Making digital patients

Julian Isla
Founder

dravet europe
Dravet Syndrome European Federation

29 foundation twenty-nine

Founder

Microsoft Consulting Services. AI

Committee on Orphan Medicinal Products

TAG. Therapeutic Advisory Group

Founder

Advisor

Evaluator

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

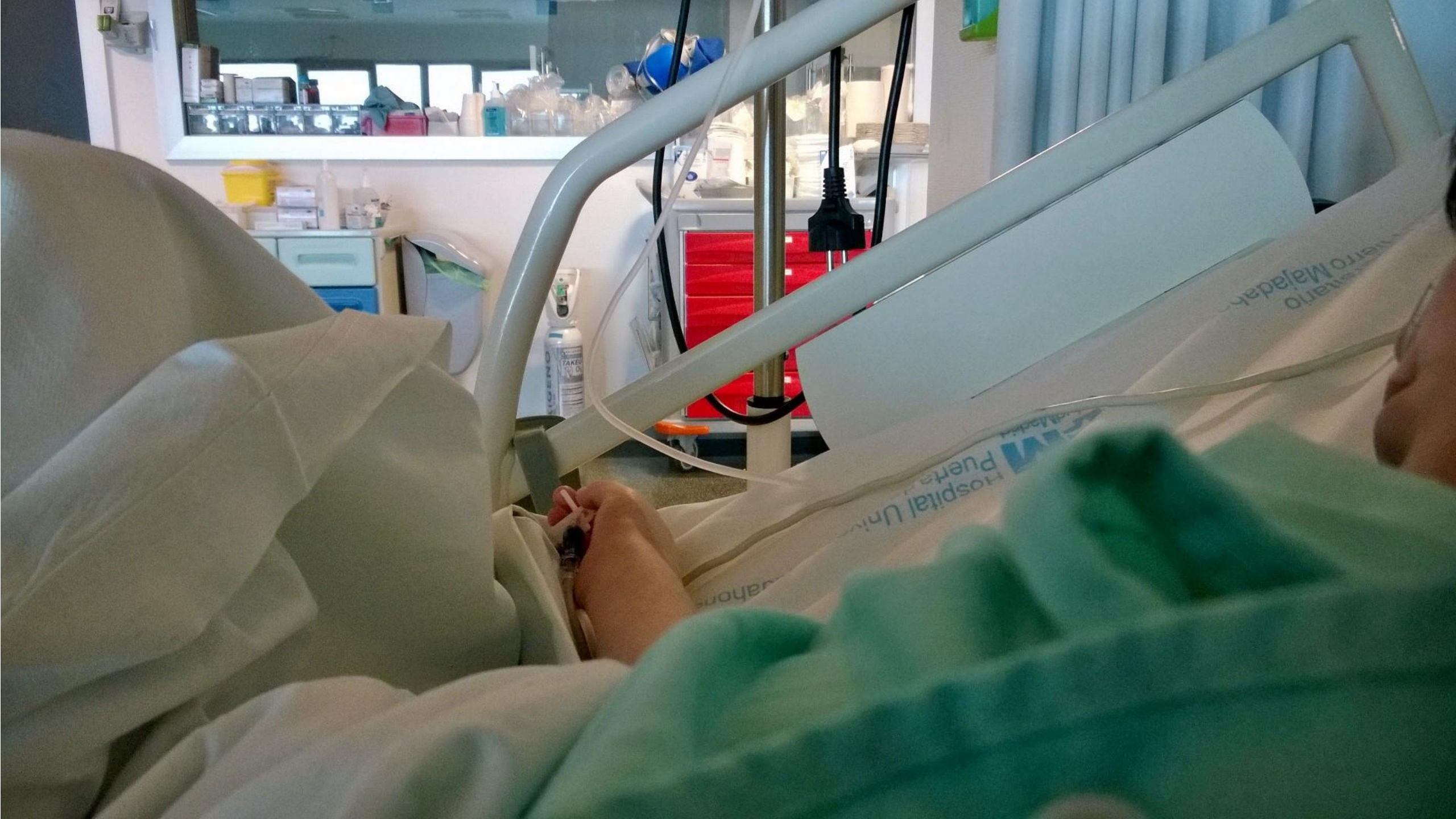
I am attending this conference as an individual expert, and I do not represent the EMA. The views expressed here are my personal opinions, and may shall not be understood or quoted on behalf of the EMA or reflect the position of the COMP

