WORKSHOP ON PERSONALISED MEDICINES

What does ‘personalised’ mean to patients and healthcare professionals

Julian Isla

Microsoft. Consulting Services

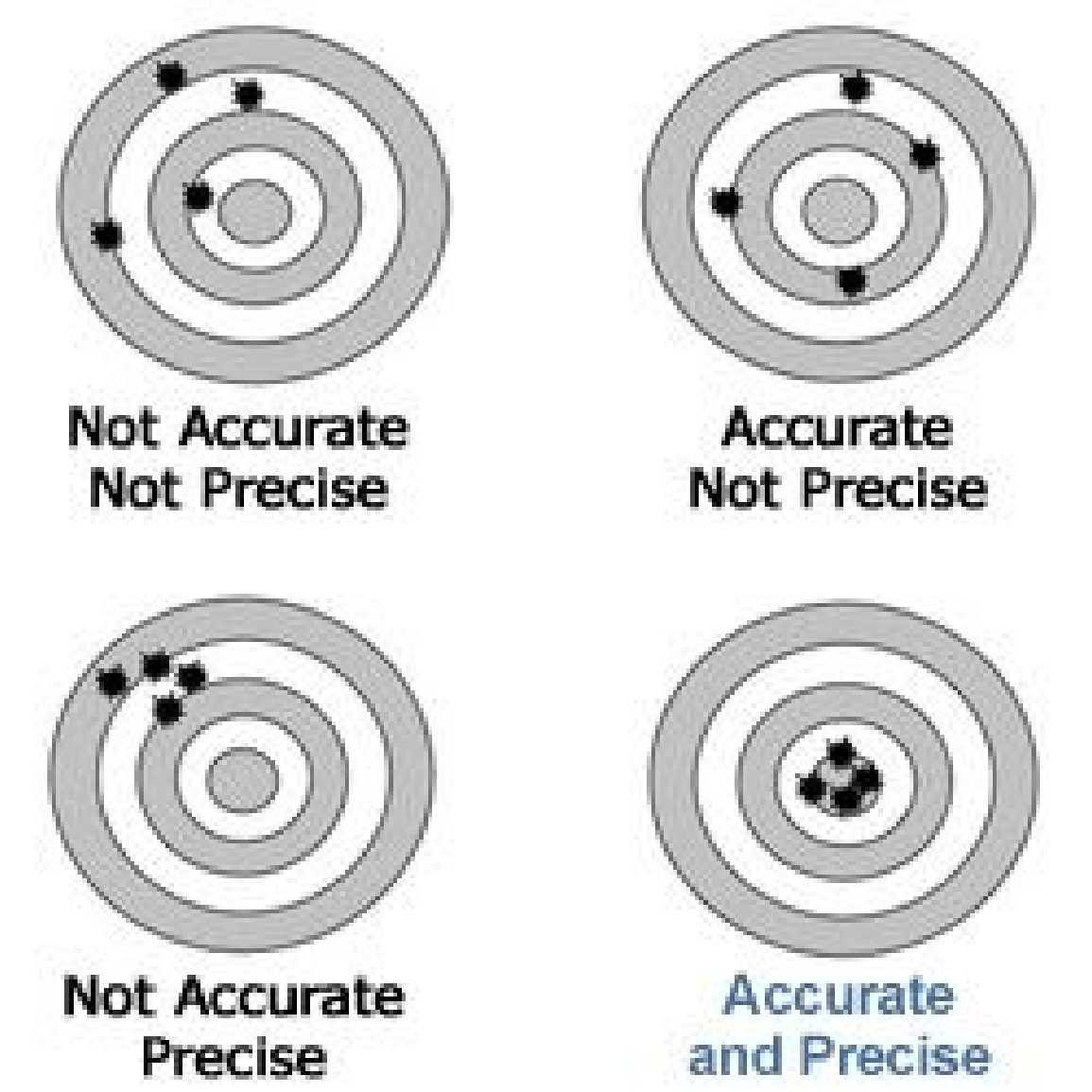
Dravet Syndrome European Federation. Chairman

EURORDIS Therapeutic Action Group

EMA delegate. COMP patient representative

Scientific Advisory Board

Personalised = Precission

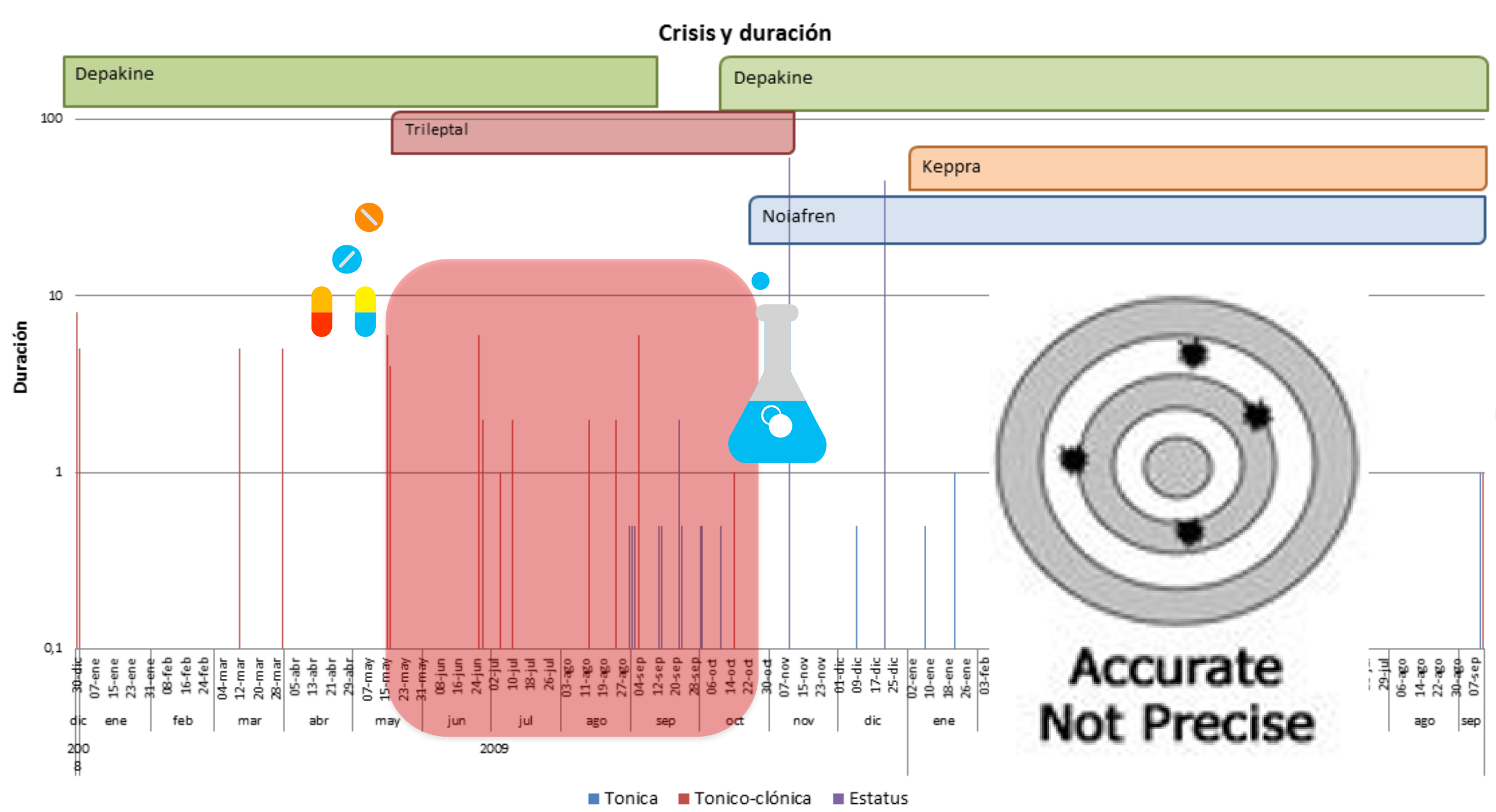


According to the National Institutes of Health (NIH), precision medicine is "*an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person*"

