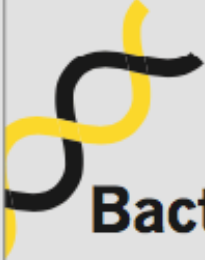
...2011220200002110100020221201112101010111022...

TRIPLETS TO DNA CODE
A synthesis machine creates strands of DNA using the trits as a guide. At each step, the next zero, one or two is translated to one of the three bases that differ from the base just used.

...TAGATGTGTACAGACTACGCGCAGCGAGATCGACTCGACT...

FROM CODE TO STORAGE

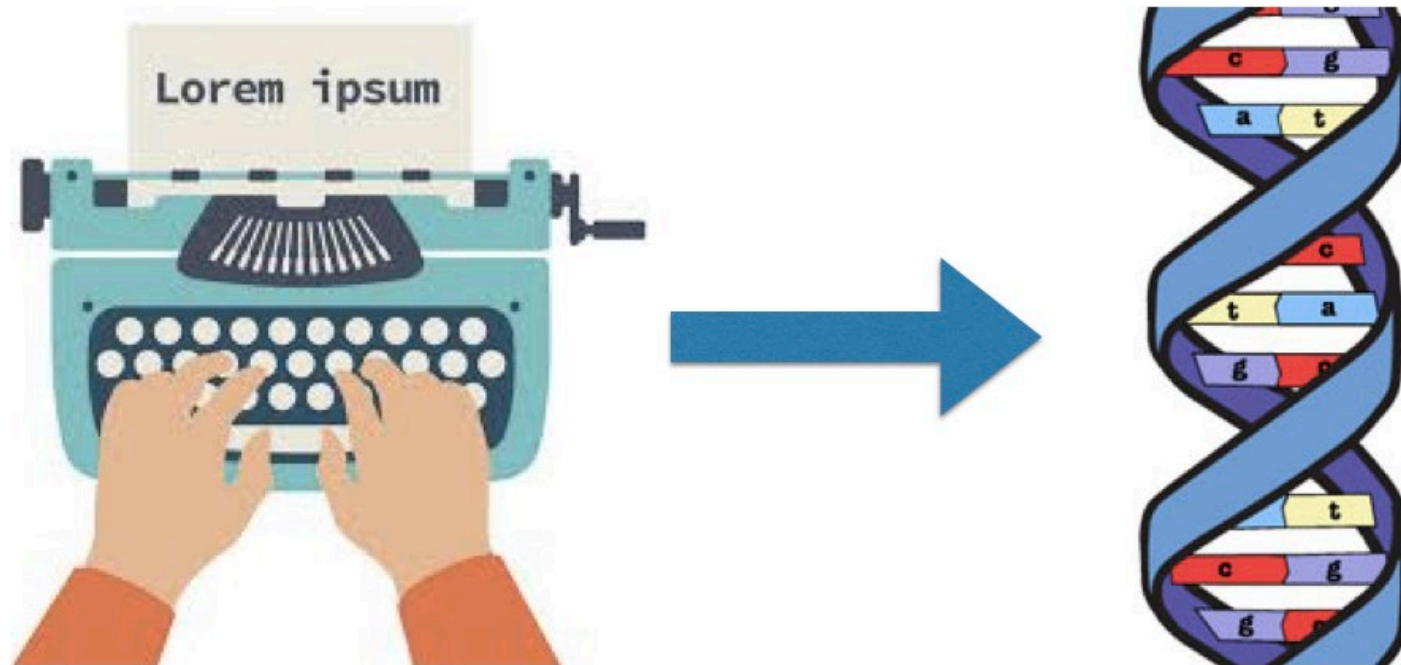|	Hard disk	Flash memory	Bacterial DNA
Read–write speed (μs per bit)	~3,000–5,000	~100	<100
Data retention (years)	>10	>10	>100
Power usage (watts per gigabyte)	~0.04	~0.01–0.04	$<10^{-10}$
Data density (bits per cm³)	$\sim 10^{13}$	$\sim 10^{16}$	$\sim 10^{19}$