CV

http://www.ema.europa.eu/docs/en_GB/document_library/contacts/islaj_CV.pdf

DOI

http://www.ema.europa.eu/docs/en_GB/document_library/contacts/islaj_DI.pdf



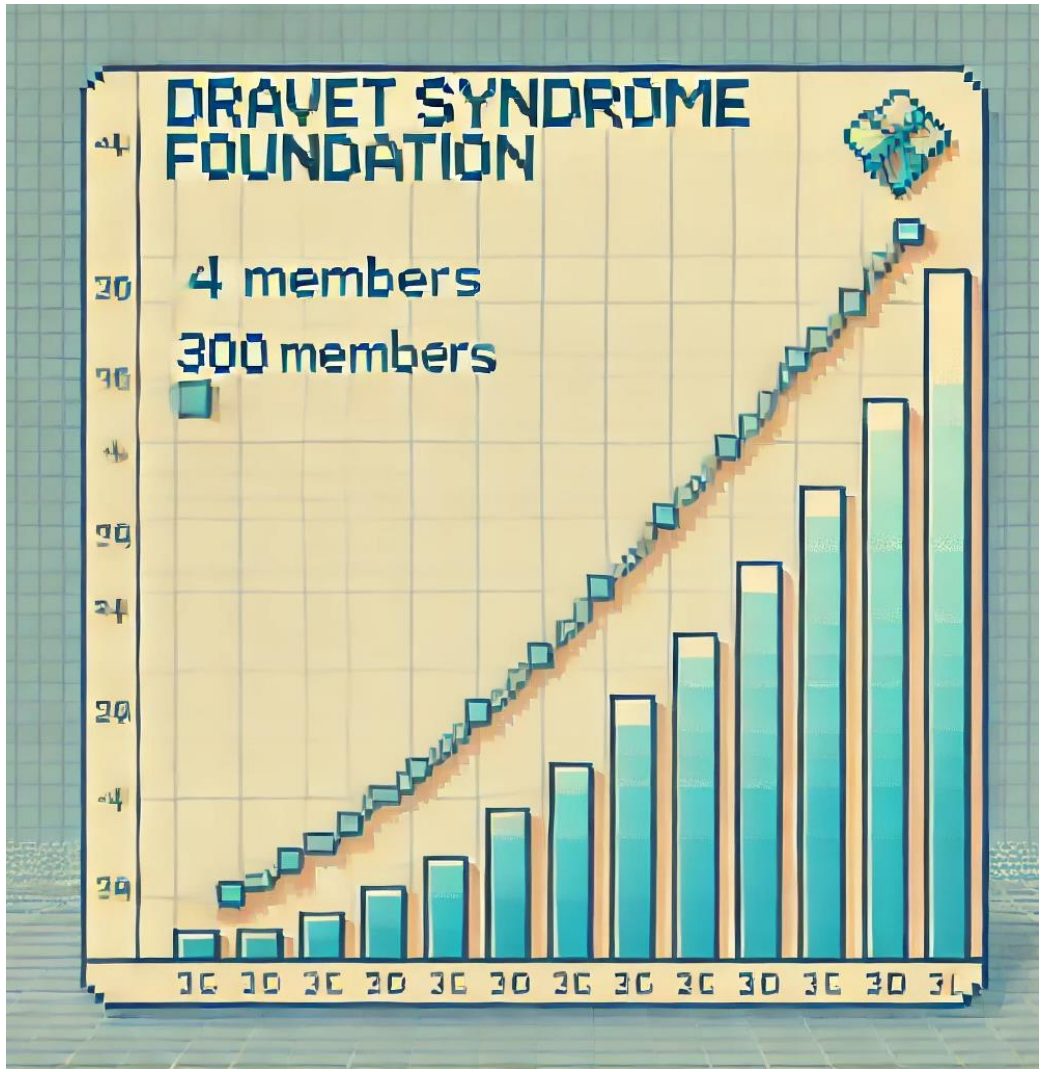
An engineer in the healthcare world

My history as a patient

Founding patient groups

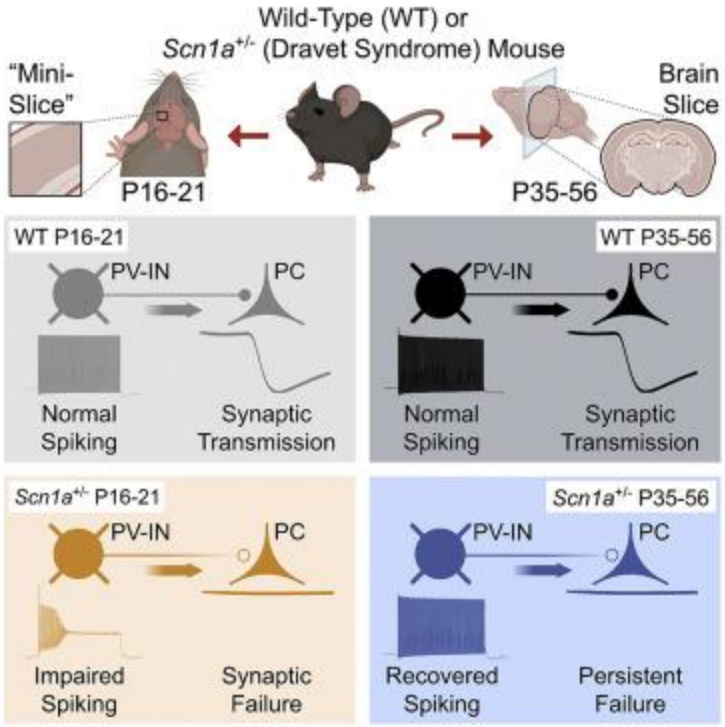
DRAVET SYNDROME
FUNDACIÓN
DELEGACIÓN EN ESPAÑA

dravetEurope
Dravet Syndrome European Federation



Science is not altruistic

William Albert Catterall (1946–2024)



I don't give a shit about your kids

The Jackson Laboratory

OVERVIEW HOW IT'S MADE H

129S1/SvImJ-*Scn1a*^{em1Dsf}/J

What Does This Nomenclature Mean?