Medical treatment

Sergio
8 years old
Dravet Syndrome

A real example of lack of precision


Crisis y duración
Depakine
Depakine
Trileptal
Keppra
Noiafren
100
10
1
0,1
Duración
Accurate
Not Precise
dic
ene
feb
mar
abr
may
jun
jul
ago
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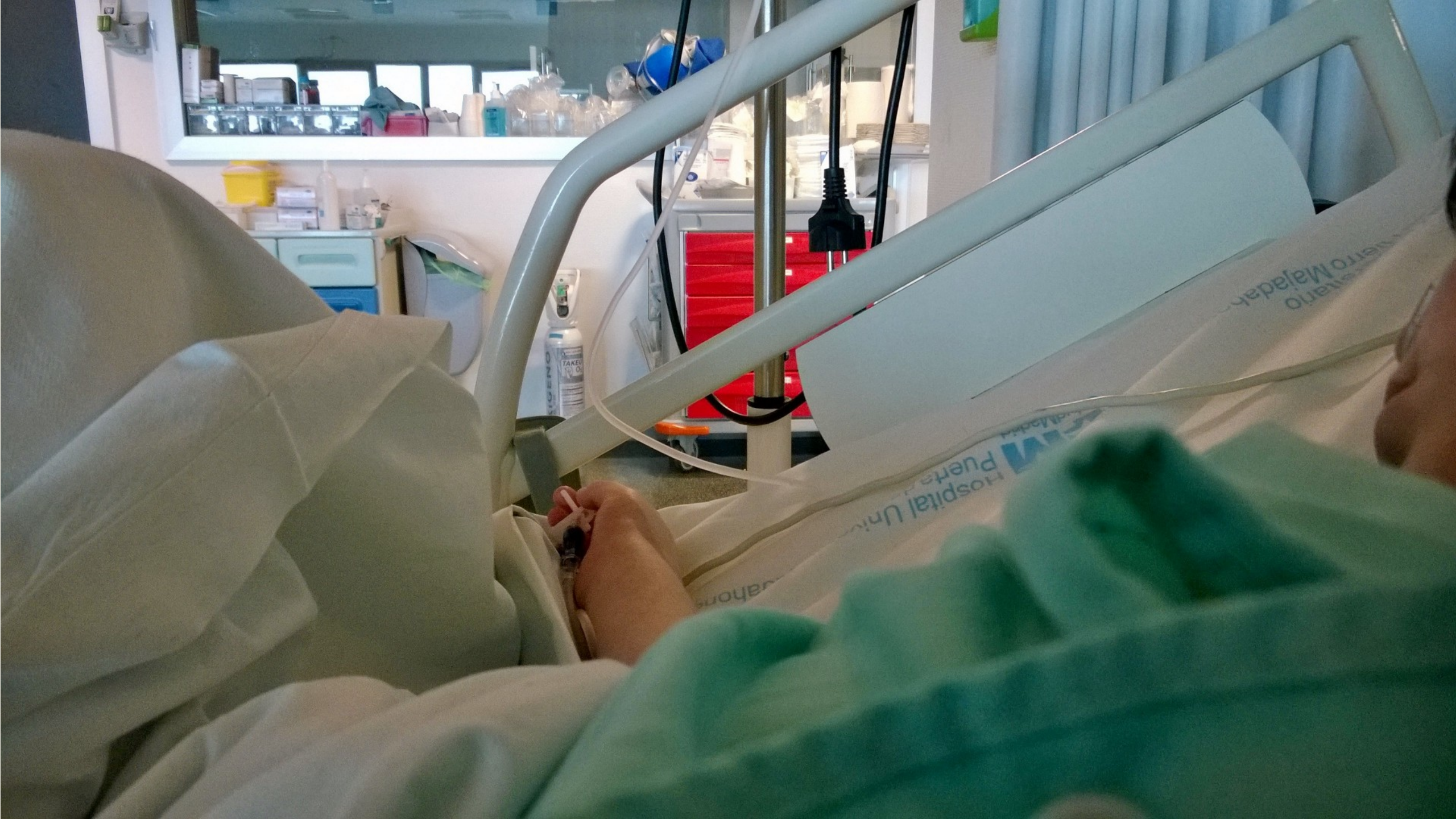
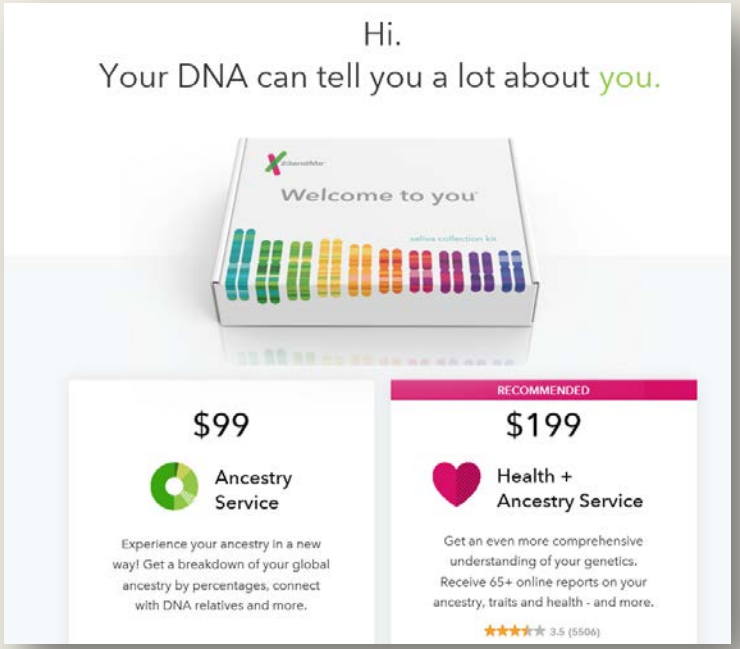
sep
2008
2009
Tonica
Tonico-clónica
Estatus

