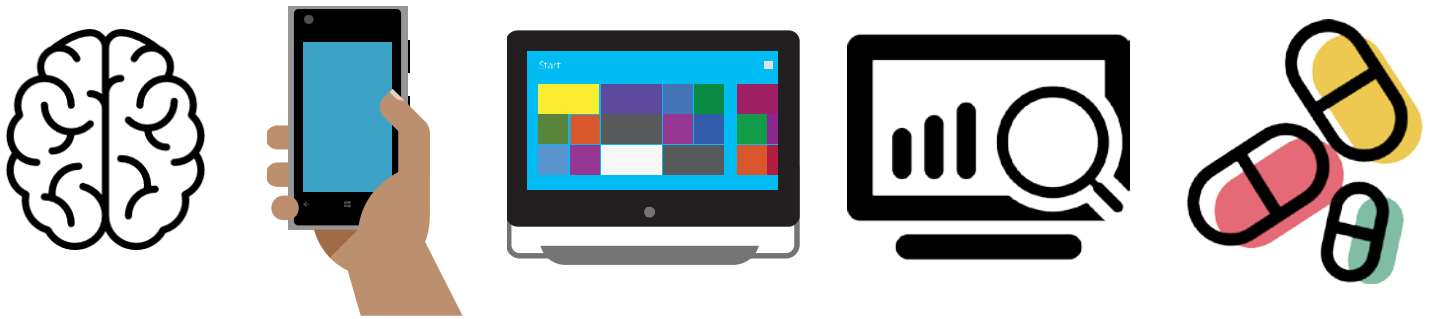
Wacean

A patient-driven innovative tool for data capture



Microsoft. Consulting Services

CTO of Dravet Syndrome Foundation

President for Dravet Syndrome European Federation

EURORDIS Therapeutic Action Group

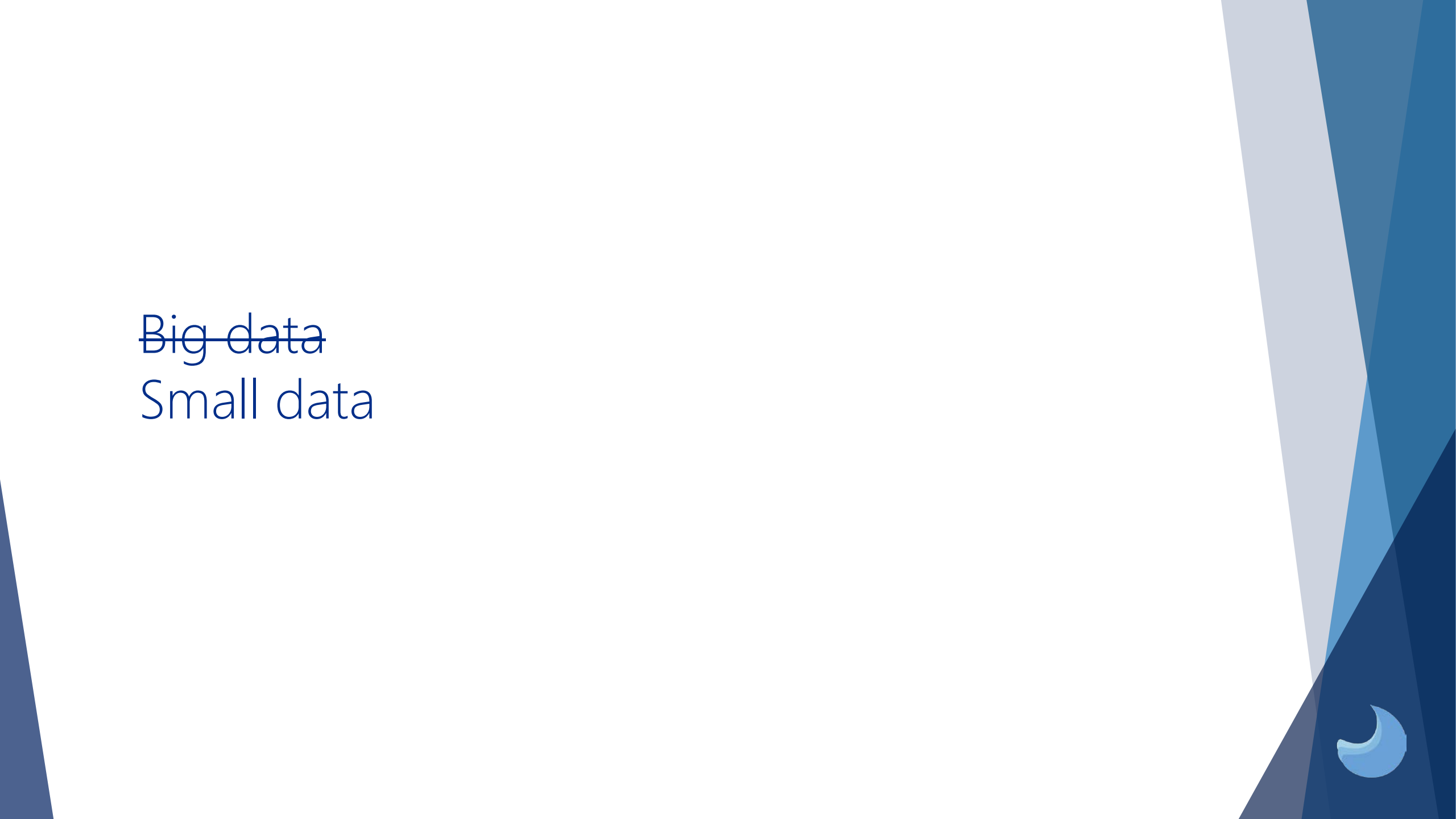
EMA delegate. COMP patient representative