Strain #:**034129**

RRID:IMSR_JAX:034129

Common Name: ***SCN1A*^{R613X}; Dravet model #10**

SCN1A^{R613X} mice carry an A to T point mutation in nucleotide 1837 (converting arginine (R) 613 to a STOP (X) codon) in addition to a silent C to T mutation at position 1833. The R613X nonsense mutation is expected to block the translation of complete protein from mRNA, enabling studies of readthrough therapeutics to restore protein function. On this 129S1/SvImJ genetic background, heterozygotes will likely demonstrate a mild (if any) Dravet Syndrome phenotype. Homozygotes are not viable.…

Read More

Donating Investigator(s)

Dravet Syndrome Foundation , Spain - null

2013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Rare Diseases Europe
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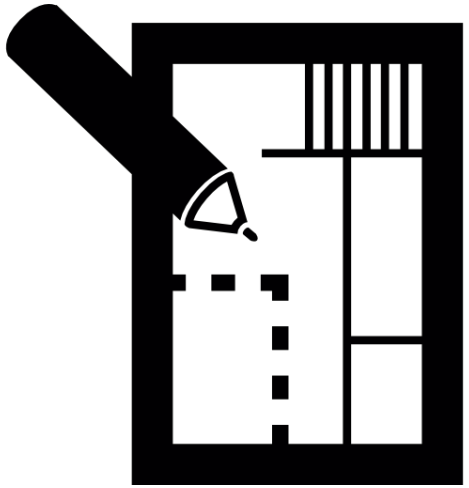
Orphan Application for the Treatment of Dravet Syndrome


cucina
FINALIST

Inclusion and exclusion criteria for fenfluramine CT

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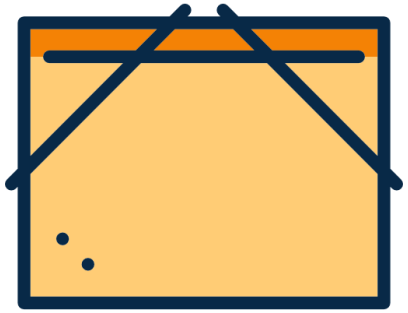

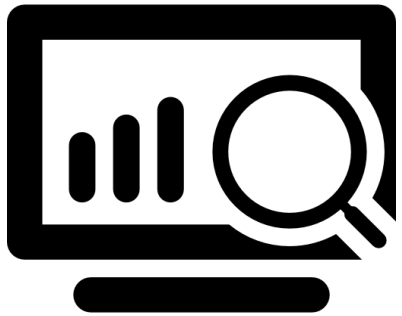

SurveyMonkey

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

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Dravet syndrome
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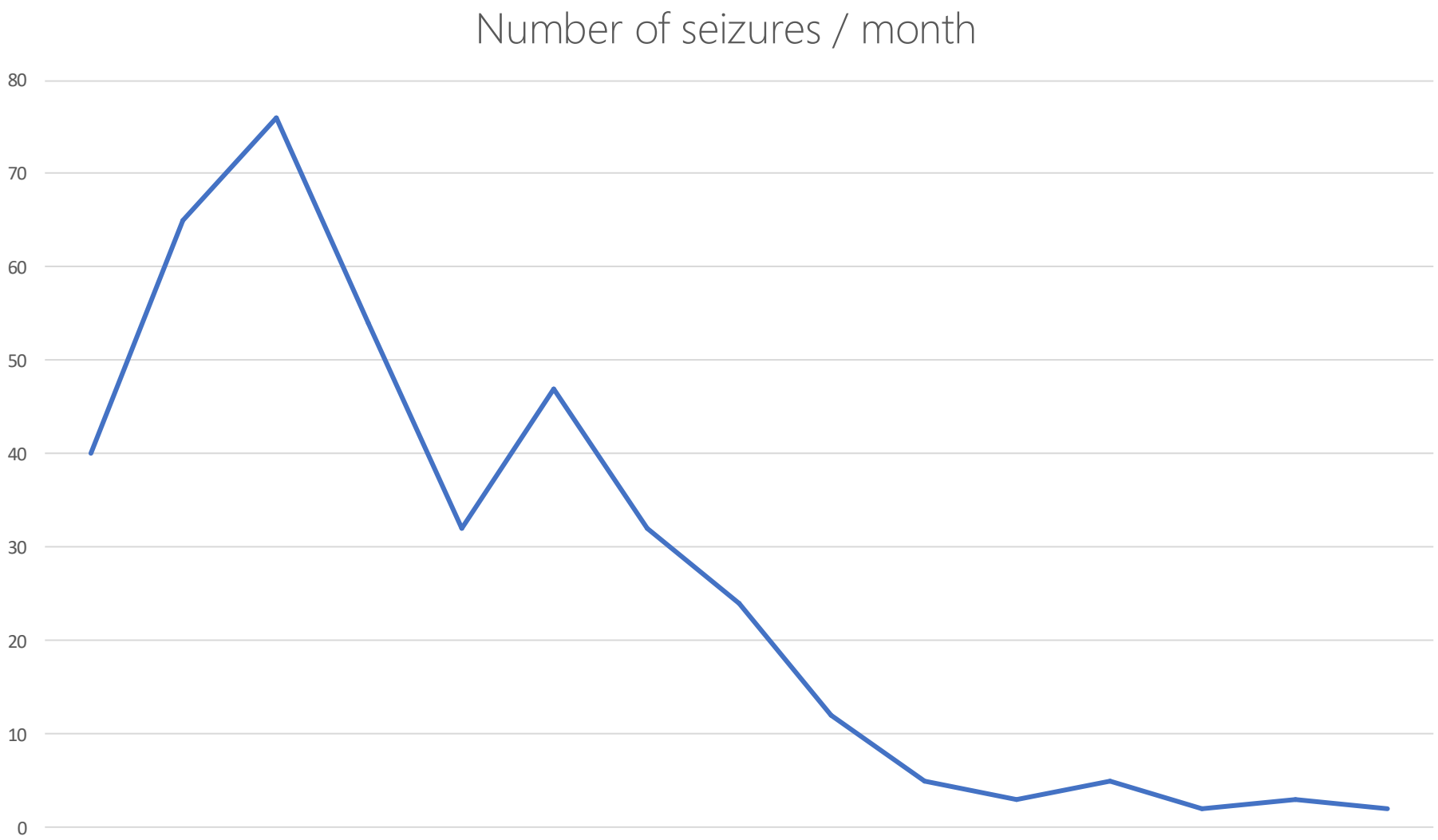
ABSTRACT