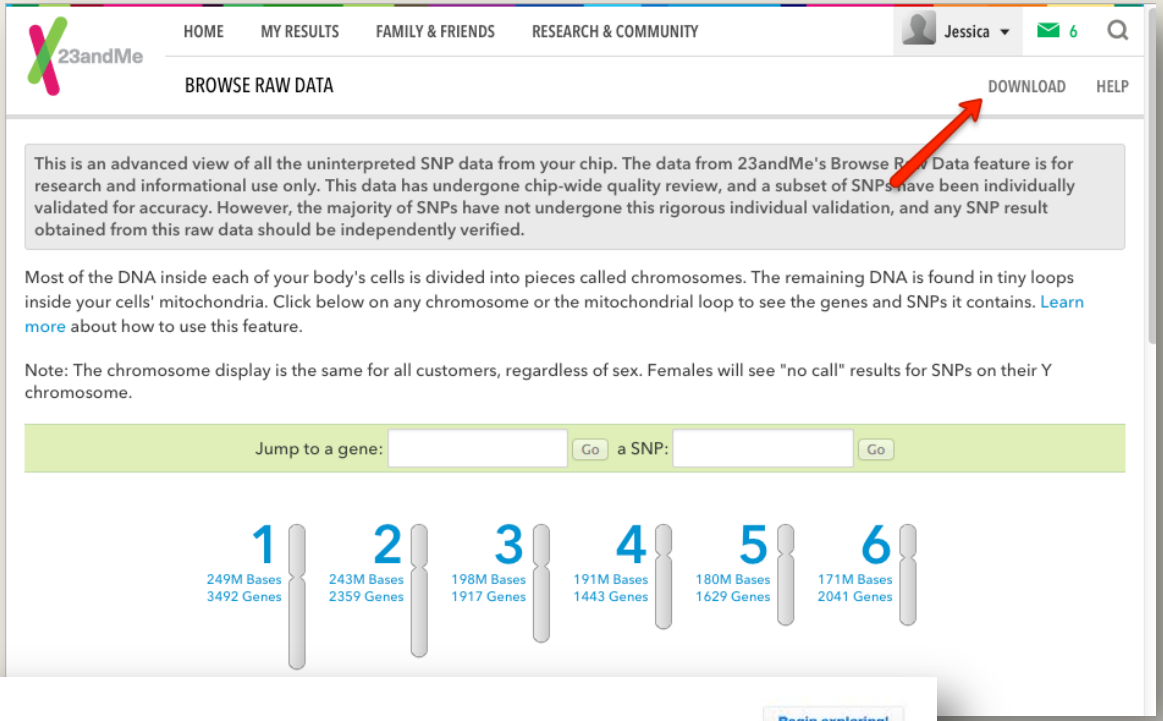


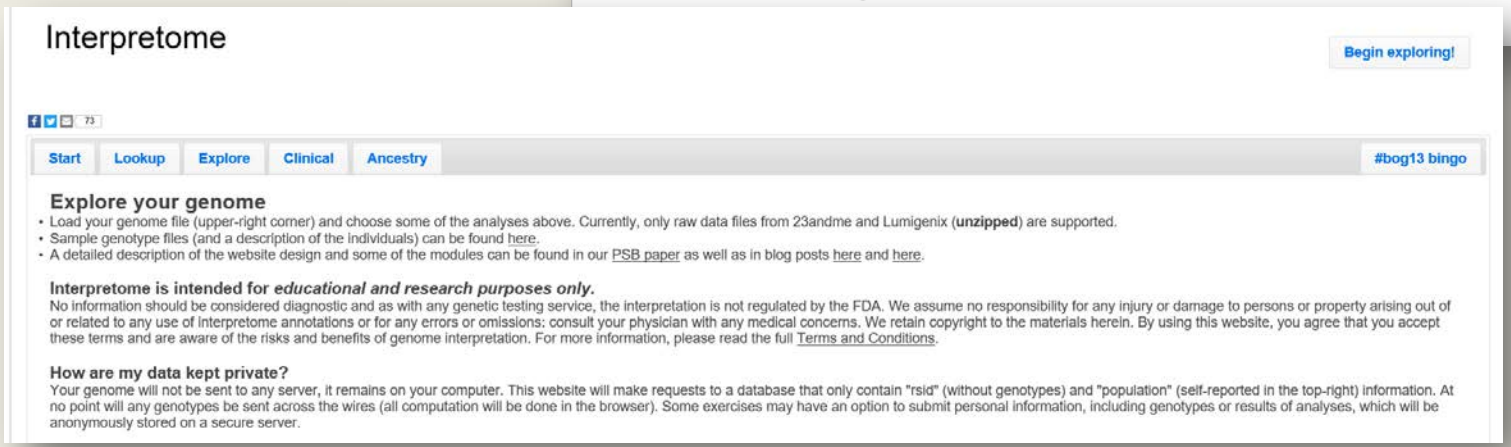

WHERE ARE WE NOW?
YOU ARE
HERE

Individual variability in genes
Environment
Lifestyle
Medical treatment

Individual variability in genes







Internet of DNA

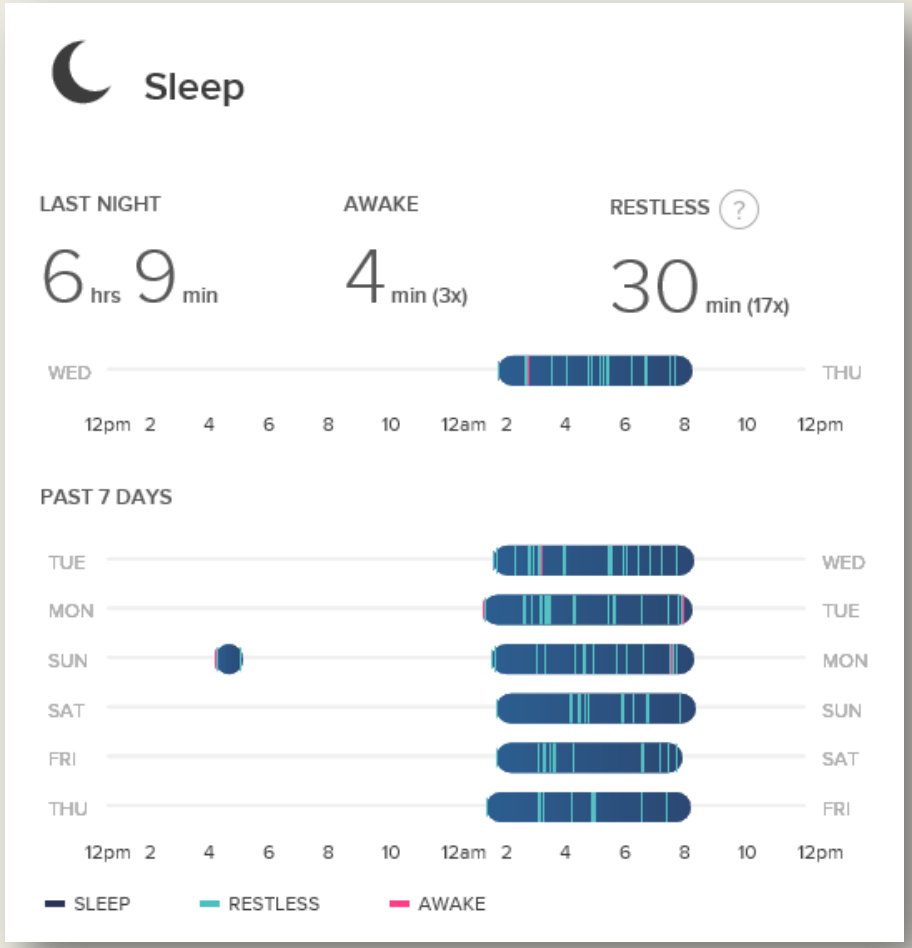
A global network of millions of genomes could be medicine's next great advance.

