

AI & PID: What do we speak of?

Nizar MAHLAOU

18/10/2024



David is 11 years old.
He weighs 60 pounds.
He is 4 feet, 6 inches tall.
He has brown hair.

His love is real.
But he is not.



ARTIFICIAL INTELLIGENCE

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Artificial Intelligence (AI)

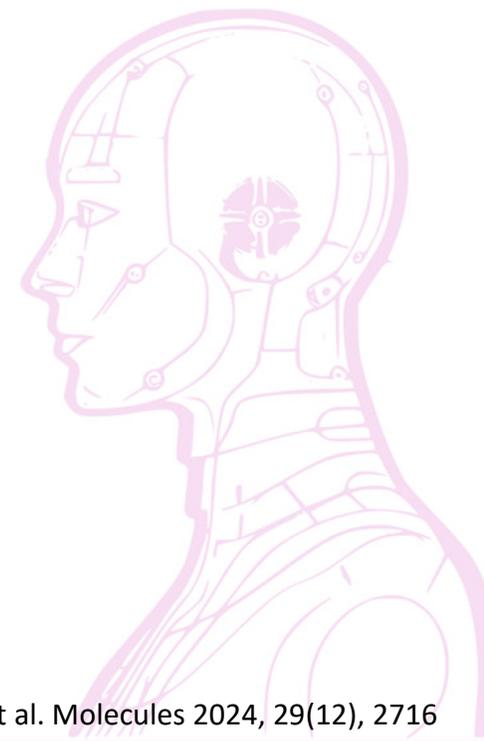
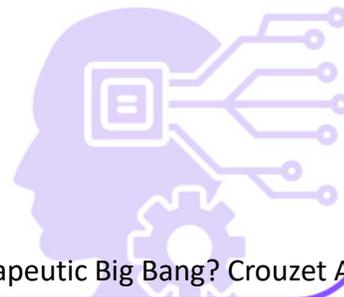
A group of **theories** and **mathematical models** implemented to **imitate** and **simulate** the **structures** and **operational principles** of the **human brain**.

Machine Learning (ML)

A set of **models** based on **mathematical concepts** and **computer science**, programmed to **autonomously** accomplish specific **supervised** or **unsupervised** tasks.

Deep Learning (DL)

A subfield of ML that employs **multi-layered neural networks** to **autonomously** learn **intricate representations from data**.



The Nobel Prize in Physics 2024

John Hopfield

“for foundational discoveries and inventions that enable machine learning with artificial neural networks”



John Hopfield. Ill. Niklas Elmehed © Nobel Prize Outreach

Geoffrey Hinton

“for foundational discoveries and inventions that enable machine learning with artificial neural networks”



Geoffrey Hinton. Ill. Niklas Elmehed © Nobel Prize Outreach

They used physics to find patterns in information

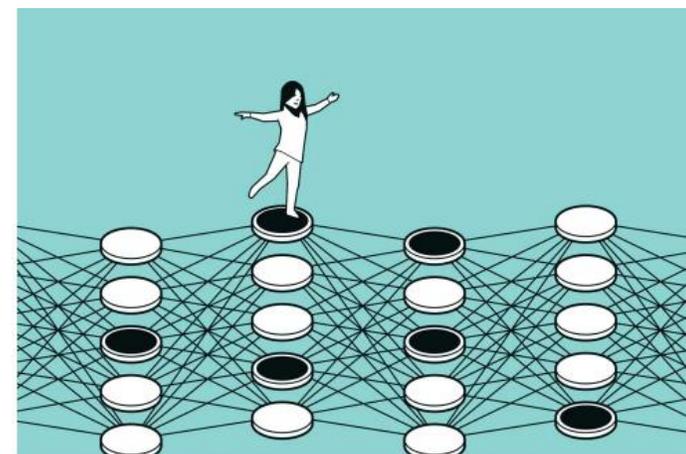
This year’s laureates used tools from physics to construct methods that helped lay the foundation for today’s powerful machine learning. John Hopfield created a structure that can store and reconstruct information. Geoffrey Hinton invented a method that can independently discover properties in data and which has become important for the large artificial neural networks now in use.