Scientific Advisory Board

Big data

~~Big data~~

Small data

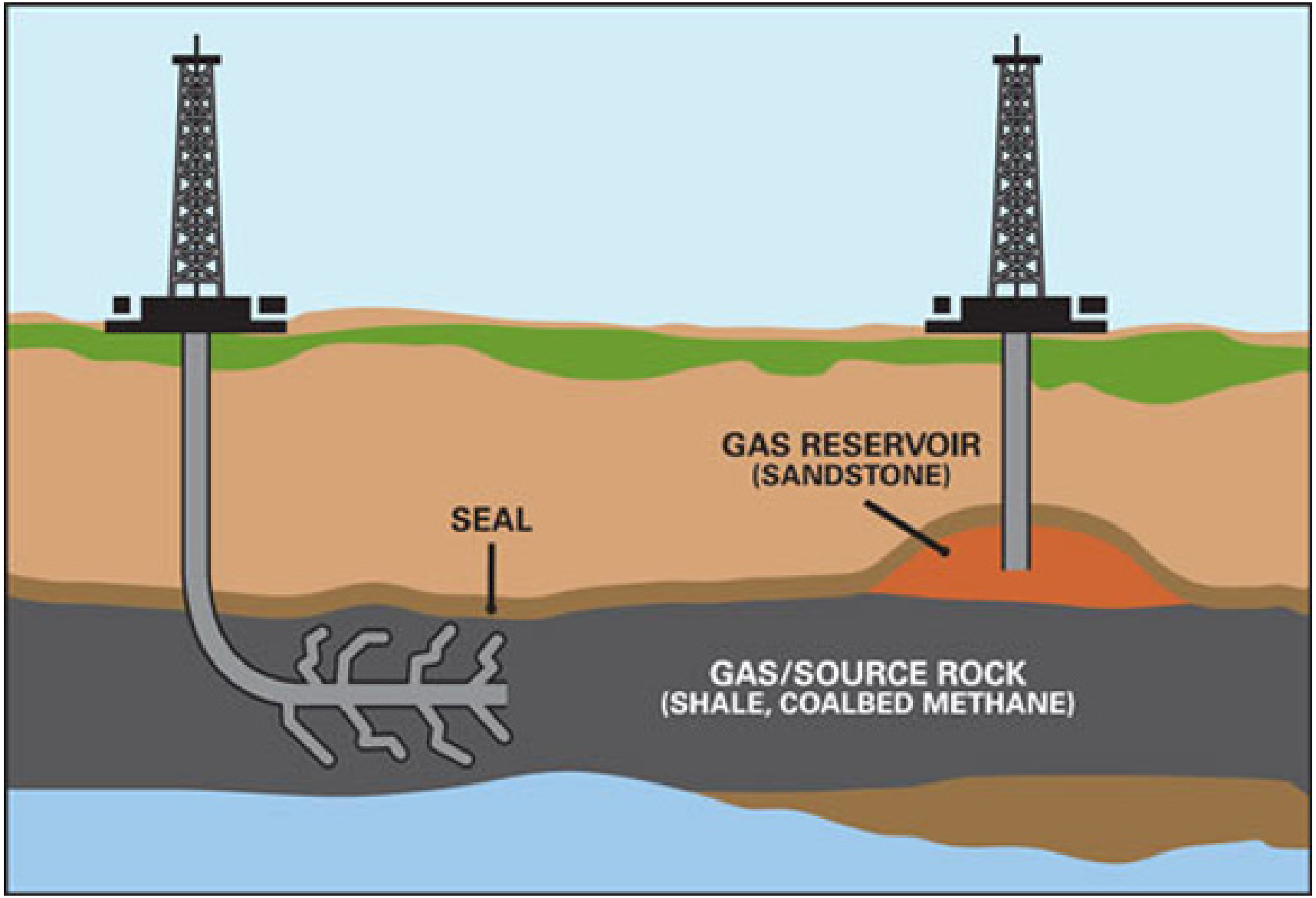


~~Big data~~

~~Small data~~

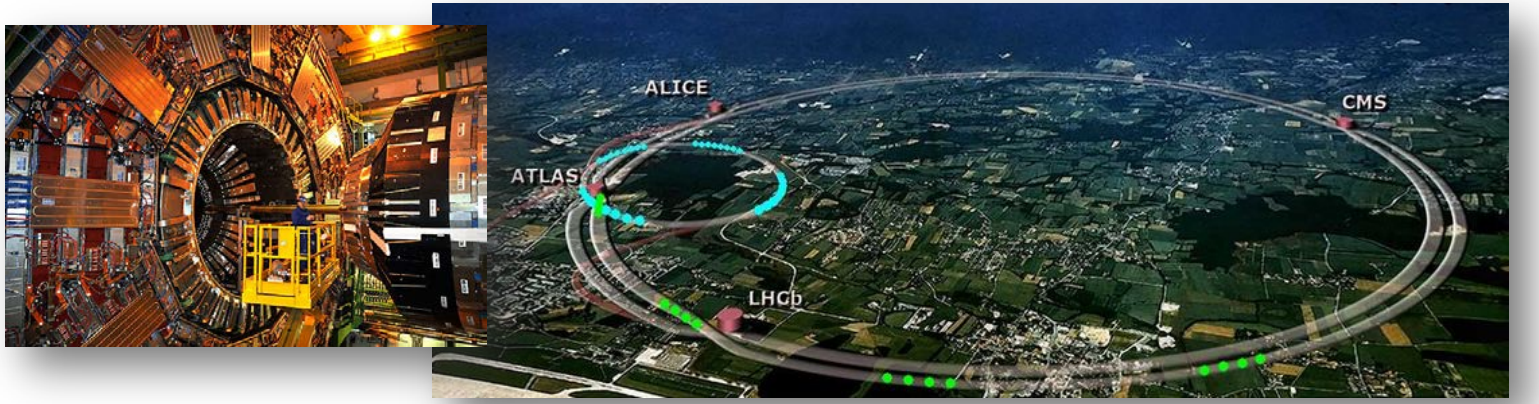
Spread data

Data mining for rare diseases



An example of real big data

CERN



30 petabytes every year

illumina®

~3

PETABYTES

Every 18 months

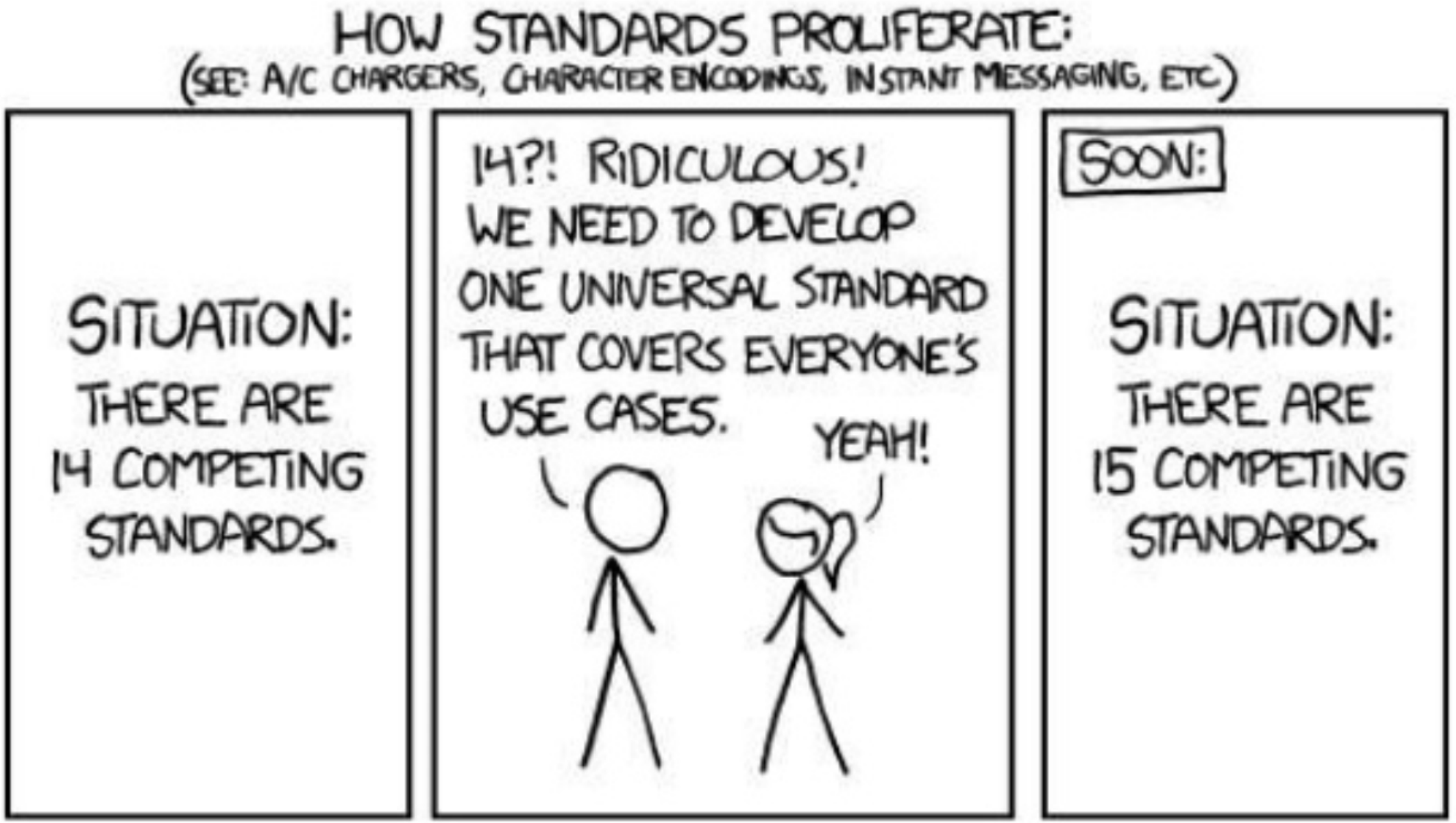
NETFLIX

3

PETABYTES

Total

Let’s create standards!


HOW STANDARDS PROLIFERATE:
(SEE: A/C CHARGERS, CHARACTER ENCODINGS, INSTANT MESSAGING, ETC)
SITUATION: THERE ARE 14 COMPETING STANDARDS.
14?! RIDICULOUS! WE NEED TO DEVELOP ONE UNIVERSAL STANDARD THAT COVERS EVERYONE'S USE CASES.
YEAH!
SOON:
SITUATION: THERE ARE 15 COMPETING STANDARDS.

Interoperability is a myth

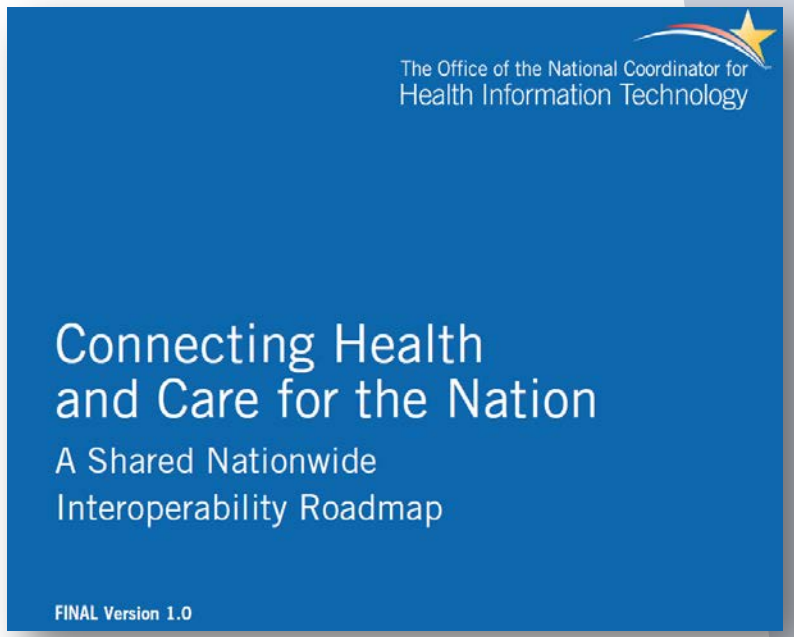
The Death of Interoperability: Is it Time for One Record?

MARCH 25, 2015 BY LEONARD KISH 0 COMMENTS

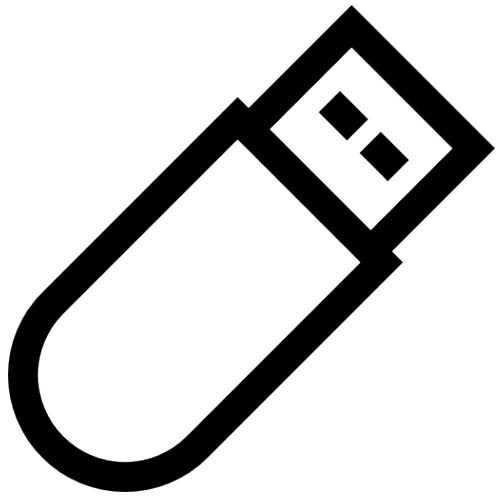
We've invested $ billions upon $ billions in interoperability. It's past time to ask, "Is it a lost cause?"

Incentives insufficient to fix market dynamics?

Don't worry folks, Congress is on the job. The problem has gotten so bad or become so old that we finally had congressional hearings last week on health record interoperability. There was predictable scolding and sound bites, and much of the testimony w
ago, or more ago, because the problem is exactly the sa



~~Interoperability~~ Portability



CNN Tech » 'Instagram for doctors' lets medics share photos to solve mystery cases International Edition + menu

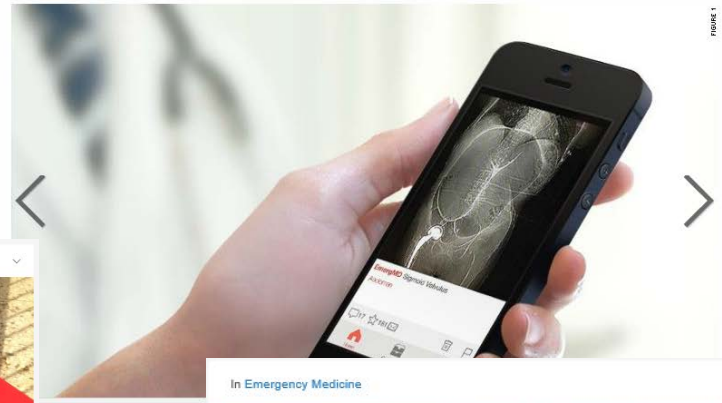
vital signs with Dr. Sanjay Gupta

IN ASSOCIATION WITH مدينة دبي الطبية Dubai Healthcare City

'Instagram for doctors' lets medics share photos to solve mystery cases

Meera Senthilingam, for CNN

Updated 1023 GMT (1823 HKT) February 10, 2015



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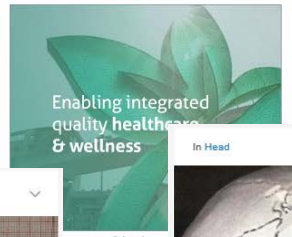
Roger Federer: 14-year streak ends

Hollywood expresses shock after voting results



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se of **cookies**,
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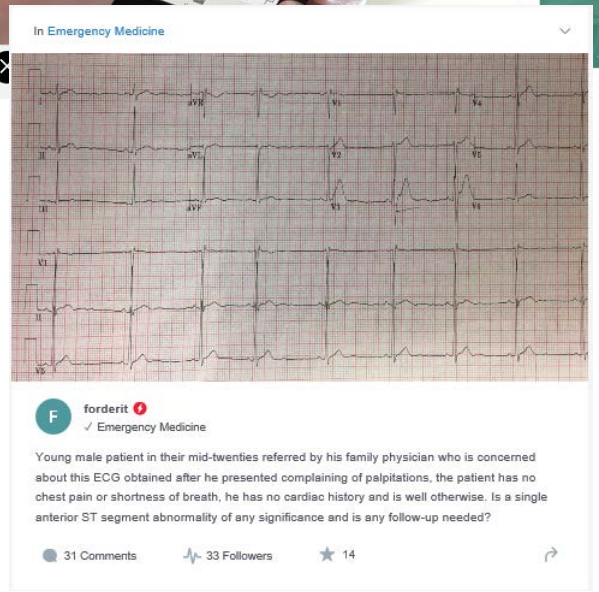