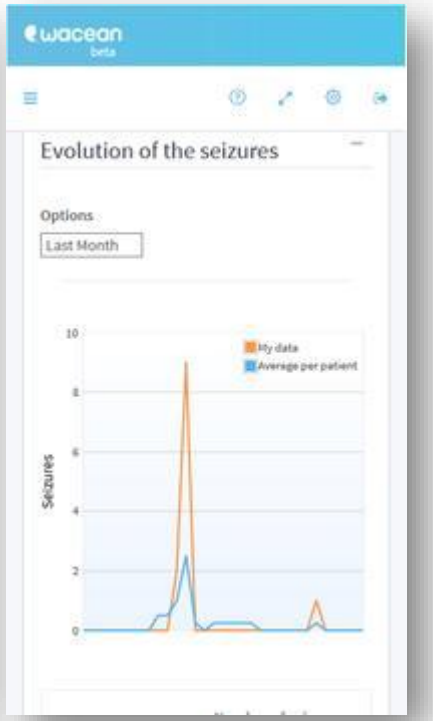
Dravet syndrome is a rare form of epilepsy largely refractory to current antiepileptic medications. The only precedents of randomized placebo-controlled trials in Dravet syndrome are the two small trials that led to the approval of stiripentol. With the arrival of new clinical trials for Dravet syndrome, 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-


Number of seizures / month
80
70
60
50
40
30
20
10
0

~~No money, no honey~~

No data, no honey

Do we have clinical data on our patients?



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

18 May 2016
EMA/327846/2016
Executive Director

Letter of support for Patient Data Platform for capturing patient-reported outcome measures for Dravet syndrome

On 09 December 2015 the applicant Dravet Syndrome Foundation Spain requested qualification opinion for Patient Data Platform as an electronic tool for capturing patient reported outcomes in paediatric epilepsies, pursuant to article 57(1)(n) of regulation (EC) 726/2004 of the European Parliament and of the Council.

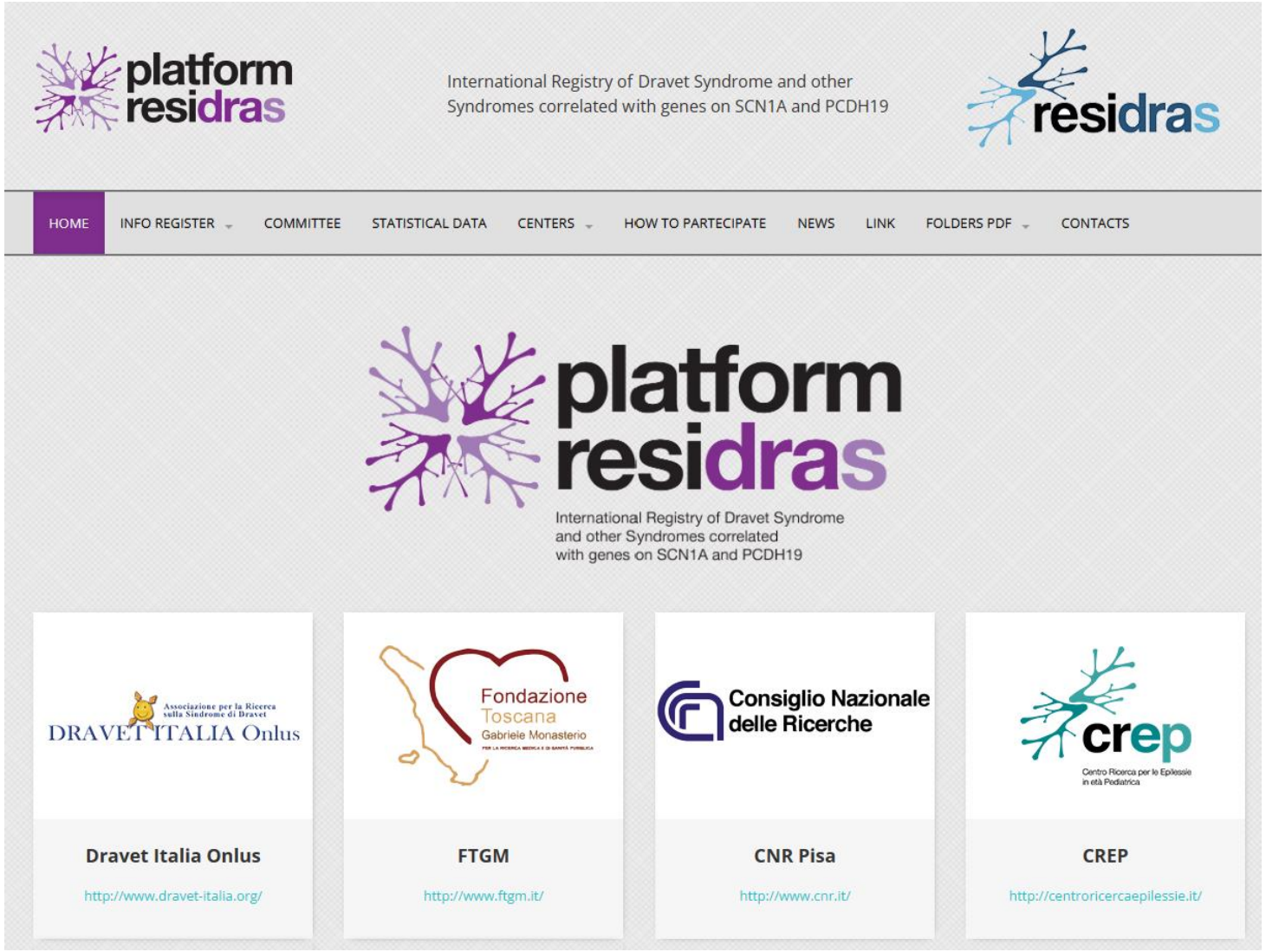
During its meeting held on 11-14 April 2016, the SAWP agreed on the qualification advice to be given to the applicant. During its meeting held on 25-28 April 2016, the CHMP adopted the advice to be given to the Applicant.