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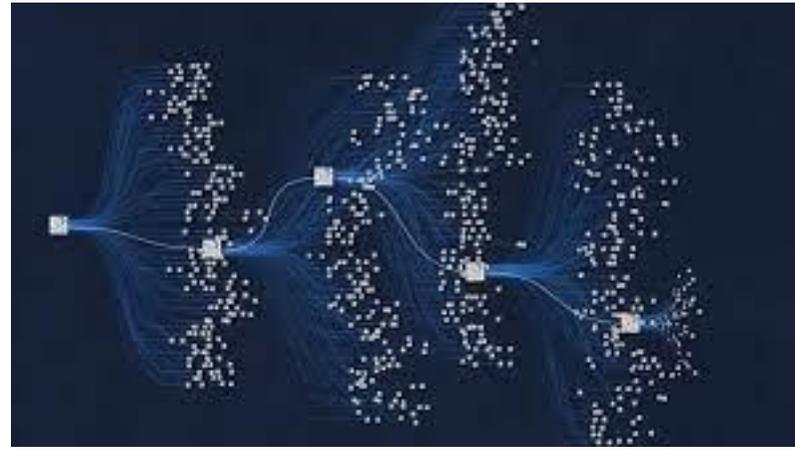
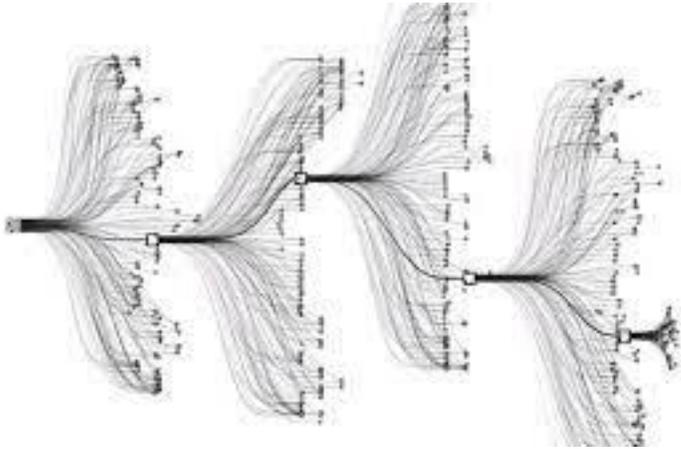


Deep Blue (IBM supercalculator, 700kg, 2m high) Garry Kasparov Feb 1996 > May 1997



IBM AI solution for Healthcare: Watson.ai

GOOGLE / ALPHABET



Verily®: prediction of the cardiovascular health by analyzing the vascularization of the eye

*** Perspectives: Quantum computers ***

We train AIs everyday without knowing it (CAPTCHAs / Images)

- **GAFAM / BATX**

Artificial Intelligence (AI) & Machine Learning (ML)

Very pervasive

All fields of human activities

“4th industrial revolution”

BIG DATA

Data Mining

Computational approaches



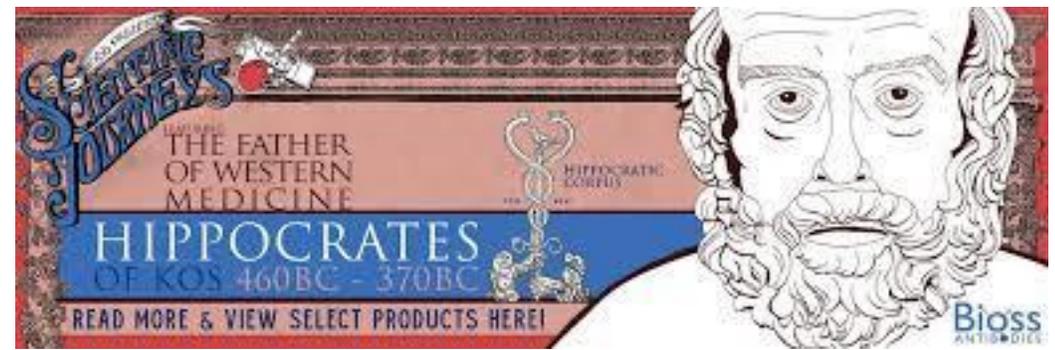
Big datasets

- Genomic data (WES/WGS)
- High Res images
- Continuous output from wearable sensors

- Only a tiny fraction (< 5%) of this amount of data has been processed.

- Advances in AI are taming the unbridled amalgamation of Big Data by putting it to work.

