



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

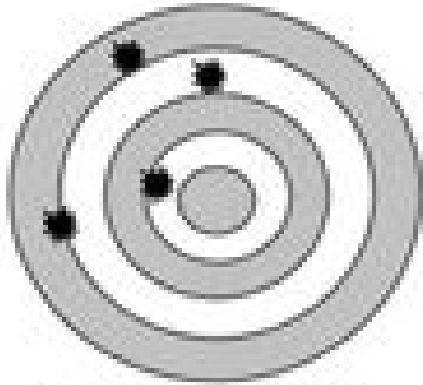


EMA delegate. COMP patient representative

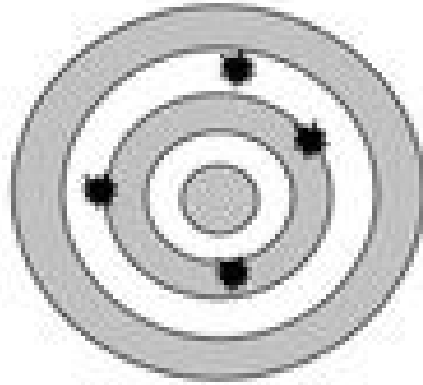


Scientific Advisory Board

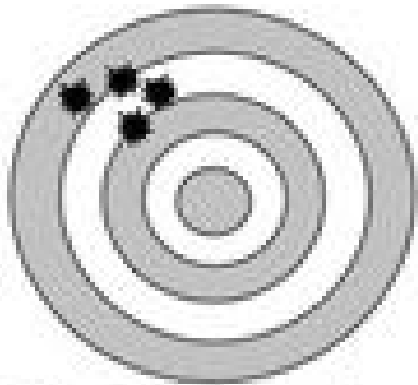
Personalised=Precision



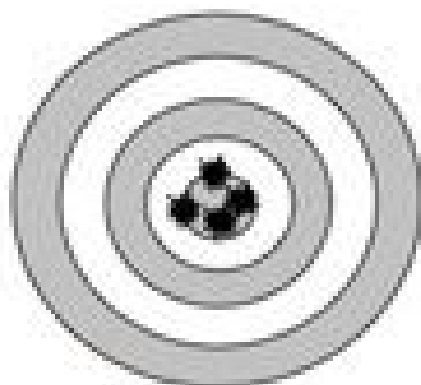
Not Accurate
Not Precise



Accurate
Not Precise



Not Accurate
Precise



Accurate
and Precise

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*"