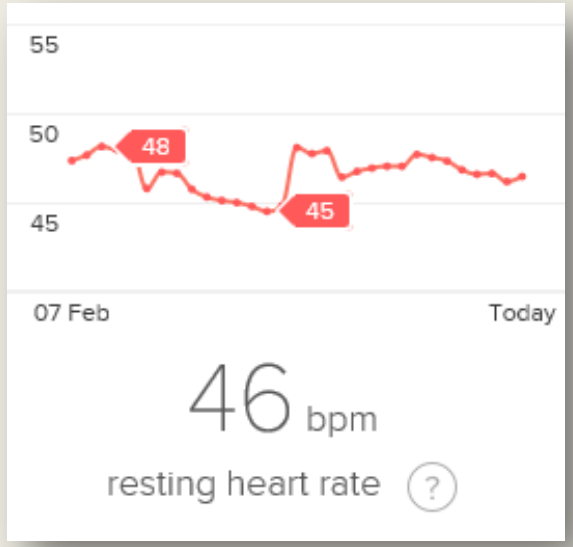
Availability: 1-2 years

Breakthrough | Why It Matters | Key Players

10 Breakthrough Technologies 2015

Environment and lifestyle





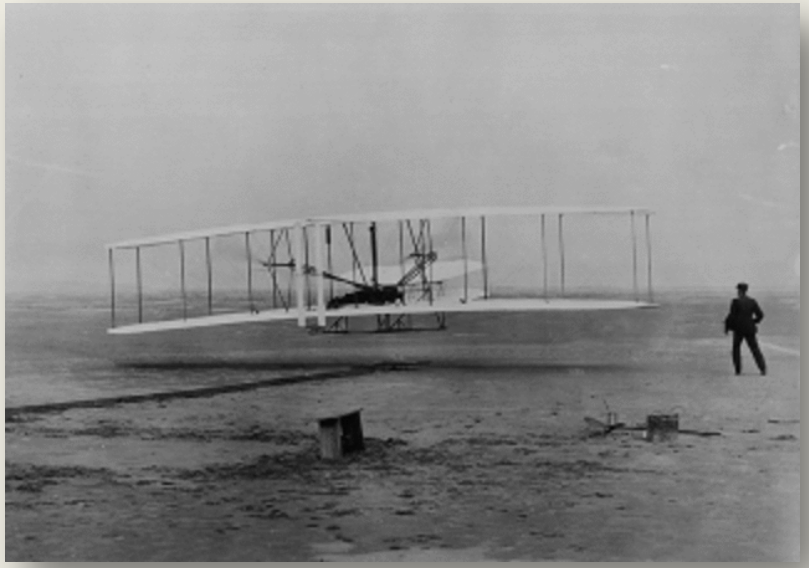
"Healthcare is not a science problem, it's an information problem"

Thomas Goetz

“Healthcare is a science **and** information problem"

Learning from mistakes

Feedback loop




Airbus A380
The A380 will be a true colossus, with models seating 555 people.
The A380 will be the world's first airliner with passenger cabins on two full decks. Airbus argues that explosive growth in air traffic will fuel a healthy demand for super-jumbo jets. Boeing disagrees, and predicts the A380 will turn into a white elephant.
Suites with beds, as shown above, or private lounge suites, below, could be options for passengers once the aircraft reaches cruising altitude.
Three options for airlines buying the A380 are retail space, a hair salon, above, and a bar, below.
Seats vs. salons
Gift shops, lounges, casinos, restaurants — Airbus is touting endless uses for the A380's capacity. But will airlines opt for seats or salons?
Airbus Industrie Photographs
Wing span 261 ft., 10 in.
A380
Length 239 ft., 3 in.
Wing span 211 ft., 5 in.
747-400ER
Length 231 ft., 10 in.
Upper deck
Main deck
Cargo area
Wing
Wing
Cross section for the A380 series
Using space
The forward and rear cargo holds could be used for passenger or crew facilities.
CREW REST SPACE
Bunks
Stairs
Passenger space combining bar, private office spaces and lavatory
Private offices
Bar
Stairs
Lavatory
ECONOMY
BUSINESS
FIRST CLASS
ECONOMY
FIRST CLASS
Cargo area
Cargo area
Cargo area
Seating
REAR
MIDDLE
FRONT
The A380 could have 10-abreast seating in coach and 6-abreast in first and business sections, depending on airlines' preference.
Sources: Airbus and Boeing

