



When patients take the lead

Julian Isla

June 5-9, Barcelona

EURORDIS.ORG



draveteurope
Dravet Syndrome European Federation



EURORDIS
Rare Diseases Europe



EUROPEAN MEDICINES AGENCY
SCIENCE · MEDICINES · HEALTH



ciberer
CENTRO DE INVESTIGACIÓN BIOMÉDICA EN RED
DE ENFERMEDADES RARAS







Catastrophic

catastrophe

ˌkæˈtæʃ.əˈtrɒfi/ ˌkɑːnɑːˈstrɒfi

1. a sudden event that causes very great trouble or destruction.
2. a bad situation

Dravet Syndrome phenotype

- Severe cognitive and motor impairment
- Frequent status epilepticus
- No effective drugs
- No effective therapy
- 20% mortality
- Care-dependent for life
- No research
- No hope

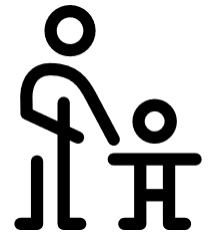
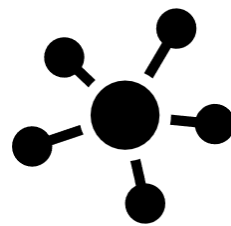
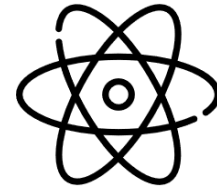
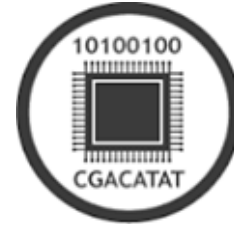
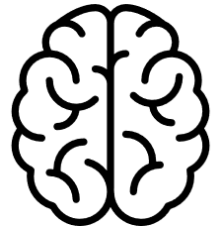




¿Patients?



We are impatient






lu. 20/05/2013 11:32

Nancy Hamilton <nancy.hamilton@eurordis.org>

EURORDIS Summer School list of participants

To Nancy Hamilton

Cc Maria Mavris

 You forwarded this message on 20/05/2013 12:13.

[Click here to download pictures.](#) To help protect your privacy, Outlook prevented automatic download of some pictures in this message.



Dear Summer School Participants,

Please find attached the list of attendees in case you would like to communicate with each other before the course begins.

I am also re-sending you the practical information sheet to help in organising your trip and your stay in Barcelona.

In the meantime, please let me know if you have any questions regarding the Summer School.

--

Nancy Hamilton
Training Manager
EURORDIS

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Capacity building



EURORDIS Summer School for Patient Advocates in Clinical Trials and Drug Development

Barcelona, Spain
June 17-21, 2013

Sponsored by

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Developed with the support of



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CERTIFICATE
for

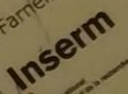
Julian Isla Gomez

32 hours of learning

EURORDIS Summer School
for Patient Advocates
in Clinical Trials and Drug Development

June 17-21, 2013
Cosmo Caixa
Barcelona

Josep Torrent-Farnell



Le Cam





EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

ubilee line
astbound
platform 2

Canary Wharf
North Greenwich
Canning Town
West Ham
Stratford

CANARY WHARF

Subline to
Westbound →
platform 1

Canary Wharf
Canning Place
Canning Lane
Canning Street
Canning Way
Canning Quay
Canning Dock
Canning Wharf

THE EXTRAORDINARY



NEL 66

2 1
Westbound
platform

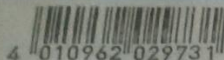


EMA

EMA



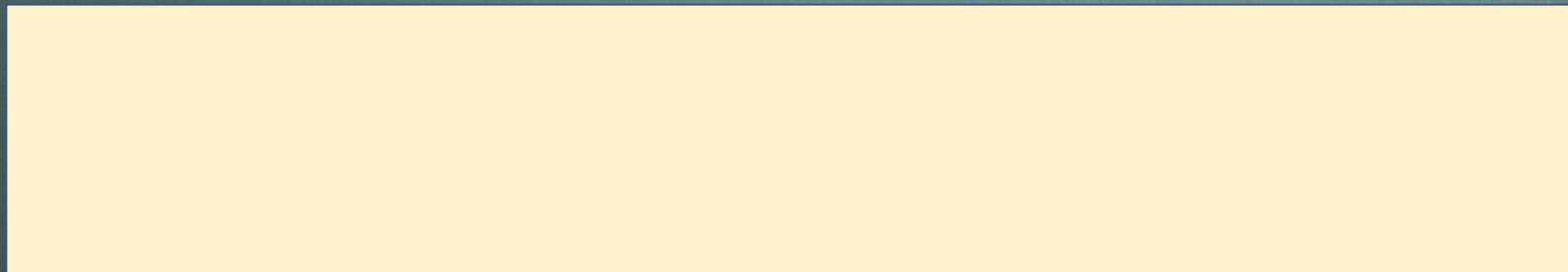
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MADE IN GERMANY