The sponsor seeks qualification opinion for their proposed "Patient Data Platform" (PDP) as a patient-reported outcome measure (PROM) to be used within drug development for paediatric epilepsies.

The Patient Data Platform has been designed by a patient organization with patient needs in mind, and is primarily a tool to improve comprehensive patient care by facilitating patient data capture and integration as well as to produce reports and summaries that can be shared with physicians. As such, it is patient-friendly and brings direct benefit to patients and caregivers. We believe that using the Platform for capturing PROs in the context of drug development will not only provide high quality patient-reported data but also reduce the burden on patients and caregivers to complete separate questionnaires or surveys during clinical trials, therefore improving compliance. (http://www.ispor.org/workpaper/patient_reported_outcomes/Coons.pdf)


STOP

A physician-based platform appeared



Who manage this data?

Significant benefit	Clinical data	Benefit/Cost
• Poorly collected by pharma. Very important for orphan drug designation	• Collected by physicians, CRO and pharma. Very controlled and regulated	• Poorly collected by pharma where benefit explanation is needed. Not centralized. Captured in silos with no strategy

Significant benefit

Article 3(1)b of Regulation EC 141/2000 states that in the case where a satisfactory method of diagnosis, prevention or treatment of the condition exists, the sponsor has to establish ‘that the medicinal product will be of significant benefit to those affected by that condition’.

EMA has limited experience getting information from patients and their patients' organizations

In 2021 The problem appeared again during fenfluramine CHMP evaluation for marketing authorization