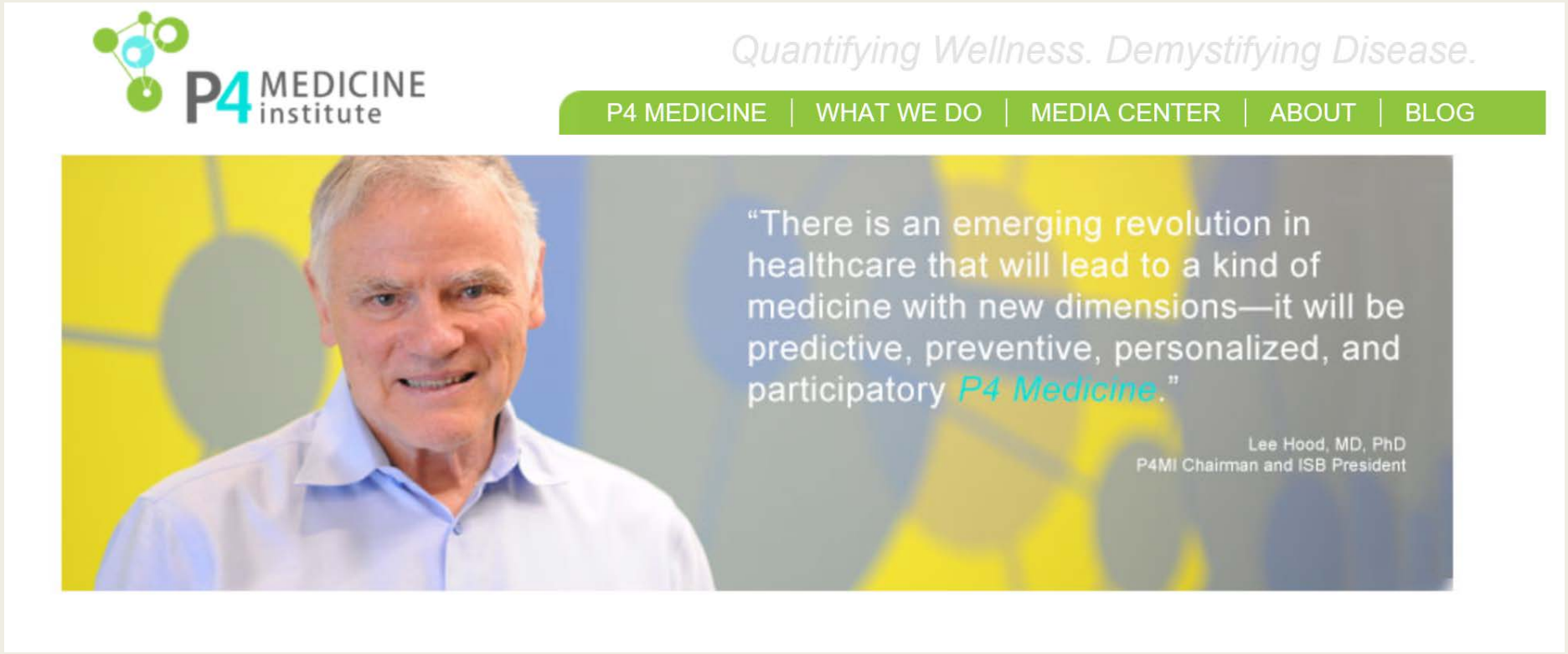
110 years

Patient digital transformation

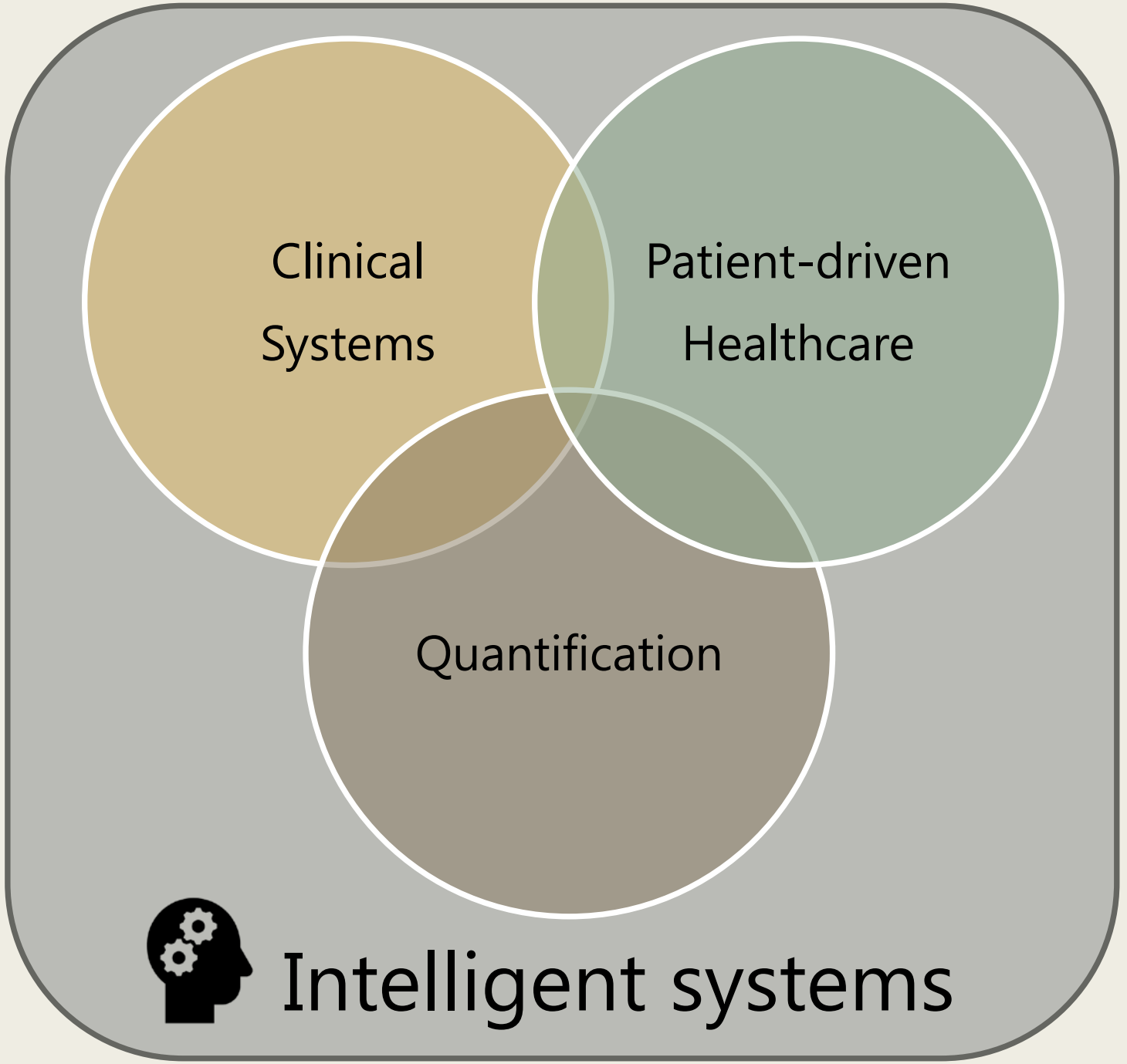
3 2581703423 4457570

P-4 medicine

predictive, preventive, personalized and participatory



Quantifying wellness is the key

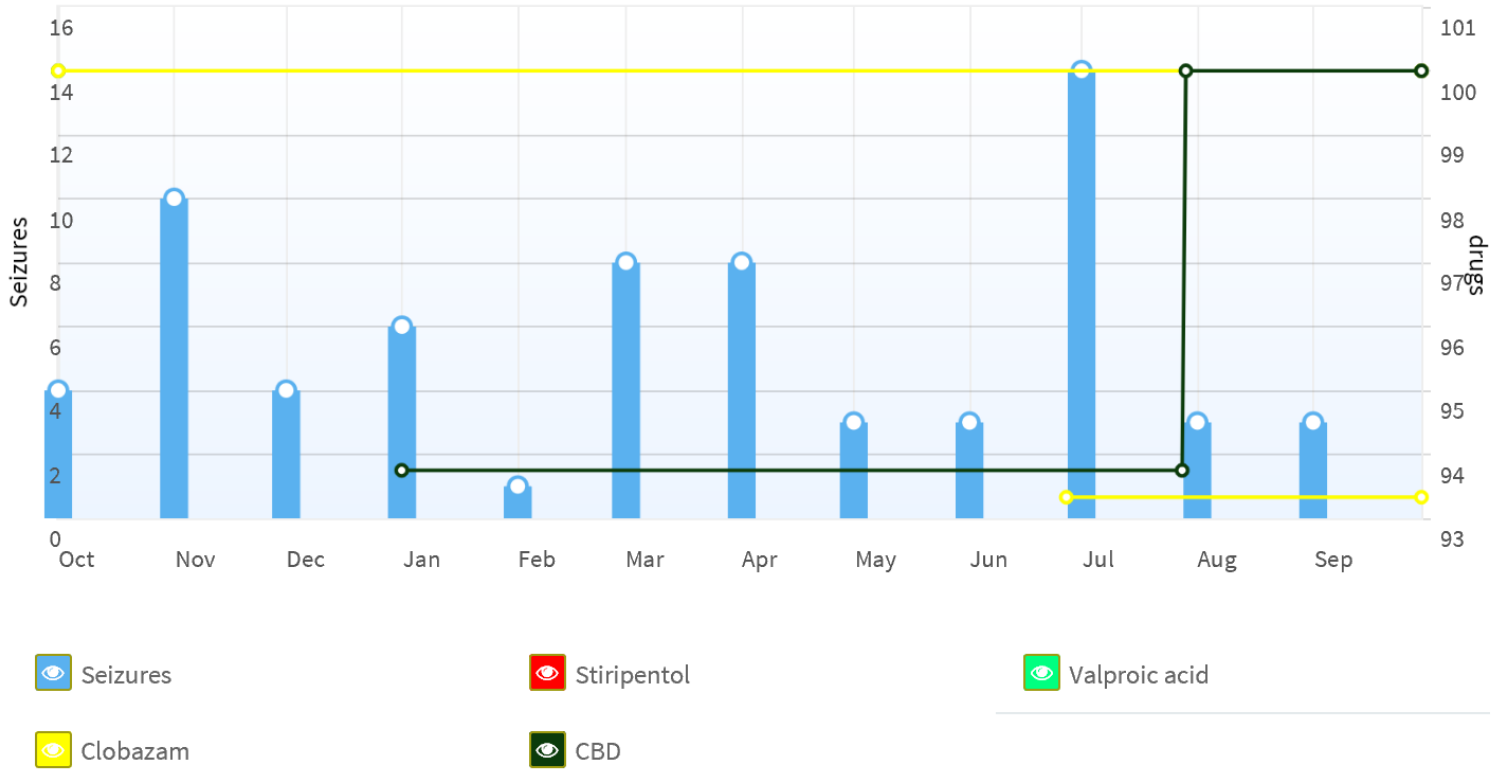

Clinical
Systems
Patient-driven
Healthcare
Quantification
Intelligent systems

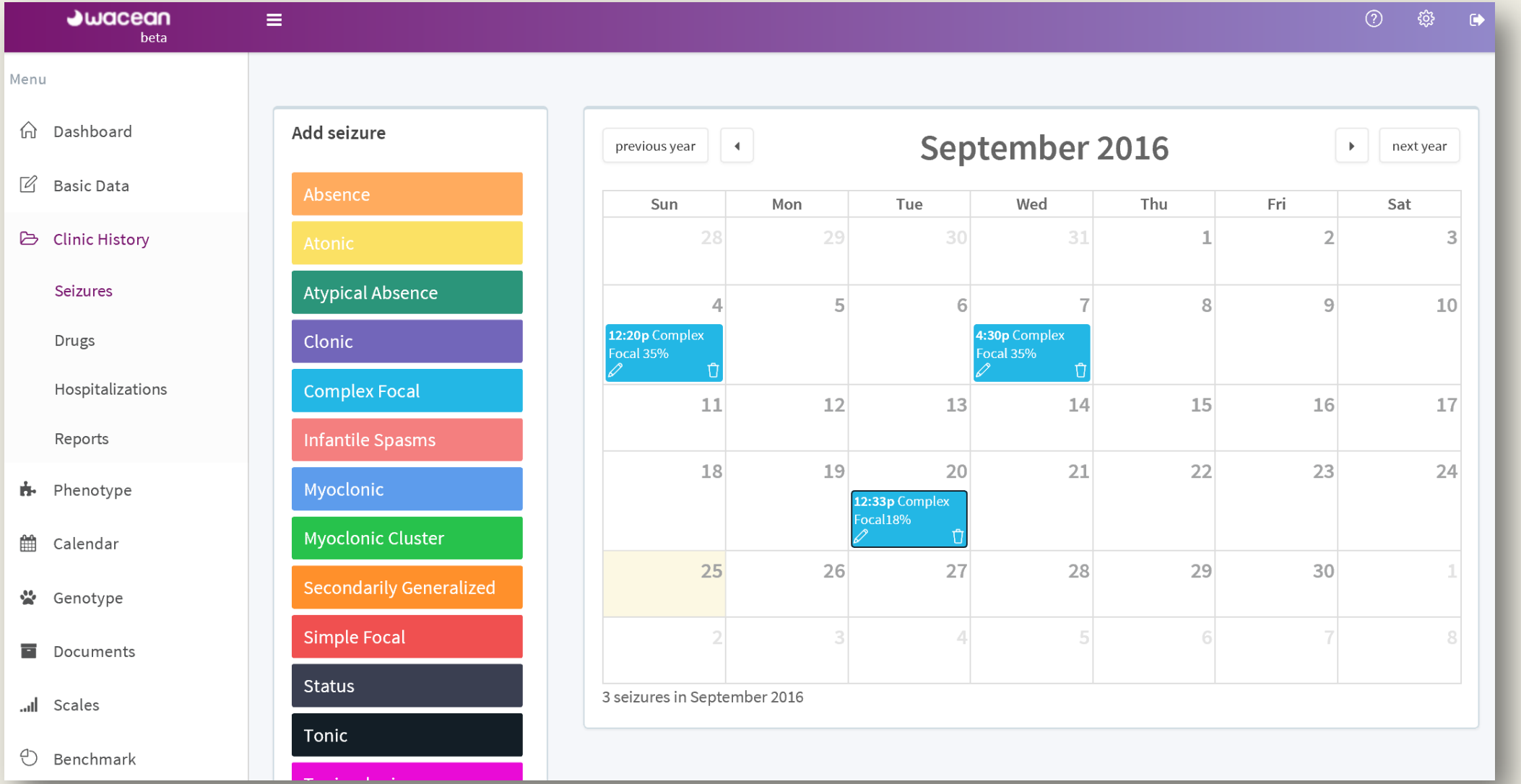
wacean beta

Menu

- Dashboard
- Basic Data
- Clinic History
- Phenotype
- Calendar
- Genotype
- Documents
- Scales
- Benchmark