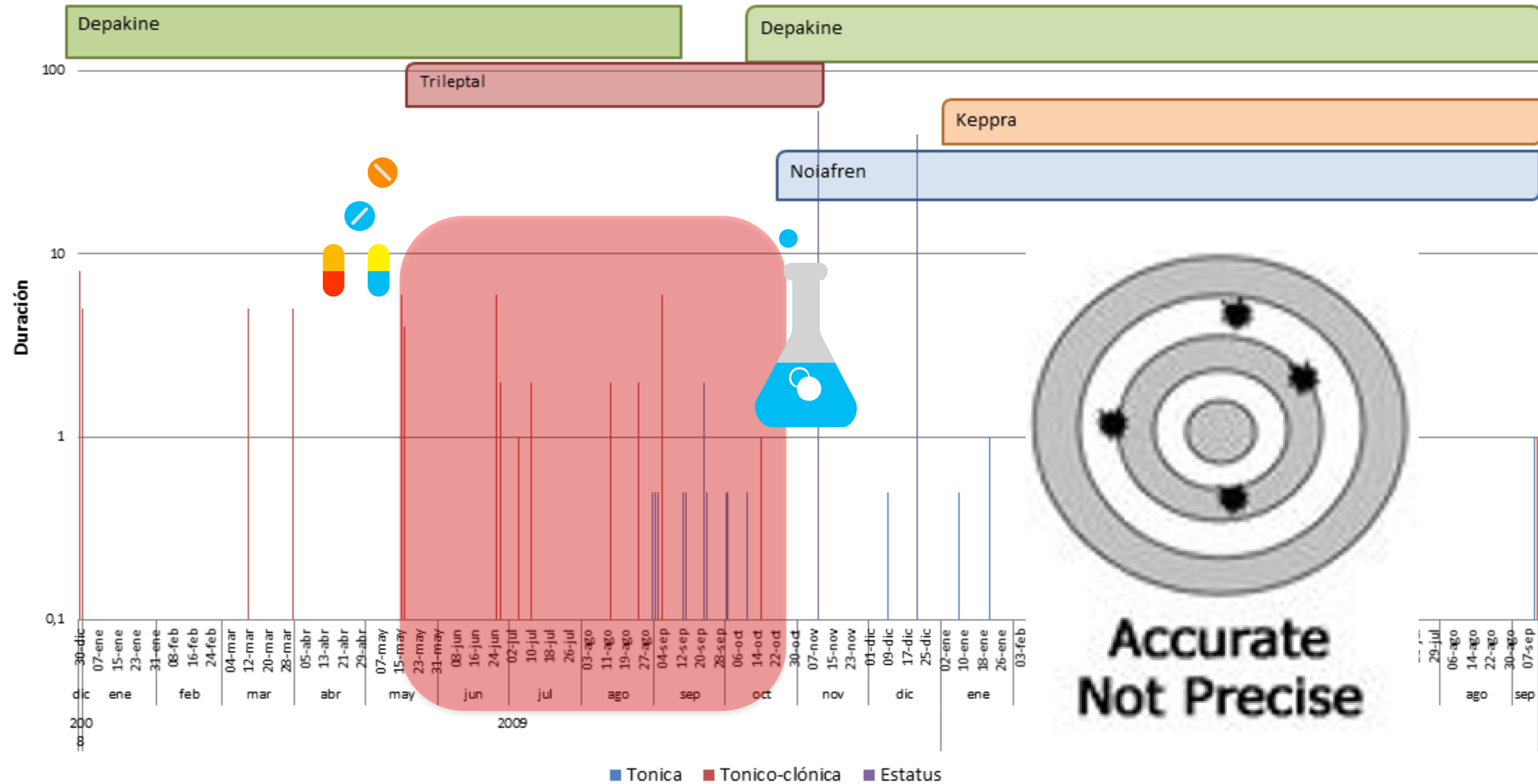
Medical treatment



Sergio
8 years old
Dravet Syndrome

A real example of lack
of precision

Crisis y duración







WHERE ARE WE NOW?

YOU ARE
HERE

Individual variability in genes

Environment

Lifestyle

Medical treatment





Individual variability in genes

Hi.
Your DNA can tell you a lot about **you**.



\$99
Ancestry Service
Experience your ancestry in a new way! Get a breakdown of your global ancestry by percentages, connect with DNA relatives and more.

RECOMMENDED
\$199
Health + Ancestry Service
Get an even more comprehensive understanding of your genetics. Receive 65+ online reports on your ancestry, traits and health - and more.
★★★★★ 3.5 (5506)

23andMe

HOME MY RESULTS FAMILY & FRIENDS RESEARCH & COMMUNITY

BROWSE RAW DATA

DOWNLOAD HELP

This is an advanced view of all the uninterpreted SNP data from your chip. The data from 23andMe's Browse Raw Data feature is for research and informational use only. This data has undergone chip-wide quality review, and a subset of SNPs have been individually validated for accuracy. However, the majority of SNPs have not undergone this rigorous individual validation, and any SNP result obtained from this raw data should be independently verified.

Most of the DNA inside each of your body's cells is divided into pieces called chromosomes. The remaining DNA is found in tiny loops inside your cells' mitochondria. Click below on any chromosome or the mitochondrial loop to see the genes and SNPs it contains. [Learn more](#) about how to use this feature.

Note: The chromosome display is the same for all customers, regardless of sex. Females will see "no call" results for SNPs on their Y chromosome.