Orphan Application for the Treatment of Dravet Syndrome





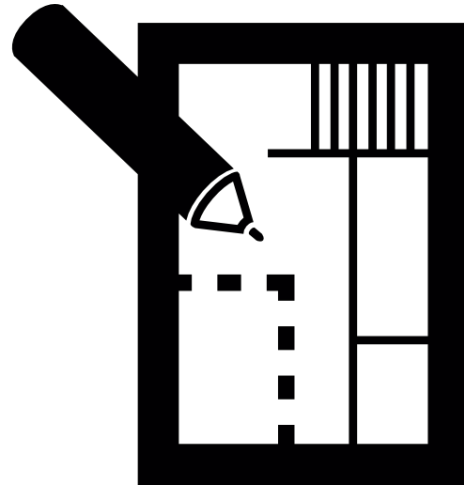
Scientific advice and protocol assistance



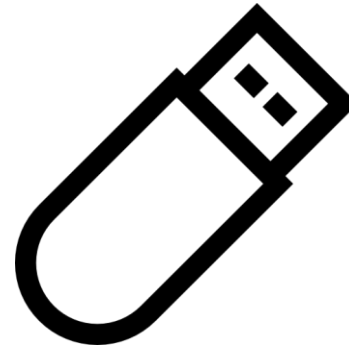
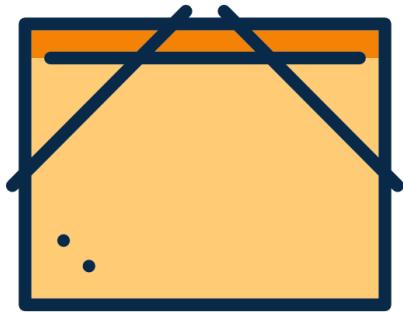
The screenshot shows the European Medicines Agency (EMA) website. The header includes the EMA logo, the text "EUROPEAN MEDICINES AGENCY SCIENCE MEDICINES HEALTH", and the European Union flag. There is a search bar, text size controls, and social media links. The navigation menu is highlighted on "Human regulatory". The main content area is titled "Scientific Advice Working Party" and includes a breadcrumb trail: Home > Committees > Working parties and other groups > CHMP > Scientific Advice Working Party. The text describes the Scientific Advice Working Party (SAWP) as a standing working party with the sole remit of providing scientific advice and protocol assistance, established by the Committee for Medicinal Products for Human Use (CHMP). It lists the members: a chairperson, 28 members including three from the Committee for Orphan Medicinal Products (COMP), one from the Paediatric Committee (PDCO), and one from the Committee for Advanced Therapies (CAT). It also lists the areas of expertise: non-clinical safety and pharmacokinetics. A sidebar on the left lists various committees and groups, with "Working parties and other groups" and "CHMP" expanded.

