THE NEW WAY TO EXPLORE THE BRAIN FUNCTIONING

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Alba Di Pardo
Centre for Neurogenetics and Rare Diseases
IRCCS Neuromed, ITALY

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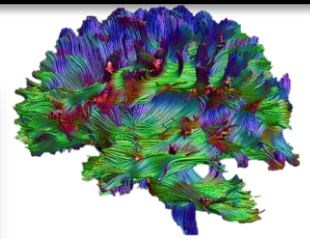
Taking care of the BRAIN

TECHNOLOGIES



BRAIN

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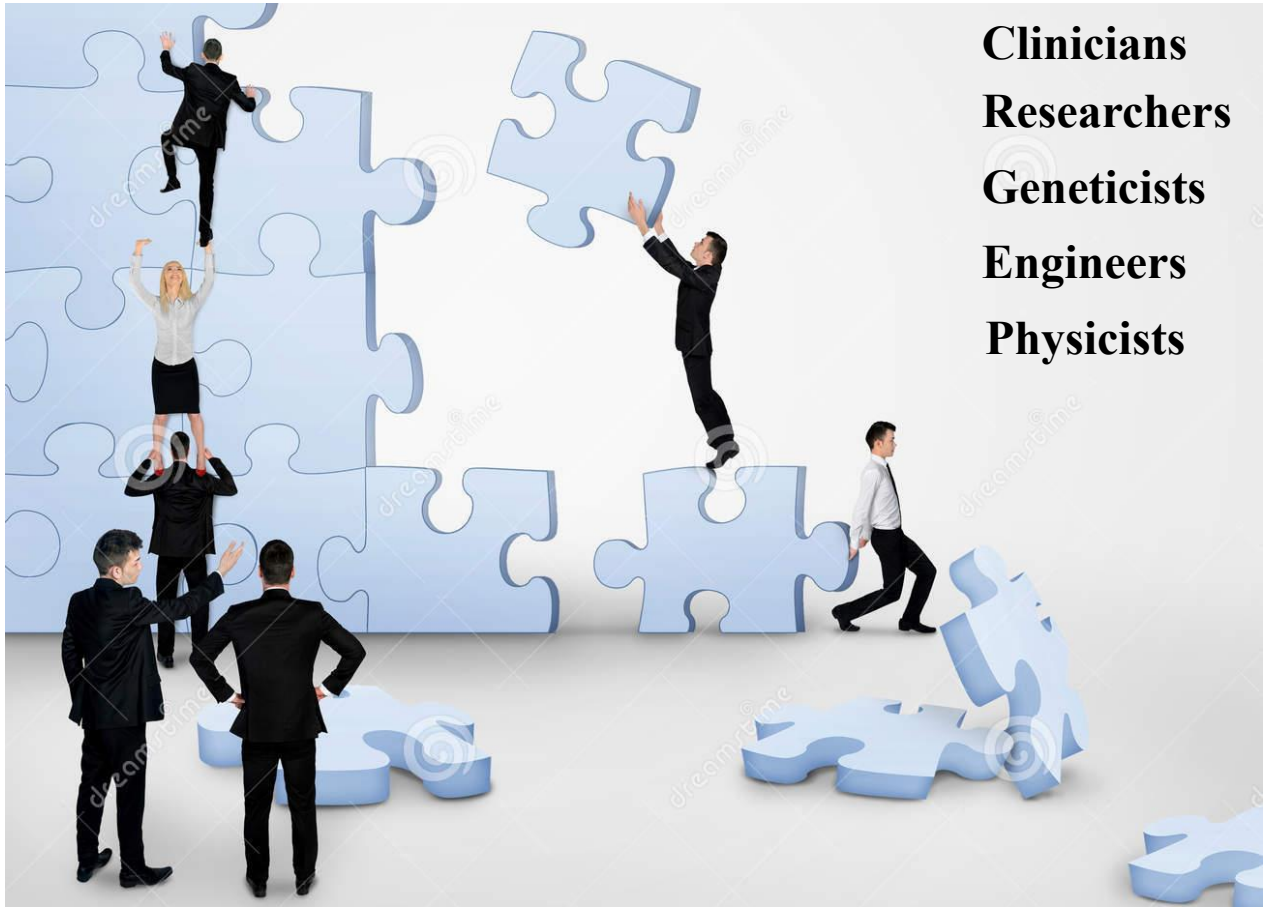
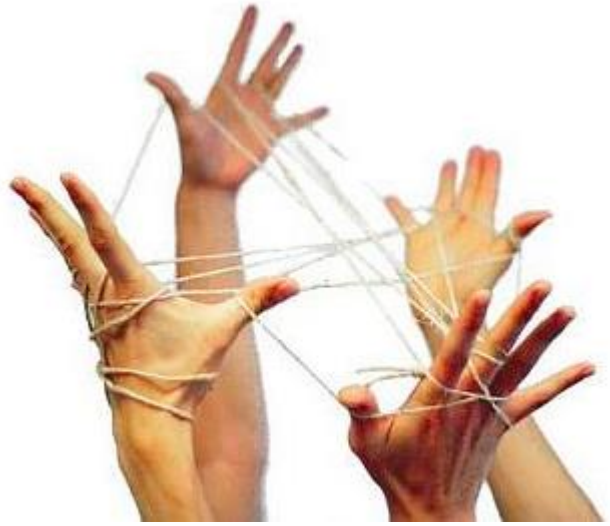
Together for the BRAIN





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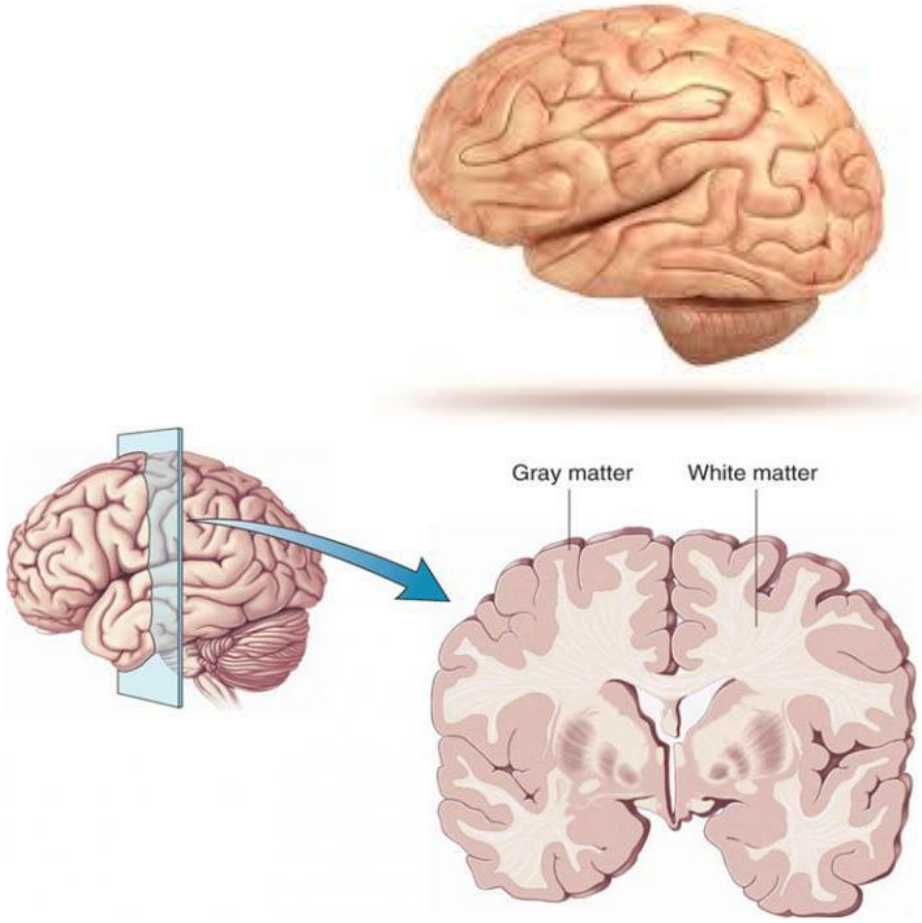
The BRAIN: the most complex human system…

…with still a lot of unexplored properties

To make the **BRAIN** understandable…a funny analogy



BRAIN:
Structure and Function

Frontal
- Personality
- Emotions and arousal
- Intelligence
- Ability to concentrate, make decisions, plan, put things in order, solve problems
- Awareness of what is around you
- Voluntary movement
- Ability to speak and write
- Behaviour control

Parietal
- Sensations: pain, touch, temperature
- Understanding and interpreting sensory information, such as size, colour and shape
- Understanding space and distance
- Math calculations

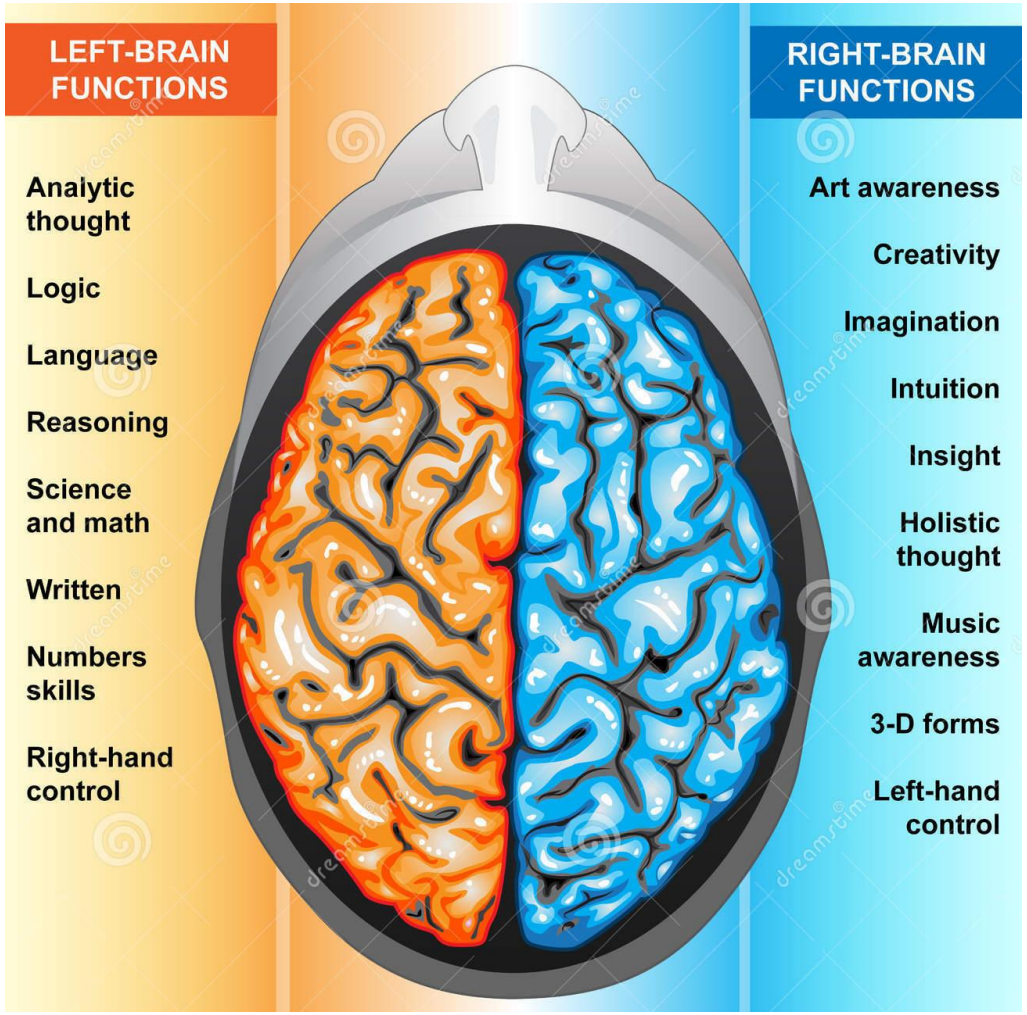
Occipital
- Vision
- Interpreting what you see

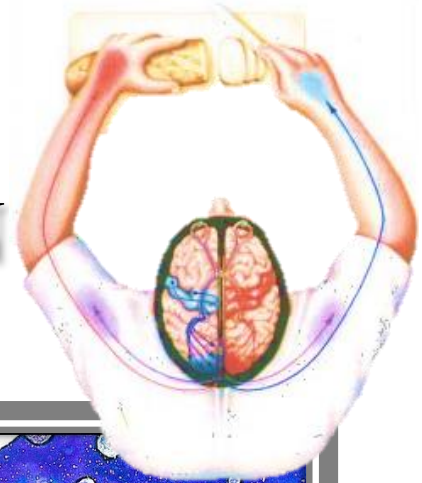
Temporal
- Ability to understand language
- Hearing
- Memory, long-term storage of memories
- Organization and planning
- Behaviour and emotions

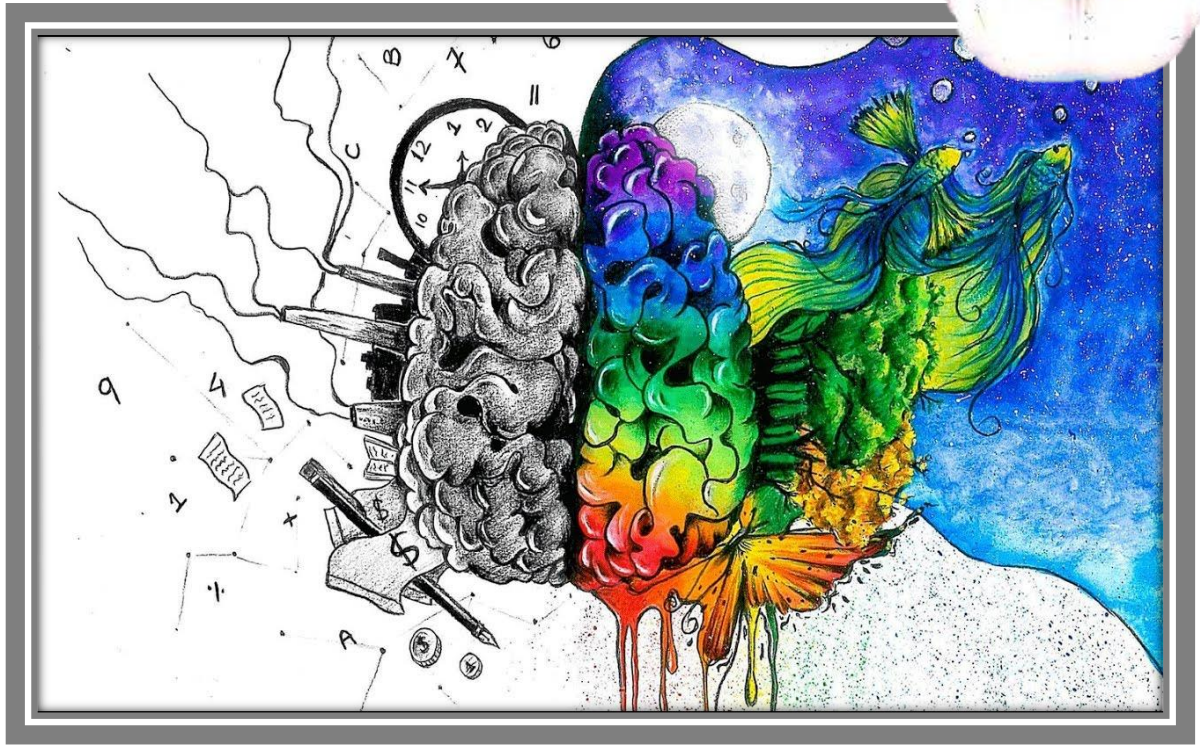
Brain stem
- Breathing
- Heart rate control
- Consciousness, alertness, wakefulness
- Swallowing
- Blood pressure
- Sweating