Jump to a gene: Go a SNP: Go

Chromosome	Bases	Genes
1	249M Bases	3492 Genes
2	243M Bases	2359 Genes
3	198M Bases	1917 Genes
4	191M Bases	1443 Genes
5	180M Bases	1629 Genes
6	171M Bases	2041 Genes

Interpretome

Begin exploring!

Start Lookup Explore Clinical Ancestry #bog13 bingo

Explore your genome

- Load your genome file (upper-right corner) and choose some of the analyses above. Currently, only raw data files from 23andme and Lumigenix (**unzipped**) are supported.
- Sample genotype files (and a description of the individuals) can be found [here](#).
- A detailed description of the website design and some of the modules can be found in our [PSB paper](#) as well as in blog posts [here](#) and [here](#).

Interpretome is intended for educational and research purposes only.
No information should be considered diagnostic and as with any genetic testing service, the interpretation is not regulated by the FDA. We assume no responsibility for any injury or damage to persons or property arising out of or related to any use of Interpretome annotations or for any errors or omissions: consult your physician with any medical concerns. We retain copyright to the materials herein. By using this website, you agree that you accept these terms and are aware of the risks and benefits of genome interpretation. For more information, please read the full [Terms and Conditions](#).

How are my data kept private?
Your genome will not be sent to any server, it remains on your computer. This website will make requests to a database that only contain "rsid" (without genotypes) and "population" (self-reported in the top-right) information. At no point will any genotypes be sent across the wires (all computation will be done in the browser). Some exercises may have an option to submit personal information, including genotypes or results of analyses, which will be anonymously stored on a secure server.