Mainly advice for clinical trials

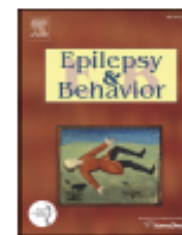
Inclusion and exclusion criteria







Data from 274 patients in just one week



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

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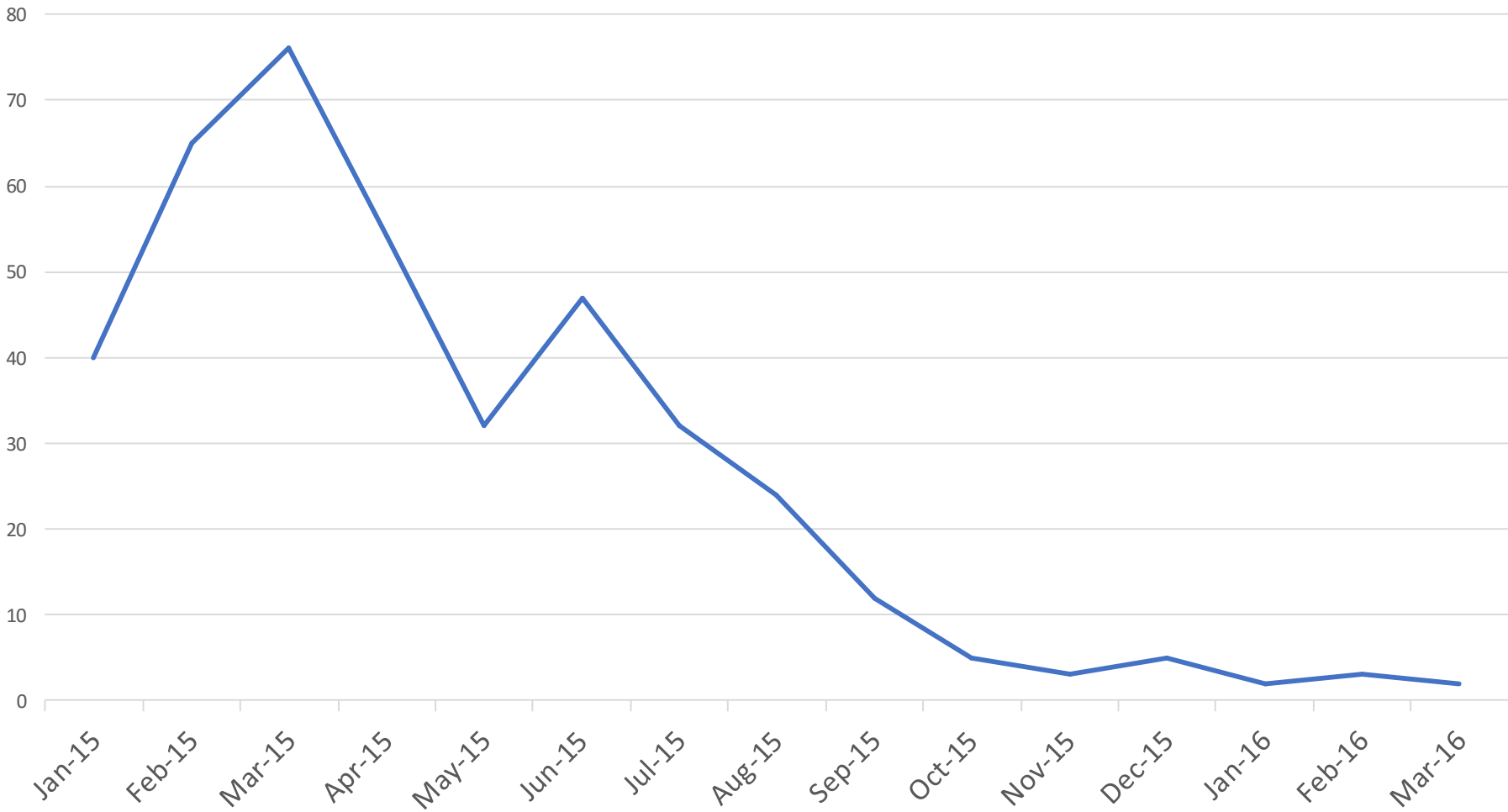
Keywords:

Dravet syndrome
Childhood epilepsy
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-