Cerebellum
- Balance
- Motor (movement) coordination
- Posture
- Fine motor skills

LOGIC vs EMOTION

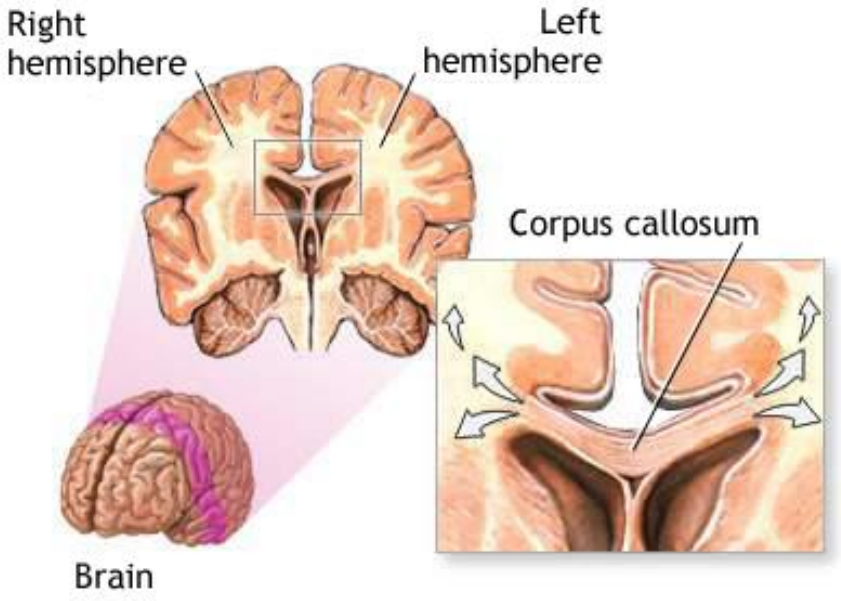




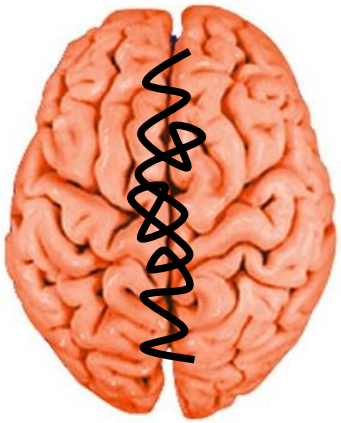


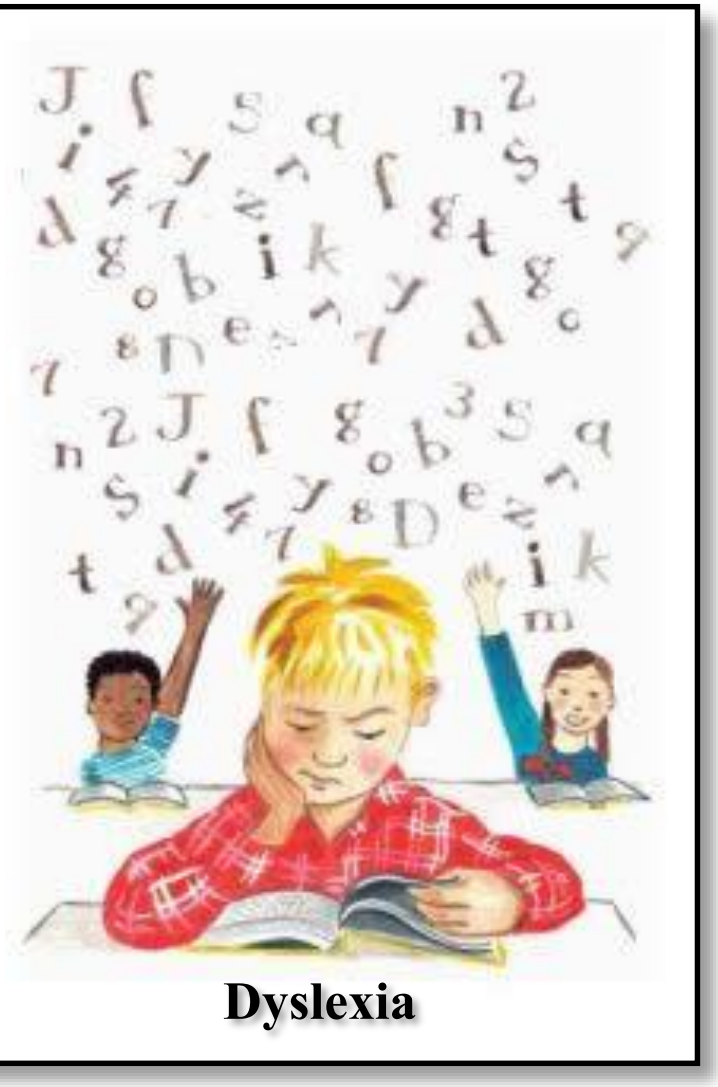
Corpus callosum:

more than just a midline structure

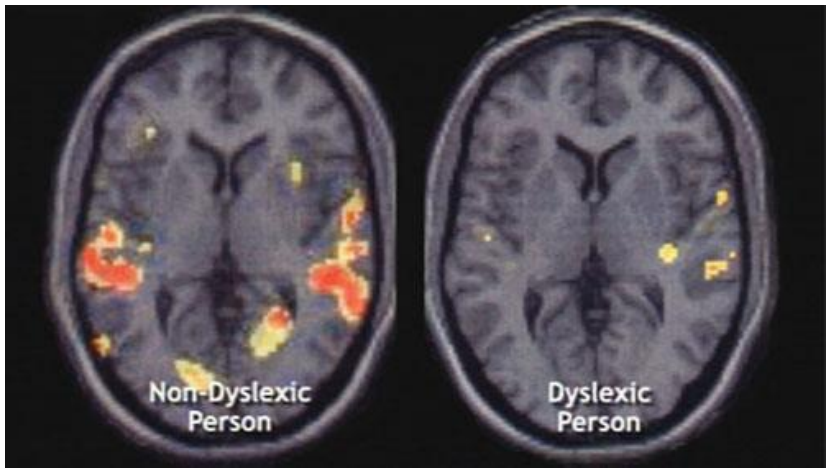


SPLIT-Brain Syndrome





Dyslexia



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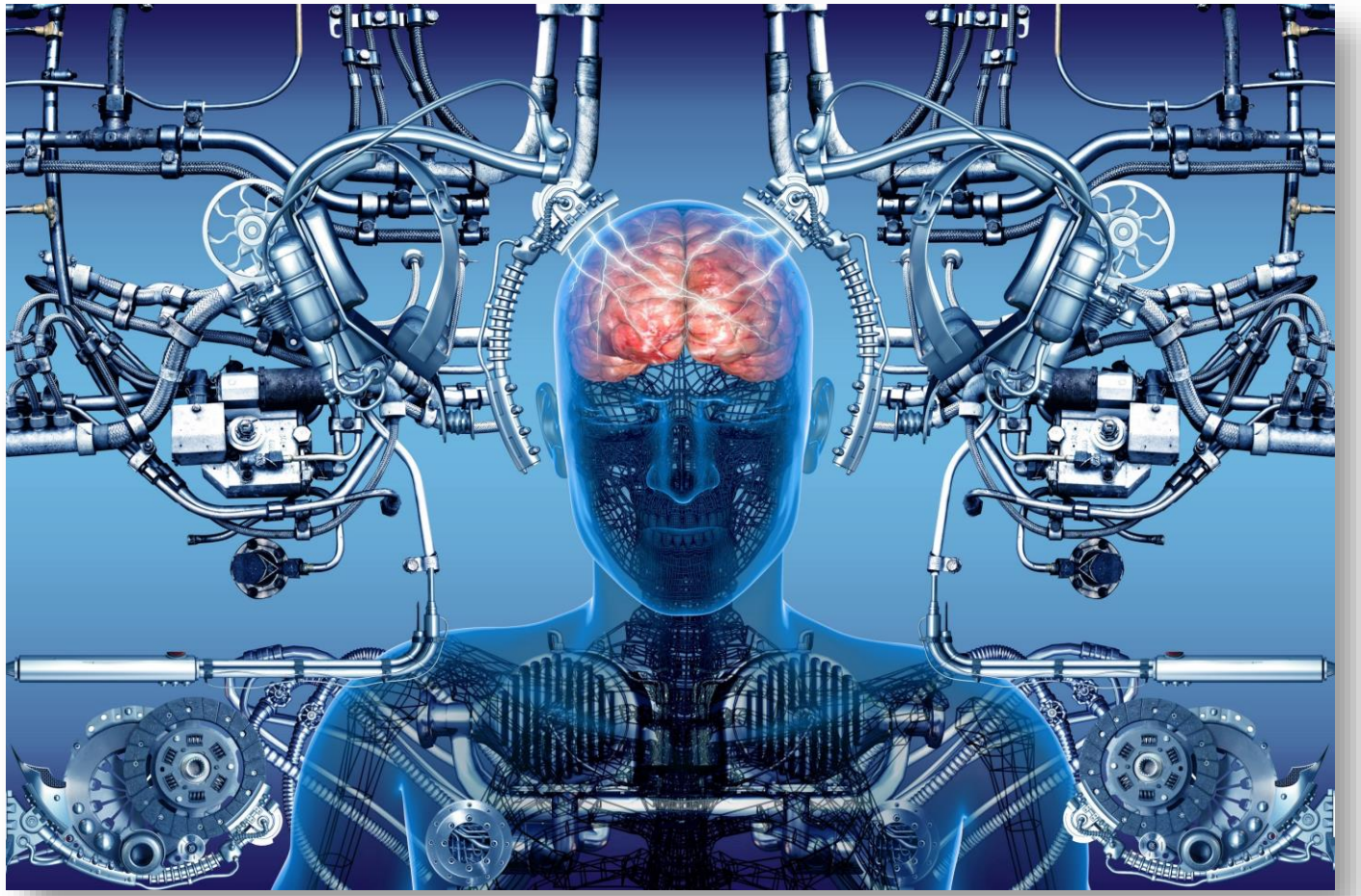
The SCIENCE behind the human BRAIN is COMPLEX

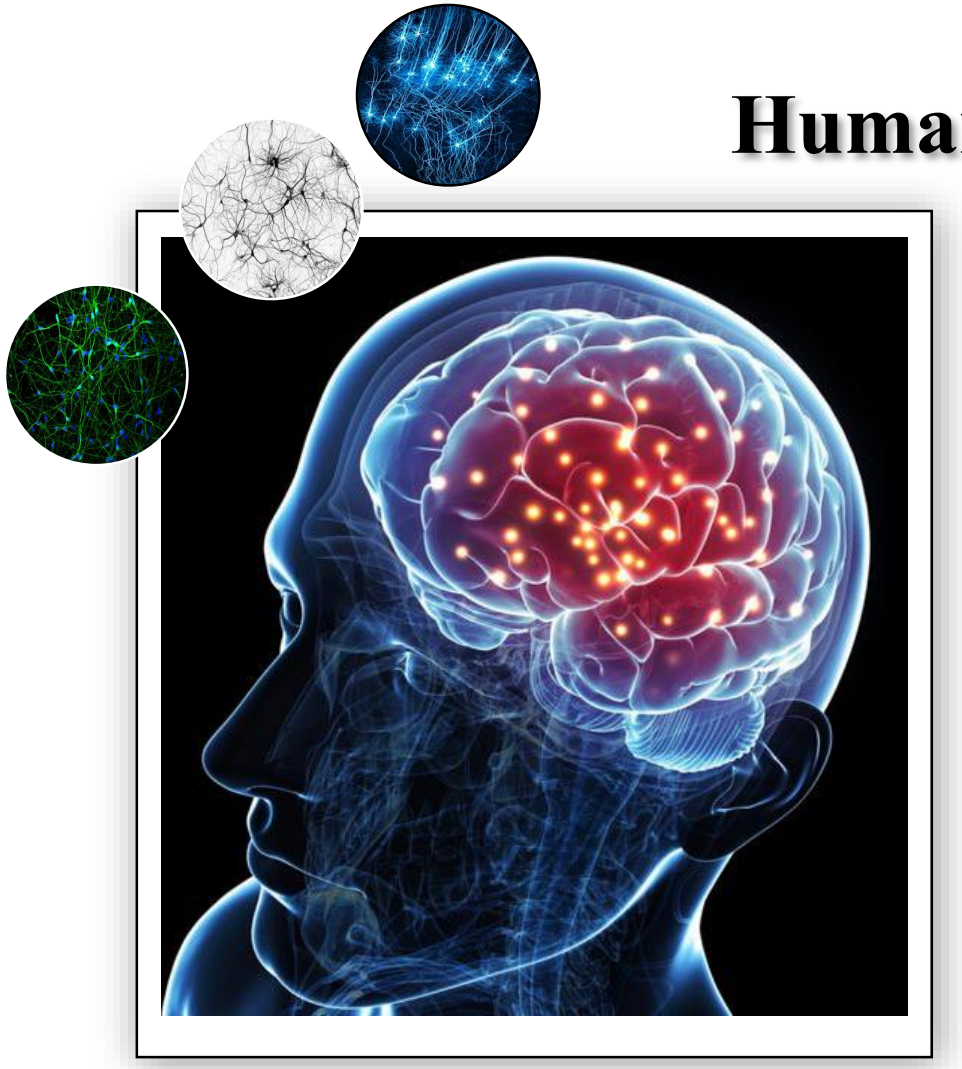


Anatomy

Complex Networking

Computational power

Human BRAIN



The human BRAIN has 100 billion Neurons, each of which is connnected to **10 thousant other neurons.**