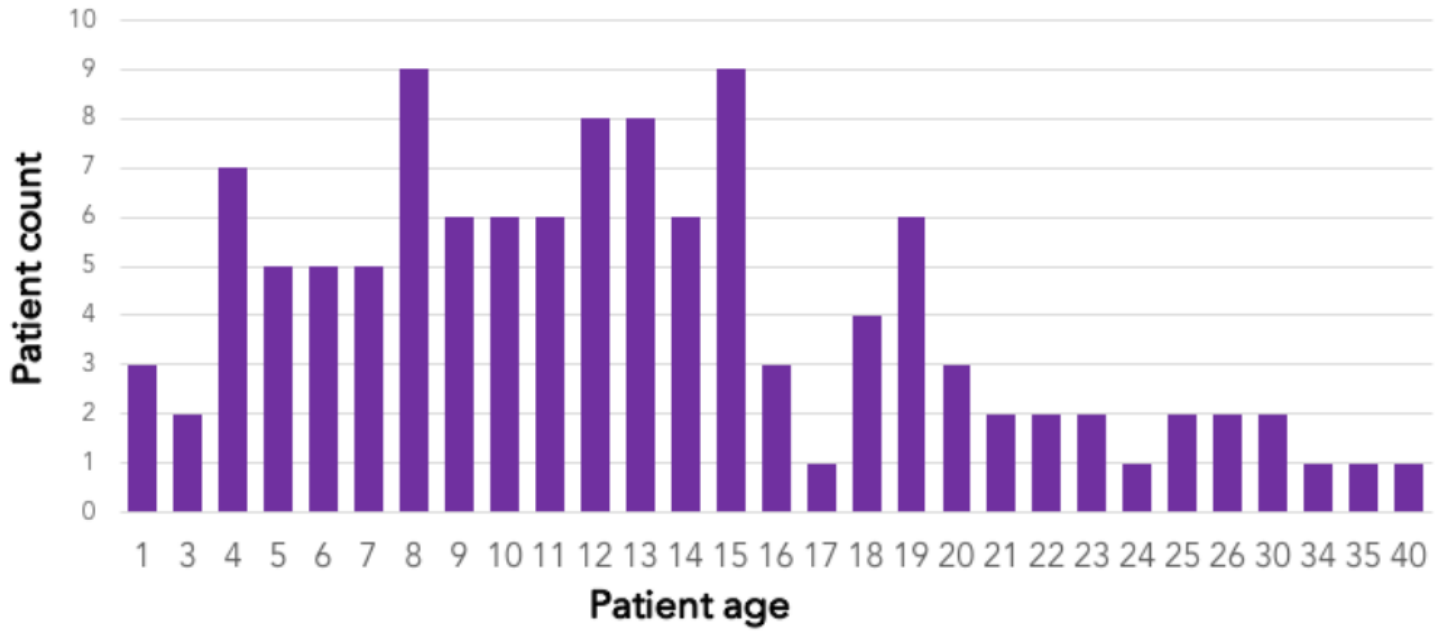

SurveyMonkey

Countries involved

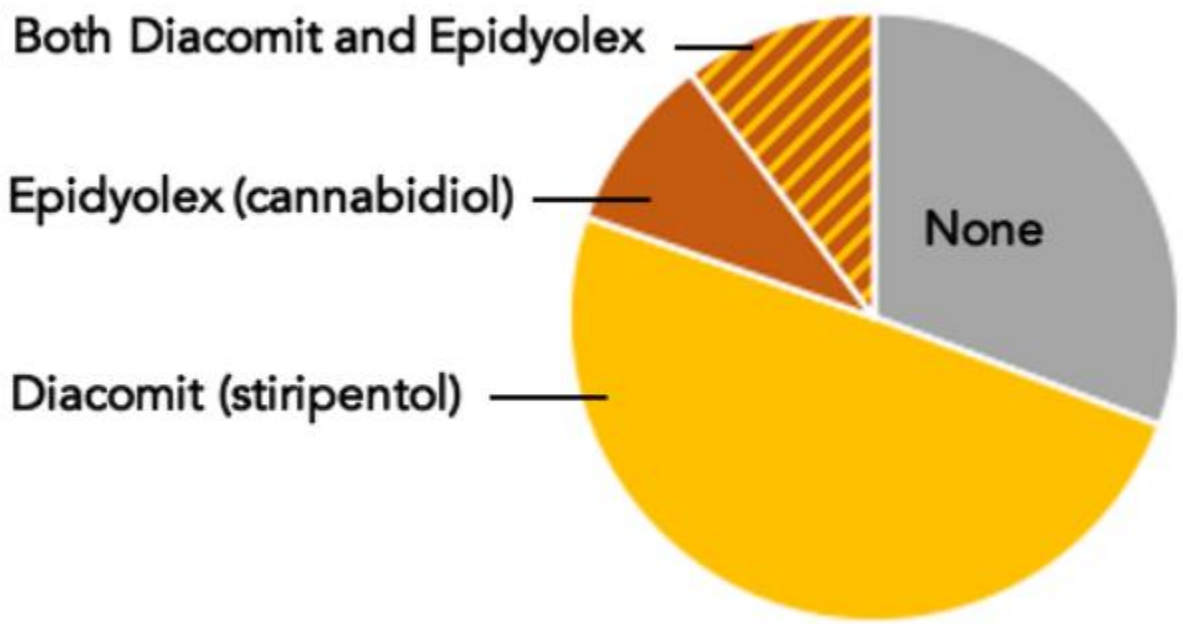
Country	Participants*	Complete responses
Germany	33	29
Italy	31	30
Netherlands	13	12
United Kingdom	12	12
Belgium	11	9
Spain	6	4
Switzerland	6	6
France	4	4
United States	2	1
Total responders	**118**	**107**

(*) Including patients with partial responses.