- Need for HIGH QUALITY data



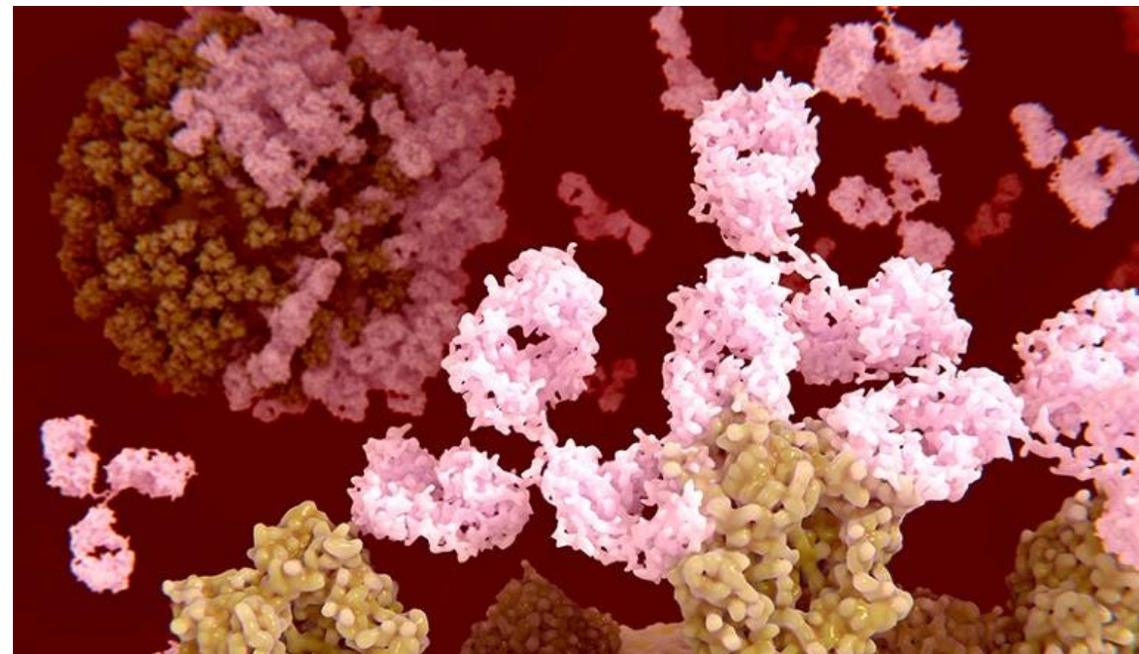
- **Hippocrates predicted big datasets in medicine**
- “It is more important to know what sort of person has a disease than to know what sort of disease has a person”
- Genome
- Proteome
- Metabolome
- Transcriptome
- Lipidome
- Microbiome

NEWS | 19 March 2024

'A landmark moment': scientists use AI to design antibodies from scratch

Modified protein-design tool could make it easier to tackle challenging drug targets – but AI antibodies are still a long way from reaching the clinic.

By [Ewen Callaway](#)



PRESS RELEASE

9 October 2024

The Nobel Prize in Chemistry 2024

The Royal Swedish Academy of Sciences has decided to award the Nobel Prize in Chemistry 2024 with one half to

David Baker

University of Washington, Seattle, WA, USA
Howard Hughes Medical Institute, USA.

Demis Hassabis

Google DeepMind, London, UK

John M. Jumper

Google DeepMind, London, UK

“for computational protein design”

“for protein structure prediction”

They cracked the code for proteins' amazing structures

The Nobel Prize in Chemistry 2024 is about proteins, life's ingenious chemical tools. David Baker has succeeded with the almost impossible feat of building entirely new kinds of proteins. Demis Hassabis and John Jumper have developed an AI model to solve a 50-year-old problem: predicting proteins' complex structures. These discoveries hold enormous potential.

The diversity of life testifies to proteins' amazing capacity as chemical tools. They control and drive all the chemical reactions that together are the basis of life. Proteins also function as hormones, signal substances, antibodies and the building blocks of different tissues.

“One of the discoveries being recognised this year concerns the construction of spectacular proteins. The other is about fulfilling a 50-year-old dream: predicting protein structures from their amino acid sequences. Both of these discoveries open up vast possibilities,”

in long strings that fold up to make a three-dimensional structure, which is decisive for the protein's function. Since the 1970s, researchers had tried to predict protein structures from amino acid sequences, but this was notoriously difficult. However, four years ago, there was a stunning breakthrough.