Sitting on your shoulders the BRAIN is the most complicated object in the known **universe.**

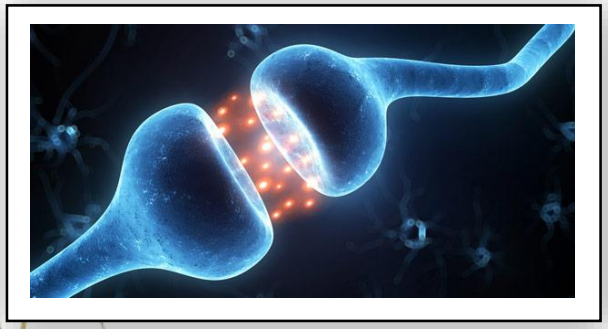
The most misterious machine

Michio Kaku

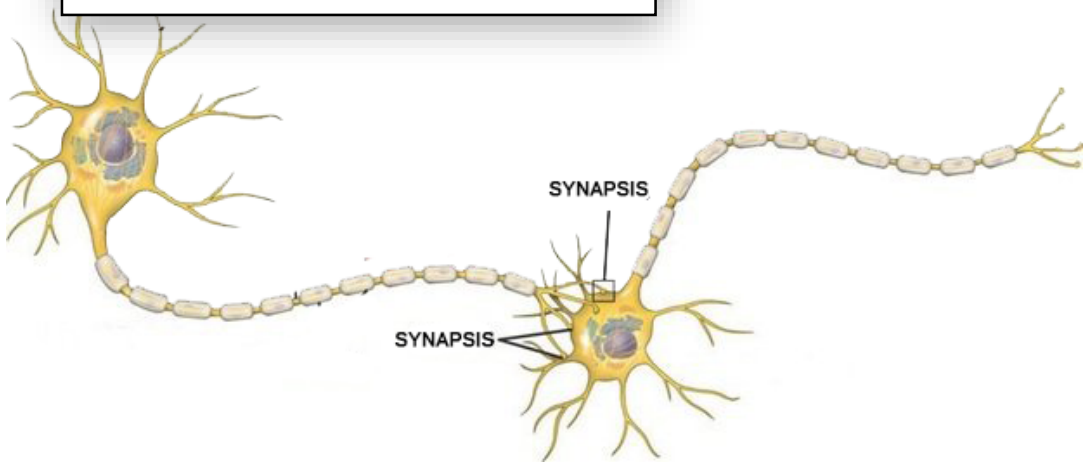
The COMPUTATIONAL Network of the BIOLOGICAL Machine

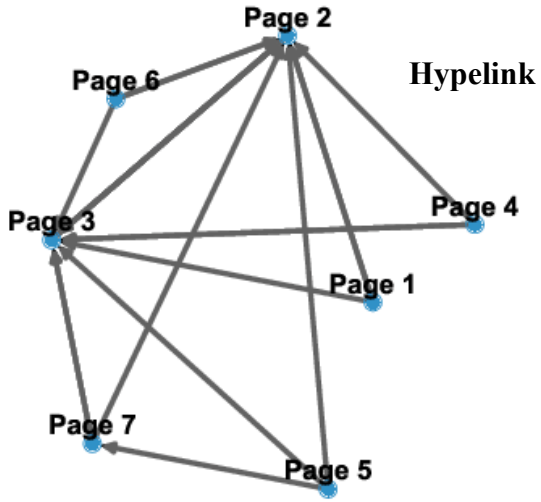
The COMPLEX computational network of human BRAIN

SYNAPSIS



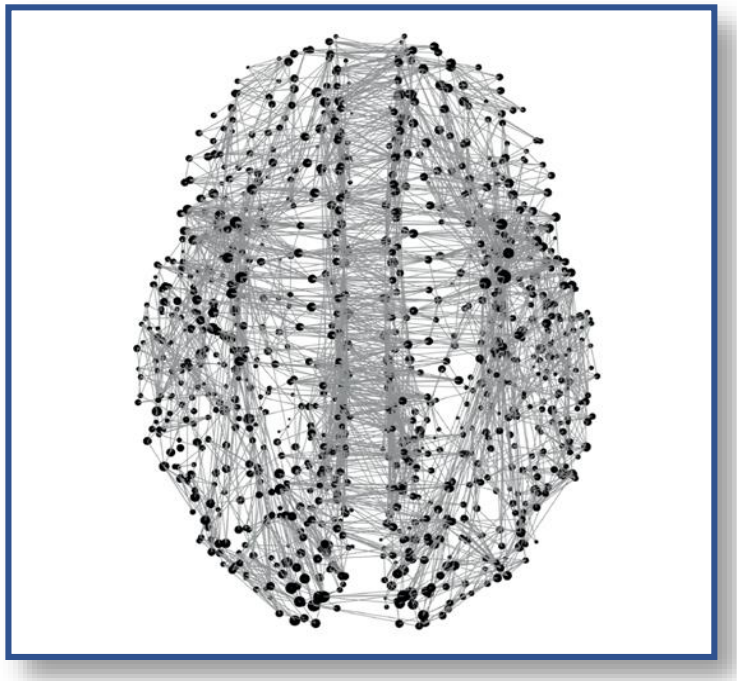
One synapse, by itself, is more like a microprocessor—with both memory-storage and information-processing elements—than a mere on/off switch. In fact, one synapse may contain on the order of 1,000 molecular-scale switches.



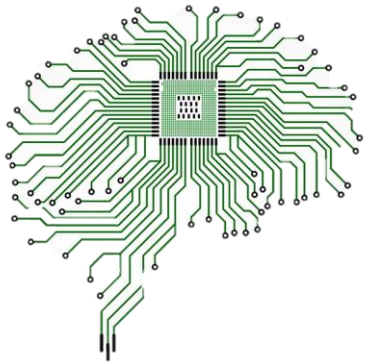


Human BRAIN has more connection than every computer on Earth combined

100 trillion **links**



300 trillion **synapses**



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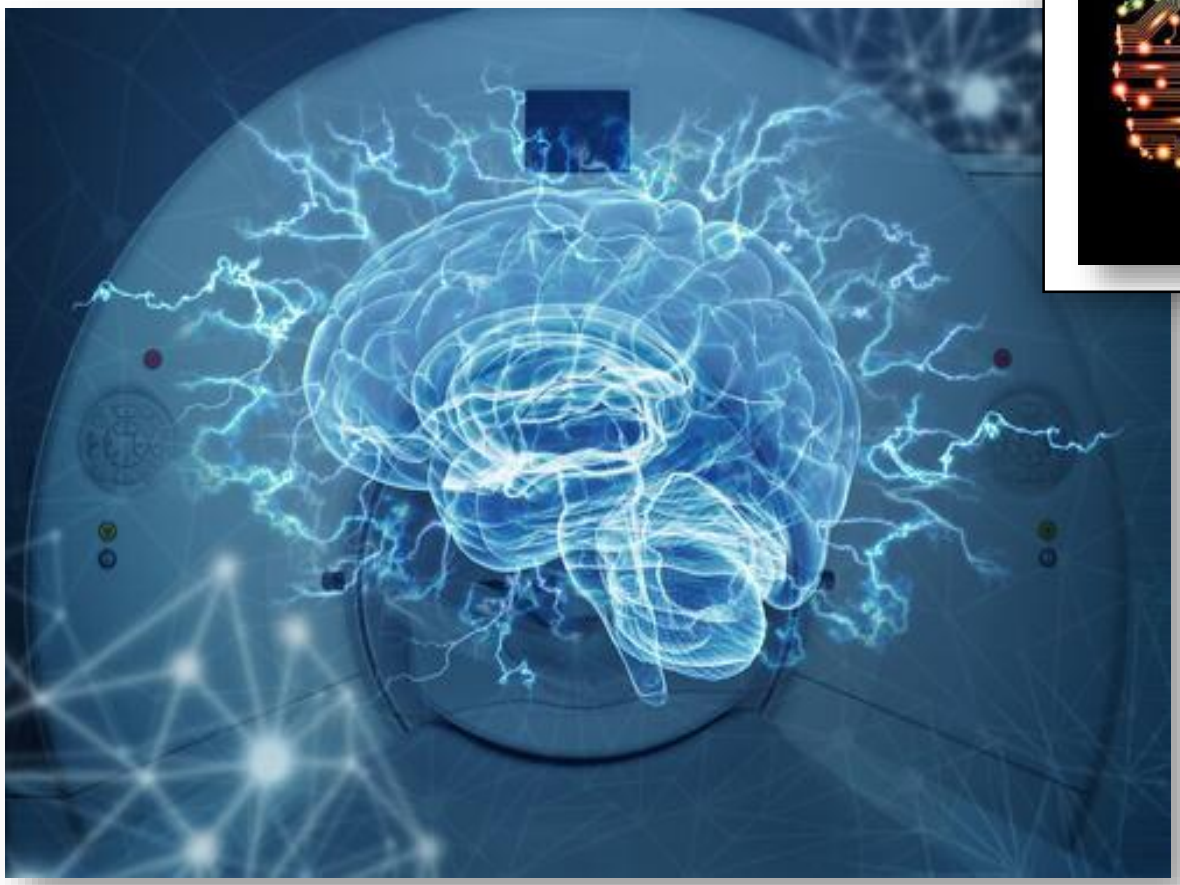
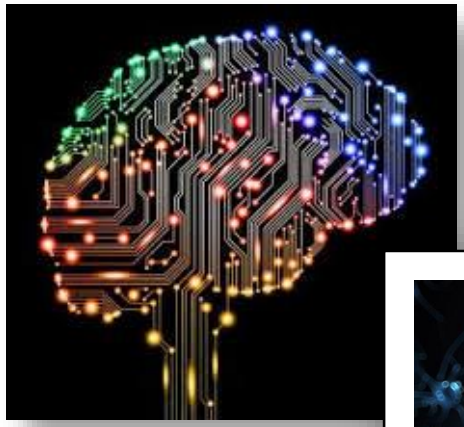
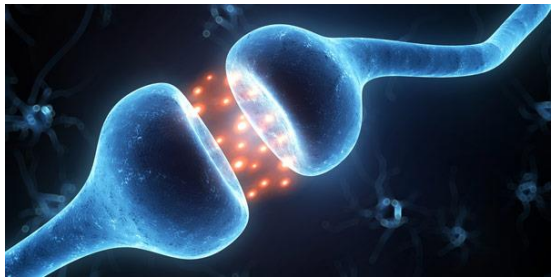
Matter of Connections