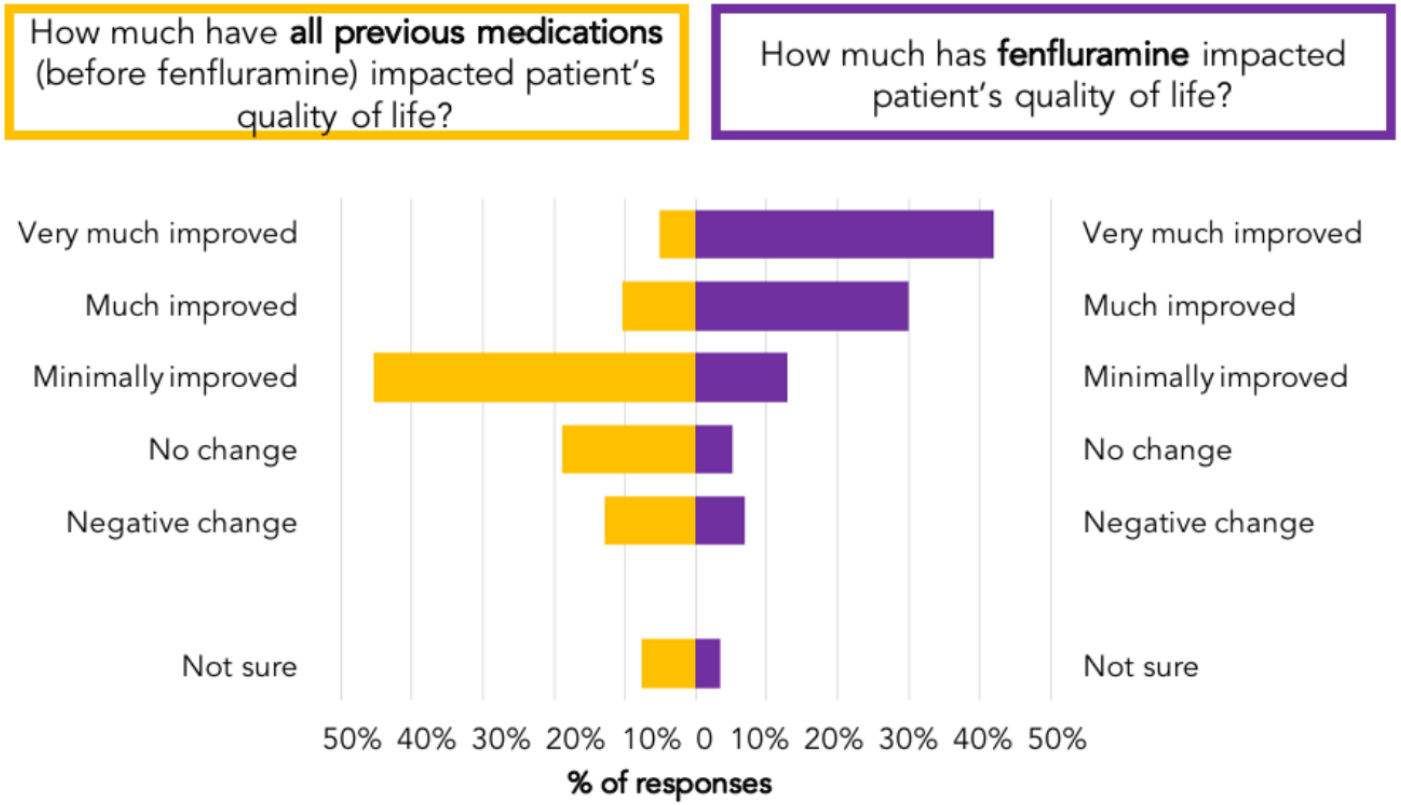
Patients by age



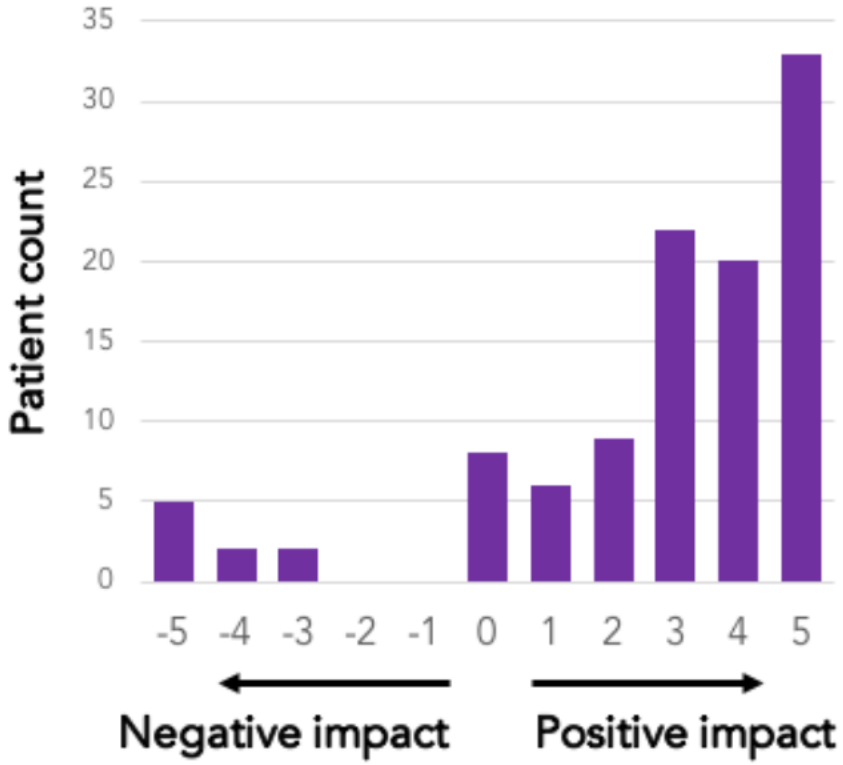
Prior treatment



Impact in patient quality of life



Significant benefit



What about adults?

The importance of collecting data

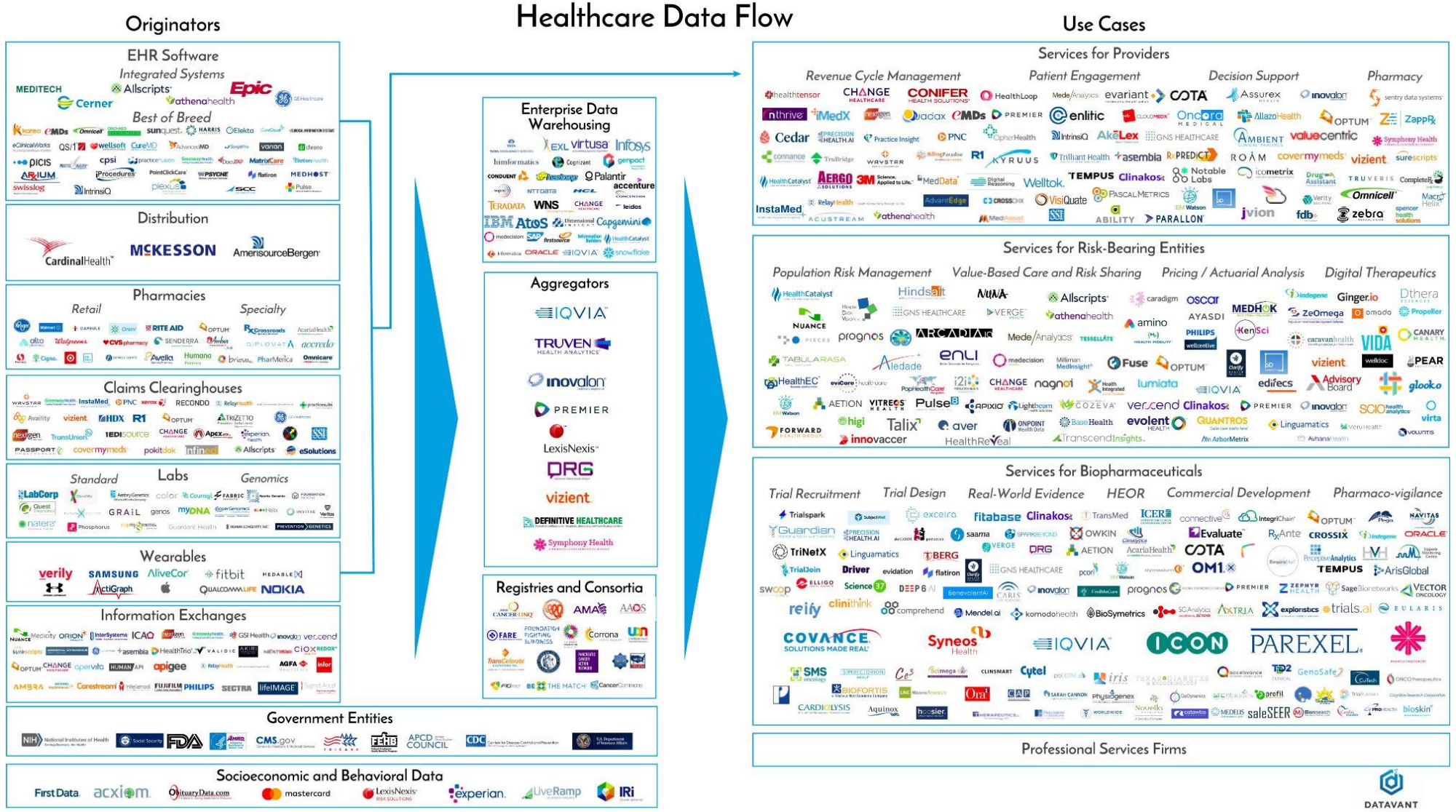
We faced the lack of data problem several timeS