In 2020, **Demis Hassabis** and **John Jumper** presented an AI model called AlphaFold2. With its help, they have been able to predict the structure of virtually all the 200 million proteins that researchers have identified. Since their breakthrough, AlphaFold2 has been used by more than two million people from 190 countries. Among a myriad of scientific applications, researchers can now better understand antibiotic resistance and create images of enzymes that can decompose plastic.

Life could not exist without proteins. That we can now predict protein structures and design our own proteins confers the greatest benefit to humankind.



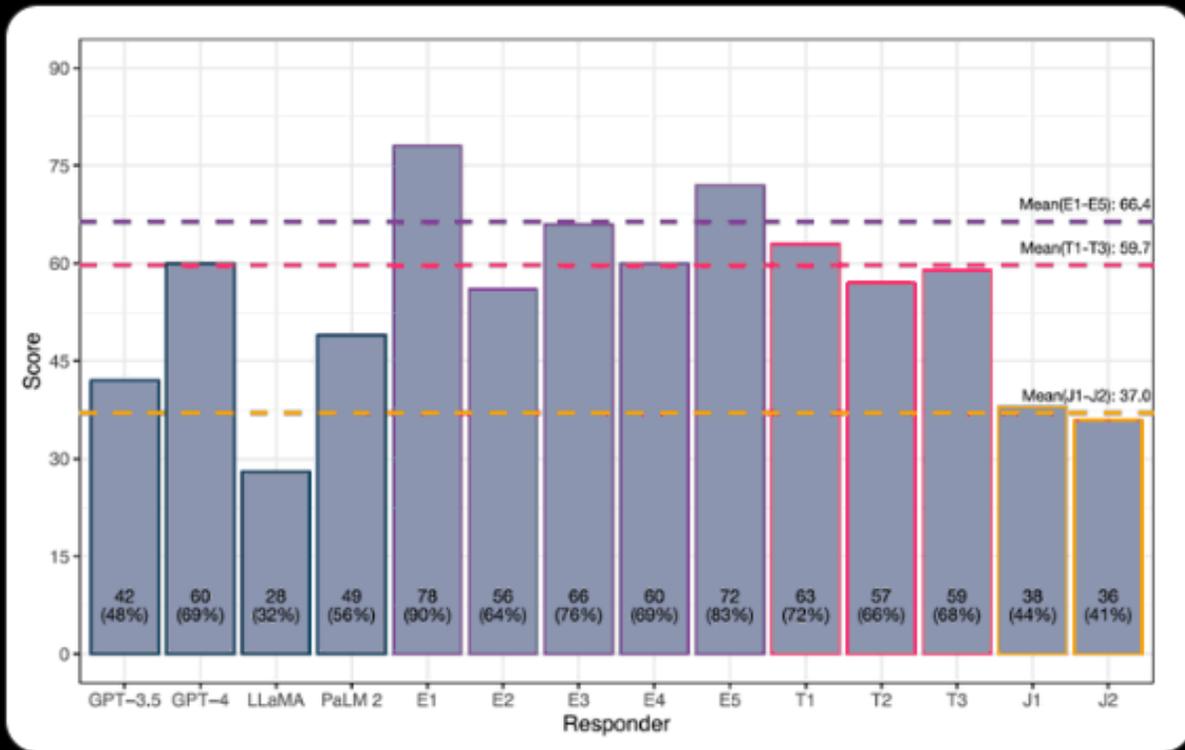
Eric Topol 
@EricTopol



"Large language models (LLMs) are approaching expert-level ophthalmological knowledge and reasoning"

[journals.plos.org/digitalhealth/...](https://journals.plos.org/digitalhealth/) @PLOS DigiHealth

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[nature](#) > [news](#) > article

NEWS | 21 March 2024

Google AI could soon use a person's cough to diagnose disease

Machine-learning system trained on millions of human audio clips shows promise for detecting COVID-19 and tuberculosis.