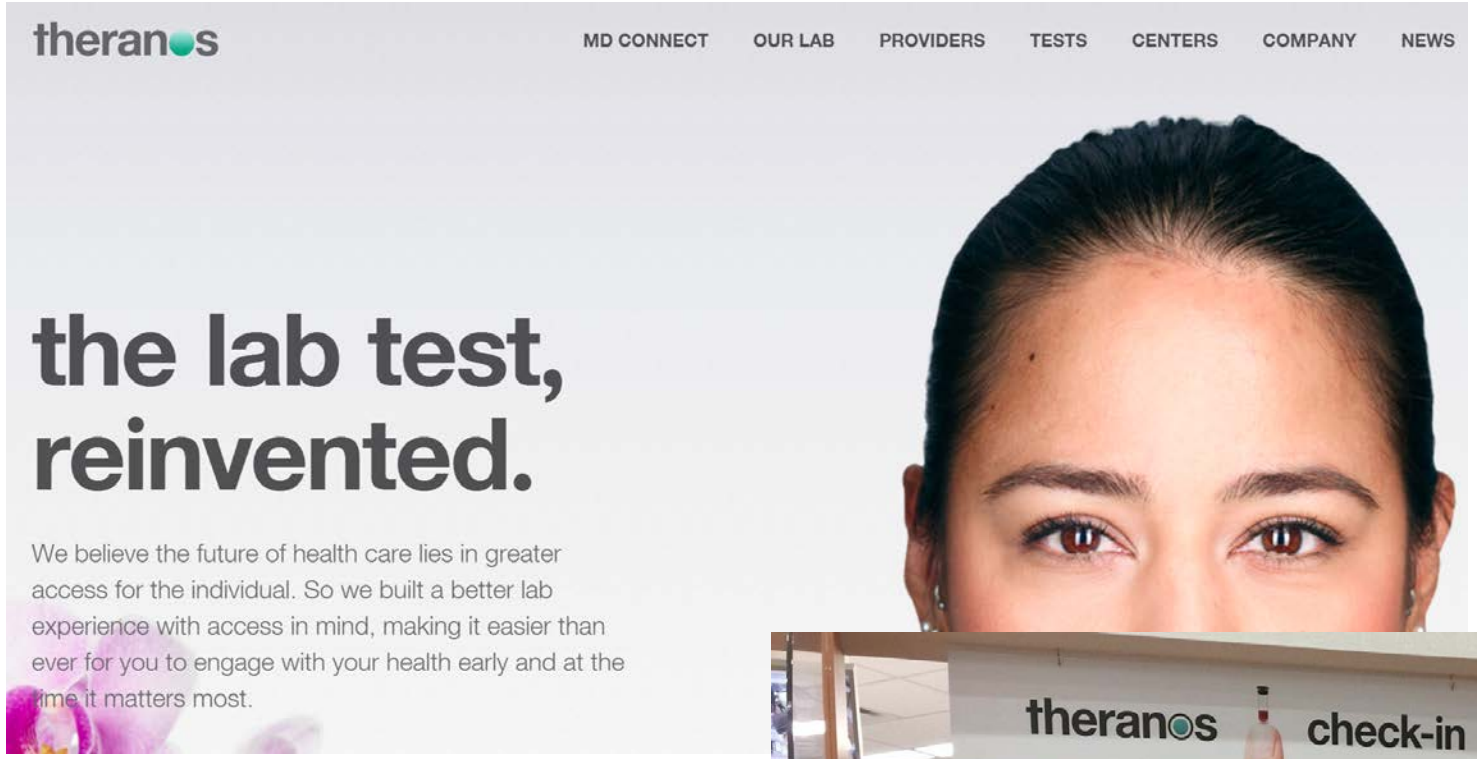
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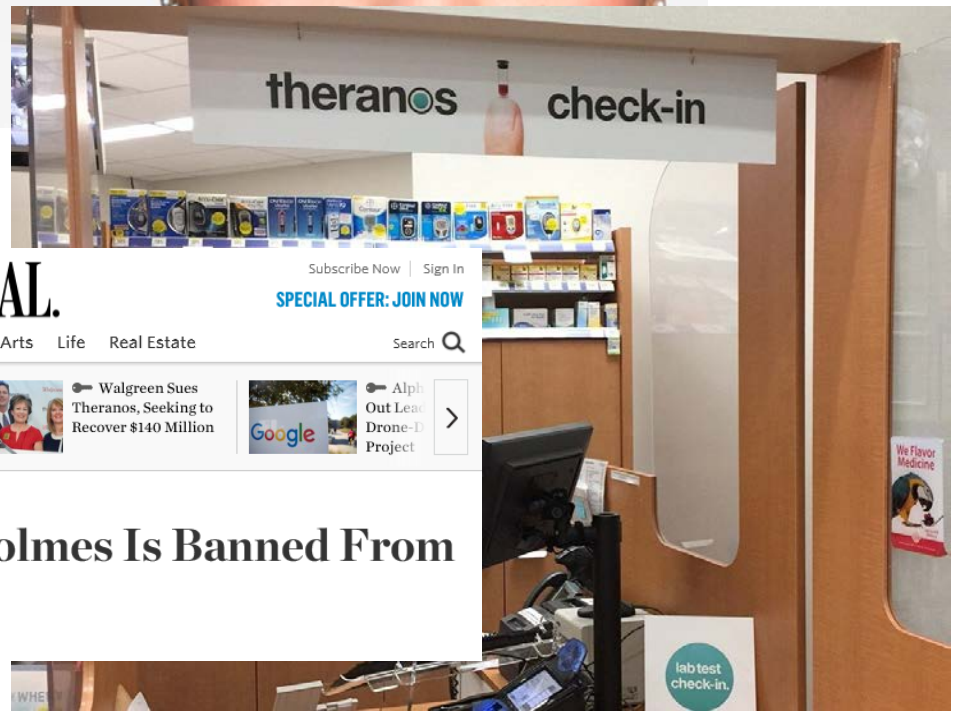
We believe the future of health care lies in greater access for the individual. So we built a better lab experience with access in mind, making it easier than ever for you to engage with your health early and at the time it matters most.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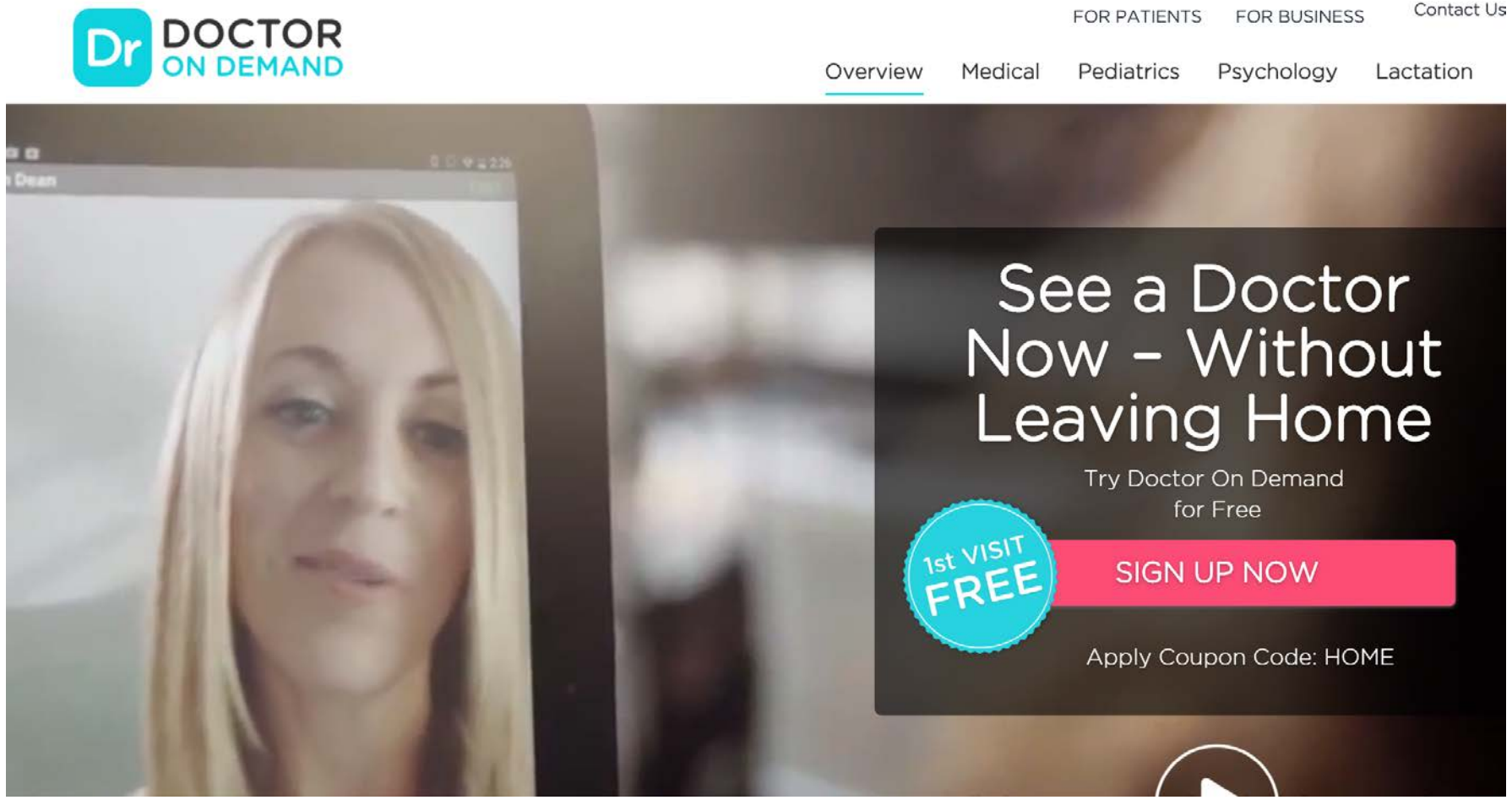
BUSINESS
Theranos Dealt Sharp Blow as Elizabeth Holmes Is Banned From Operating Labs
Company also remains subject of criminal probe into whether it misled investors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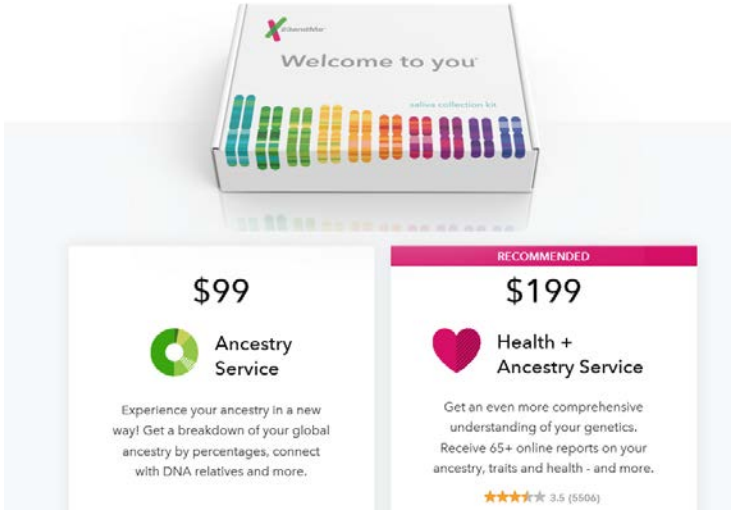
Hi.
Your DNA can tell you a lot about you.
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RECOMMENDED
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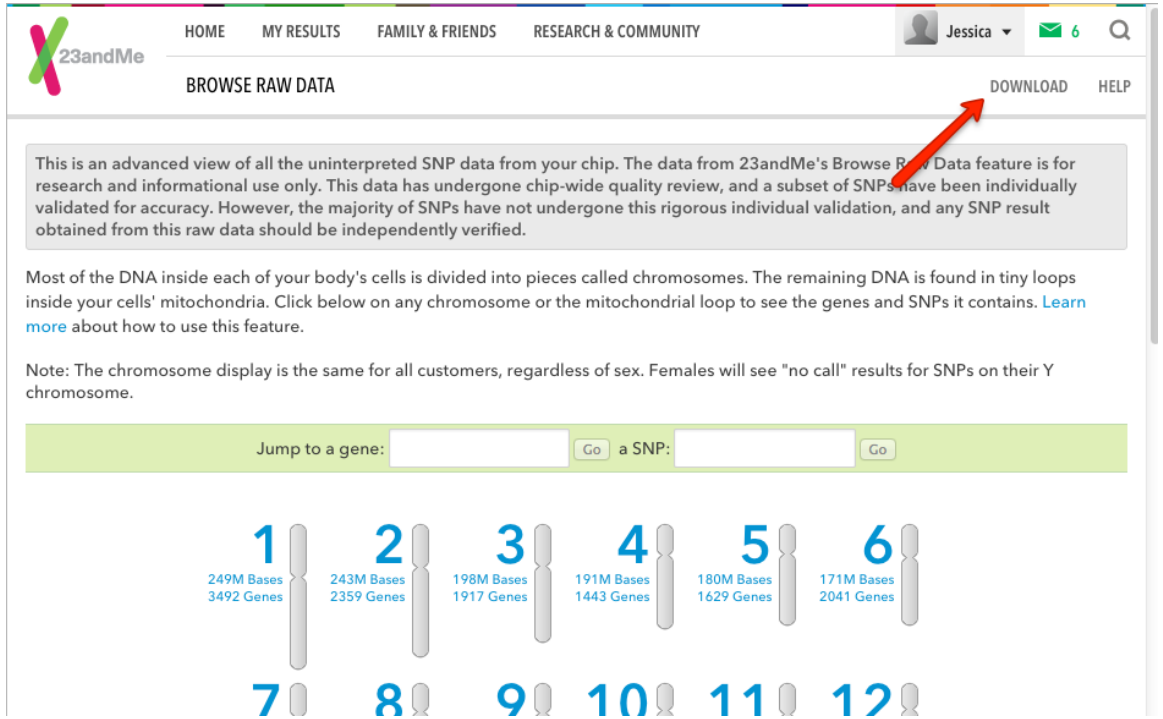
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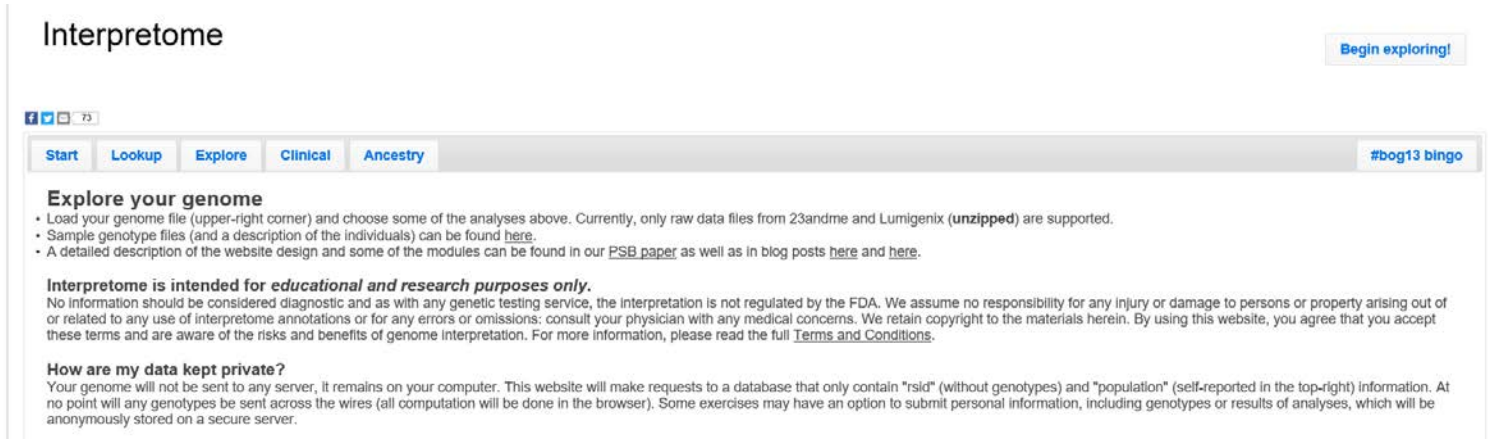
FAMILY & FRIENDS
RESEARCH & COMMUNITY
Jessica
6
BROWSE RAW DATA
DOWNLOAD
HELP
This is an advanced view of all the uninterpreted SNP data from your chip. The data from 23andMe's Browse Raw Data feature is for research and informational use only. This data has undergone chip-wide quality review, and a 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
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
7
8
9
10
11
12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Begin exploring!
Start
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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.