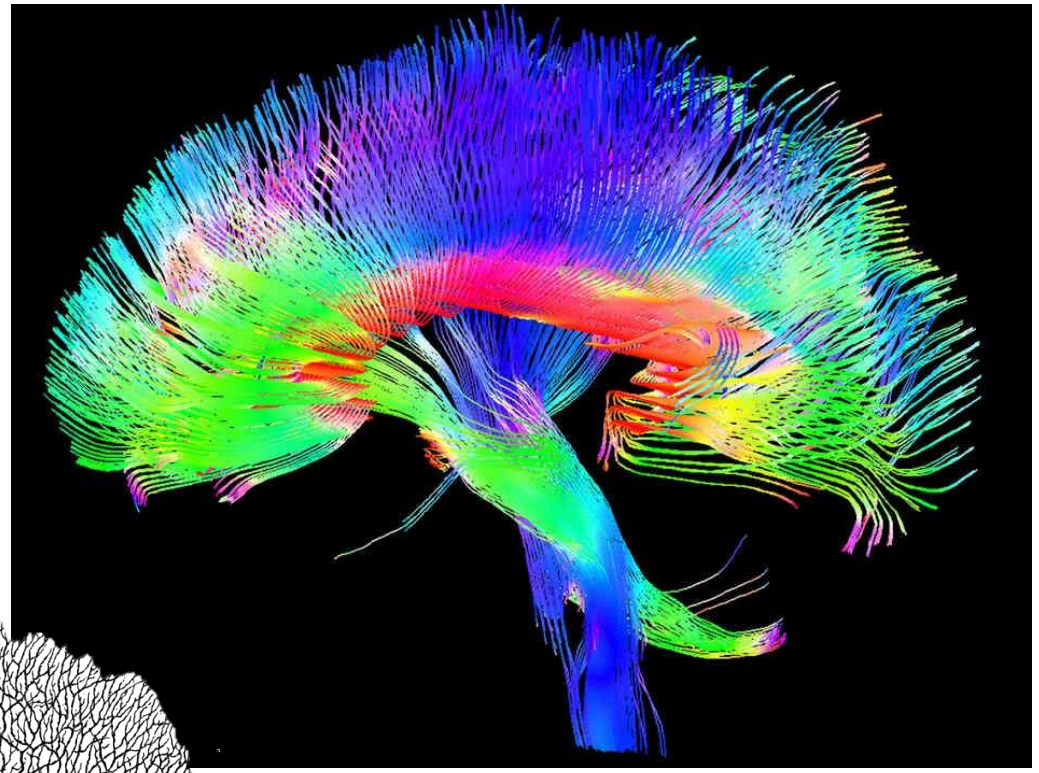


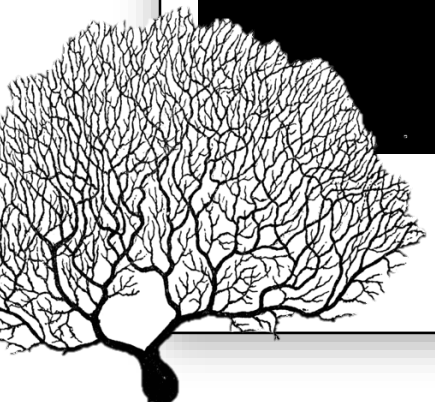
REASON

MEMORY

EMOTION

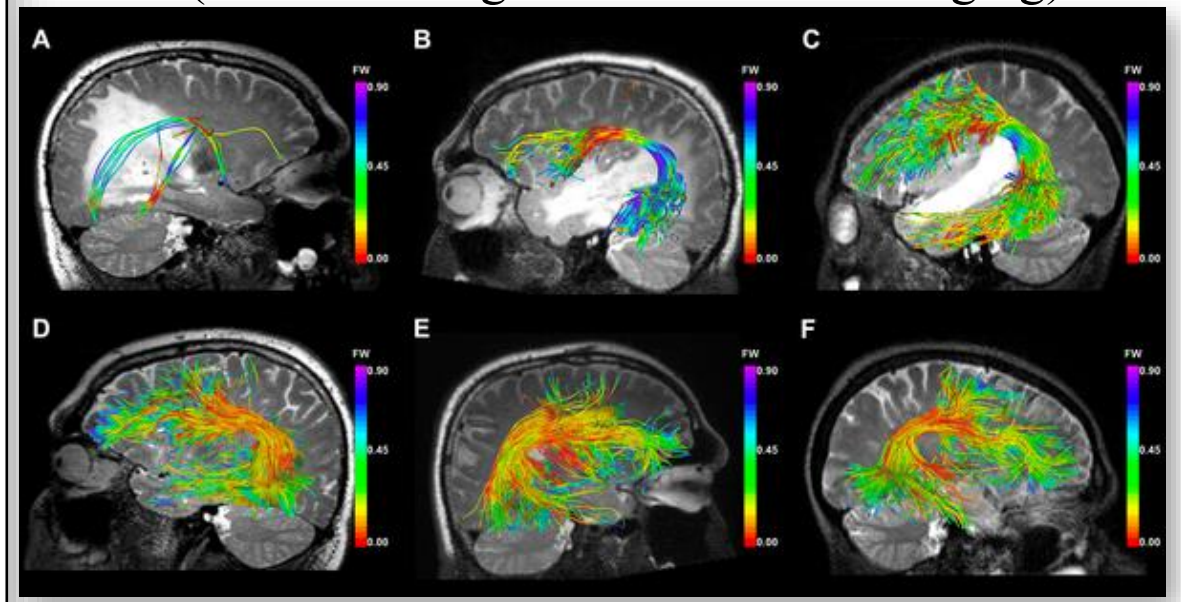
BRAIN WIRING diagram and CONNECTOME





The physic circuitry of the average brain contains trillions of synapsis and this complex network is known as CONNECTOME

DMRI (diffusion Magnetic Resonance Imaging)



to accurate map the BRAIN architecture and connections in HEATHY and DISEASE condition

(new insights for autism and schizofrenia)

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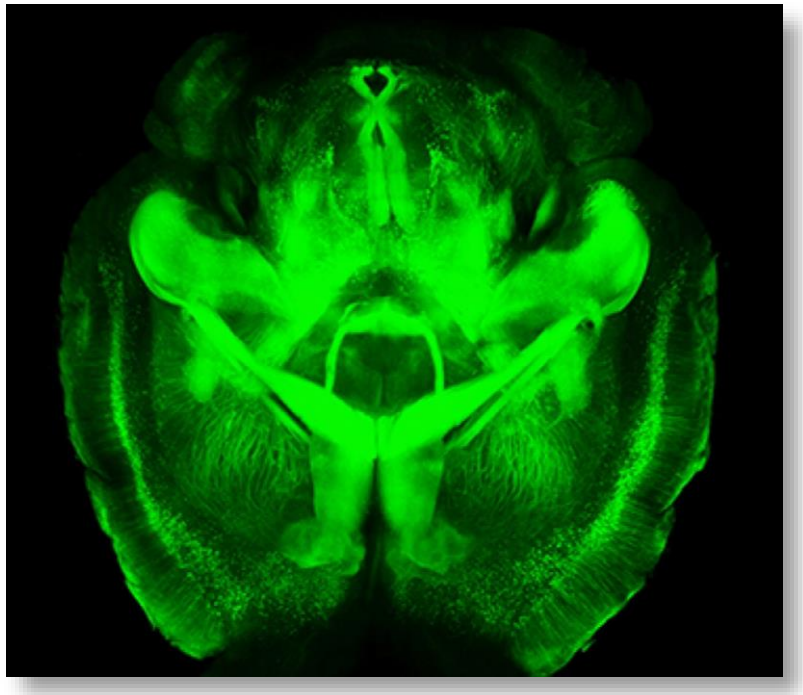
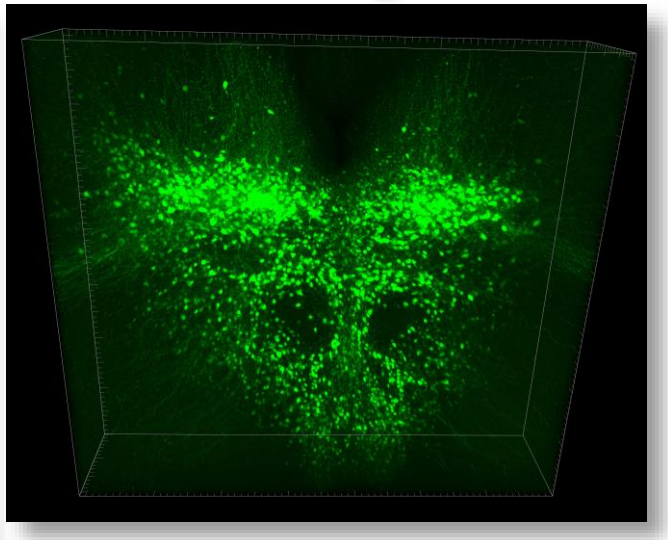
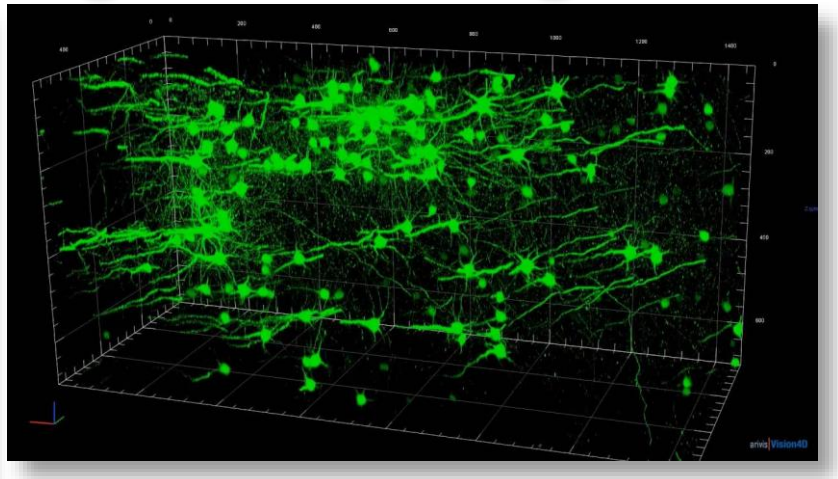
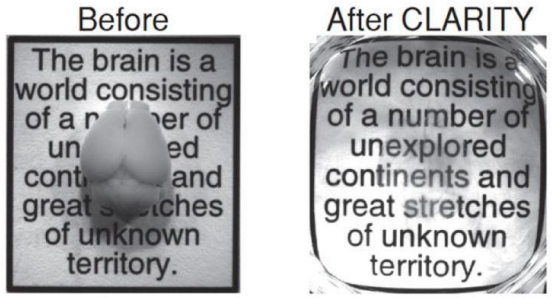
CLARITY: to visualize neurological wiring in nonliving brain

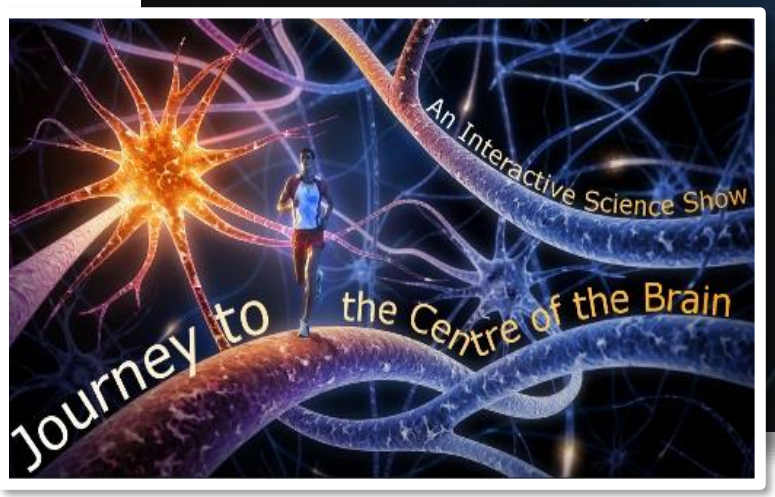


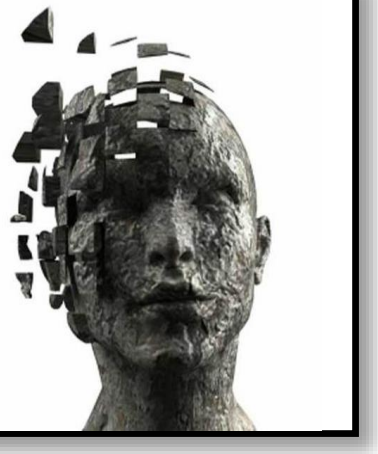




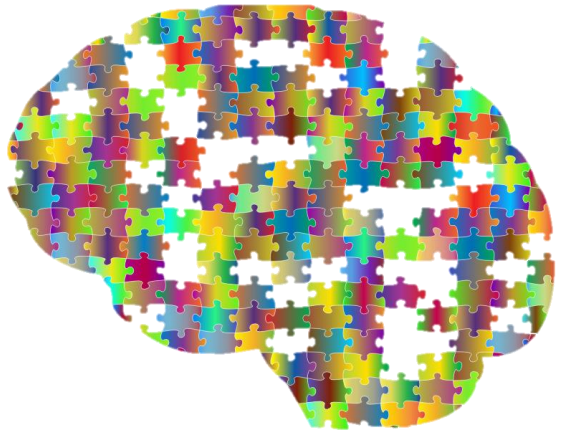


DIVING INTO THE UNKNOWN


An Interactive Science Show
Journey to
the Centre of the Brain



*The big issue in **Neuroscience** is to understand how pieces of the brain fit and work together…*



*The big issue in **Medicine** is to objectively diagnose and treat brain disorders*

Why so much interest in Human Brain?