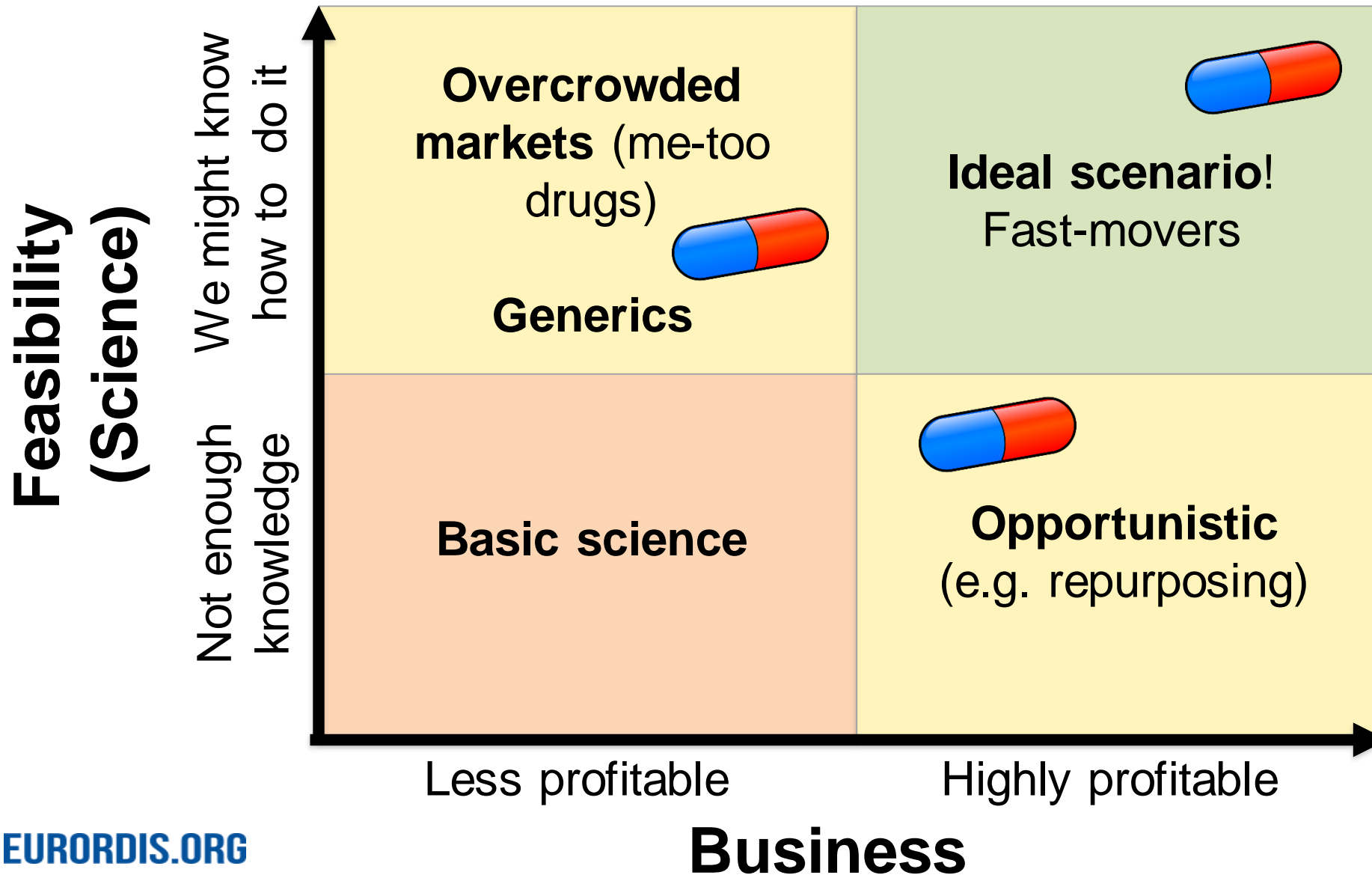
Seizures





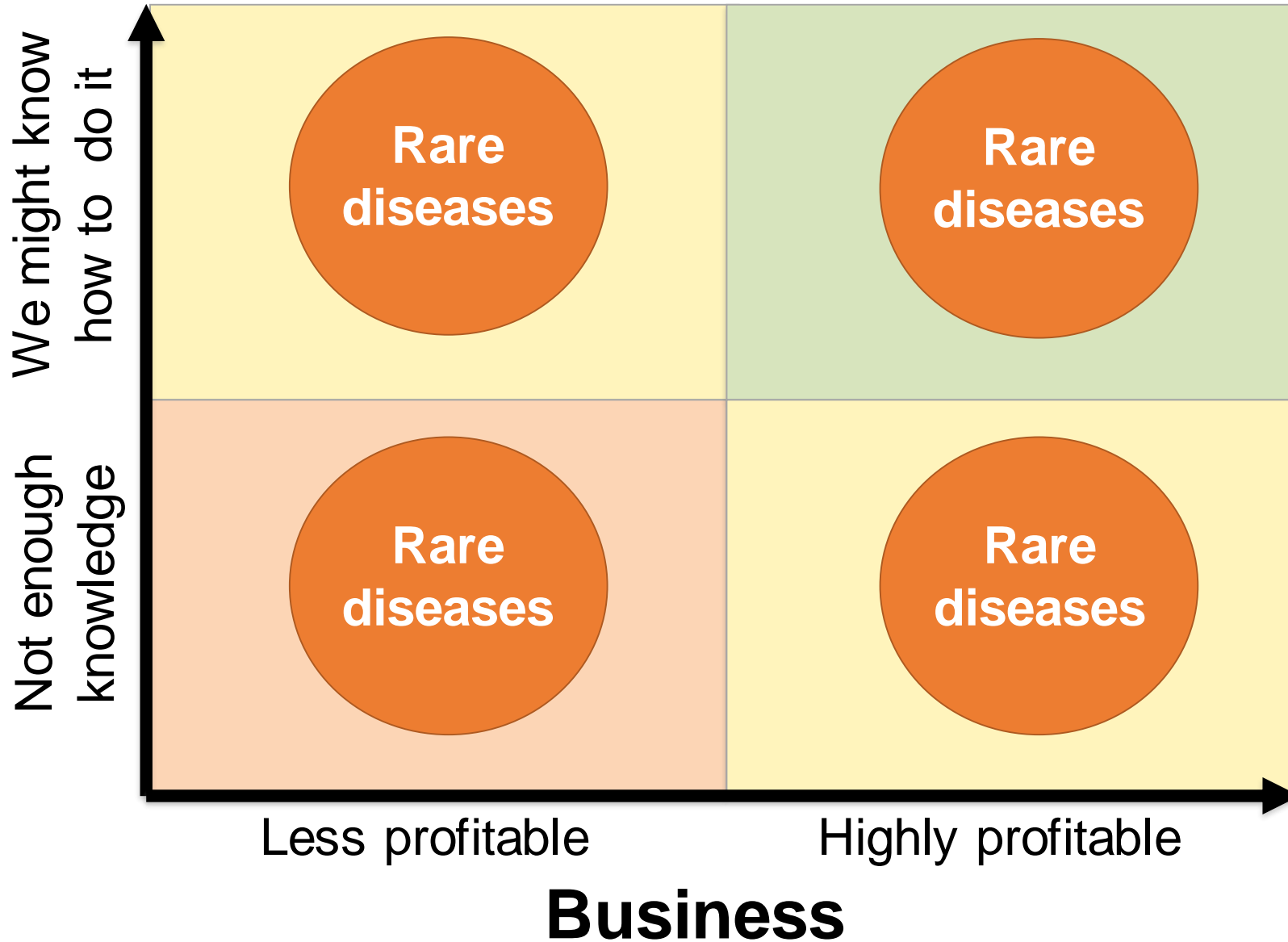
Choosing a disease