Dravet support group

14 de septiembre · [redacted], Estados Unidos

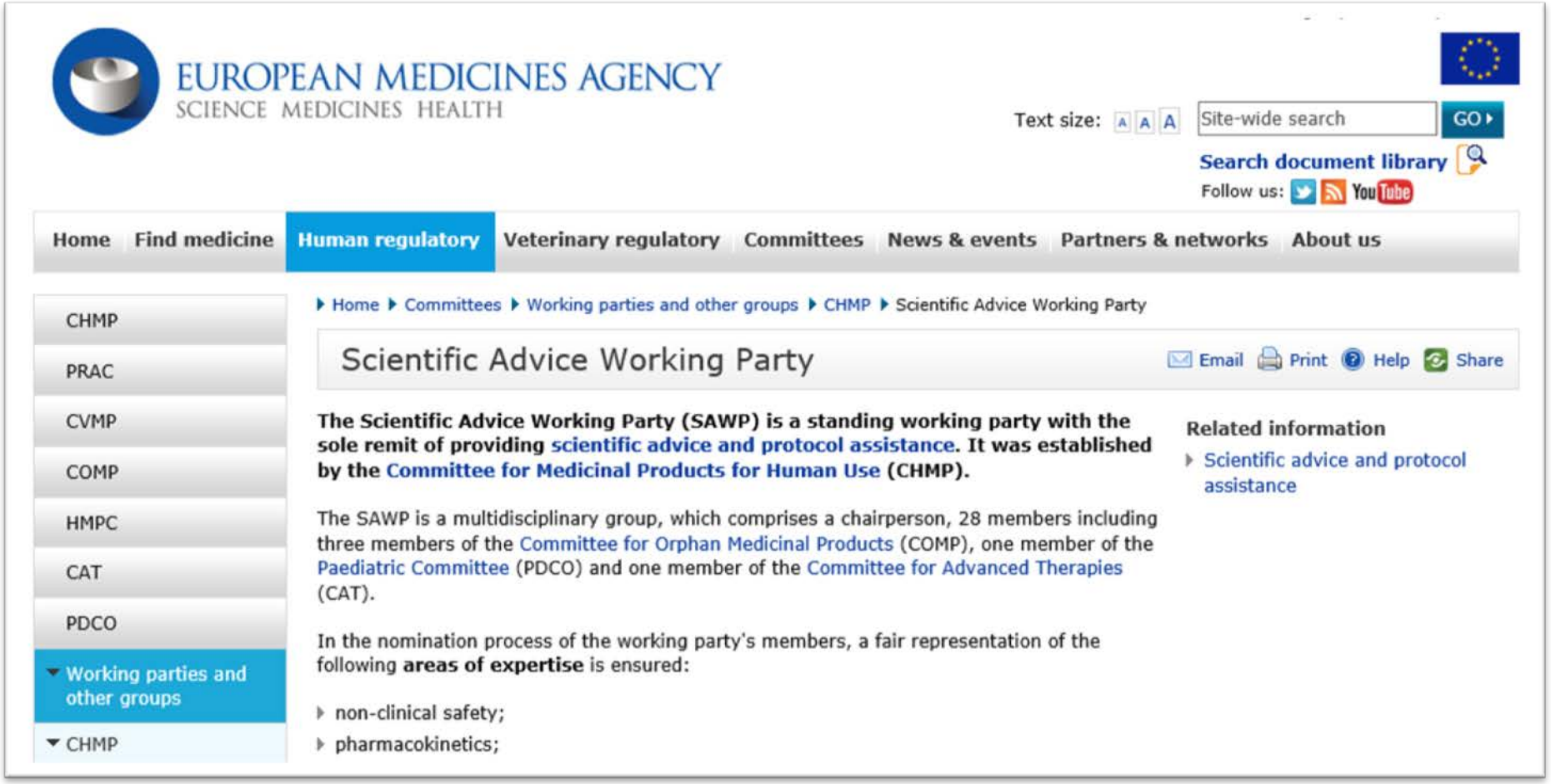
I thought it worthwhile to share a personal reflection I had at [redacted] appointment today: When we thought about enrolling him in Zogenix's fenfluramine trial, we were nervous about the length of time we'd be "out of control" of his medication (4 weeks of no med changes before screening, then 6 weeks of baseline, and then 14 weeks of treatment with placebo, low dose, or high dose, not knowing which group he was in). That seemed like such a long time and we worried about what could happen to him. I was also somewhat skeptical of a pharmaceutical company being in control of the medication, something I've handled for the past 10 years. But he is significantly past the half way point, and in 8 short weeks we'll finish the blind part of the trial and will be able to start him on open-label fenfluramine, which we've been waiting to do for over 3 years. It is amazing how quickly it has flown by. I can't say how he's doing because I don't want to compromise the trial, but if anyone is on the fence about whether or not to start, think of the potential payoff (getting access to a med not otherwise available) for a relatively short time commitment in the grand scheme of things. (I speak only about our personal experience - not everyone will feel the same, of course!)

[redacted] y 52 personas más 23 comentarios

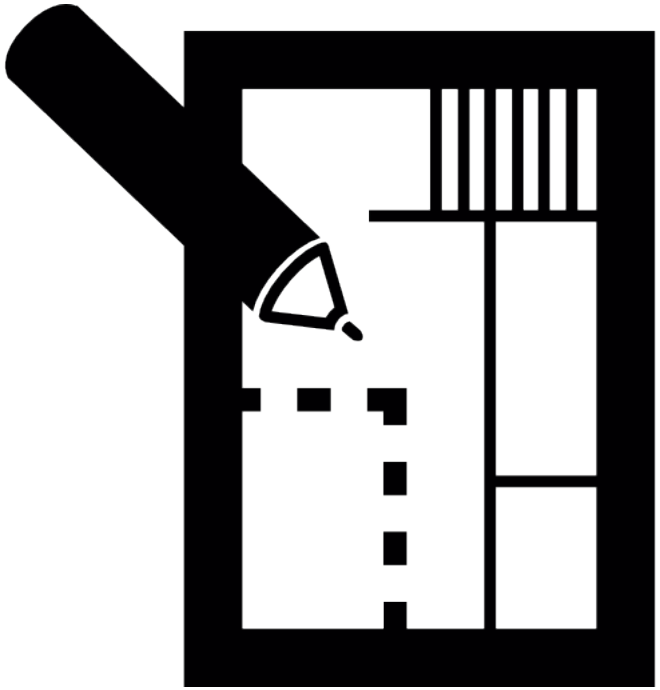
Me gusta Comentar

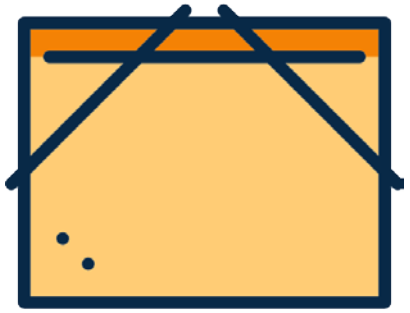
Patient needs

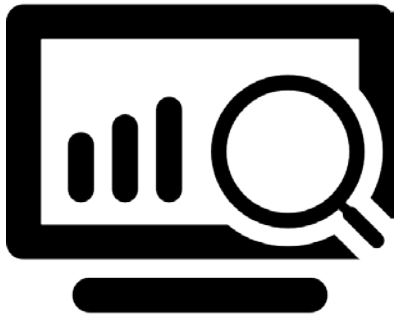
Scientific advice and protocol assistance



Dravet Syndrome Drugs







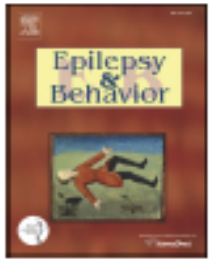
SurveyMonkey

Data from 274 patients in just one week

Contents lists available at ScienceDirect

Epilepsy & Behavior

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



The European patient with Dravet syndrome: Results from a parent-reported survey on antiepileptic drug use in the European population with Dravet syndrome

Luis Miguel Aras, Julián Isla, Ana Mingorance-Le Meur *

Dravet Syndrome Foundation Spain, Madrid, Spain

ARTICLE INFO

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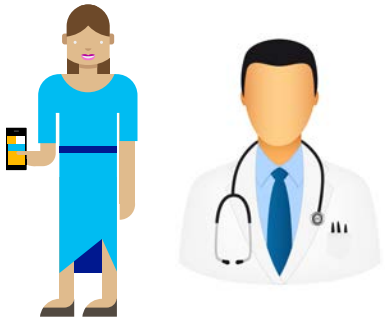
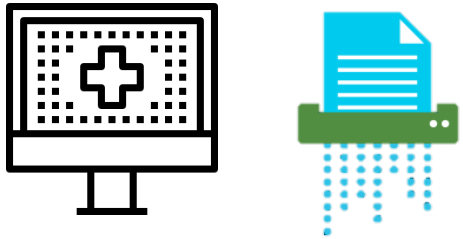
Antiepileptic drug
Orphan drug
Stiripentol
Clinical trials

ABSTRACT

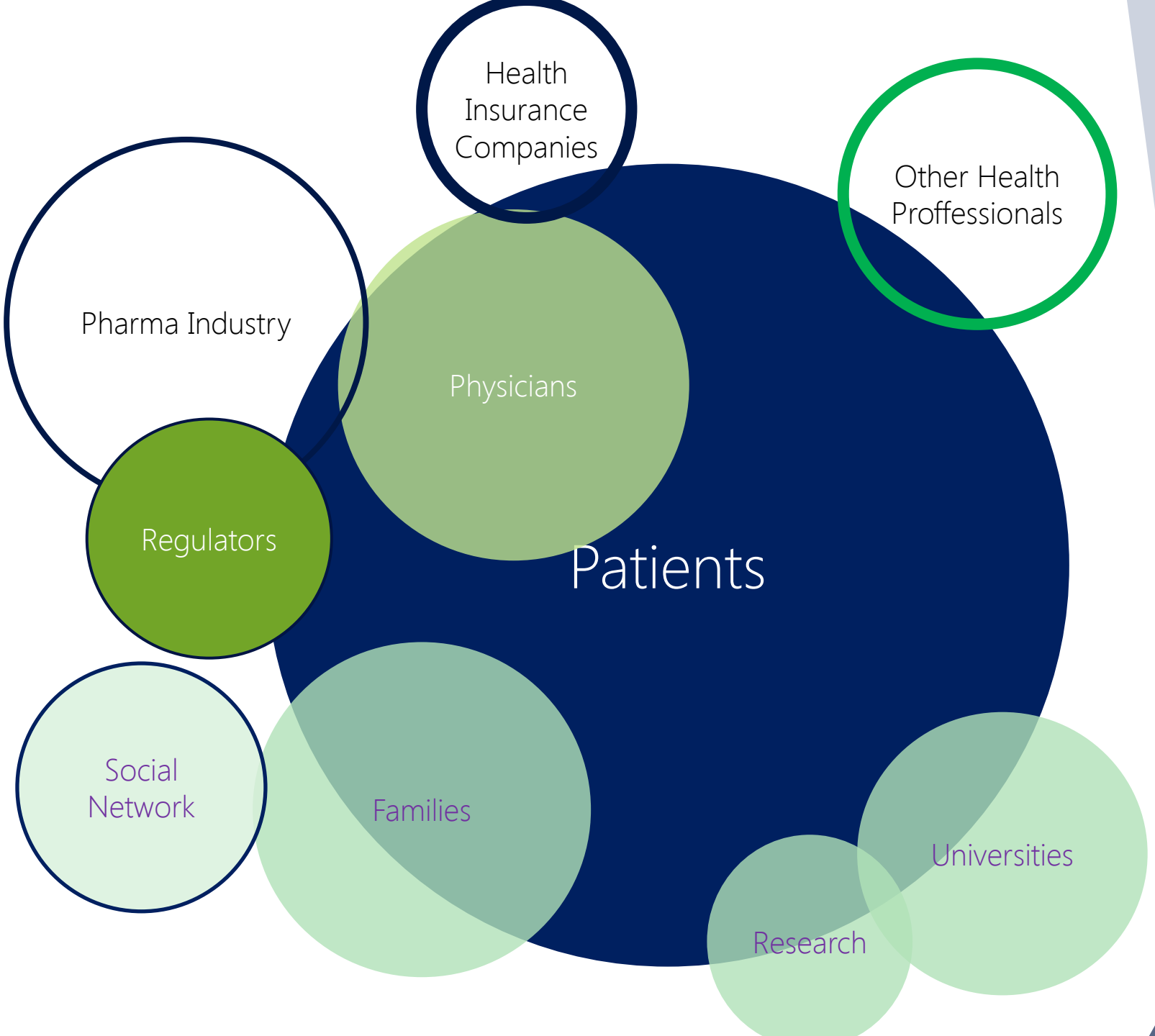
Dravet syndrome is a rare form of epilepsy largely refractory to current antiepileptic medications. The only precedents of randomized placebo-controlled trials in Dravet syndrome are the two small trials that led to the approval of stiripentol. With the arrival of new clinical trials for Dravet syndrome, we sought to determine the characteristics of the patient population with Dravet syndrome in Europe today, which has possibly evolved subsequent to the approval of stiripentol and the ability to diagnose milder clinical cases via genetic testing. From May to June 2014, we conducted an online parent-reported survey to collect information about the demographics, disease-specific clinical characteristics, as well as current and past use of antiepileptic medications by European patients with Dravet syndrome. We present data from 274 patients with Dravet syndrome from 15 European countries. Most patients were between 4 and 8 years of age, and 90% had known mutations in *SCN1A*. Their epilepsy was characterized by multiple seizure types, although only 45% had more than 4 tonic–clonic seizures per month on average. The most common drug combination was valproate, clobazam, and stiripentol, with 42% of the total population currently taking stiripentol. Over a third of patients with Dravet syn-