Human BRAIN

SOCIETY

Animals **EVOLUTION**

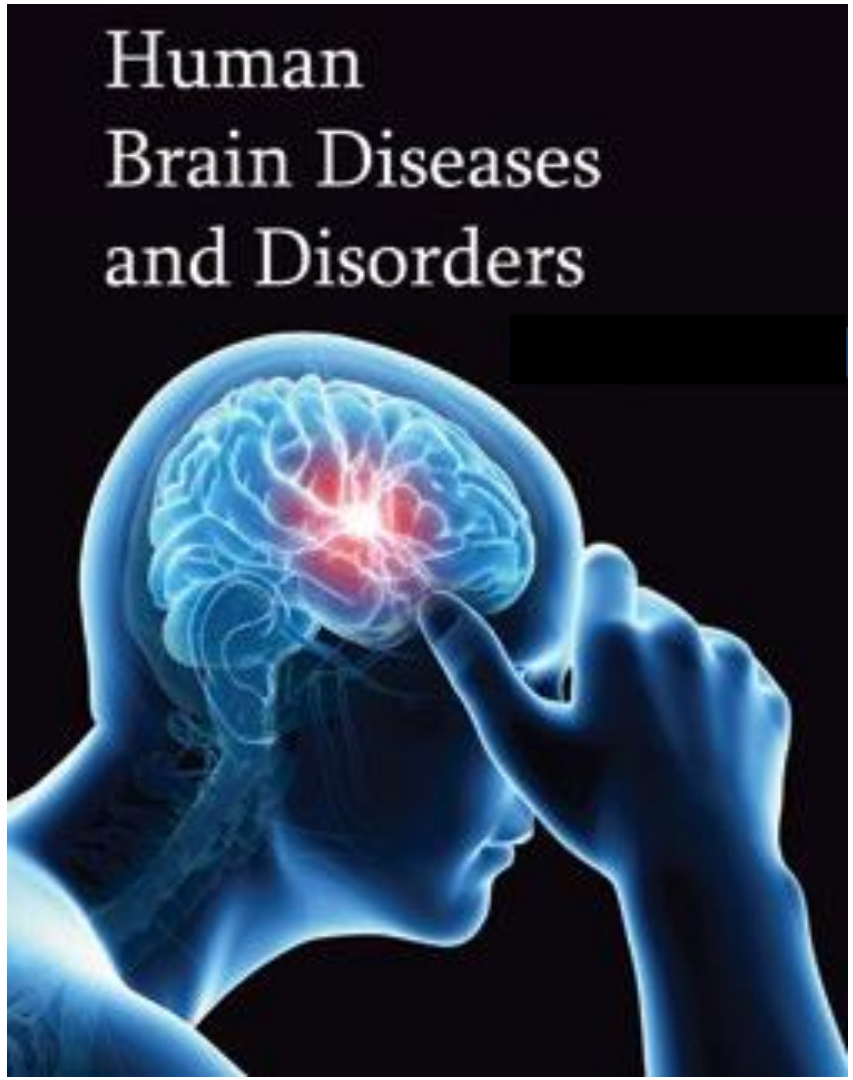
WORKING model **DISEASES**

Knowledge **Experimentation**

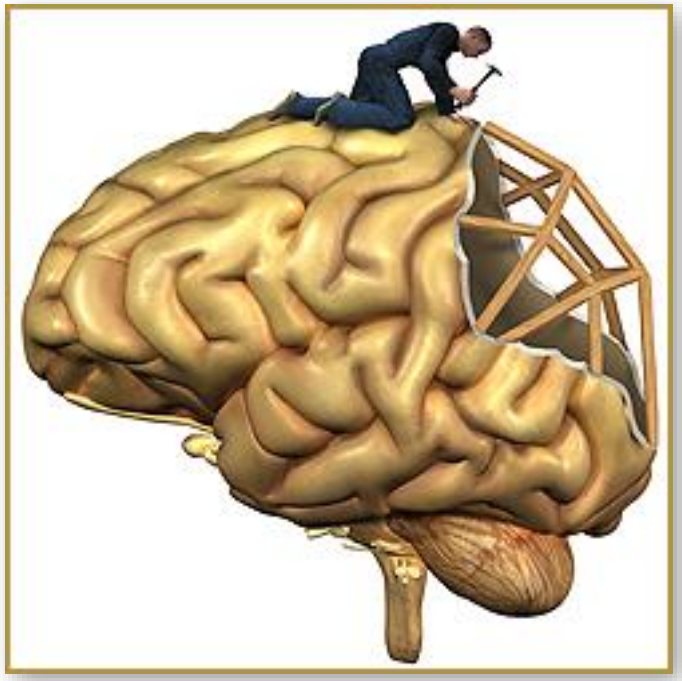
DATA

> 2 billions of patients

The urgent NEED

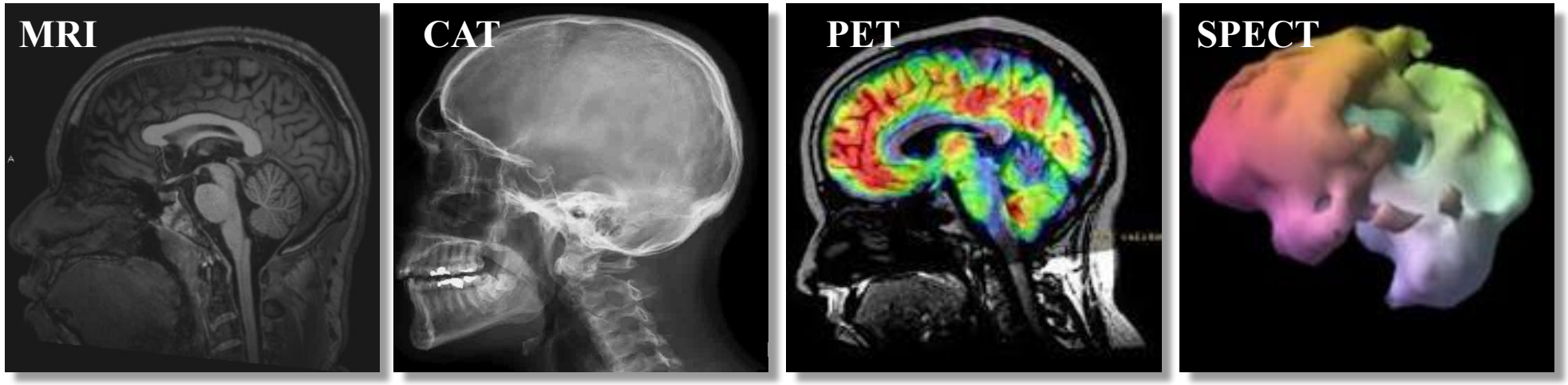


Looking INSIDE the BRAIN to DIAGNOSE mental diseases



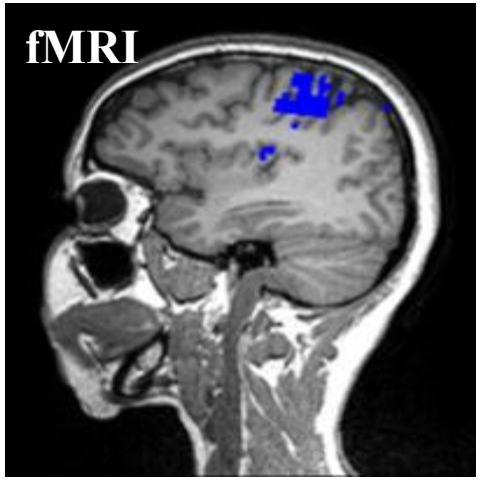
NOT sufficient to make diagnosis

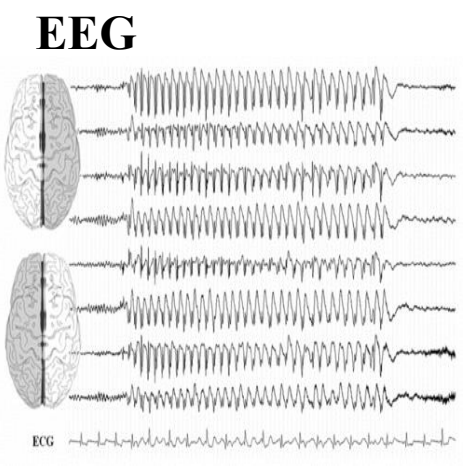
Brain Imaging Technologies to look at STRUCTURE …



In structural imaging, machines take snapshots of the brain's large-scale anatomy and allow researchers to look inside the brain.

…and FUNCTION





Functional imaging provides a dynamic view of the brain, showing which areas are active during thinking and perception.

The fMRI and EEG are very important tools mostly used in diagnosis of different neurological condition that affect the normal functioning of the brain also without any apparent structural alteration.

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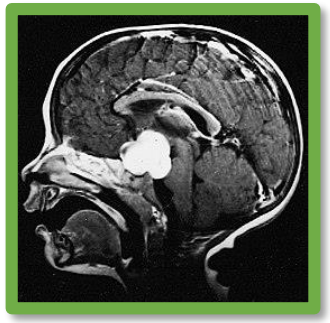
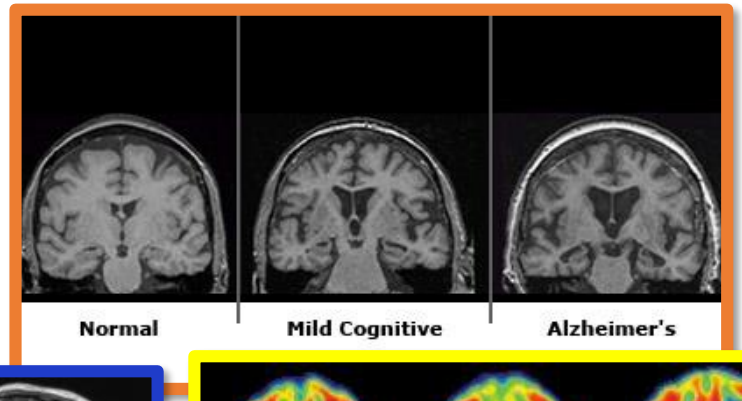
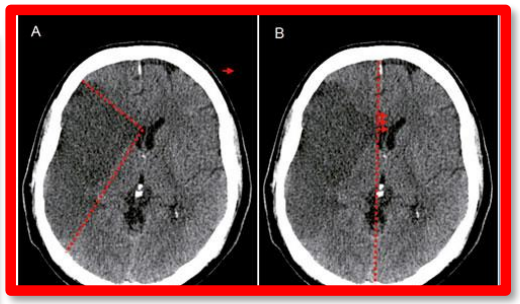
… to learn about BRAIN disorders

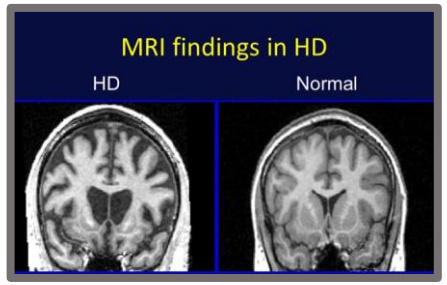


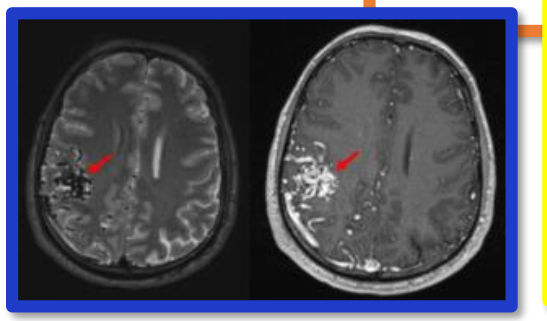
Brain cancer

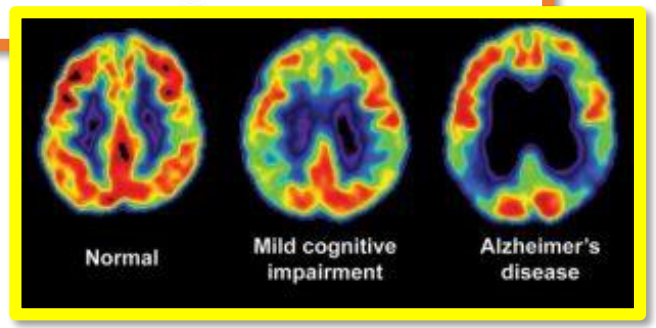


Ischemic stroke

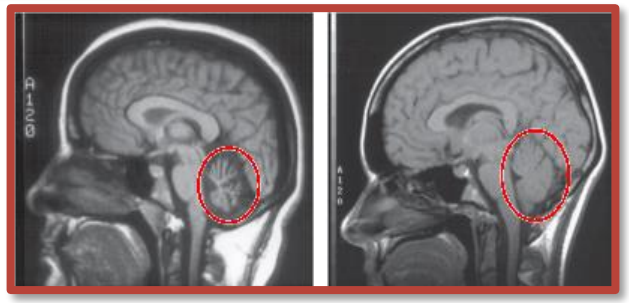






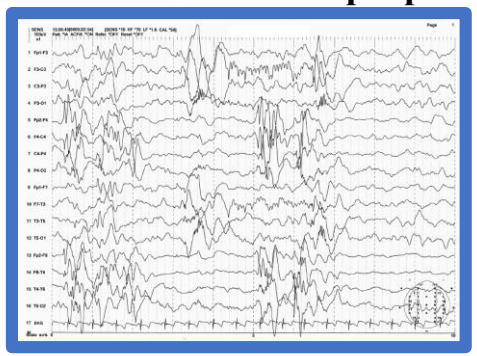


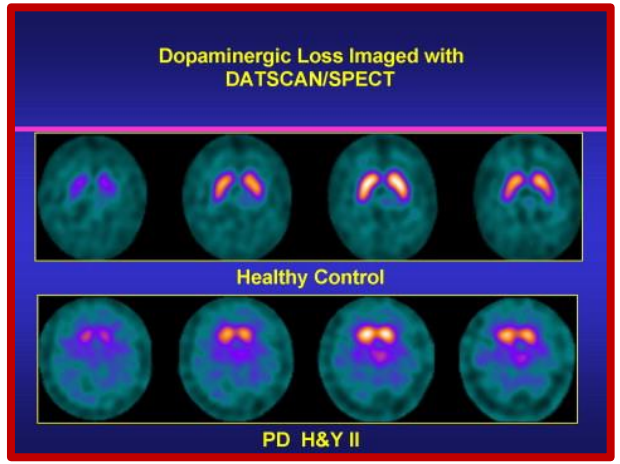
Arteriovenous malformation



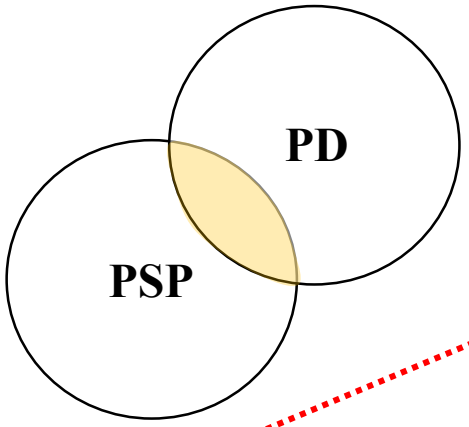
Atrophy of the Cerebellum

Epilepsy





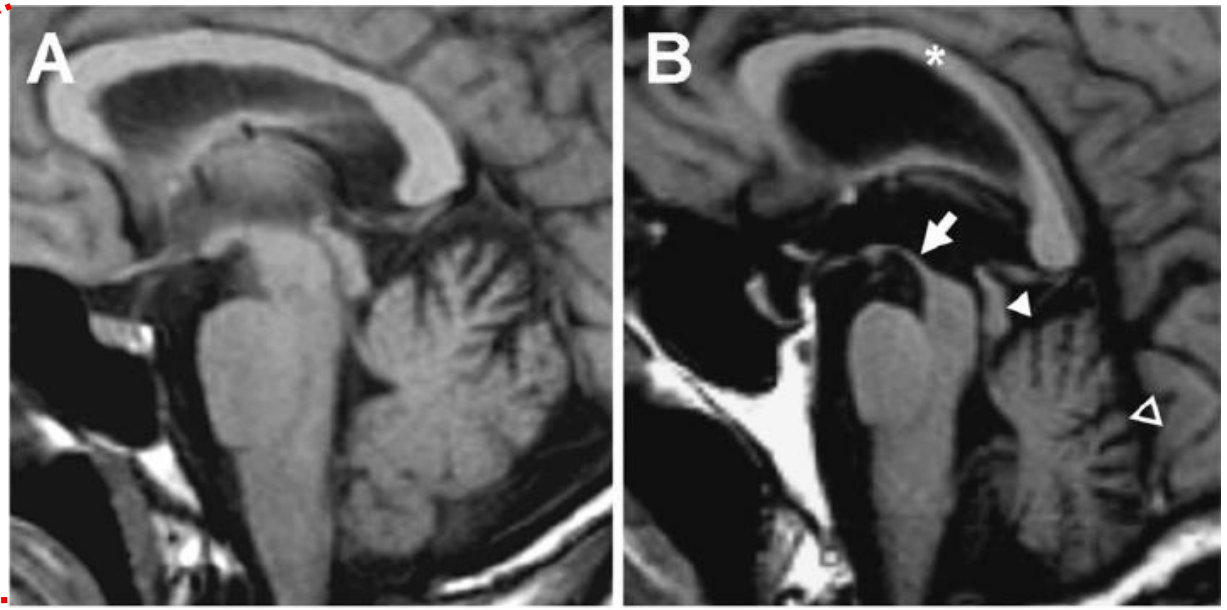
… to make a differencial diagnosis between BRAIN diseases



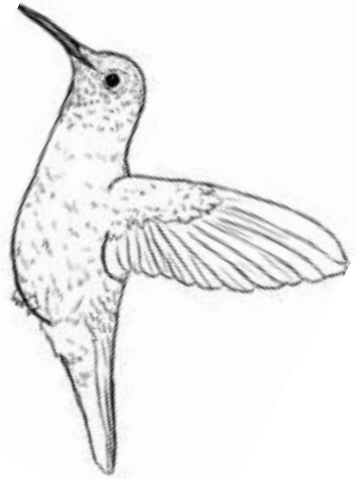
RESEARCH HIGHLIGHTS

MOVEMENT DISORDERS

Imaging differentiates progressive supranuclear palsy from Parkinson disease



PD PSP

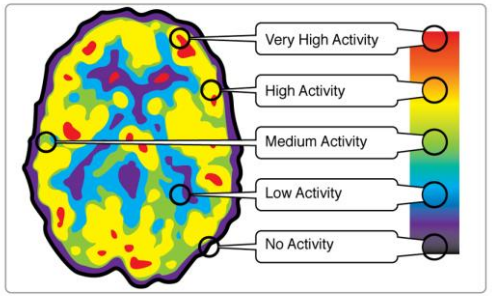


…and to make a diagnosis before the appearance of disease symptoms in RARE diseases

Eur J Nucl Med Mol Imaging. 2012 Jun;39(6):1030-6. doi: 10.1007/s00259-012-2114-z. Epub 2012 Apr 12.