By [Mariana Lenharo](#)



AI & Rare Diseases:

The Disruptive Momentum might be around the corner



Eric Topol

@EricTopol

When it comes to the differential diagnosis for a rare disease case, [#GPT4](#) outperforms physician specialists and other LLMs

arxiv.org/abs/2402.06341

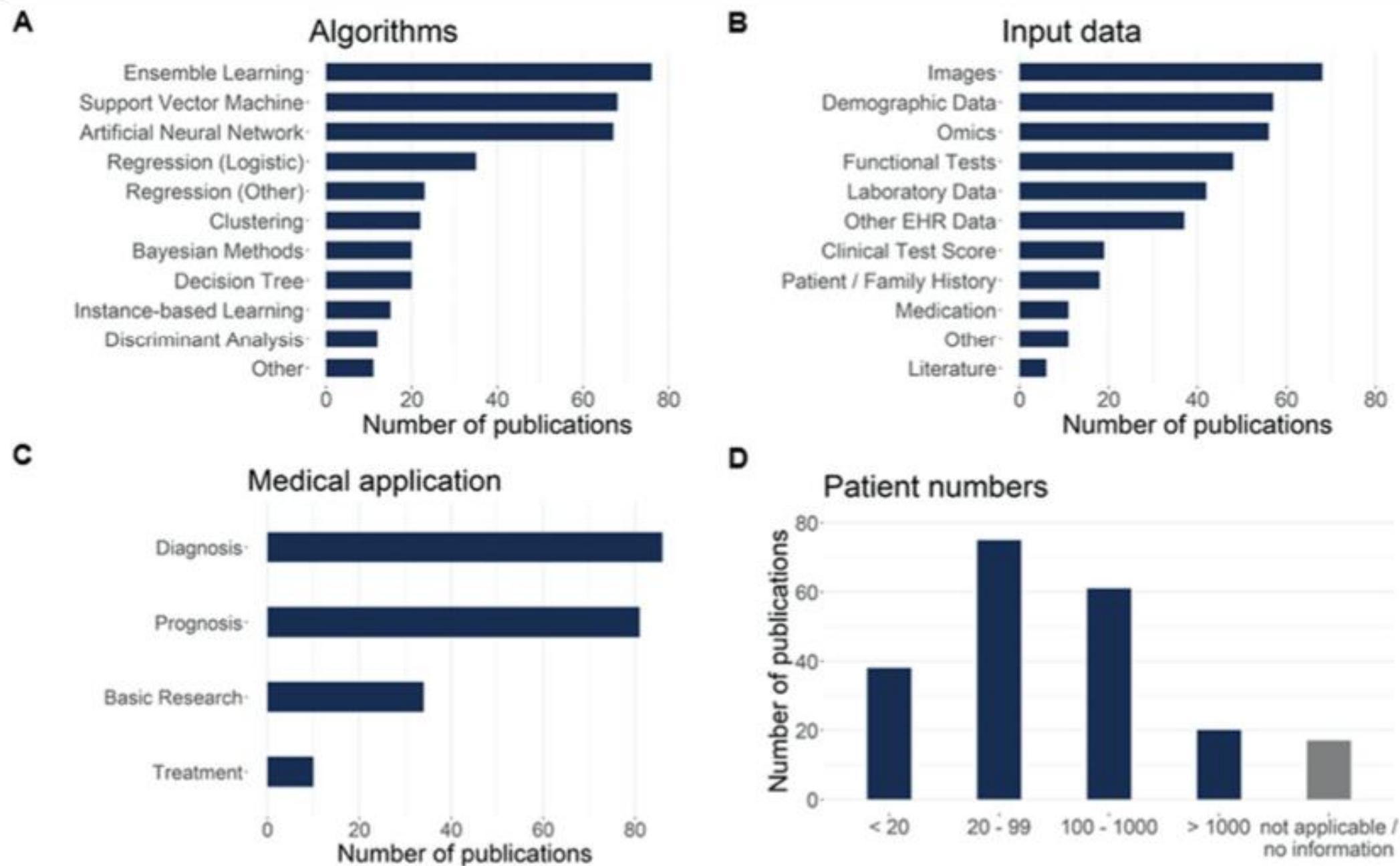
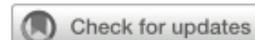


Fig. 4 Types of algorithms used in the studies (a); input data (b); medical application (c); number of patients (d). Studies using more than one type of algorithm or input data are listed in more than one category

PID: Stakes — General considerations

- Lack of awareness
- Lack of information of the public
- Lack of training of medical students, physicians, nurses and all HCPs
- Lack of consideration of the concept of holistic approach
 - Silos / Hyper-specializations
 - Too much “doctor ego”
 - It’s OK for a doctor not to know everything about everything
 - It’s OK If the patient knows some things better than the doctors
- Lack of understanding of the patient journey
- Lack of human resources and time:
 - Listening carefully is critical but it demands time and effort