Paradigm shift



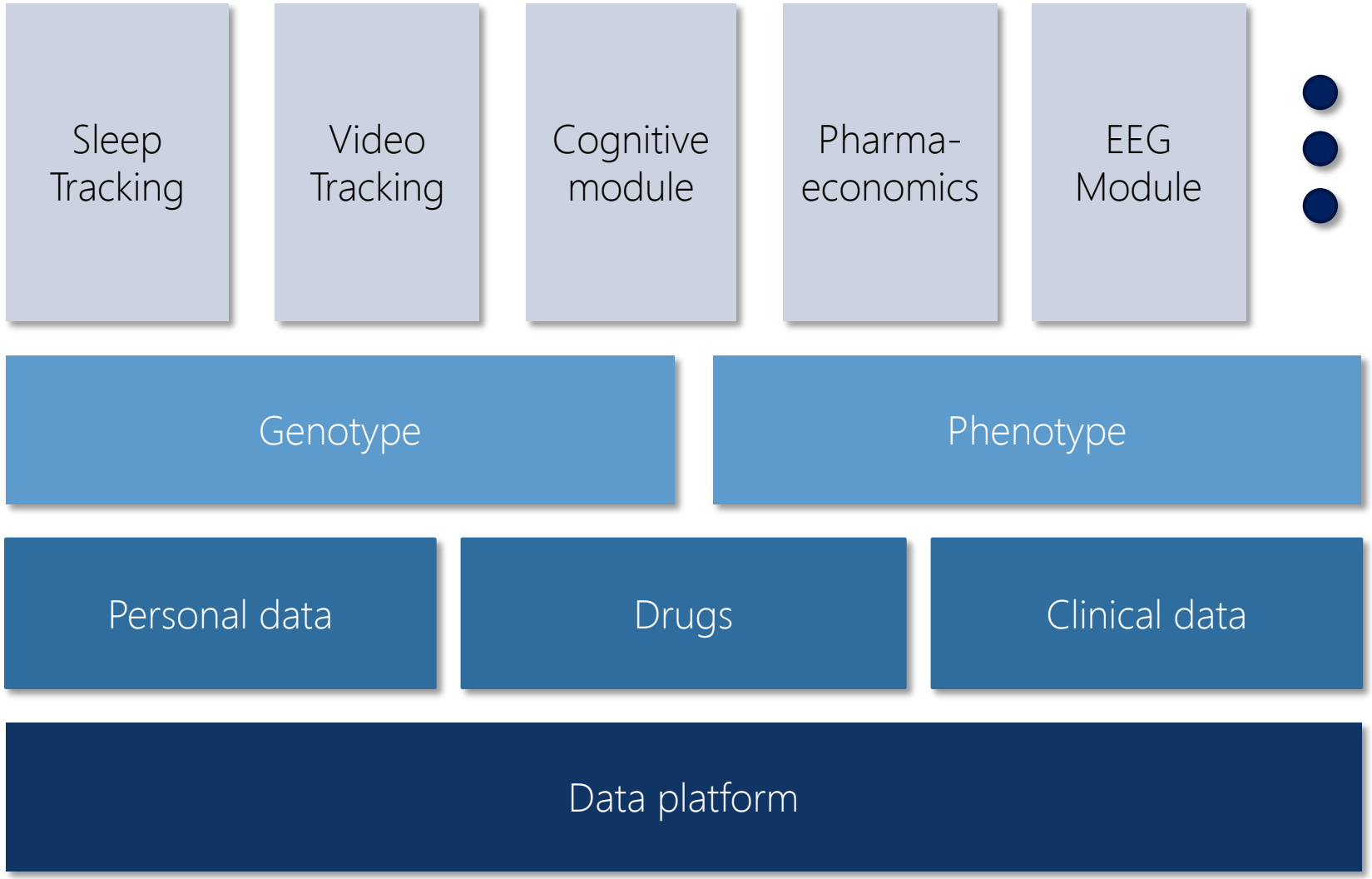


Ecosystem


Health Insurance Companies
Other Health Proffessionals
Pharma Industry
Physicians
Regulators
Patients
Social Network
Families
Universities
Research

Why PROMs?

- They are the reflection of patient-centeredness in clinical research;
- Patient Reported Outcomes are measurements based on data provided by patients regarding their health condition without amendment or interpretation of the patient's response by a clinician or anyone else;
- Such as the other types of Patient Relevant Outcome Measures, they have to be convincing to satisfy the requirements of both regulators and HTA during the assessment of a product.
- However, appropriate and validated outcome measures of disease activity, or disease progression, still do not exist for the vast majority of rare diseases, even diseases for which medicines are already approved, or for which therapies are under development.


Sleep Tracking
Video Tracking
Cognitive module
Pharma-economics
EEG Module
Genotype
Phenotype
Personal data
Drugs
Clinical data
Data platform