18F-FDG PET uptake in the pre-Huntington disease caudate affects the time-to-onset independently of CAG expansion size.

Ciarmiello A[1], Giovacchini G, Orobello S, Bruselli L, Elifani F, Squitieri F.



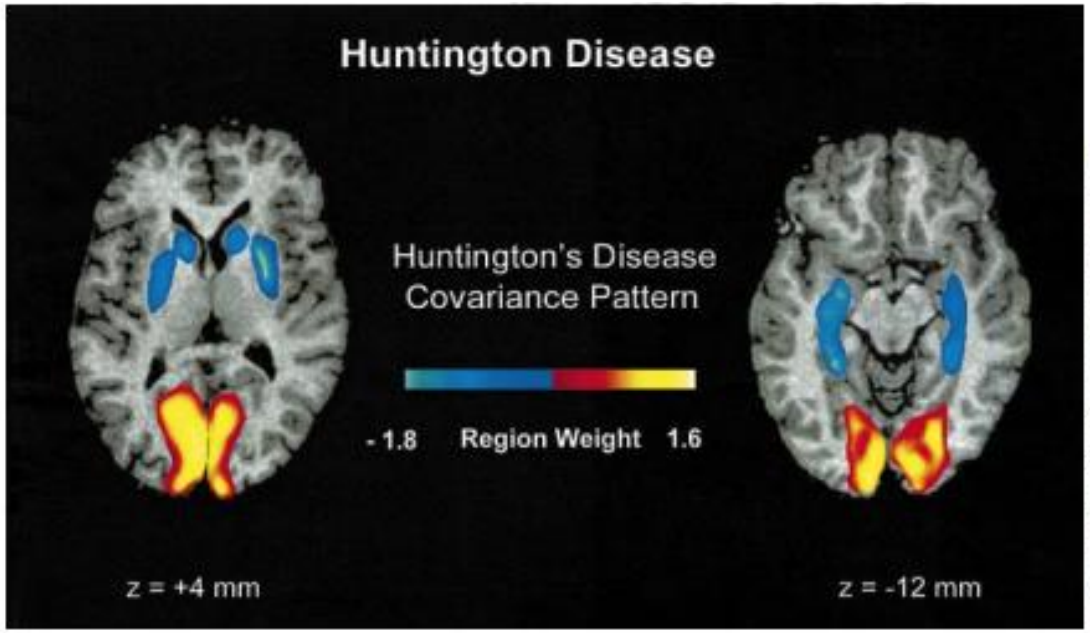


FIGURE 1. Display of region weights of scaled SSM topography associated with HD overlaid on standardized Talairach MRI sections (*13,20*). Regional metabolic covariance pattern was identified in PET data from combined group comprised of 6 neurologically normal, presymptomatic HD gene carriers with normal striatal D_2 receptor binding and 8 age-matched, gene-negative control subjects. Topography was characterized by relative decreases in striatal and mediotemporal metabolism covarying with metabolic increases in occipital region. We designated this topography as HDRP.

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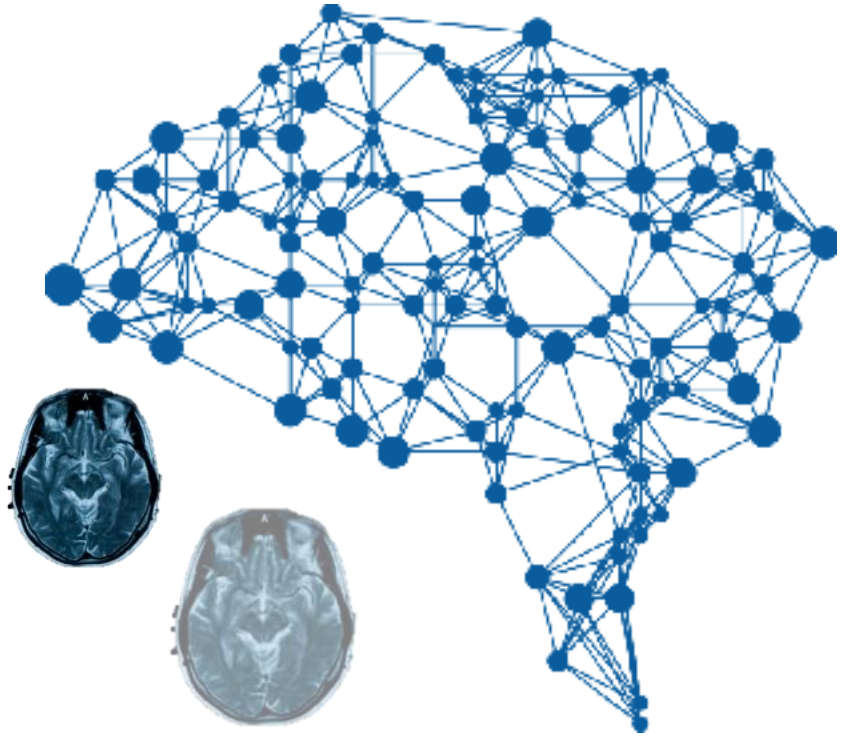
Is the DEEP LEARNING



the Solution ?

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DEEP LEARNING and its increasing popularity in medical images

SCIENTIFIC REPORTS

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Computer-Aided Diagnosis with Deep Learning Architecture: Applications to Breast Lesions in US ... Pulmonary Nodules in

Medical Image Analysis 35 (2017) 303–312

Contents lists available at ScienceDirect

Medical Image Analysis

journal homepage: www.elsevier.com/locate/media

ELSEVIER

Large scale deep learning for computer aided detection of mammographic lesions

Thijs Kooi[a,*], Geert Litjens[a], Bram van Ginneken[a], Albert Gubern-Mérida[a], Clara I. Sánchez[a], Ritse Mann[a], Ard den Heeten[b], Nico Karssemeijer[a]

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ABSTRACT

Recent advances in machine learning yielded new te
sulted in highly successful applications in many patt
speech recognition. In this paper we provide a hear
mammography CAD system, relying on a manually
work (CNN), aiming for a system that can ultimately
trained on a large data set of around 45,000 images

Neurocomputing 216 (2016) 700–708

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SCIENTIFIC REPORTS

OPEN

Deep learning as a tool for increased accuracy and efficiency of histopathological diagnosis

Geert Litjens[1], Clara I. Sánchez[2], Nadya Timofeeva[1], Iringo Kovacs[3], Christina Hulsbergen-van de Kaa[3], Jeroen van der Laagtegaal[1], Jeroen van der Laak[1,2] & Bram van Ginneken[2] &

NeuroImage: Clinical 15 (2017) 633–643

Contents lists available at ScienceDirect

NeuroImage: Clinical

journal homepage: www.elsevier.com/locate/ynicl

ELSEVIER

Fully automatic acute ischemic lesion segmentation in DWI using convolutional neural networks

Liang Chen[a,b,*], Paul Bentley[b], Daniel Rueckert[a]

[a] BioMedIA Group, Department of Computing, Imperial College London, 180 Queen's Gate, London SW7 2AZ
[b] Division of Brain Sciences, Department of Medicine, Imperial College London, Fulham

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Still little is known about the BRAIN

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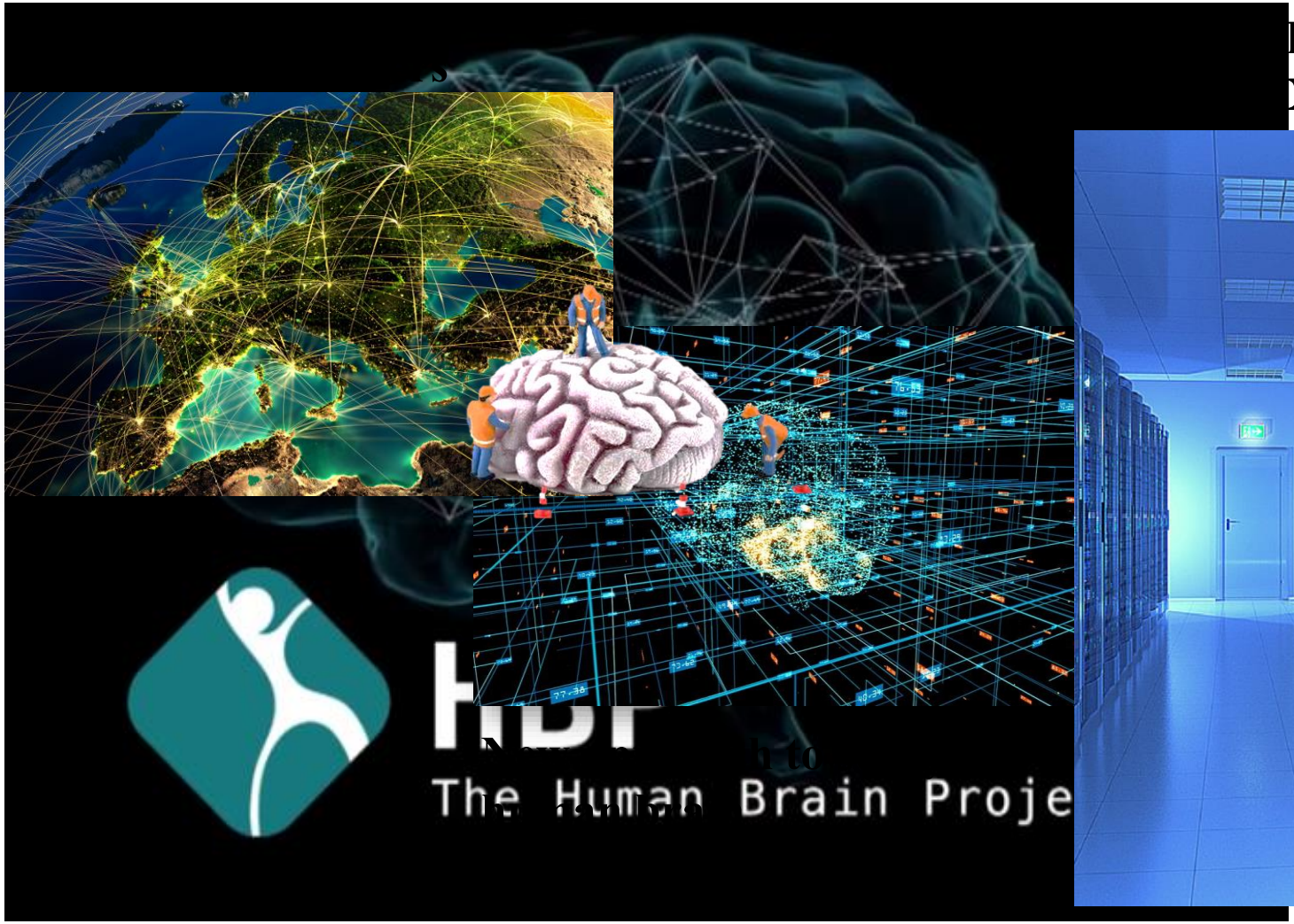
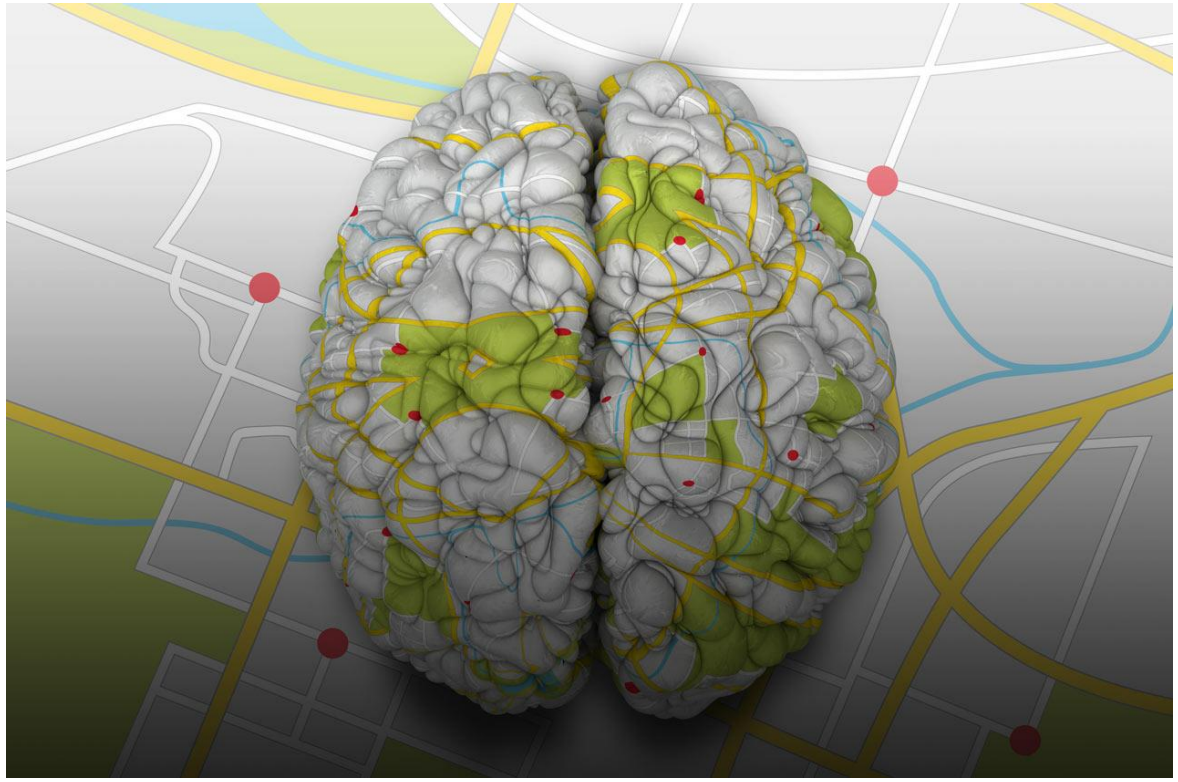
Computer model of Human brain