or drug indication

Drug development choices



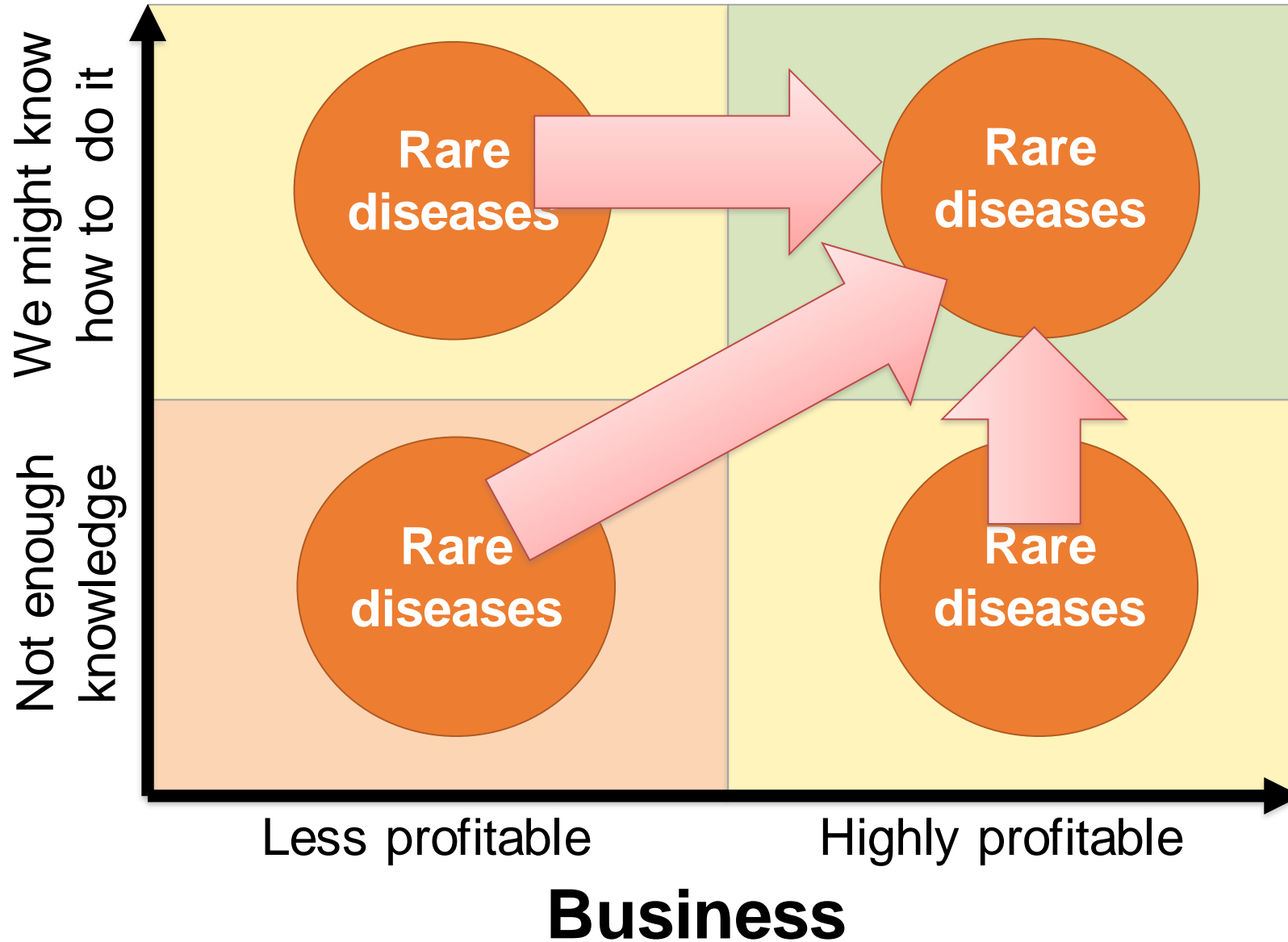
Know where your disease is
in the matrix