Sleep Tracking

Video Tracking

Cognitive module

Pharma-economi cs

EEG Module

Genotype

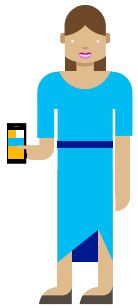
Phenotype

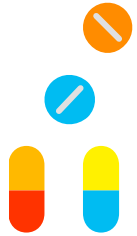
Personal data

Drugs

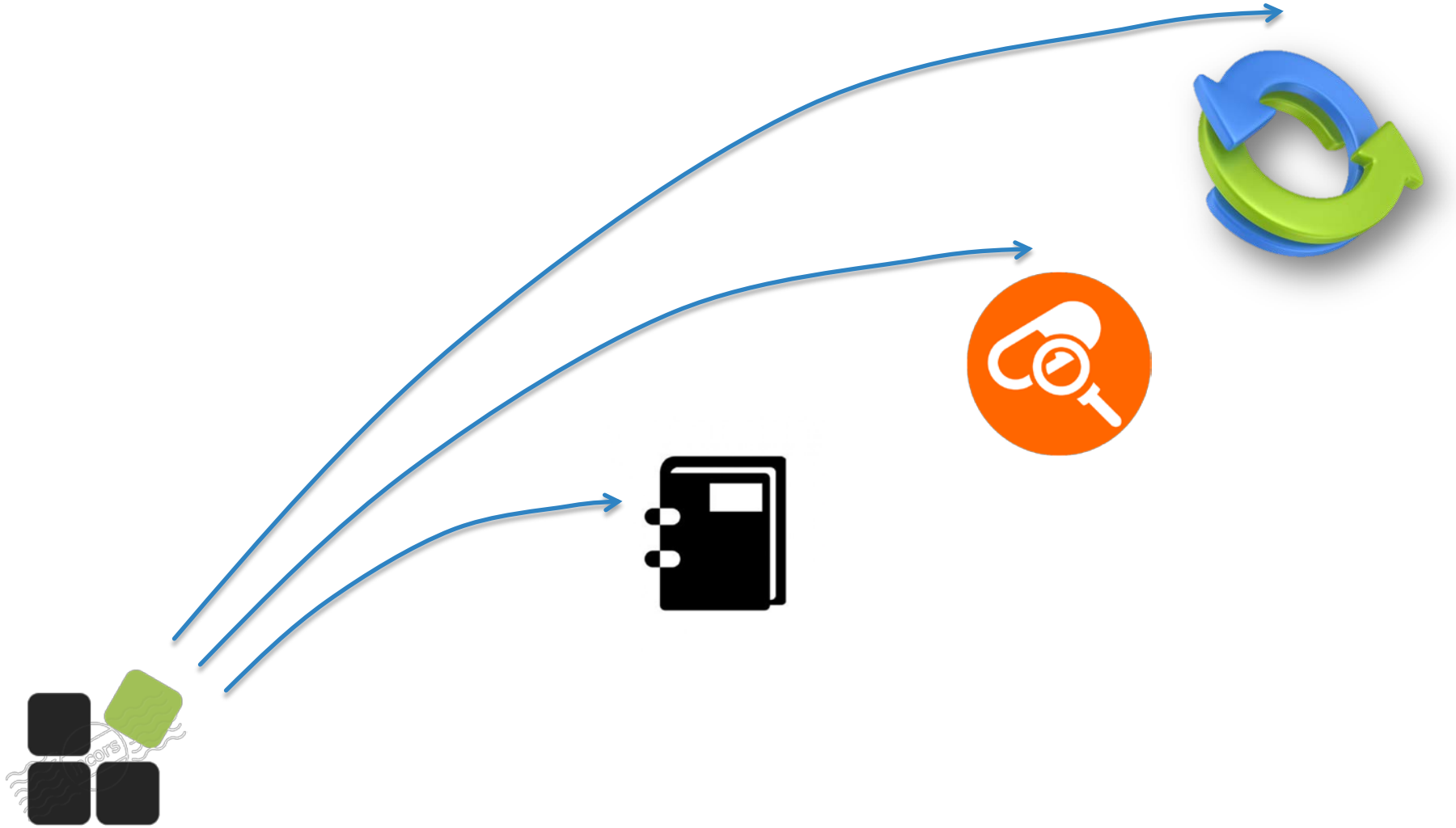
Clinical data

Data platform





Our vision



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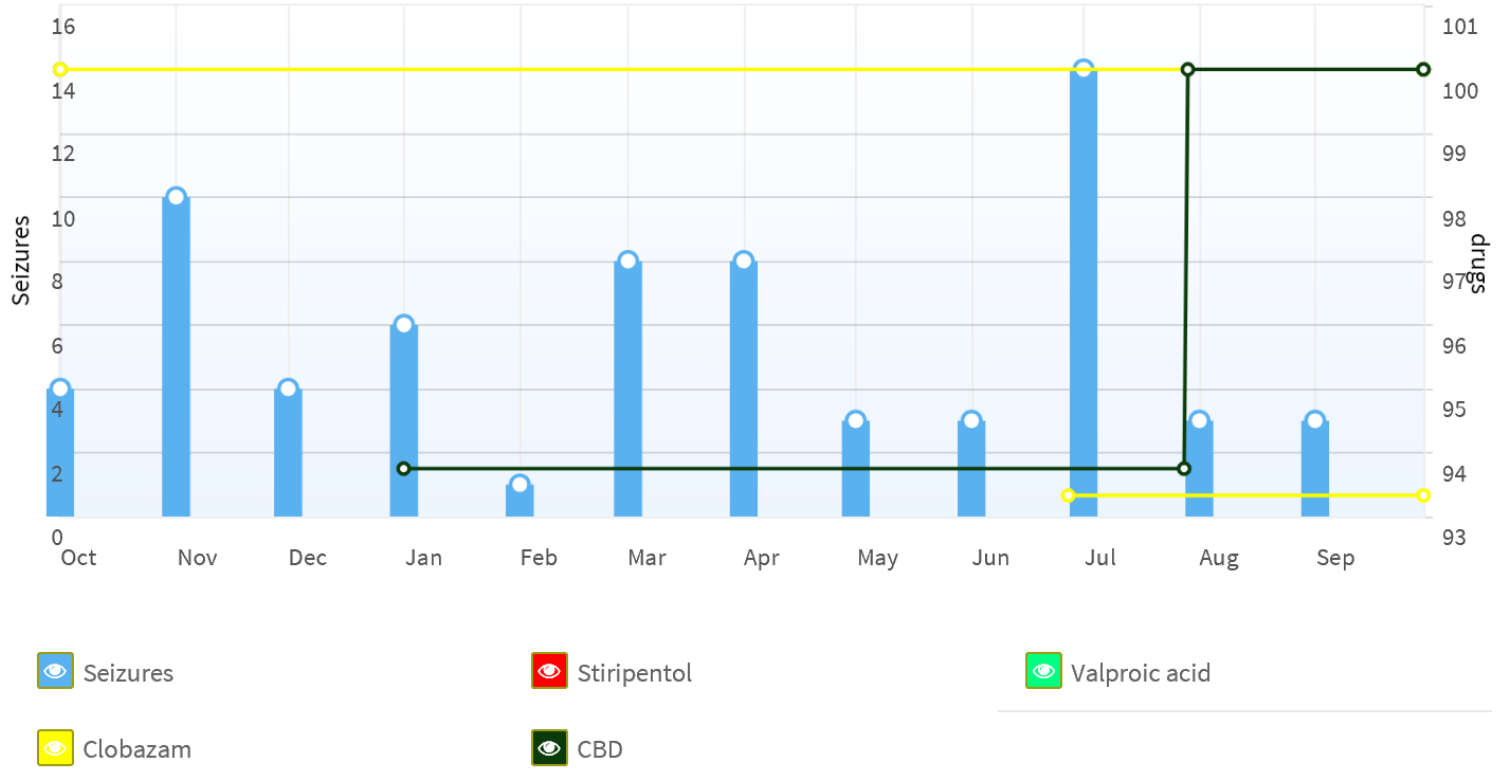
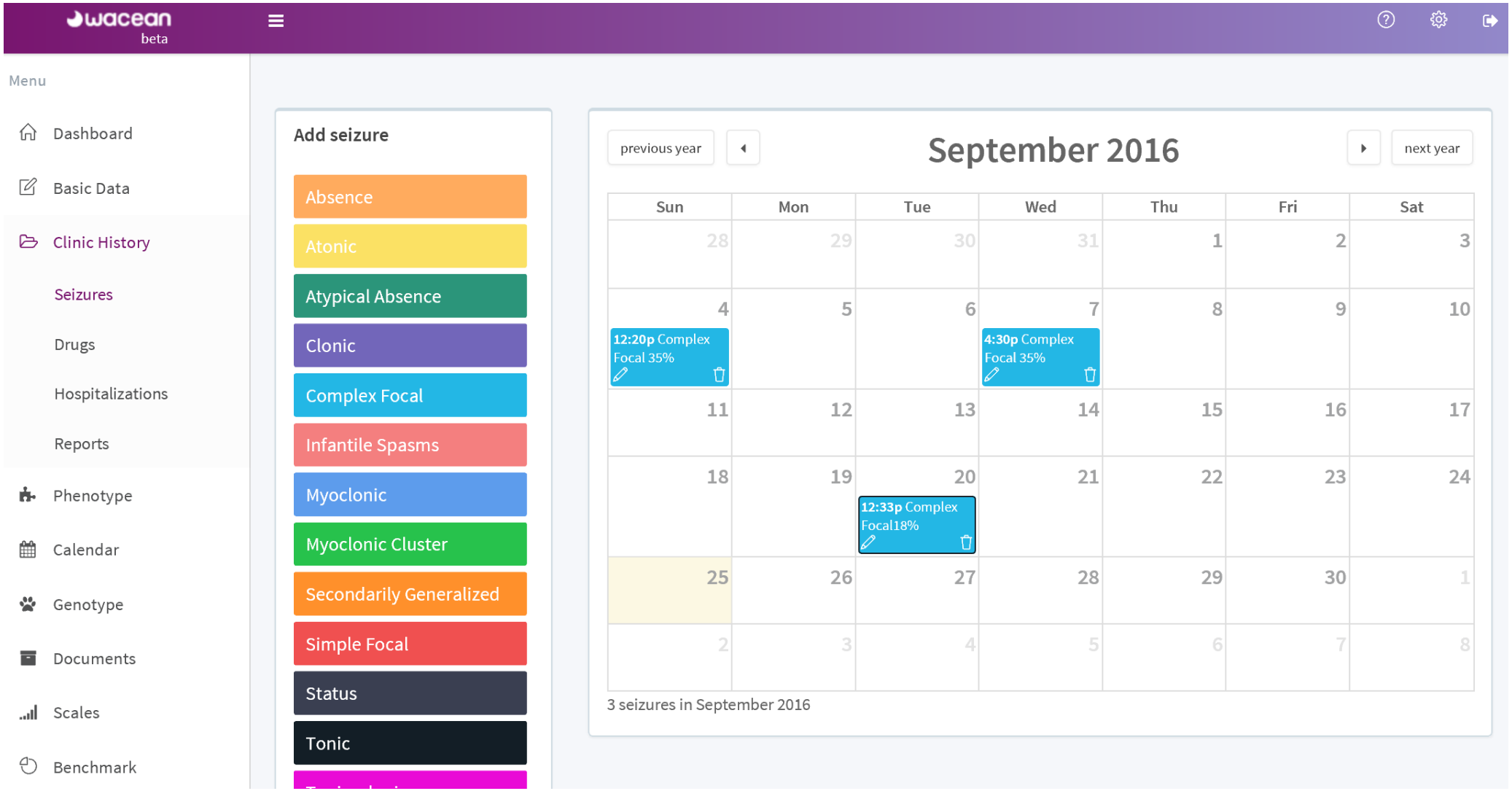
Menu

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

Seizures and drugs

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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
next year
September 2016
Sun
Mon
Tue
Wed
Thu
Fri
Sat
12:20p Complex Focal 35%
4:30p Complex Focal 35%
12:33p Complex Focal18%
3 seizures in September 2016

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Menu

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- Phenotype
- Calendar
- Genotype
- Documents
- Scales
- Benchmark