Seizures and drugs

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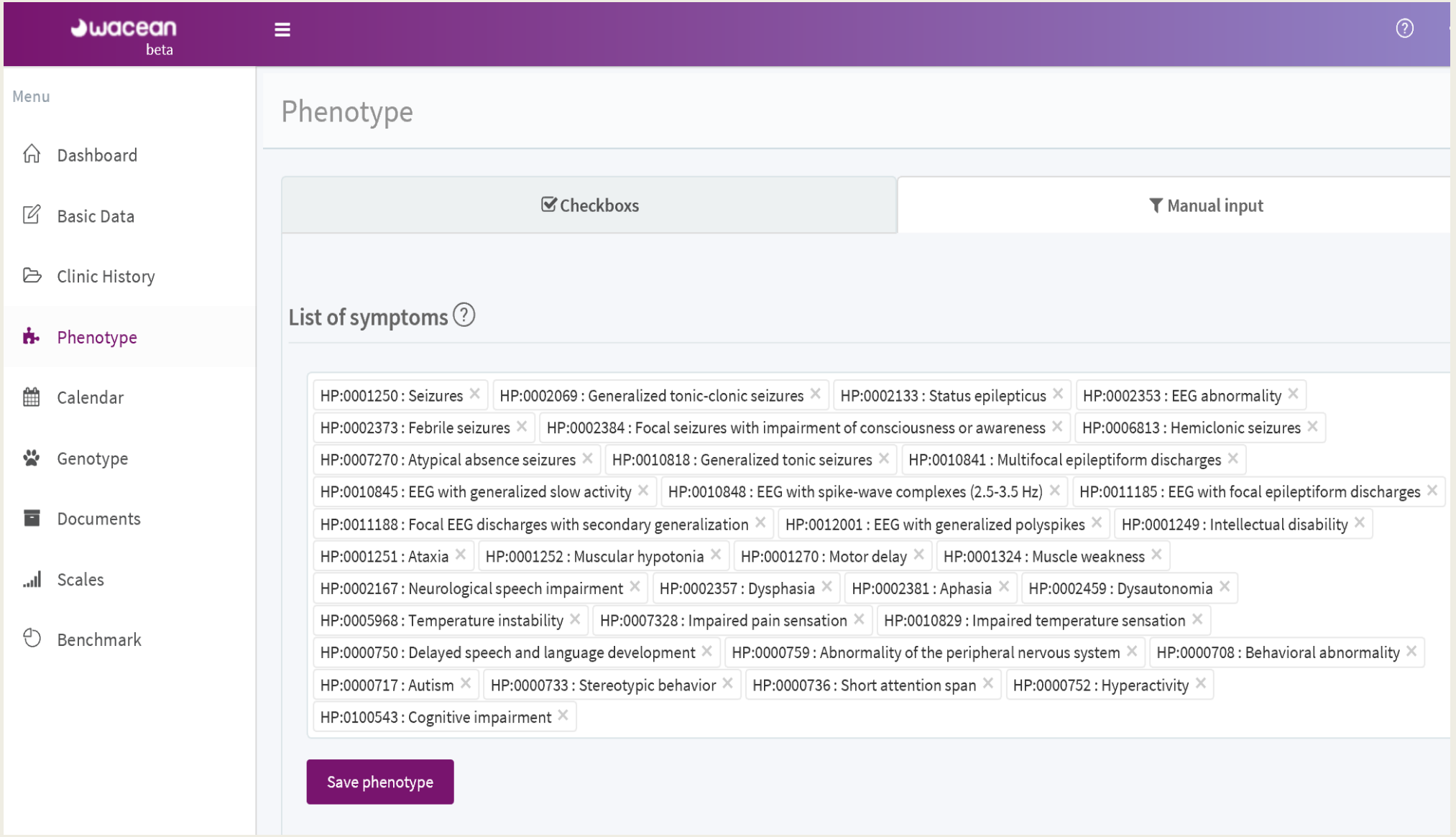
beta
Menu
Dashboard
Basic Data
Clinic History
Seizures
Drugs
Hospitalizations
Reports
Phenotype
Calendar
Genotype
Documents
Scales
Benchmark
Add seizure
Absence
Atonic
Atypical Absence
Clonic
Complex Focal
Infantile Spasms
Myoclonic
Myoclonic Cluster
Secondarily Generalized
Simple Focal
Status
Tonic
previous year
next year
September 2016
Sun
Mon
Tue
Wed
Thu
Fri
Sat
28
29
30
31
1
2
3
4
5
6
7
8
9
10
12:20p Complex Focal 35%
4:30p Complex Focal 35%
11
12
13
14
15
16
17
18
19
20
21
22
23
24
12:33p Complex Focal18%
25
26
27
28
29
30
1
2
3
4
5
6
7
8
3 seizures in September 2016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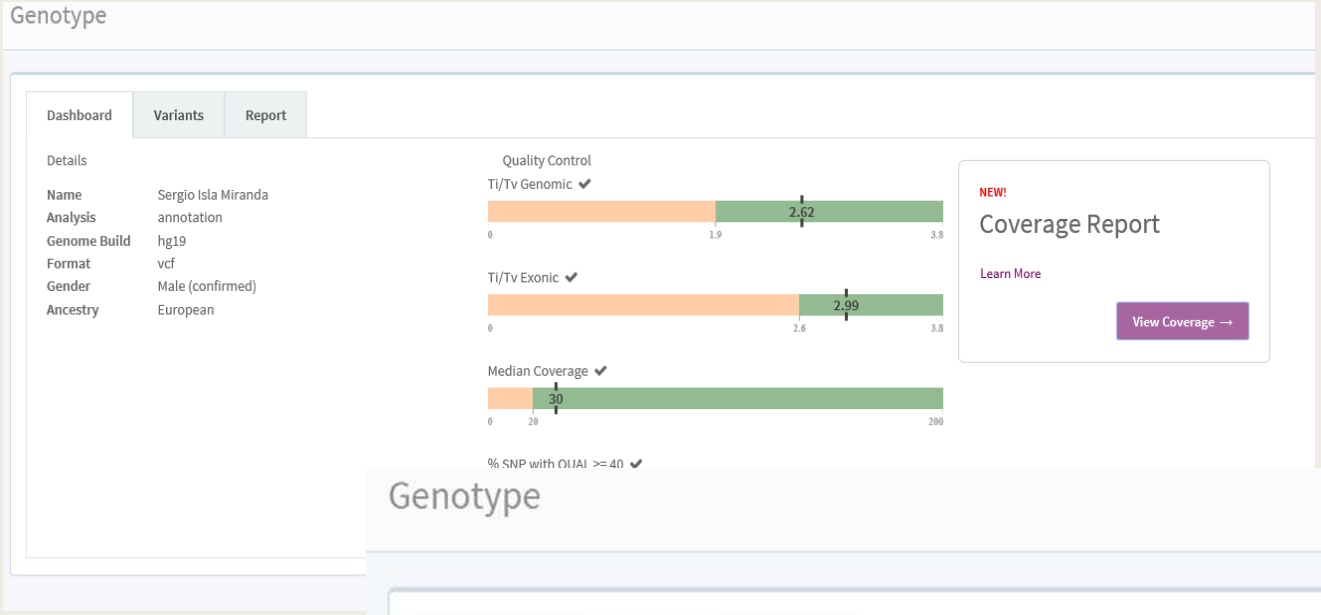
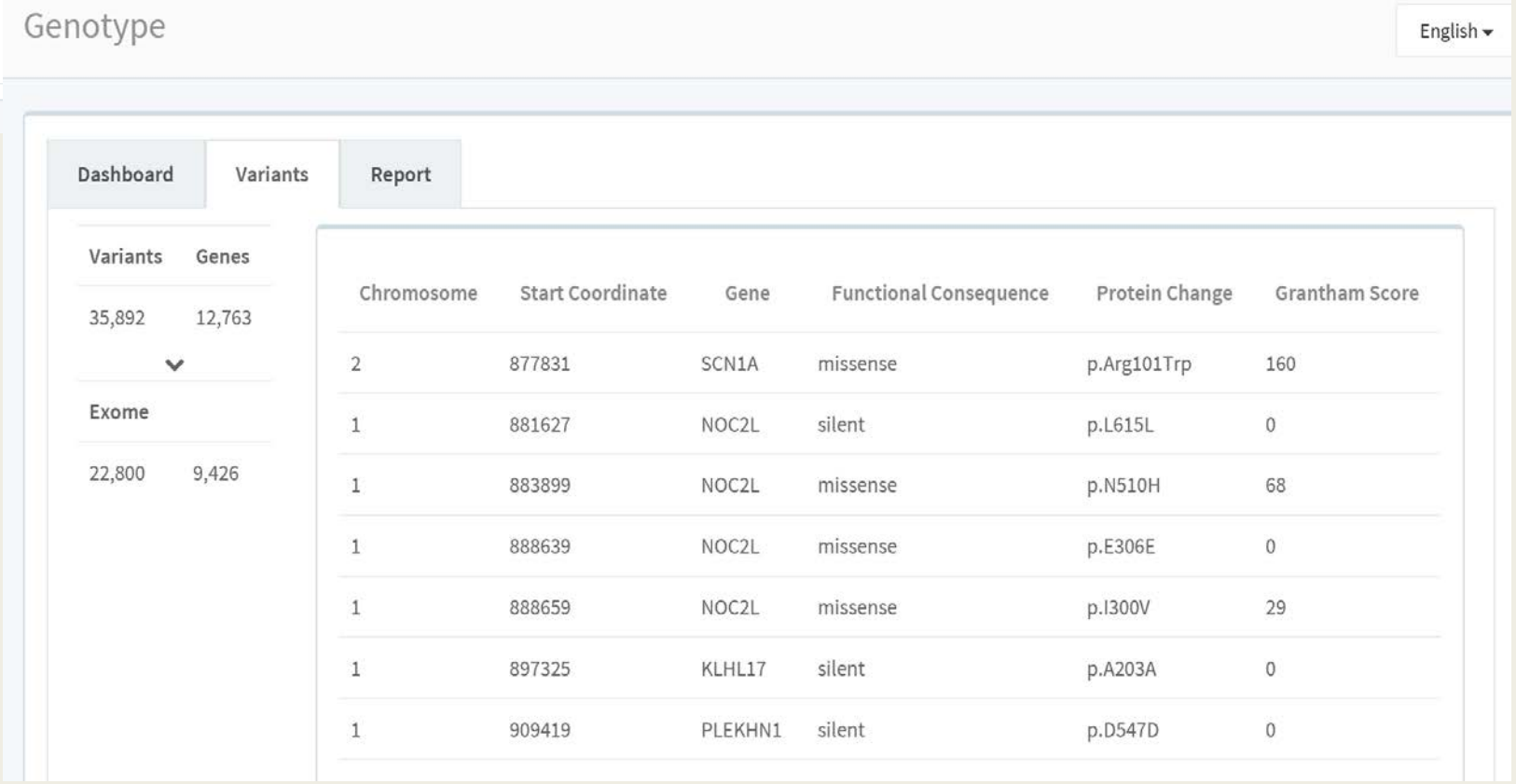
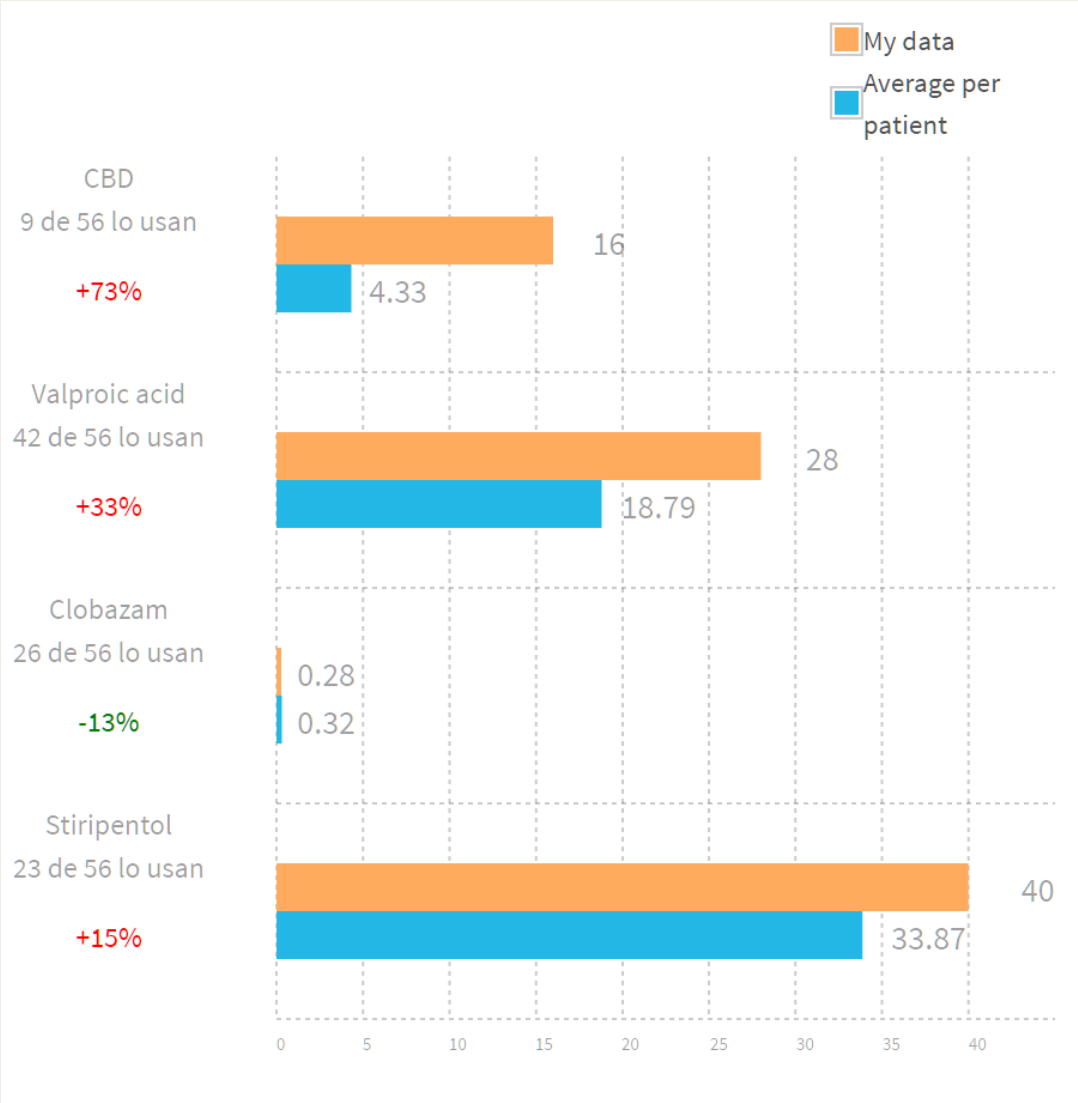
Menu
Dashboard
Basic Data
Clinic History
Phenotype
Calendar
Genotype
Documents
Scales
Benchmark
Phenotype
Checkboxs
Manual input
List of symptoms
HP:0001250 : Seizures
HP:0002069 : Generalized tonic-clonic seizures
HP:0002133 : Status epilepticus
HP:0002353 : EEG abnormality
HP:0002373 : Febrile seizures