Feasibility (Science)



Have a strategy to
get into the feasible and
profitable box

Feasibility (Science)

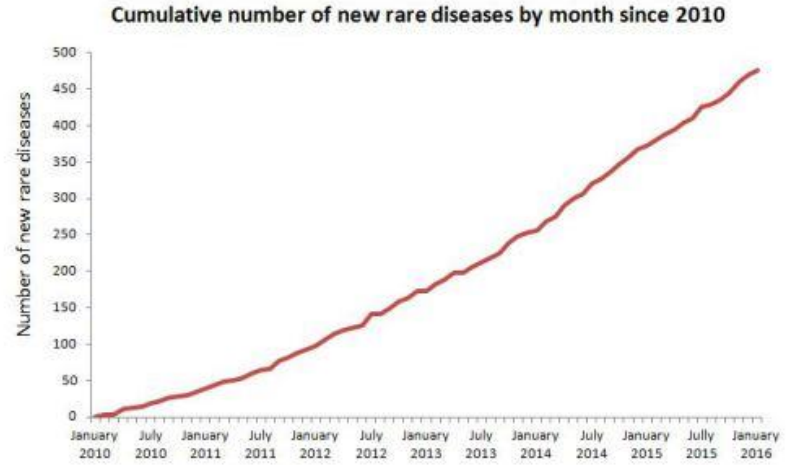


disruption

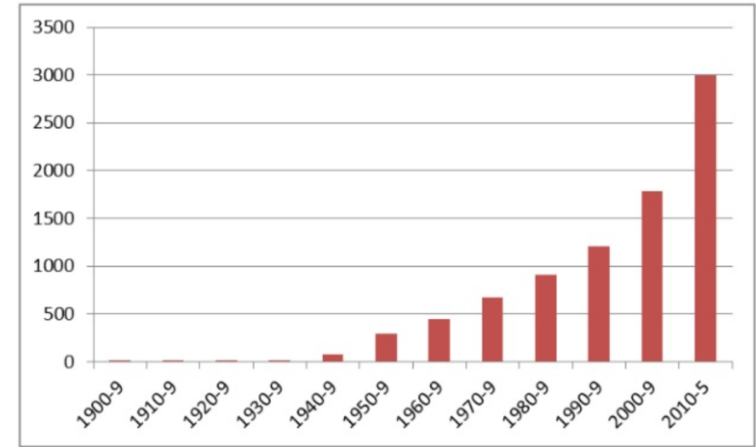
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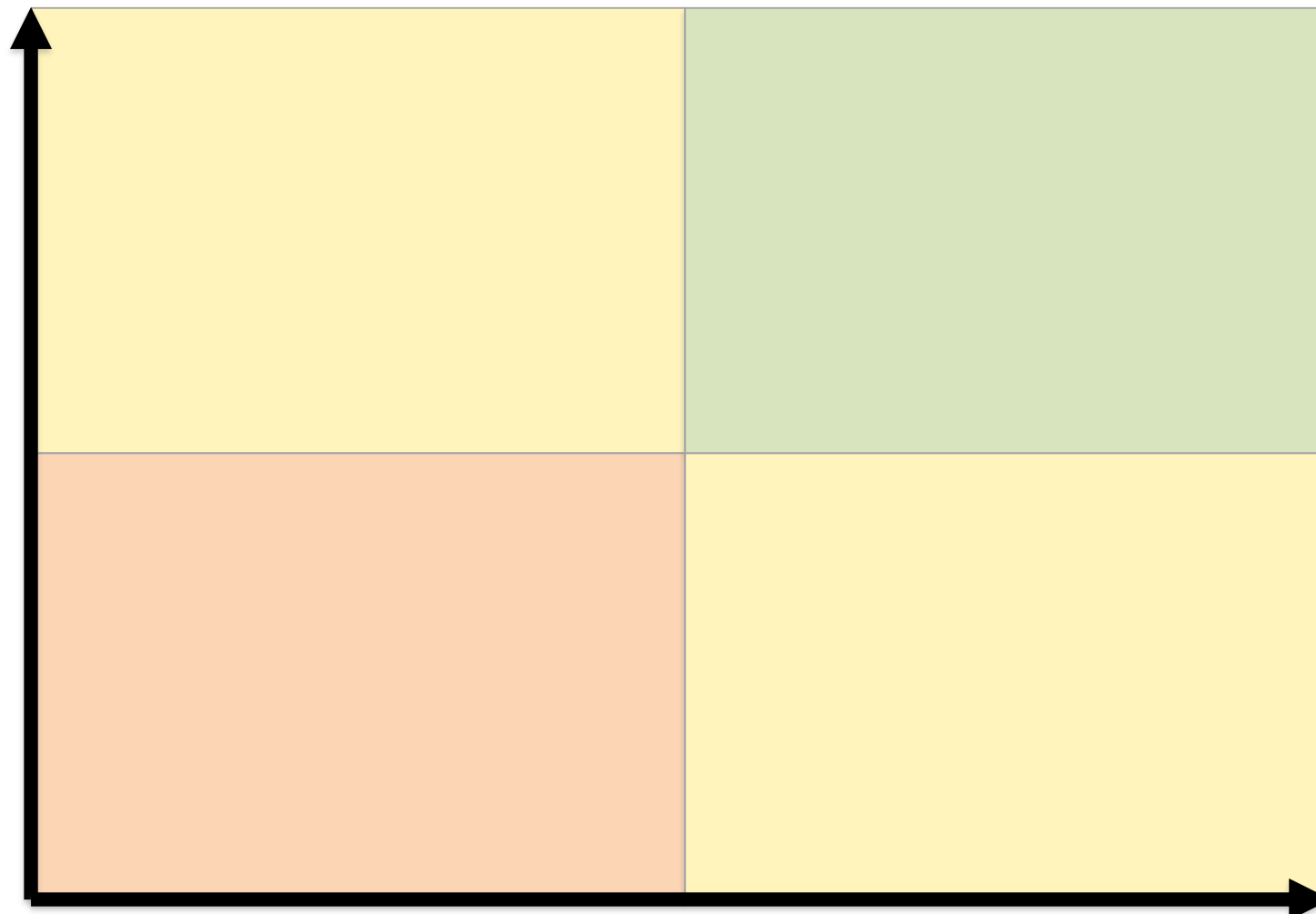
x35