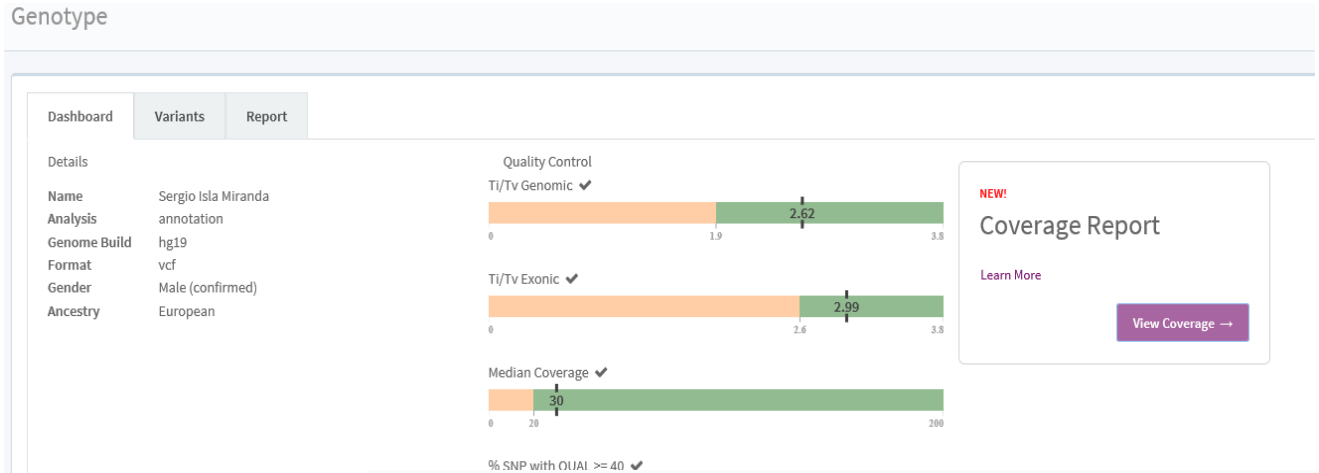
Phenotype

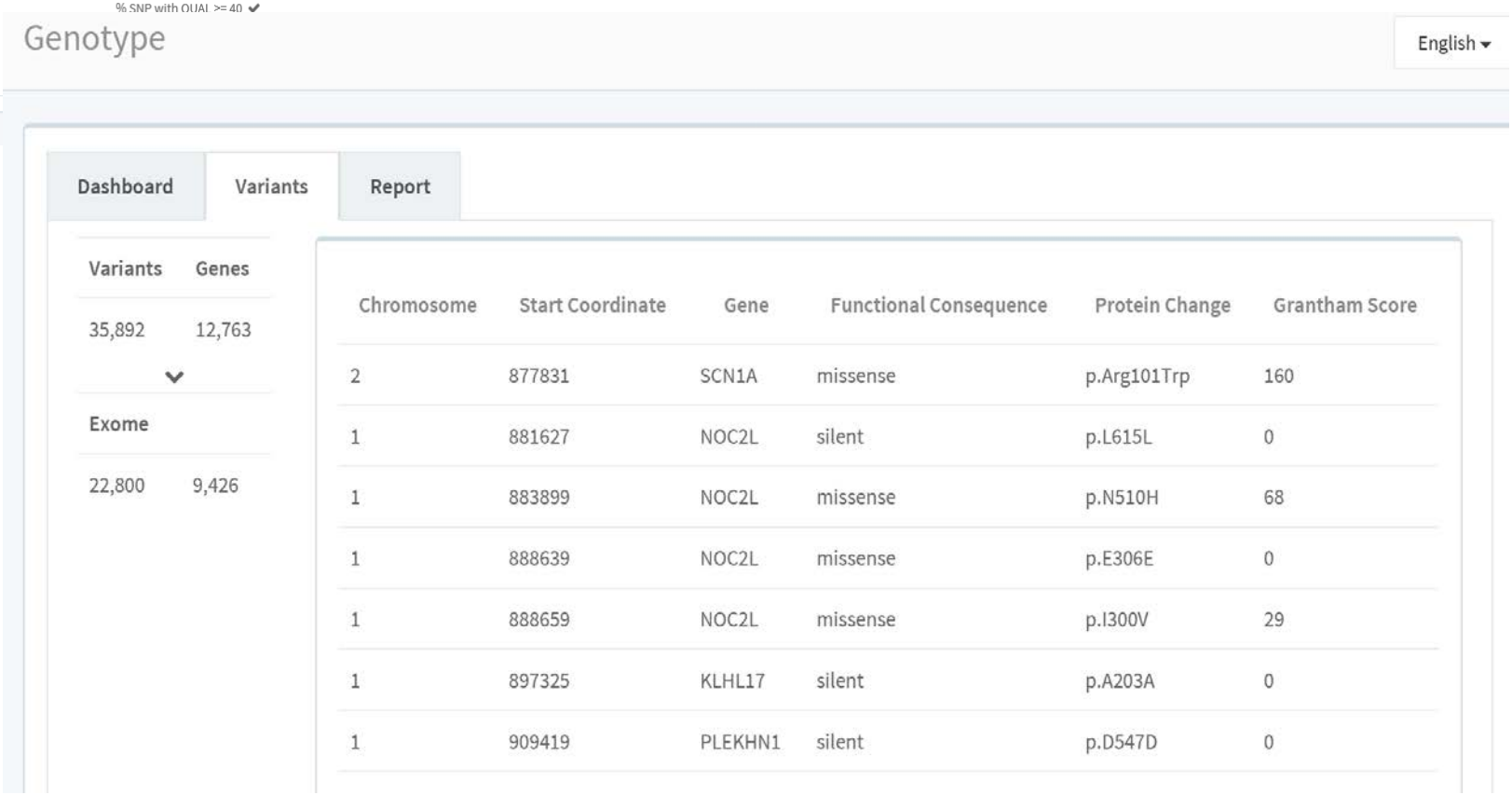
Checkboxes | Manual input

List of symptoms

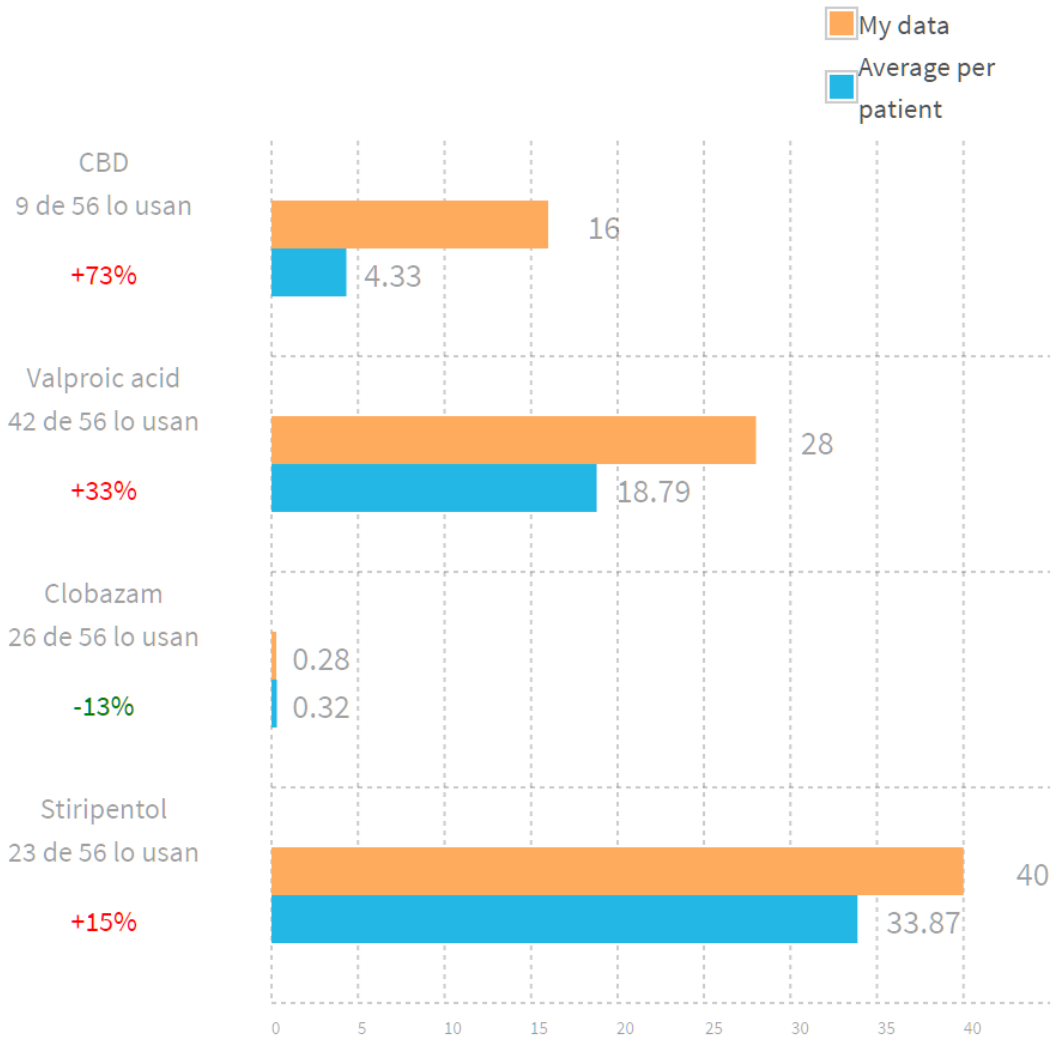
HP:0001250 : Seizures ×
HP:0002069 : Generalized tonic-clonic seizures ×
HP:0002133 : Status epilepticus ×
HP:0002353 : EEG abnormality ×
HP:0002373 : Febrile seizures ×
HP:0002384 : Focal seizures with impairment of consciousness or awareness ×
HP:0006813 : Hemiclonic seizures ×
HP:0007270 : Atypical 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



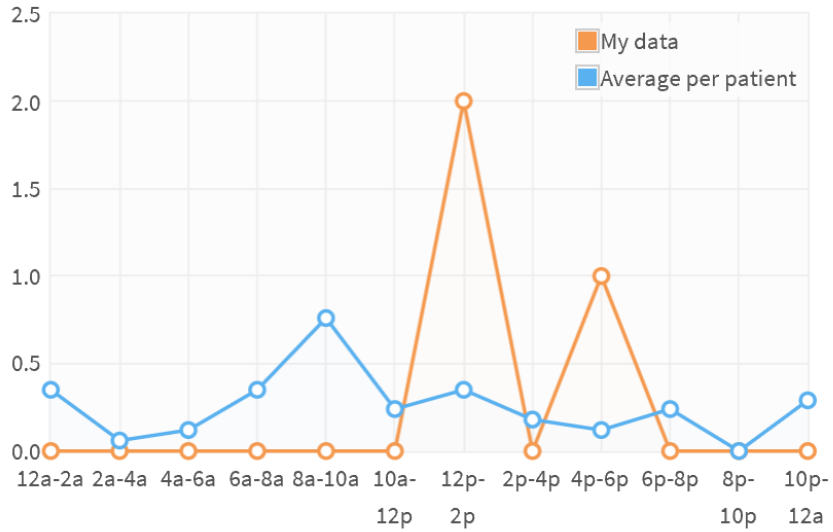


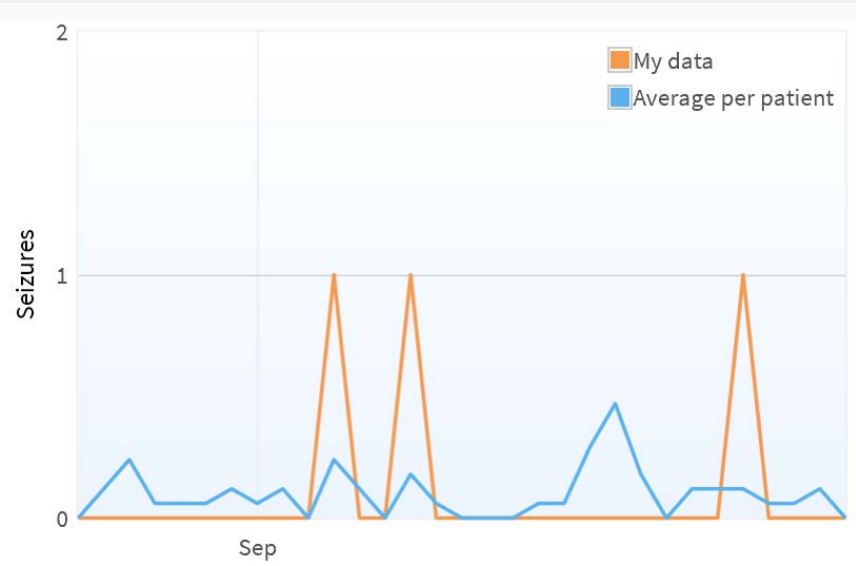
Chromosome	Start Coordinate	Gene	Functional Consequence	Protein Change	Grantham Score
2	877831	SCN1A	missense	p.Arg101Trp	160
1	881627	NOC2L	silent	p.L615L	0
1	883899	NOC2L	missense	p.N510H	68
1	888639	NOC2L	missense	p.E306E	0
1	888659	NOC2L	missense	p.I300V	29
1	897325	KLHL17	silent	p.A203A	0
1	909419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