Internet of DNA

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

Availability: 1-2 years

10 Breakthrough Technologies 2015

Introduction

Magic Leap



Nano-Architecture



Car-to-Car Communication



Project Loon



Liquid Biopsy



Megascale Desalination



Apple Pay

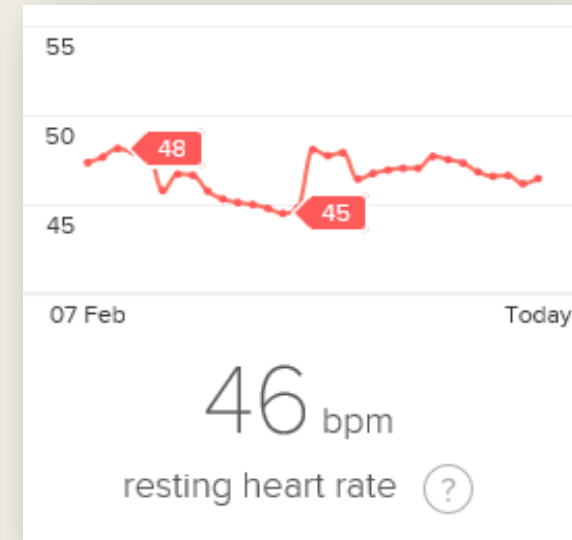


Breakthrough

Why It Matters

Key Players

Environment and lifestyle





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