Number of new articles in PubMed by decades



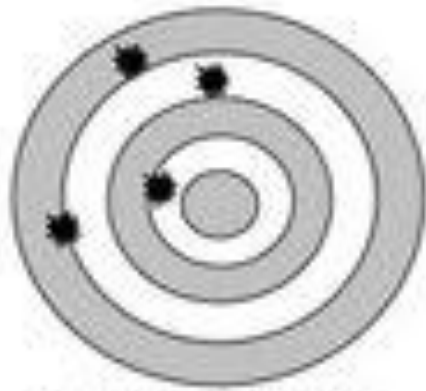
Knowledge



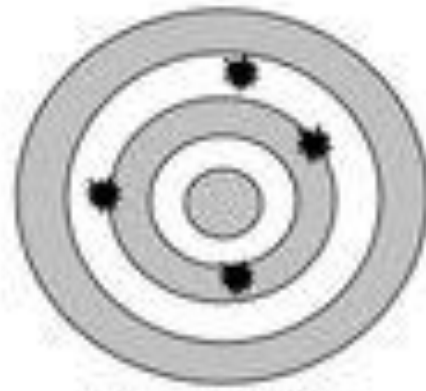
Less profitable

Highly profitable

Business



**Not Accurate
Not Precise**



**Accurate
Not Precise**



**Not Accurate
Precise**



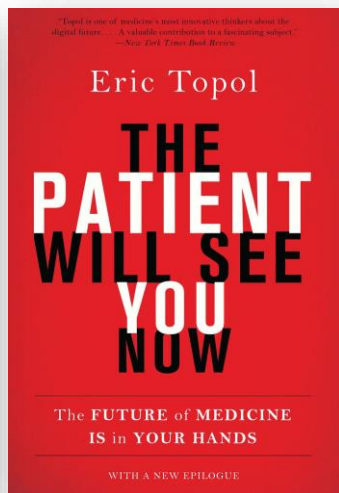
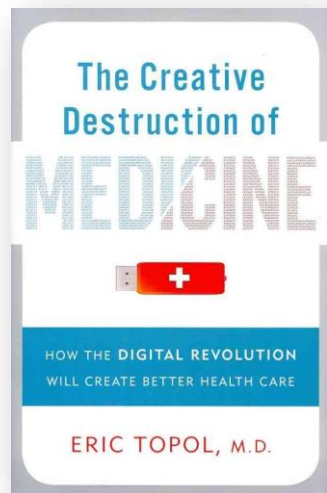
**Accurate
and Precise**

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

disruption

Eric Topol



Old medicine	New medicine
Population based	Individualized
One-Off, Doctor's office	Real-time streaming, real world data
Doctor ordered data	Patient generated data
Doctor's notes, unshared	Our notes, patient edited
Information owned by doctors and hospitals	Information owner by rightful owner
Expensive, big-ticket tech	Cheap chips, cloud
Data limited	Panoramic