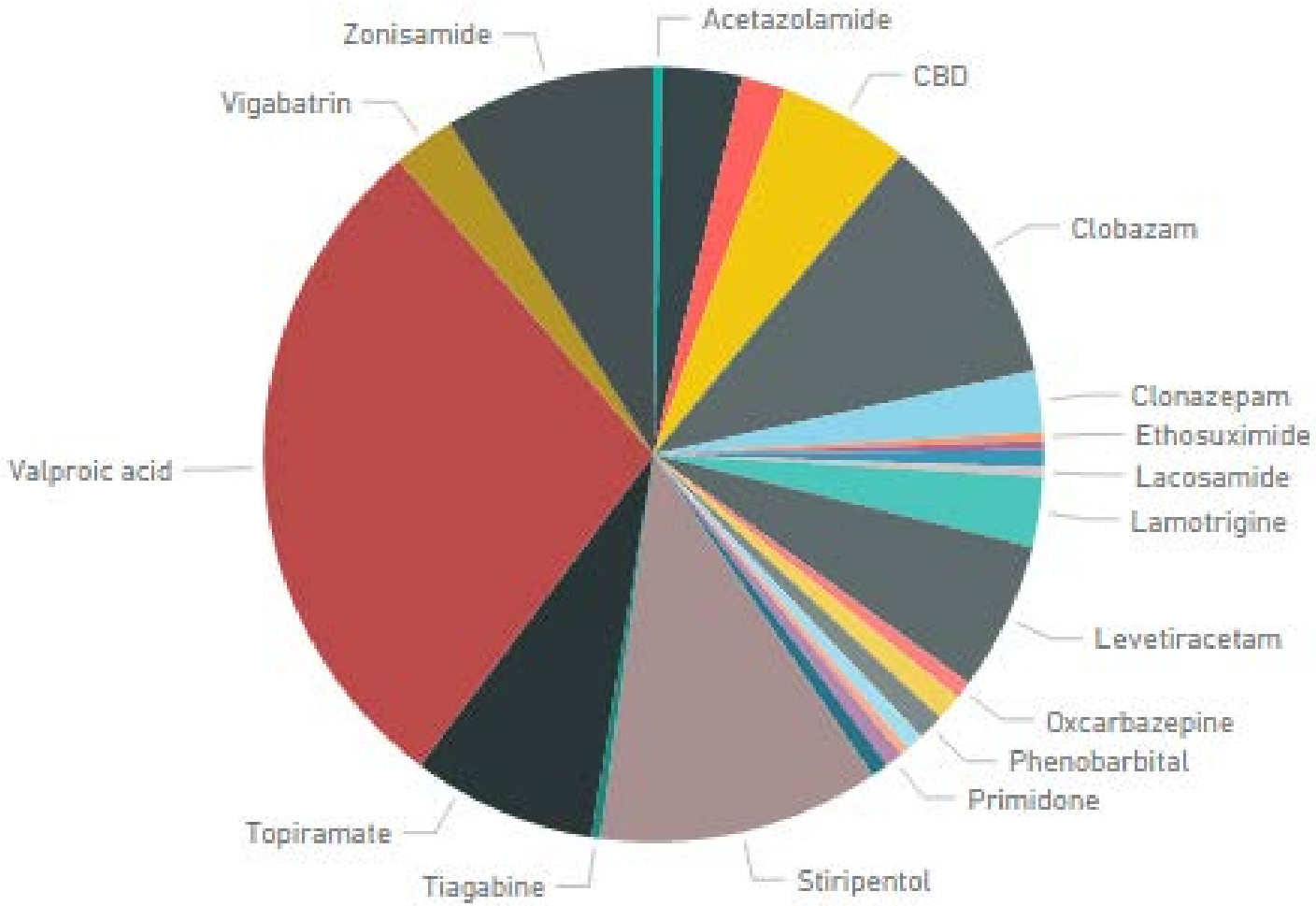


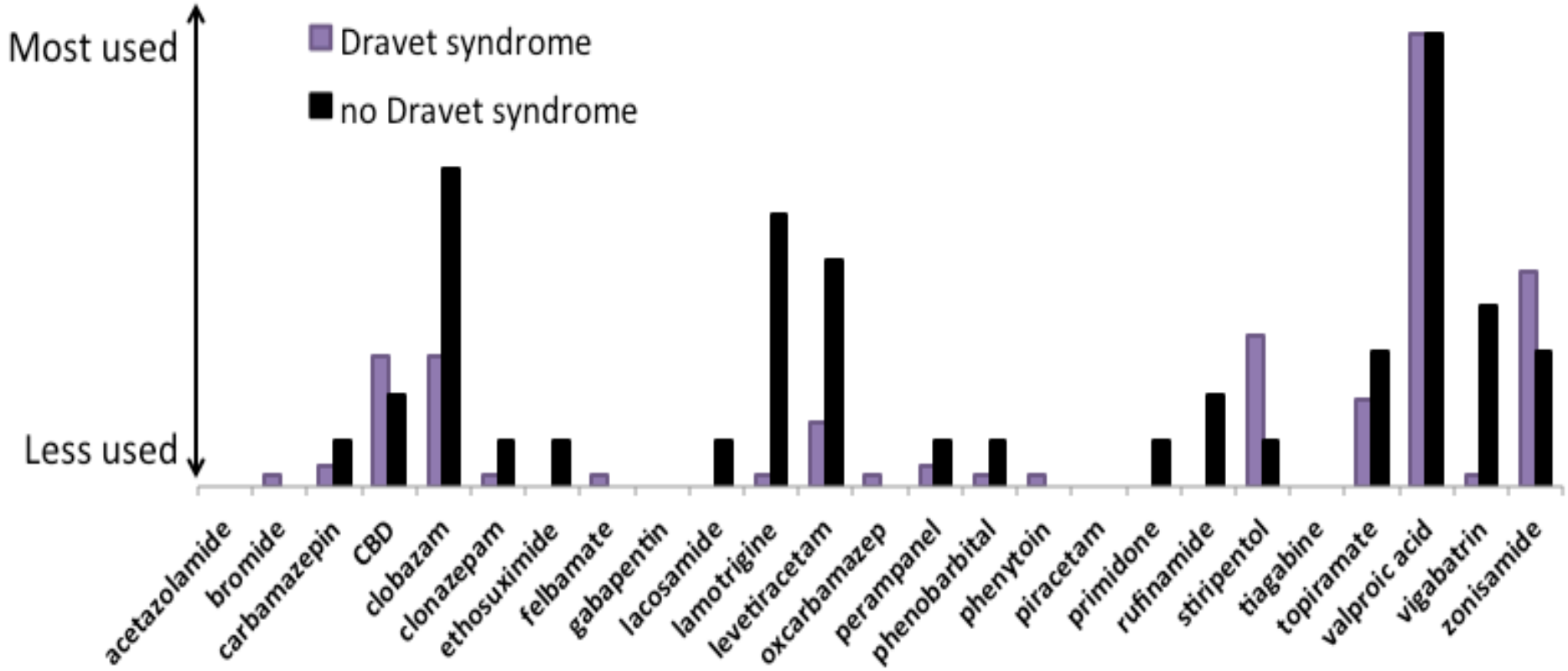
Seizures by time of day

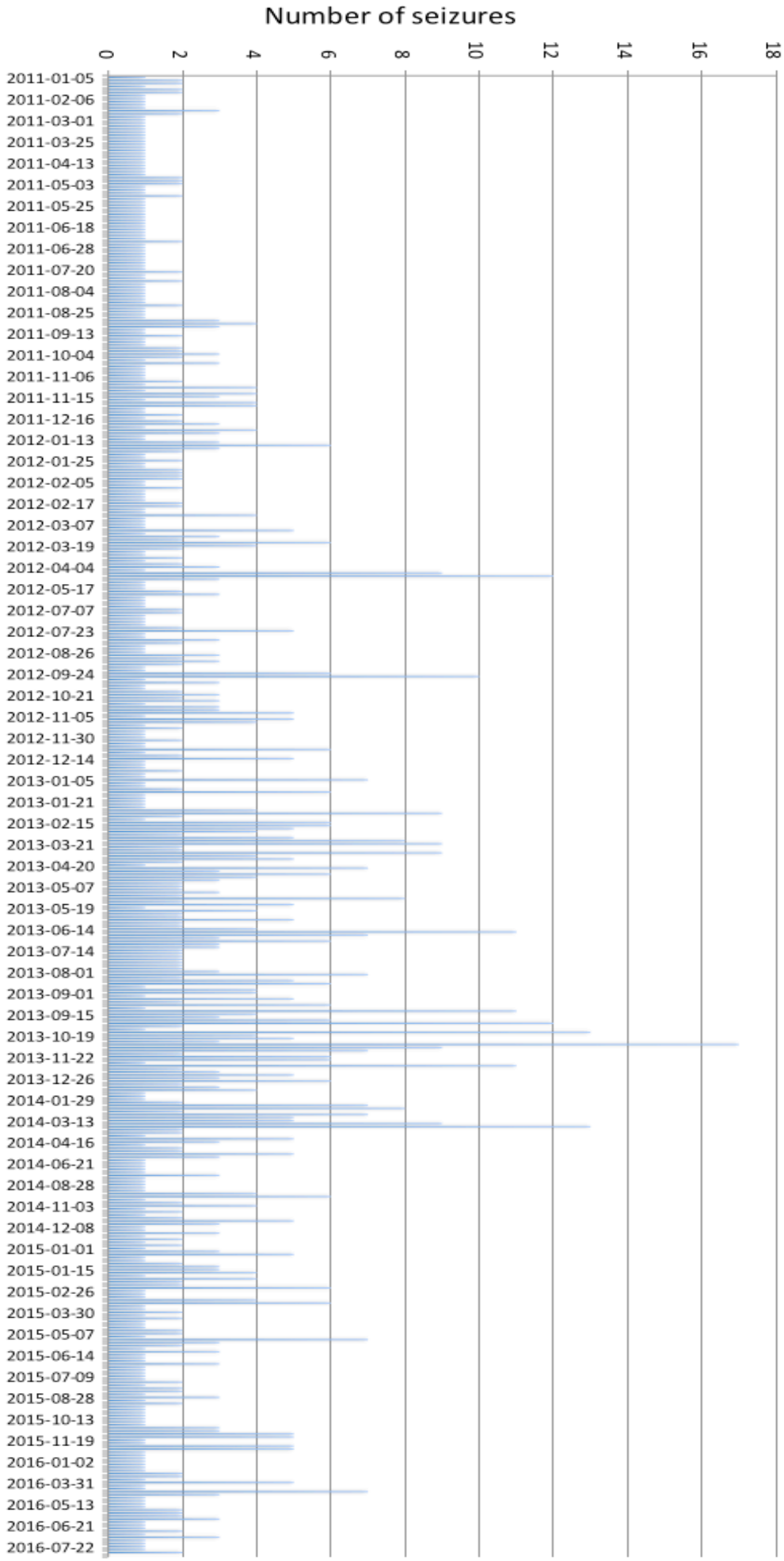
Options Last Month






Acetazolamide
CBD
Clobazam
Clonazepam
Ethosuximide
Lacosamide
Lamotrigine
Levetiracetam
Oxcarbazepine
Phenobarbital
Primidone
Stiripentol
Tiagabine
Topiramate
Valproic acid
Vigabatrin
Zonisamide


Most used
Less used
Dravet syndrome
no Dravet syndrome
acetazolamide
bromide
carbamazepin
CBD
clobazam
clonazepam
ethosuximide
felbamate
gabapentin
lacosamide
lamotrigine
levetiracetam
oxcarbamazep
perampanel
phenobarbital
phenytoin
piracetam
primidone
rufinamide
stiripentol
tiagabine
topiramate
valproic acid
vigabatrin
zonisamide


Number of seizures
0
2
4
6
8
10
12
14
16
18
2011-01-05
2011-02-06
2011-03-01
2011-03-25
2011-04-13
2011-05-03
2011-05-25
2011-06-18
2011-06-28
2011-07-20
2011-08-04
2011-08-25
2011-09-13
2011-10-04
2011-11-06
2011-11-15
2011-12-16
2012-01-13
2012-01-25
2012-02-05
2012-02-17
2012-03-07
2012-03-19
2012-04-04
2012-05-17
2012-07-07
2012-07-23
2012-08-26
2012-09-24
2012-10-21
2012-11-05
2012-11-30
2012-12-14
2013-01-05
2013-01-21
2013-02-15
2013-03-21
2013-04-20
2013-05-07
2013-05-19
2013-06-14
2013-07-14
2013-08-01
2013-09-01
2013-09-15
2013-10-19
2013-11-22
2013-12-26
2014-01-29
2014-03-13
2014-04-16
2014-06-21
2014-08-28
2014-11-03
2014-12-08
2015-01-01
2015-01-15
2015-02-26
2015-03-30
2015-05-07
2015-06-14
2015-07-09
2015-08-28
2015-10-13
2015-11-19
2016-01-02
2016-03-31
2016-05-13
2016-06-21
2016-07-22

360º care
Registry
Clinical treatment

Clinical trial
Significant benefit
Efficacy
Safety

HTA
Pay per efficacy

Potential uses

- Electronic patient record system for clinical trials
- Compasionate use data capture
- Post-marketing studies. Capture real world evidences

Data privacy

- ISO 27001, HIPAA, FedRAMP, SOC 1 and SOC 2
- Australia IRAP , UK G-Cloud, Singapore MTCS
- EU Standard Contractual Clauses / EU's Article 29 Working Party for contractual commitments

Working on a federated mode

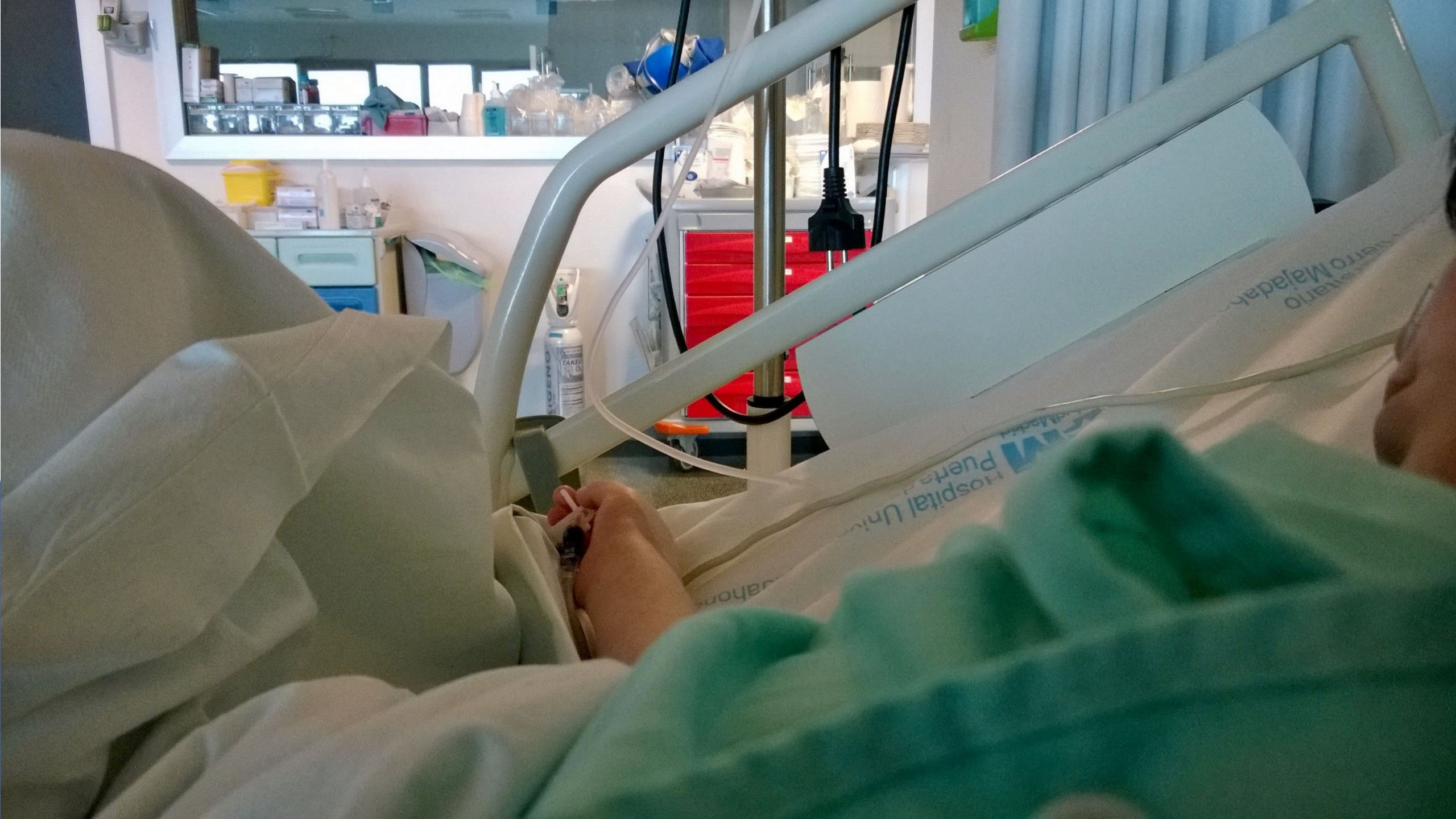
RareConnect.org
A EURORDIS INITIATIVE

RD Connect

Matchmaker Exchange
Genomic discovery through the exchange of phenotypic & genotypic profiles

PHENOME CENTRAL

Human Phenotype Ontology



Thank you

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