To explore and better undestand human brain and its diseases and use this knowledge to build new computing technologies

The BIG Challenge



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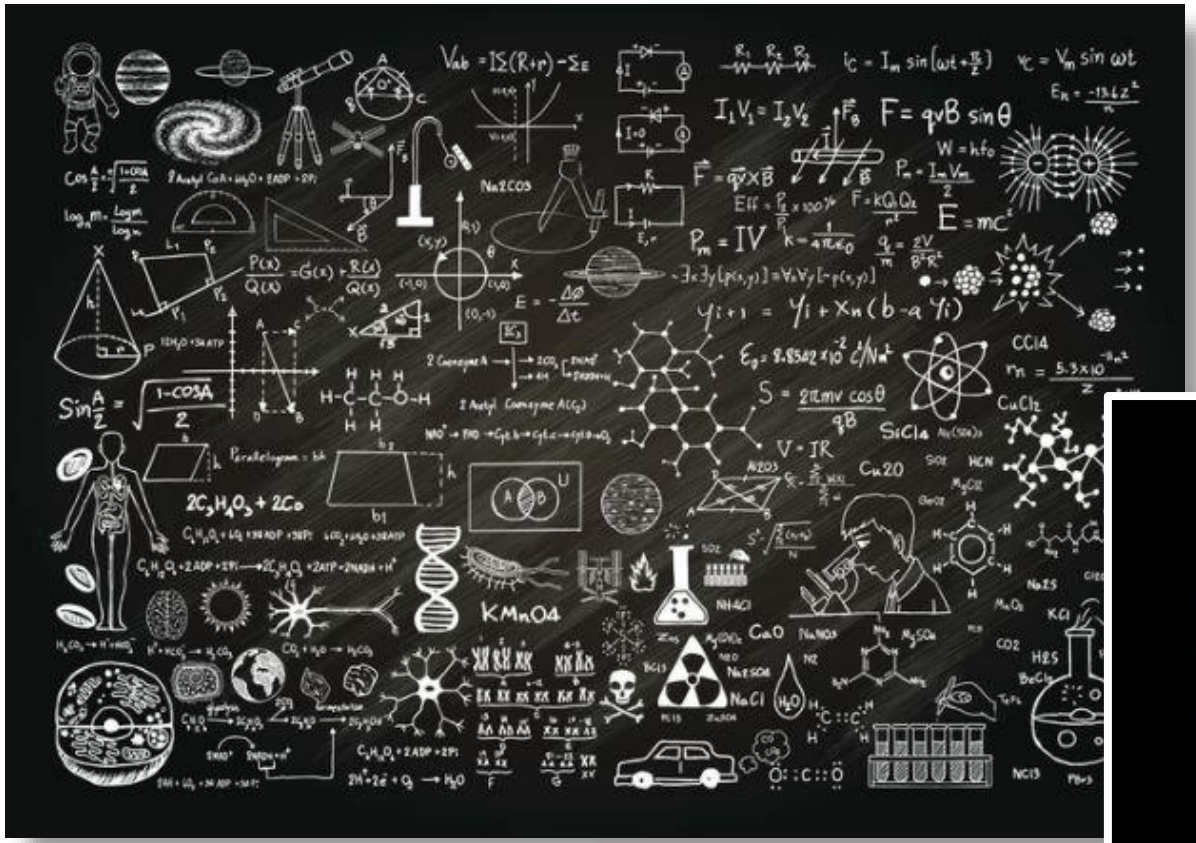
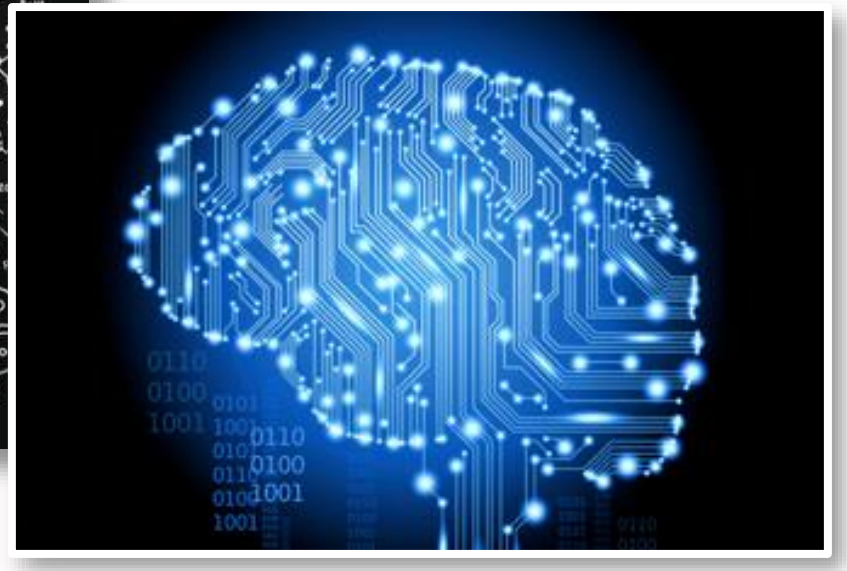
To achieve the goal of more appropriate diagnosis….



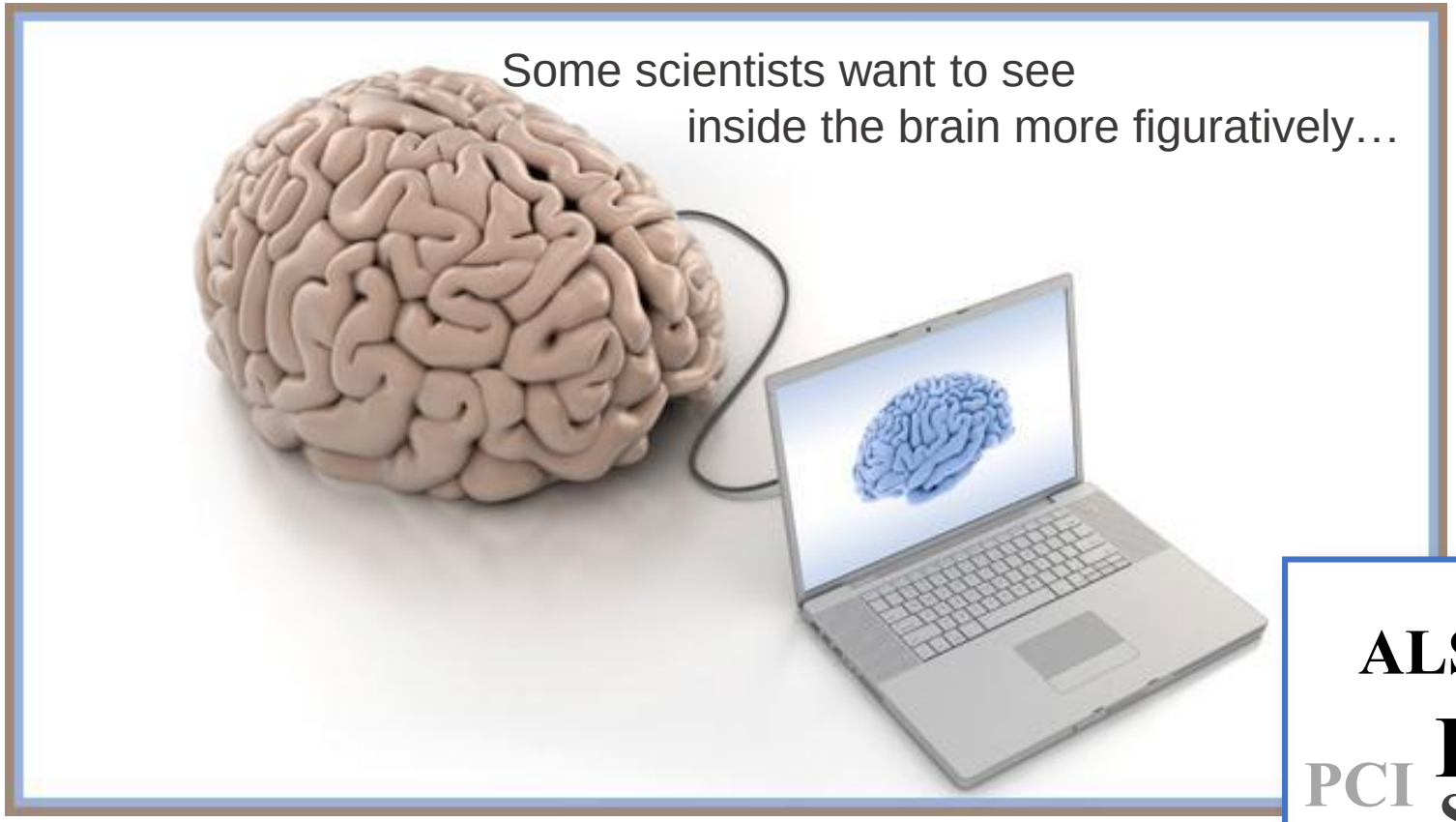
Medical Science needs your help….



we **count** on you

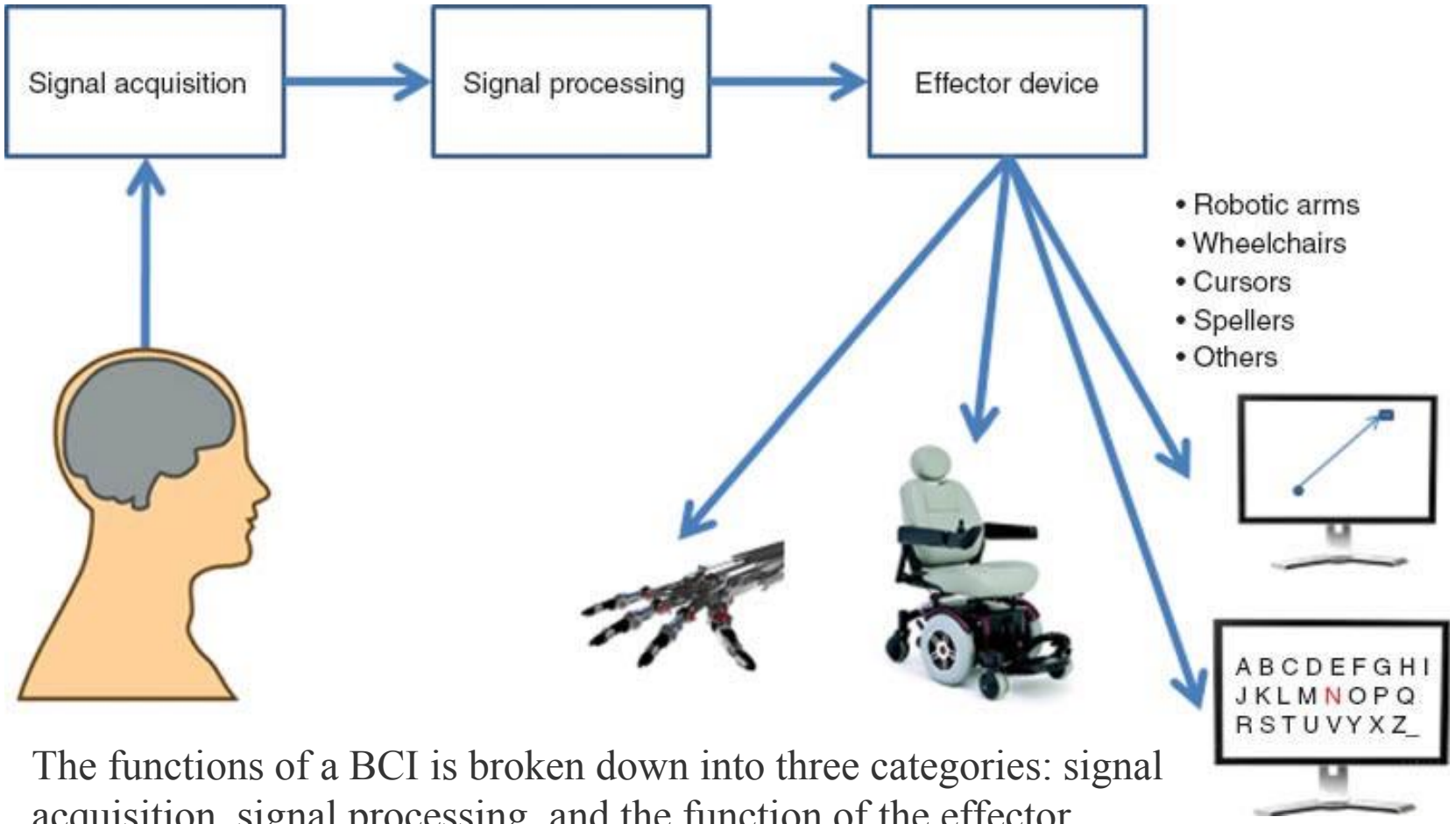
DECODING THOUGHTS

BRAIN-COMPUTER INTERFACE (BCI)



Paraplegia
ALS PD
HD DOC
PCI
Stroke

BCI systems are designed to acquire electrical signals from the **BRAIN** and process them into commands for **effector devices** to perform the patient's desired action.



The functions of a BCI is broken down into three categories: signal acquisition, signal processing, and the function of the effector device. The strenght of this technology is that a signal coming from one single neuron may be converted in a complete action.

BCI and its success to provide patient-controlled compensation for the loss of muscle movement and communication

BCIs can now even allow humans with paraplegia (paralysis of all four limbs) to control a robotic arm through thought alone, or allow users to spell out words on a computer screen using just their mind.