Thomas Goetz

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

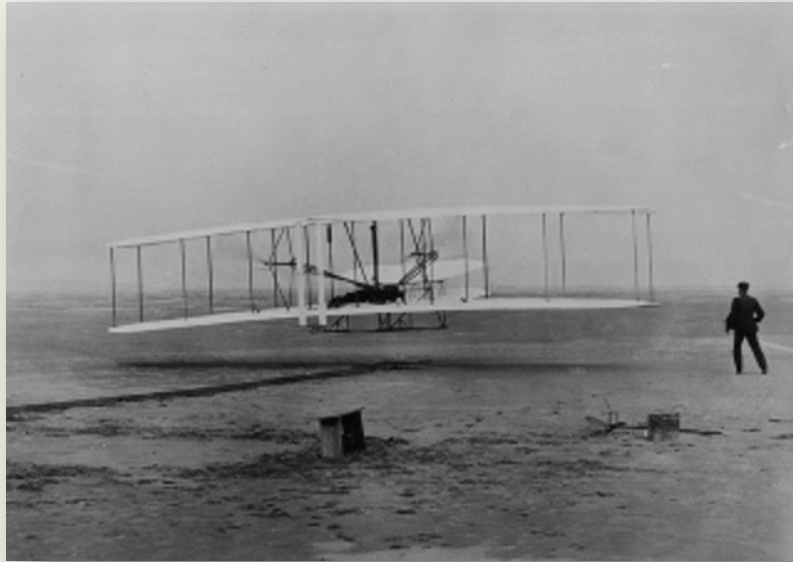


Learning from mistakes



Feedback loop





110 years

Patient digital transformation



3 2581703423 4457570

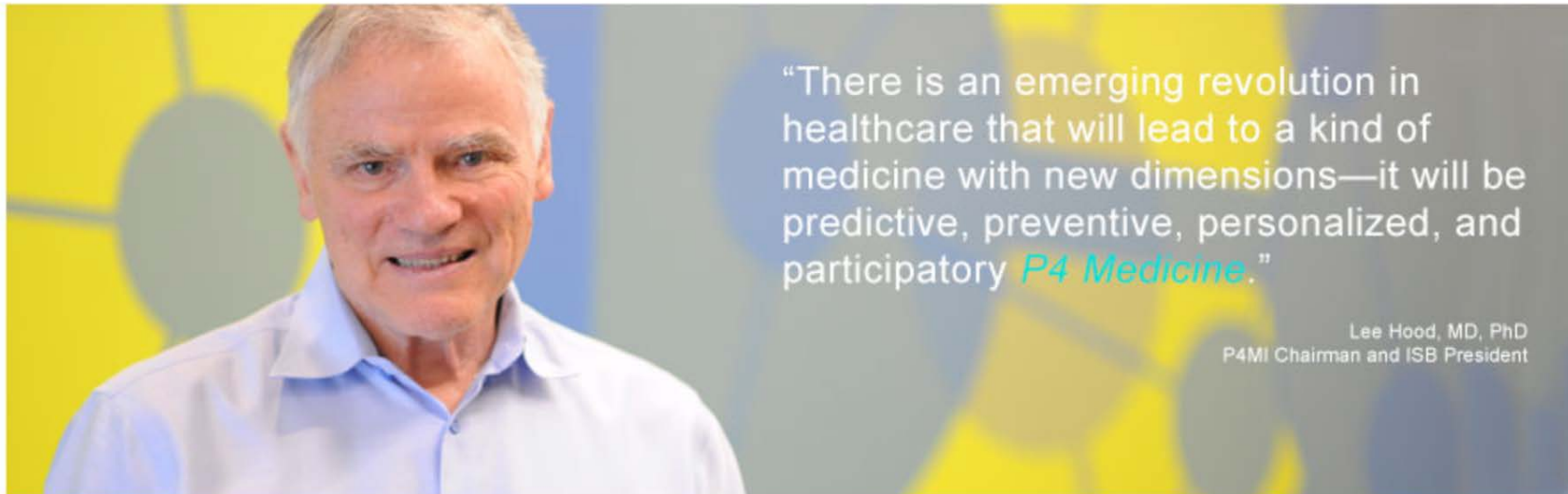
P-4 medicine

predictive, preventive, personalized and participatory



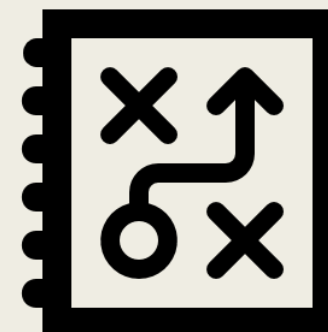
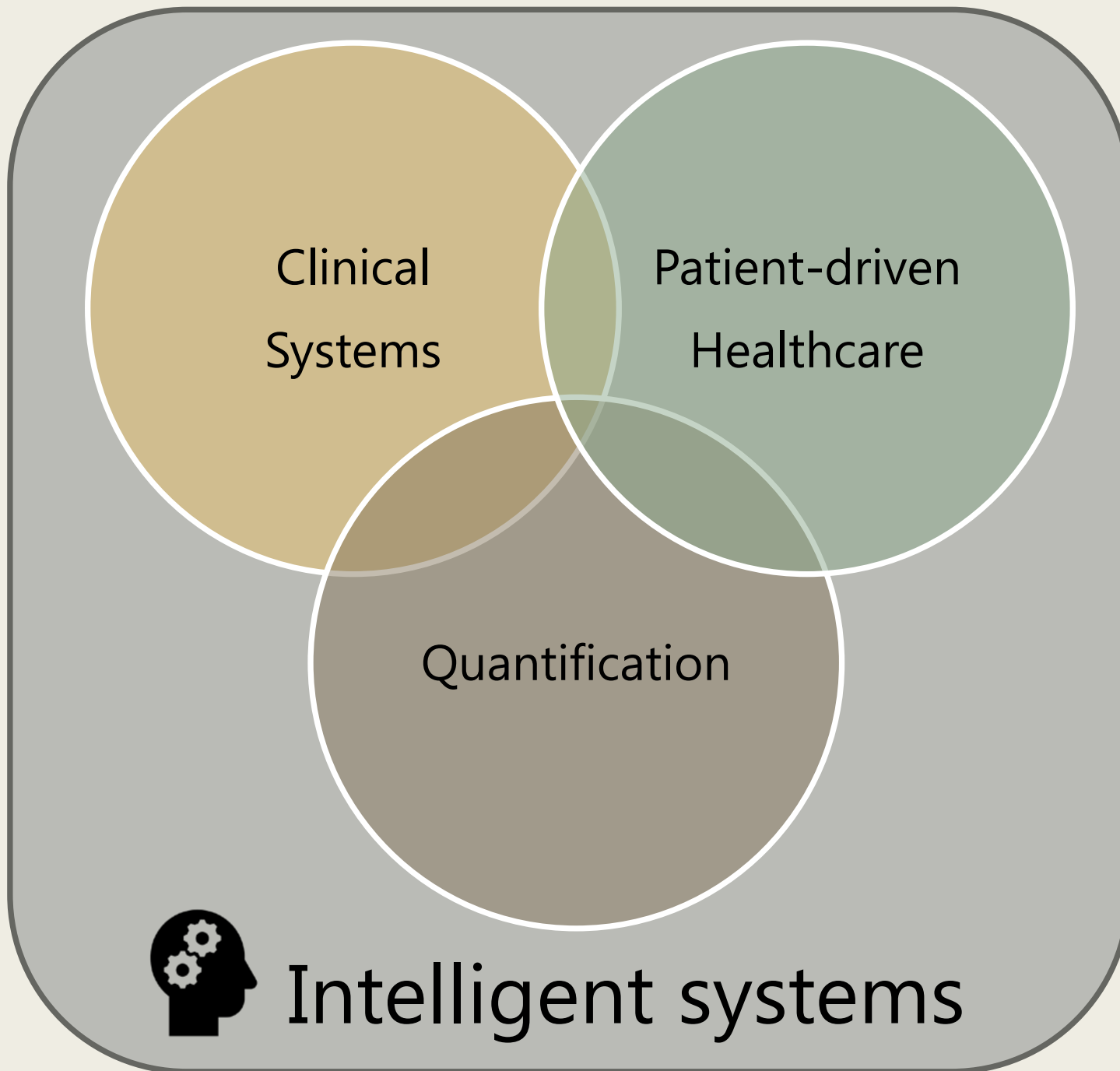
Quantifying Wellness. Demystifying Disease.