HP:0002384 : Focal seizures with impairment of consciousness or awareness
HP:0006813 : Hemiclonic seizures
HP:0007270 : Atypical absence seizures
HP:0010818 : Generalized tonic seizures
HP:0010841 : Multifocal epileptiform discharges
HP:0010845 : EEG with generalized slow activity
HP:0010848 : EEG with spike-wave complexes (2.5-3.5 Hz)
HP:0011185 : EEG with focal epileptiform discharges
HP:0011188 : Focal EEG discharges with secondary generalization
HP:0012001 : EEG with generalized polyspikes
HP:0001249 : Intellectual disability
HP:0001251 : Ataxia
HP:0001252 : Muscular hypotonia
HP:0001270 : Motor delay
HP:0001324 : Muscle weakness
HP:0002167 : Neurological speech impairment
HP:0002357 : Dysphasia
HP:0002381 : Aphasia
HP:0002459 : Dysautonomia
HP:0005968 : Temperature instability
HP:0007328 : Impaired pain sensation
HP:0010829 : Impaired temperature sensation
HP:0000750 : Delayed speech and language development
HP:0000759 : Abnormality of the peripheral nervous system
HP:0000708 : Behavioral abnormality
HP:0000717 : Autism
HP:0000733 : Stereotypic behavior
HP:0000736 : Short attention span
HP:0000752 : Hyperactivity
HP:0100543 : Cognitive impairment
Save phenotype