Proceedings from the inaugural Artificial Intelligence in Primary Immune Deficiencies (AIPID) conference



Jacques G. Rivière, MD,^{a,b,c,d} Pere Soler Palacín, MD, PhD, MS,^{a,b,c,d} and Manish J. Butte, MD, PhD^{e,f,g}

Barcelona,

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RIVIÈRE, SOLER PALACÍN, AND BUTTE 639

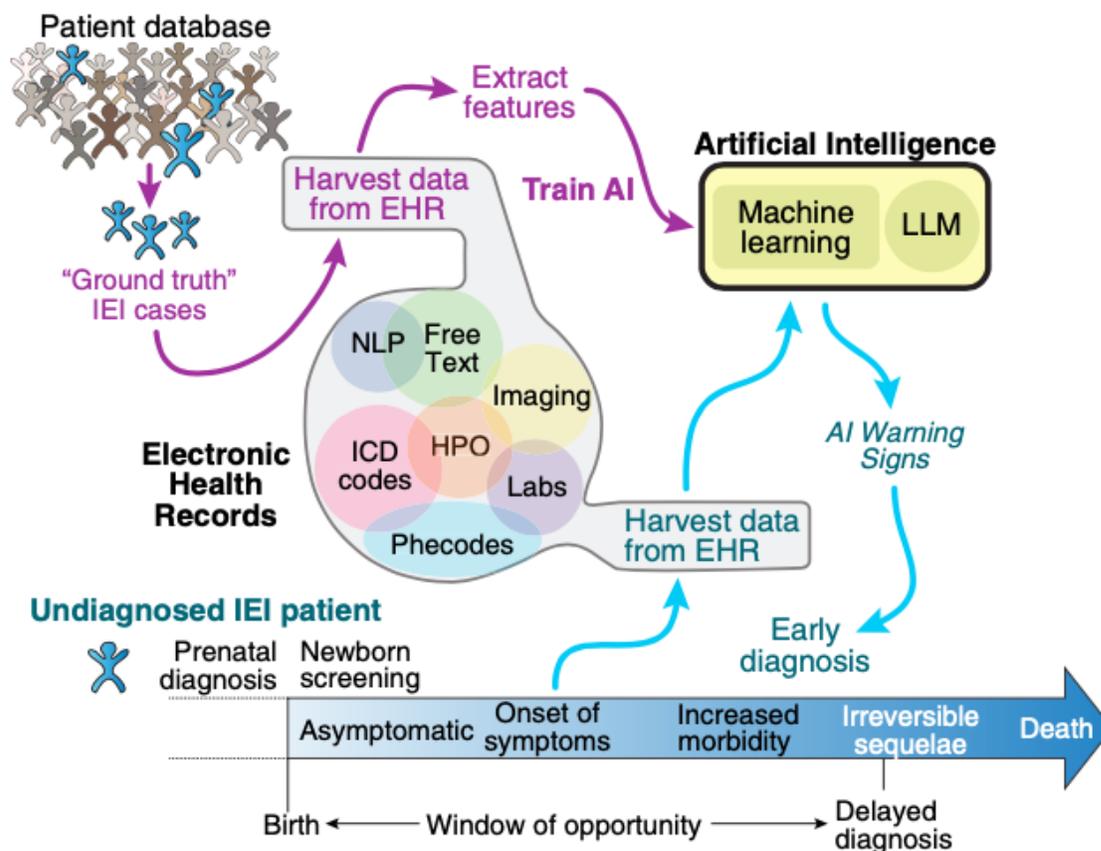


FIG 1. Opportunity for AI to accelerate the diagnosis of IEs.

- Diagnosis
- Management of disease
- Predictive, modelling AI Research
- Data

AI & PID — Computational approaches

Still early age

- Disease detection
- Management of disease
- Genetic analysis
- Drug Discovery and Development
- Remote Monitoring
- Professional Education and Training
- Concept learning
 - Improve diagnostic rates
 - Understand immunologic diseases
 - Reduce Dx delay
 - Uncover atypical manifestations in PID
 - Contribution to more accurate descriptions of PIDD
 - Contribution to so-called 'soft' science (*fatigue, neuropsychology — autism, depression, anxiety, stress*)
 - Other markers (environment, nutrition, physical activity, microbiota...)
 - Better deciphering PIDs
 - Better discrimination between PID and SID
- Better treatments/management/outcomes