[P4 MEDICINE](#) | [WHAT WE DO](#) | [MEDIA CENTER](#) | [ABOUT](#) | [BLOG](#)



Quantifying
wellness is
the key





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📄 Basic Data

📁 Clinic History

🧩 Phenotype

📅 Calendar

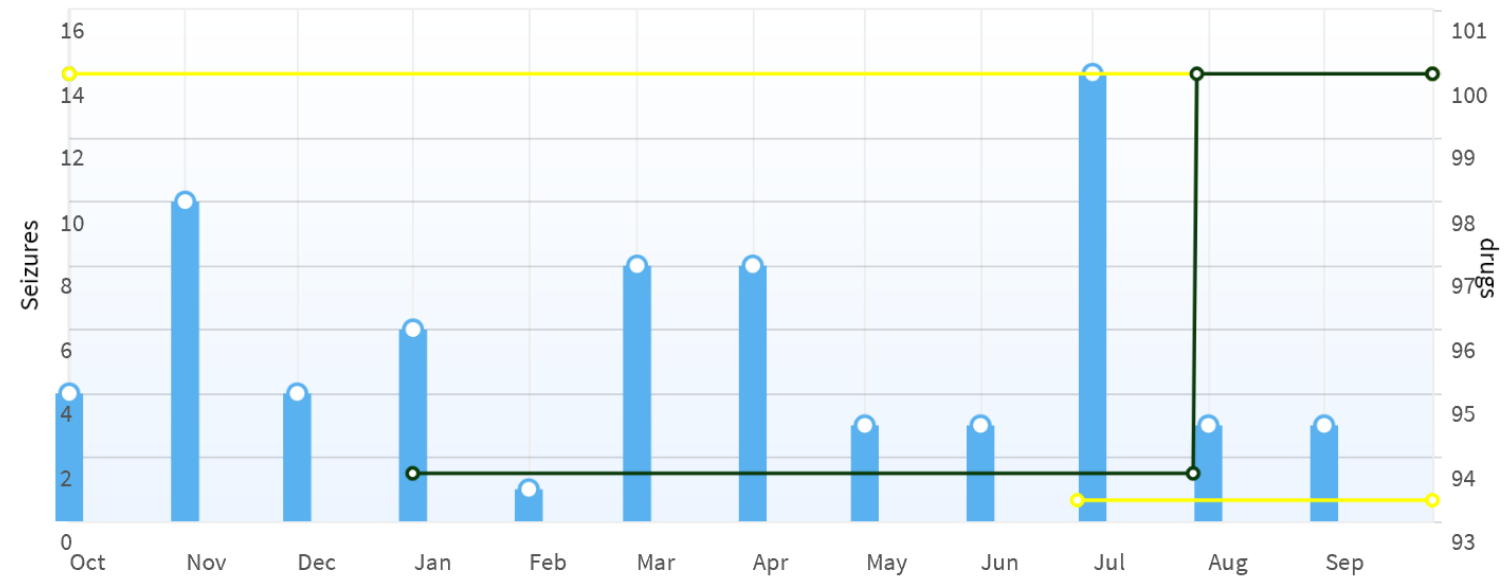
🐾 Genotype

📁 Documents

📶 Scales

🕒 Benchmark

Seizures and drugs



Seizures



Stiripentol



Valproic acid



Clobazam



CBD

wacean

beta

?

⚙

↗

Menu

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Add seizure

Absence

Atonic

Atypical Absence

Clonic

Complex Focal

Infantile Spasms

Myoclonic

Myoclonic Cluster

Secondarily Generalized

Simple Focal

Status

Tonic

previous year

◀

September 2016

▶

next year

Sun	Mon	Tue	Wed	Thu	Fri	Sat
28	29	30	31	1	2	3
4 12:20p Complex Focal 35%	5	6	7 4:30p Complex Focal 35%	8	9	10
11	12	13	14	15	16	17
18	19	20 12:33p Complex Focal 18%	21	22	23	24
25	26	27	28	29	30	1
2	3	4	5	6	7	8