DATA STORAGE

44 x 10^{21} bytes (2020) > 3 x 10^{24} bytes (2040)

DNA TYPEWRITER

Choi et al. Nature, 4 August 2022, Washington University

CRISPR-CAS9

Clustered Regularly Interspaced Short Palindromic Repeats

Opportunities | Challenges

Targeted medicines
Biofuels
Disease-resistant crops
New industrial products

Safety
Ethical concerns
Regulatory challenges

Source: GAO. | GAO-20-478SP

MOVING FROM DIAGNOSTICS TO CARE & CURE

- Making genomics truly equitable

- Genome sequencing at population scale

  - FAIR (findable, accessible, interoperable and reusable)

- Decoding multifactorial phenotypes

  - shift from reverse to forward genetics

- Long non-coding RNA's

  - 60,000 LncRNA's vs 20,000 protein coding genes

- Integrating genomics in medicine

Percent of human genome sequence released
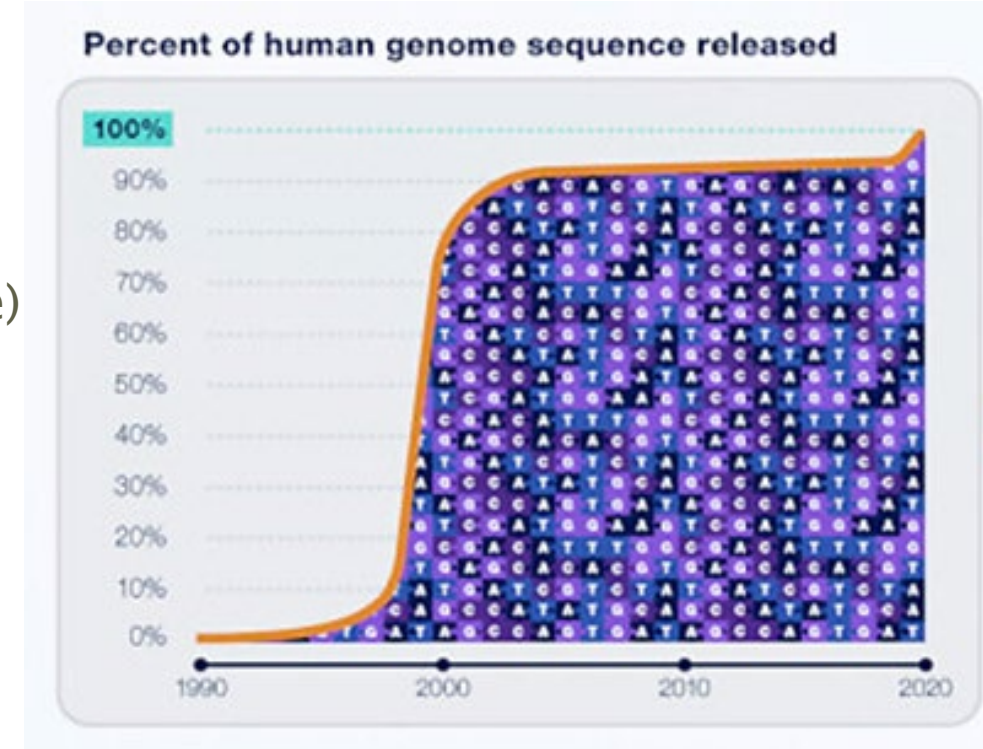
Nurk et al. The complete sequence of a human genome.
Science 31 March 2022