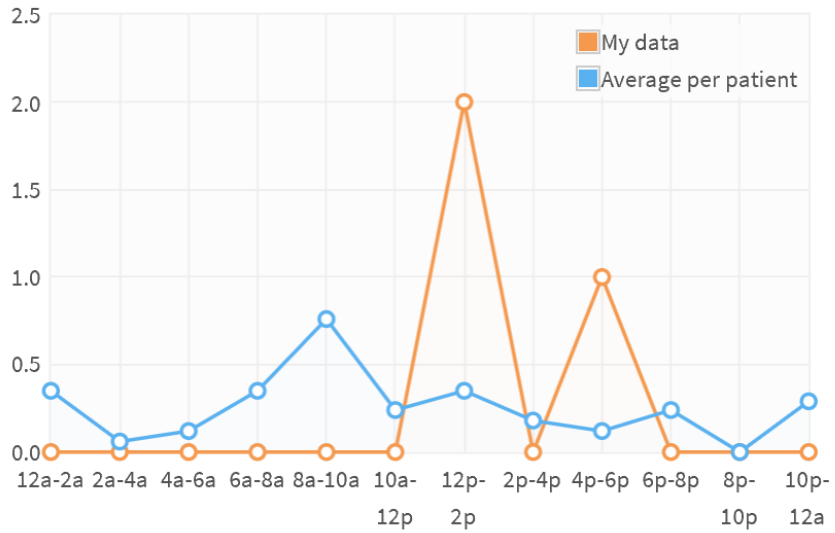


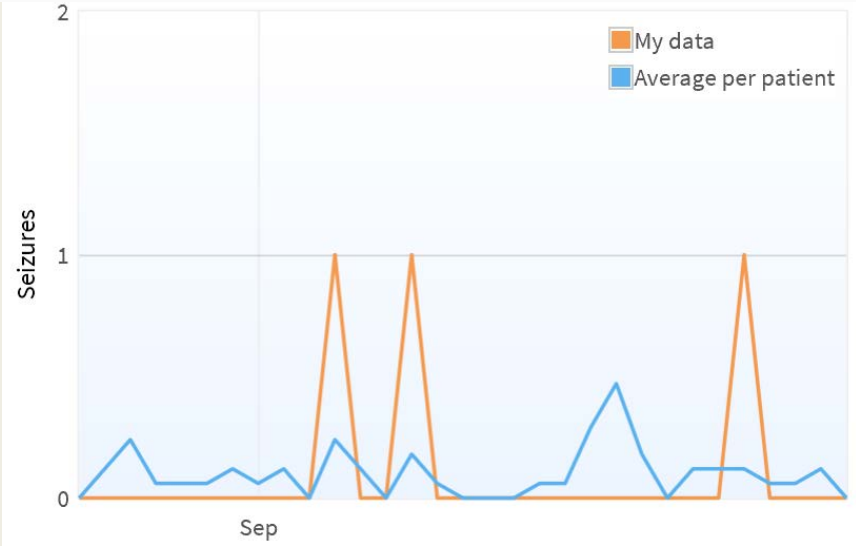
Chromosome	Start Coordinate	Gene	Functional Consequence	Protein Change	Grantham Score
2	877831	SCN1A	missense	p.Arg101Trp	160
1	881627	NOC2L	silent	p.L615L	0
1	883899	NOC2L	missense	p.N510H	68
1	888639	NOC2L	missense	p.E306E	0
1	888659	NOC2L	missense	p.I300V	29
1	897325	KLHL17	silent	p.A203A	0
1	909419	PLEKHN1	silent	p.D547D	0



Seizures by time of day

Options Last Month





Questions?

Thank you

Julian.isla@dravetfoundation.eu
@julianig