3 seizures in September 2016

Menu

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

Phenotype

☒ Checkboxes 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

Dashboard

Variants

Report

Details

Name

Sergio Isla Miranda

Analysis

annotation

Genome Build

hg19

Format

vcf

Gender

Male (confirmed)

Ancestry

European

Quality Control

Ti/Tv Genomic ✓

0

1.9

2.62

3.8

Ti/Tv Exonic ✓

0

2.6

2.99

3.8

Median Coverage ✓

0

20

30

200

% SNP with QIYI >= 40 ✓

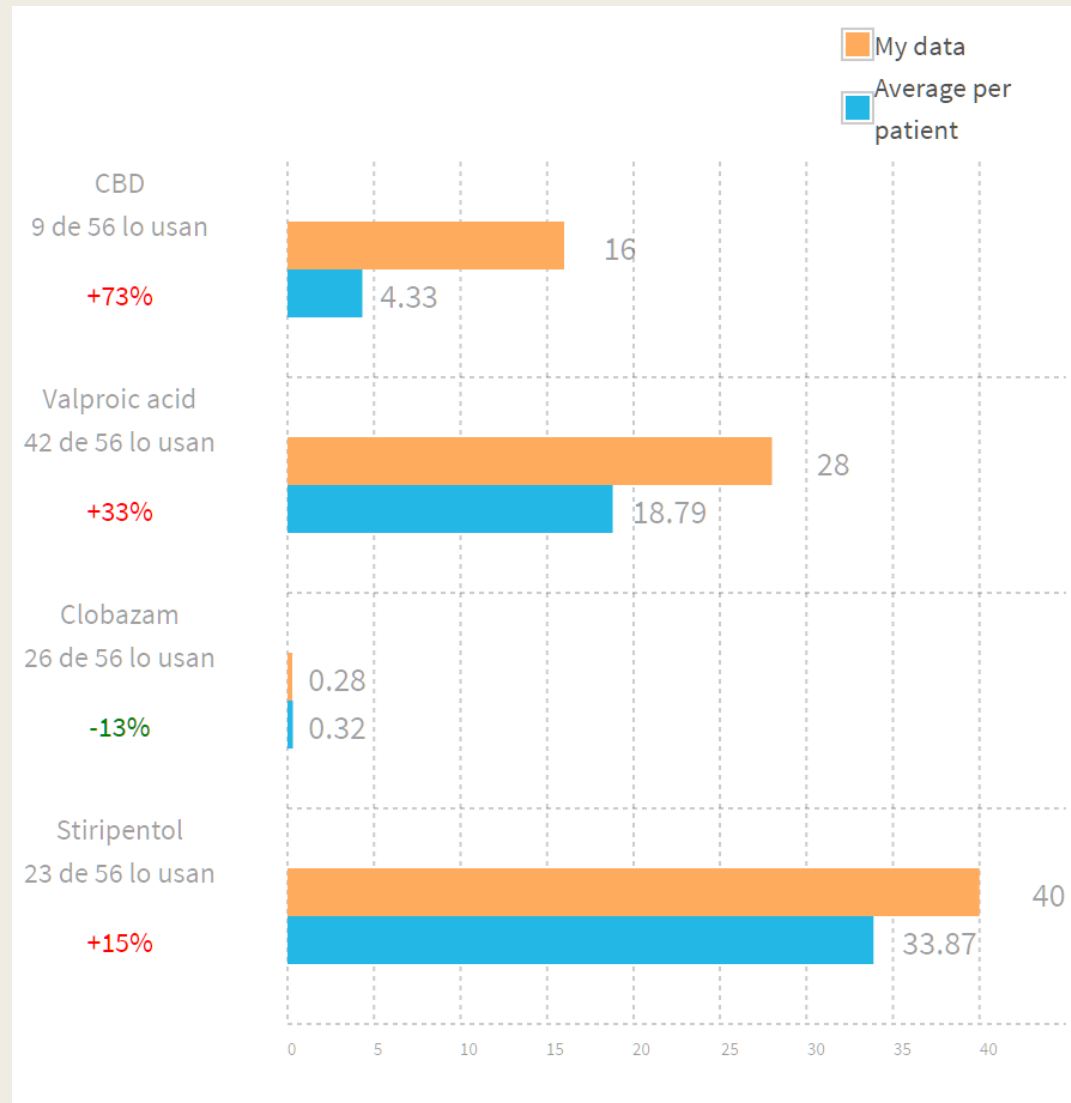
NEW!