GENETICS FIRST

2020

2040

http://www.thetgmi.org/genetics/changing-direction-genetic-testing/
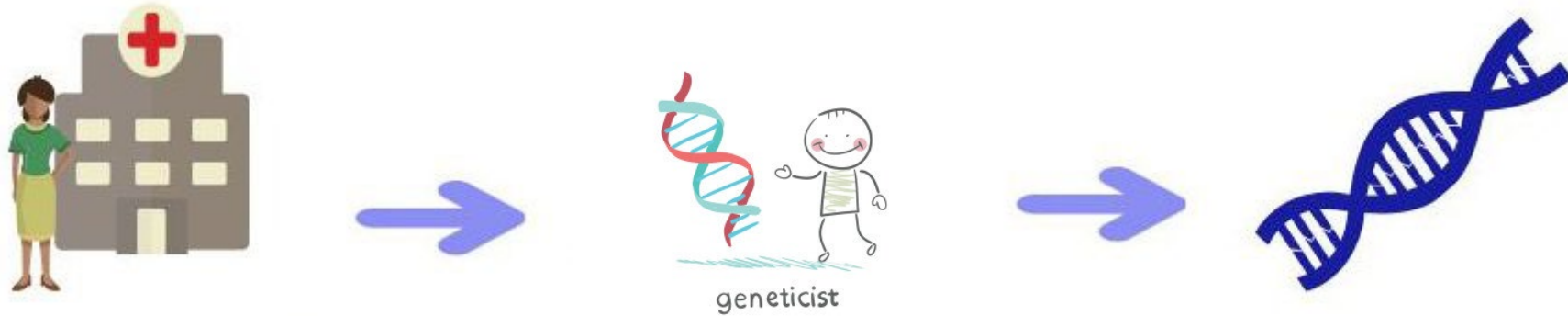
PARADIGM SHIFT
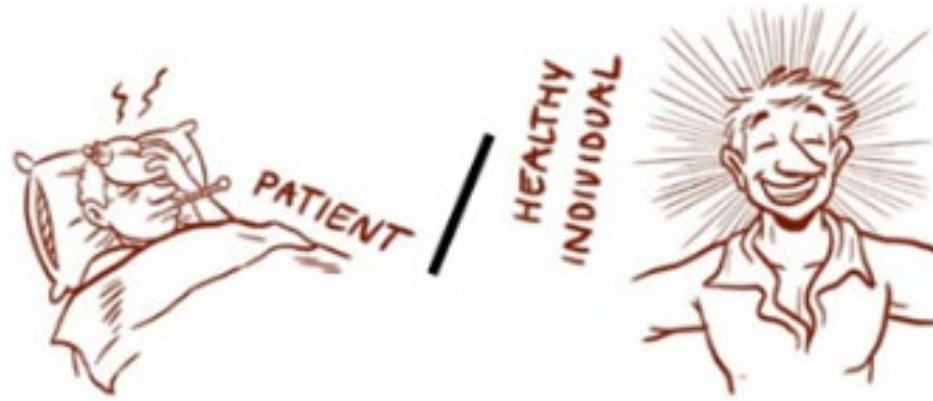
Go see the doctor	Cure / Prevention 80% / 20%	**2020**
Go see the patient	Cure / Prevention 50% / 50%	**2030**
Go check health data	Cure / Prevention 20% / 80%	**2040**

THE HOSPITAL OF THE FUTURE …

Is a network built around quality of care, organised in multidisciplinary care trajectories, with healthcare professionals, in and outside the hospital, and together with citizens, formly known as patients …"

ERIC TOPOL
THE FUTURE OF MEDICINE

SAGESCIENTISTS
SYMPOSIUM2014

GENETICS IN HEALTHCARE

Universitair Ziekenhuis Brussel

Centrum voor Medische Genetica

VUB

EDUCATION IN GENETIC TESTING

Credits to: Sonia Van Dooren, Kathelijn Keymolen and Edgard Eeckman

https://www.uzbrussel.be/web/centrum-voor-medische-genetica/brochures

PATIENT CARE, RESEARCH
But maybe education first …

IF YOU WANT TO STAY UPRIGHT,
you have to move forward

CMG
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Universitair Ziekenhuis Brussel

Centrum voor Medische Genetica

Remco Evenepoel viert zijn overwinning op het WK
Beeld BELGA

VUB

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