At-home devices have huge implications to reduce costs for hospitals and insurance companies, and—if the technology is up to par—results in a more natural set of data for the patient.

ideally

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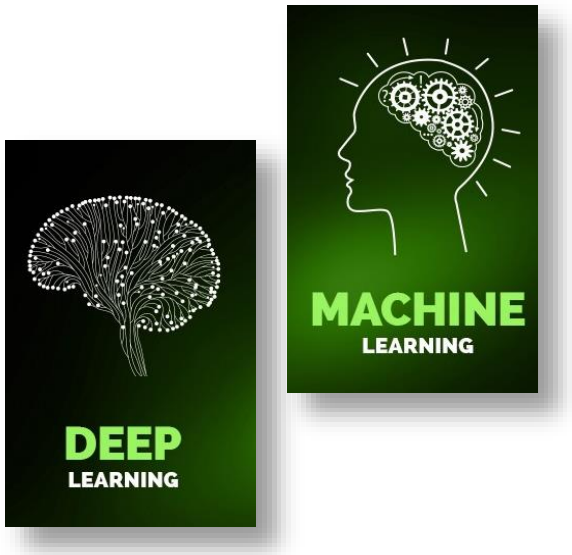


The REVOLUTION in the diagnosis of RARE DISEASES

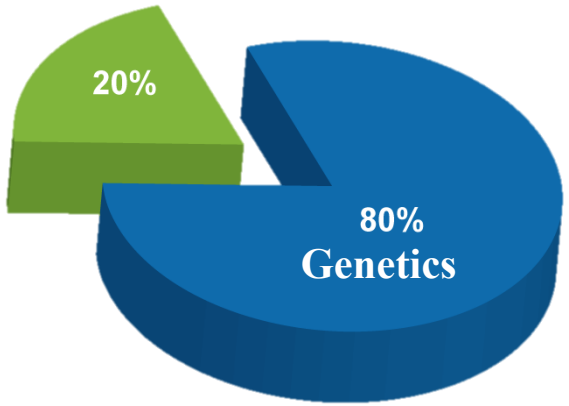
More than

300 million

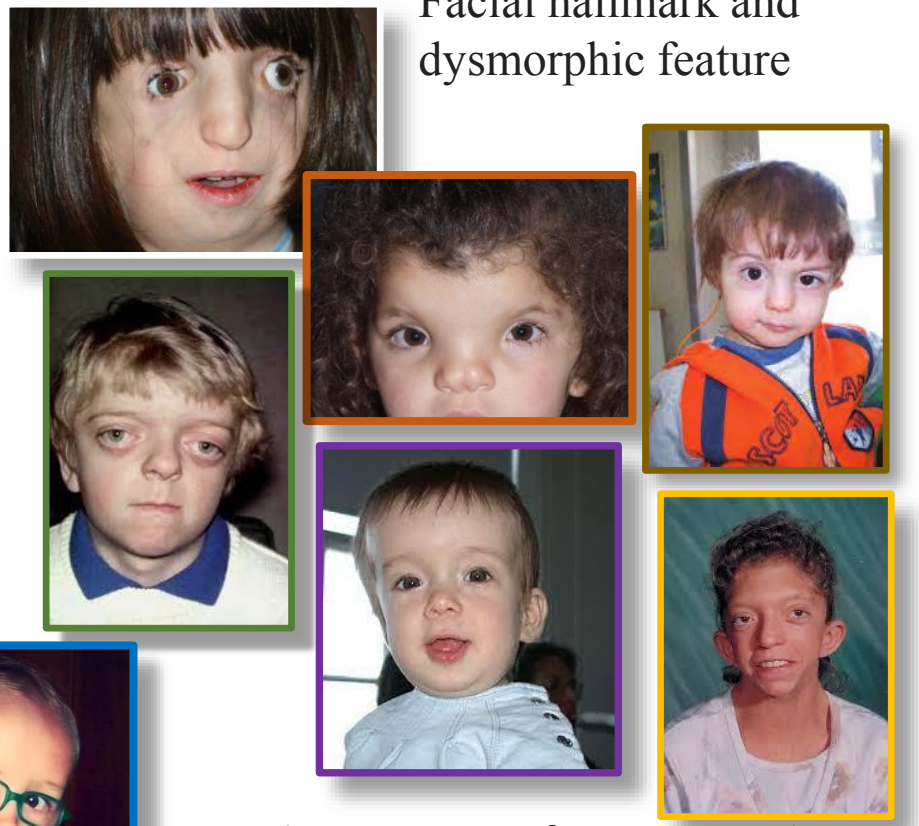
people worldwide

are affected by RARE DISEASE

Rare Diseases



Facial hallmark and dysmorphic feature



Facial signatures of some rare diseases can be subtle and difficult to diagnose, even for a trained professional

> *Rare diseases often go* **unrecognized** *precisely because of their rarity, resulting in a delay in diagnosis.*

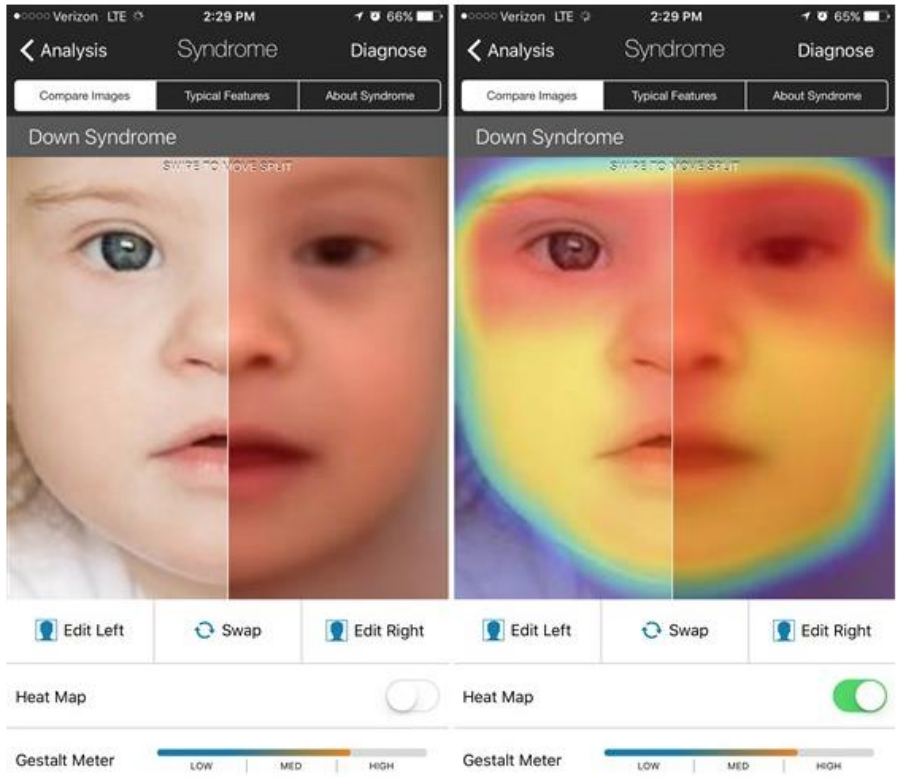
Most of patients with **rare diseases** often suffer a signficant **delay** in **diagnosis**. Many diseases are so rare that a general physician is unlikely to see a single case in his career.

The symptoms of rare diseases are often atypical and can point in many different directions. As a result, the correct diagnosis is often delayed for several years.

LETTERS | FOCUS
https://doi.org/10.1038/s41591-018-0279-0
nature medicine

Identifying facial phenotypes of genetic disorders using deep learning

Yaron Gurovich[1*], Yair Hanani[1], Omri Bar[1], Guy Nadav[1], Nicole Fleischer[1], Dekel Gelbman[1], Lina Basel-Salmon[2,3], Peter M. Krawitz[4], Susanne B. Kamphausen[5], Martin Zenker[5], Lynne M. Bird[6,7] and Karen W. Gripp[8]



Facial recognition app Face2Gene is being used by doctors to diagnose rare diseases. Courtesy FDNA

Deep learning-based new technology that can help clinicians and geneticists to diagnose rare disease based on patient facial phenotypes

The technology converts a patient photo into de-identified mathematical facial descriptors (facial descriptors)

The latest medical technology that leverages the power of big data to make better diagnoses and more accurate predictions.

Face2Gene's system uses a machine-learning algorithm. It learns from every new face it scans. The more data it acquires through its use, the more accurate should be the diagnoses.

Snapping a quick photo of the child's face within a matter of seconds, the app generated a list of potential diagnoses.

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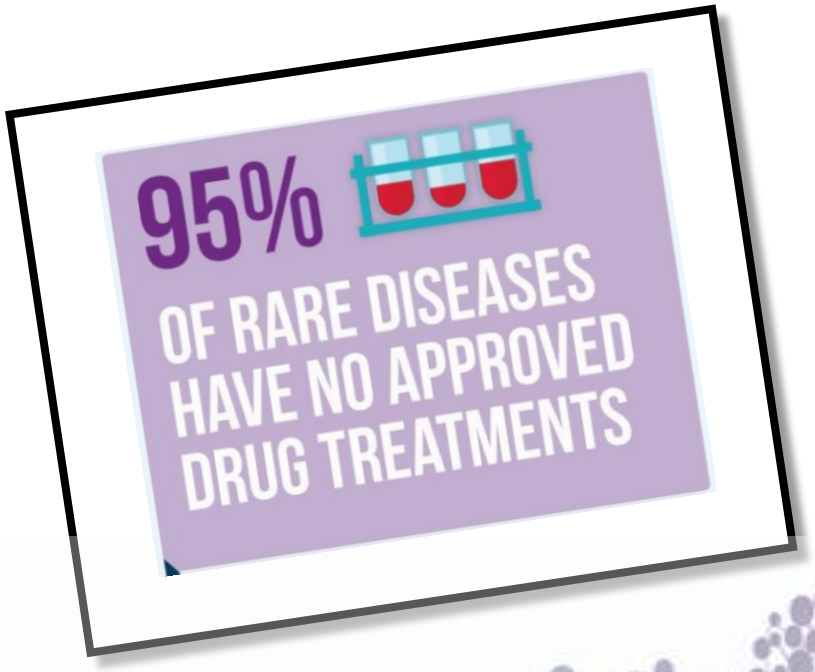
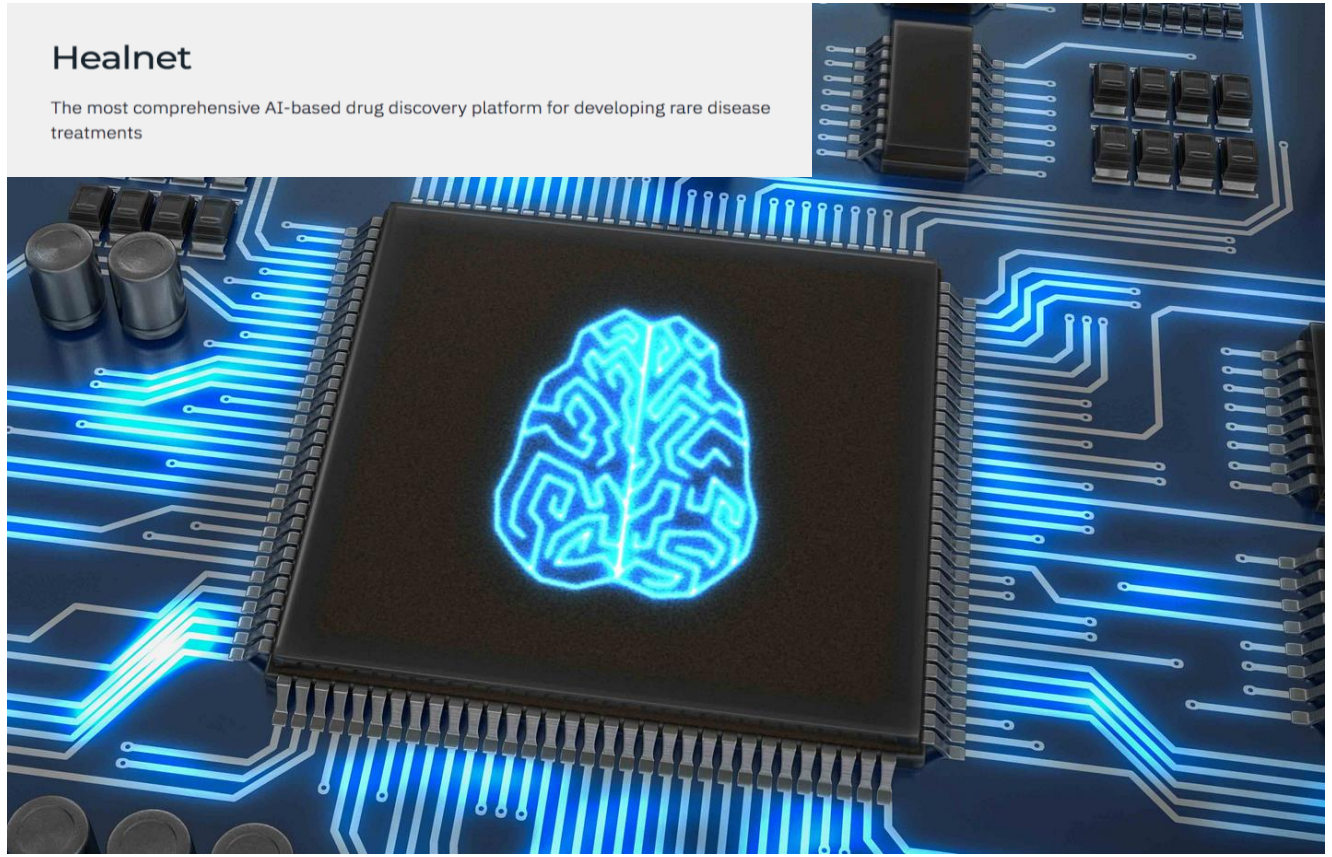


RARE DISEASES do not have cure

A Startup Using Artificial Intelligence To Find Cures For some Rare Diseases

Healx is a tech company from the Cambridge Cluster (UK), focussed on accelerating treatments for rare diseases. It integrates artificial intelligence with deep pharmacology to translate therapies into the clinic within 24 months. Thereby, dramatically reducing the time and cost compared to conventional drug discovery. To achieve this goal, Healx developed the most comprehensive AI-based drug discovery platform for rare diseases: Healnet. With the objective to translate 100 rare disease treatments towards the clinic by 2025.



"Our mission was to apply our technology and approach to identify and validate already approved drugs that may treat some of the most severe symptoms of the disease"

Take home message…

Human Brain is waiting for you to give us all the best it kept hidden up to now….

Fondazione NEUROMED

Thank You For The Attention