Coverage Report

Learn More

View Coverage →

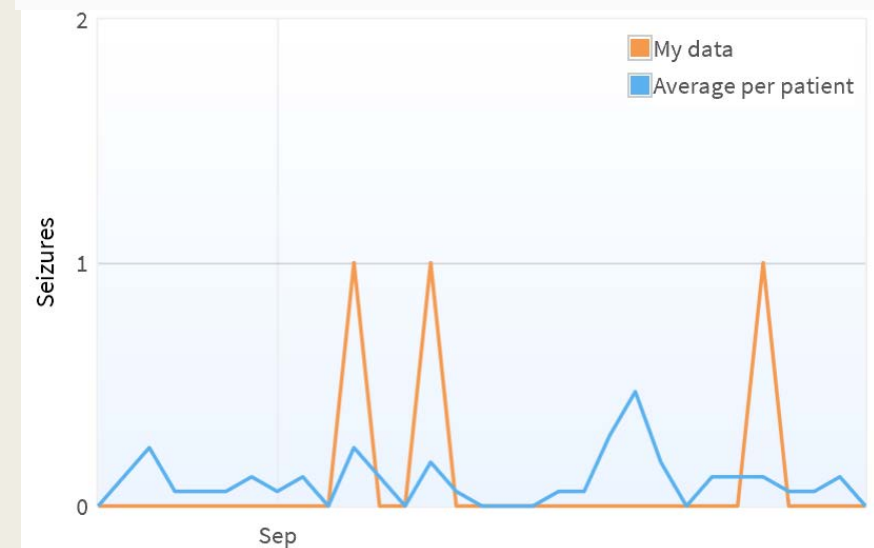
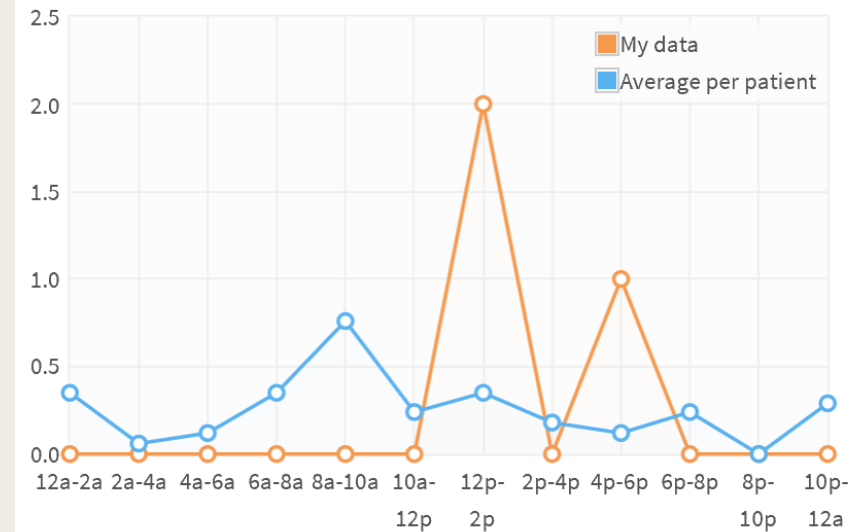
Dashboard		Variants		Report			
Variants	Genes						
35,892	12,763						
▼							
Exome							
22,800	9,426						
		Chromosome	Start Coordinate	Gene	Functional Consequence	Protein Change	Grantham Score
		2	877831	SCN1A	missense	p.Arg101Trp	160
		1	881627	NOC2L	silent	p.L615L	0
		1	883899	NOC2L	missense	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

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