“Healthcare is not a science problem, it’s an information problem”

Thomas Goetz



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

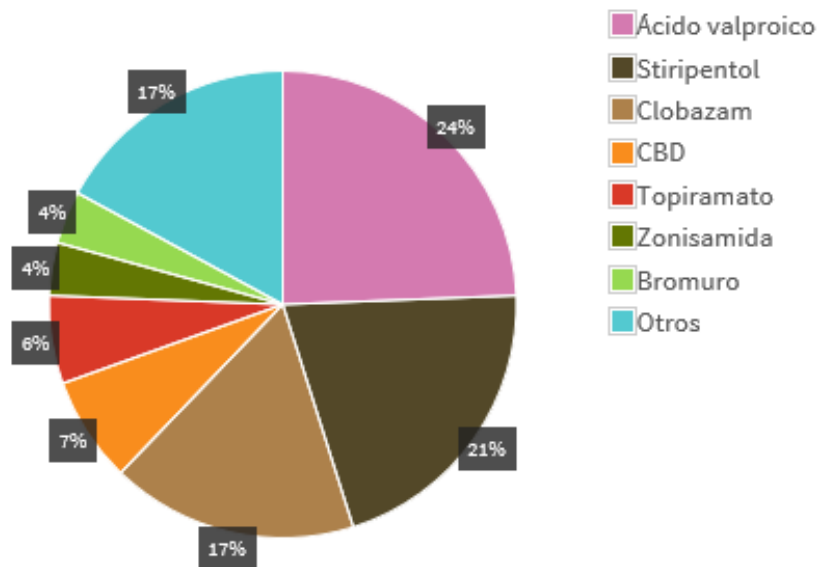
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Comparativa

Fármacos más comunes

Opciones Último mes



N = 29 Pacientes

PDP

Portal del paciente
Gestor de registros. Este es el portal de acceso. Puede configurarlo como desea.

José Perez
responsable@bbk.es

Mi cuenta

5
Tasa de actividad

Máximo de visitas

2
Número de visitas

Recibir un correo

Mi perfil completado

Crisis y medicamentación

PDP

Calendario de crisis
Añadir o quitar el tipo de crisis, o pulsar sobre el día para crear una crisis.

dicembre 2015

dom.	lun.	mar.	mié.	jue.	vie.	sáb.	dom.
		1	2	3	4	5	6
7	8	9	10	11	12	13	14
15	16	17	18	19	20	21	22
23	24	25	26	27	28	29	30
31							

Importar datos de seizuretracker.com en formato JSON

© 2015 - PDP

PDP

Informes

Ver crisis
Crisis libre

Crisis y medicamentos

Crisis
 Carbapentil
 Nitroazepan
 LAM
 Vigabatrin
 Carbamazepin
 Acedo valproico
 Medicamentos desnormalizados

Crisis por hora del día

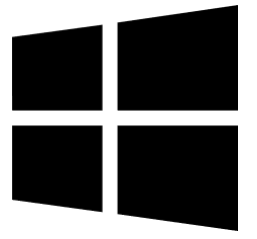
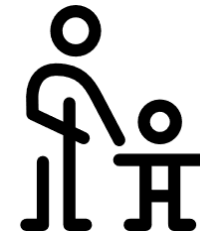
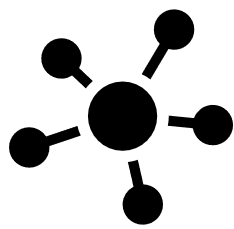
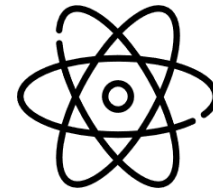
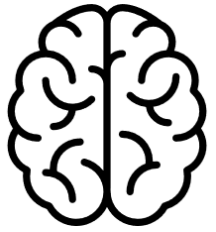
PDP

Crisis agrupadas por semanas

Tabla de crisis

1 de abril de 2015 - 31 de marzo de 2016 - Número de crisis: 106

Fecha	Hora	Tipo de crisis	Duración (horas)
5 de abril de 2015	18:30	Tónico-clónicas	00:30
24 de abril de 2015	18:42	Tónico-clónicas	00:50
21 de mayo de 2015	18:25	Tónico-clónicas	00:50
30 de mayo de 2015	18:14	Tónico-clónicas	00:58
22 de junio de 2015	13:30	Tónico-clónicas	00:50
30 de junio de 2015	23:40	Tónico-clónicas	00:55
4 de julio de 2015	04:30	Tónico-clónicas	00:50
7 de julio de 2015	18:00	Tónico-clónicas	00:50





Hospital Universitario
P.R. Puerto Rico

Centro de Diagnóstico y Referencia Epidemiológica





Thank you for your attention.

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