The lack of stable data forced to us to use ad-hoc surveys

Caregivers are starting to suffer “survey fatigue”

The problem is still there

Healthcare data is fragmented

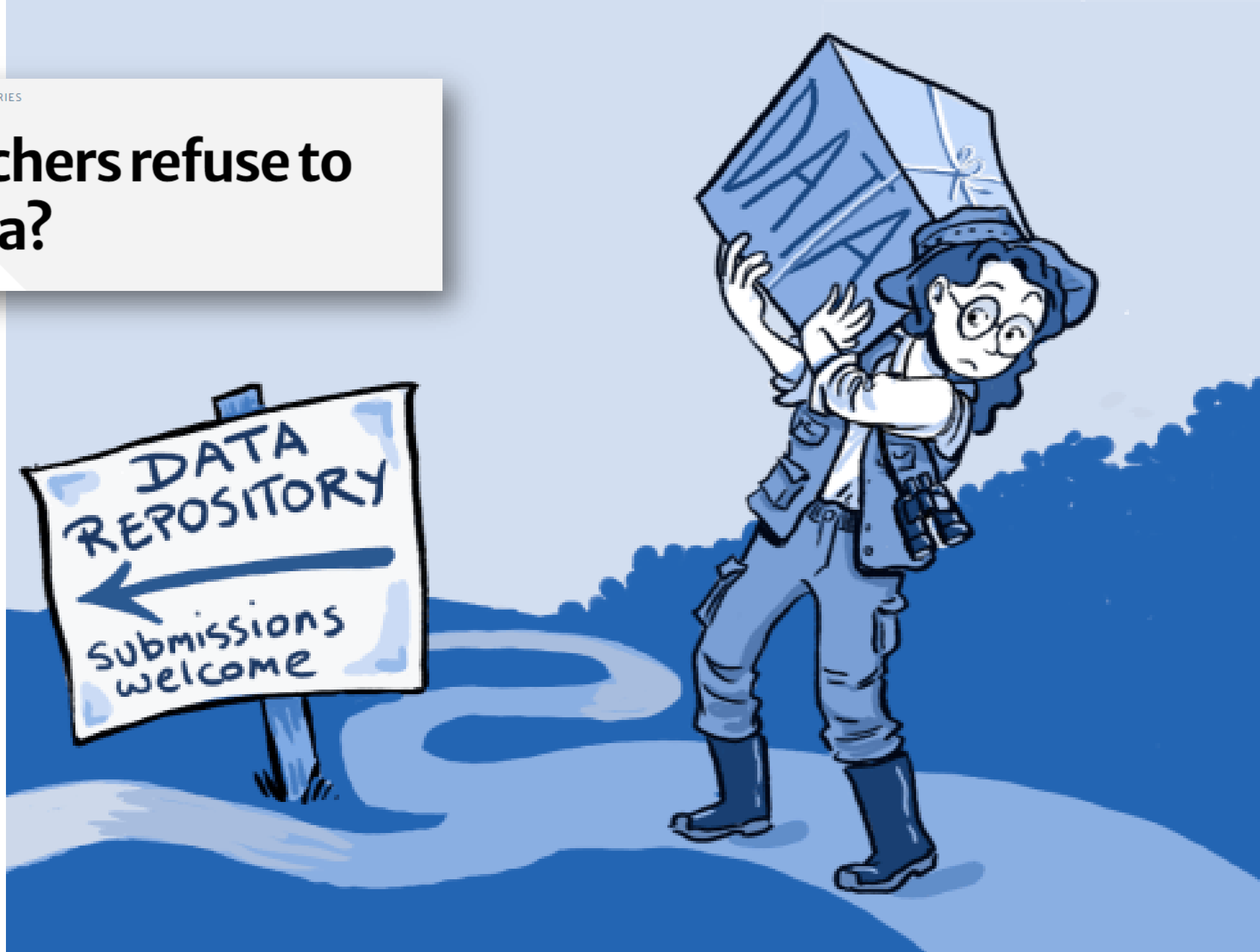


[The Fragmentation of Health Data. A Survey of The Health Data Ecosystem | by Travis May | Datavant | Medium]

POSTED MAY 13, 2016 / 1 COMMENT / CLINICAL TRIAL REGISTRIES

Why do researchers refuse to share their data?

90% don't share their data



[Open Access to Data: An Ideal Professed but Not Practised | Request PDF (researchgate.net)]

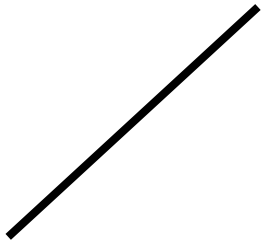
I've been trying to do patient registries for thirteen years

So far I have failed

Causes

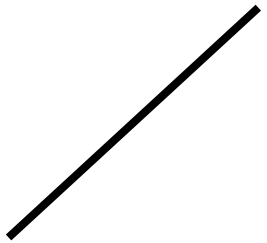
- Conflicts of interest
- Technology gap
- Lack of business model
- Usability

I invest a lot of time



The tool doesn't give me anything

I invest a lot of time



The tool doesn't give me anything

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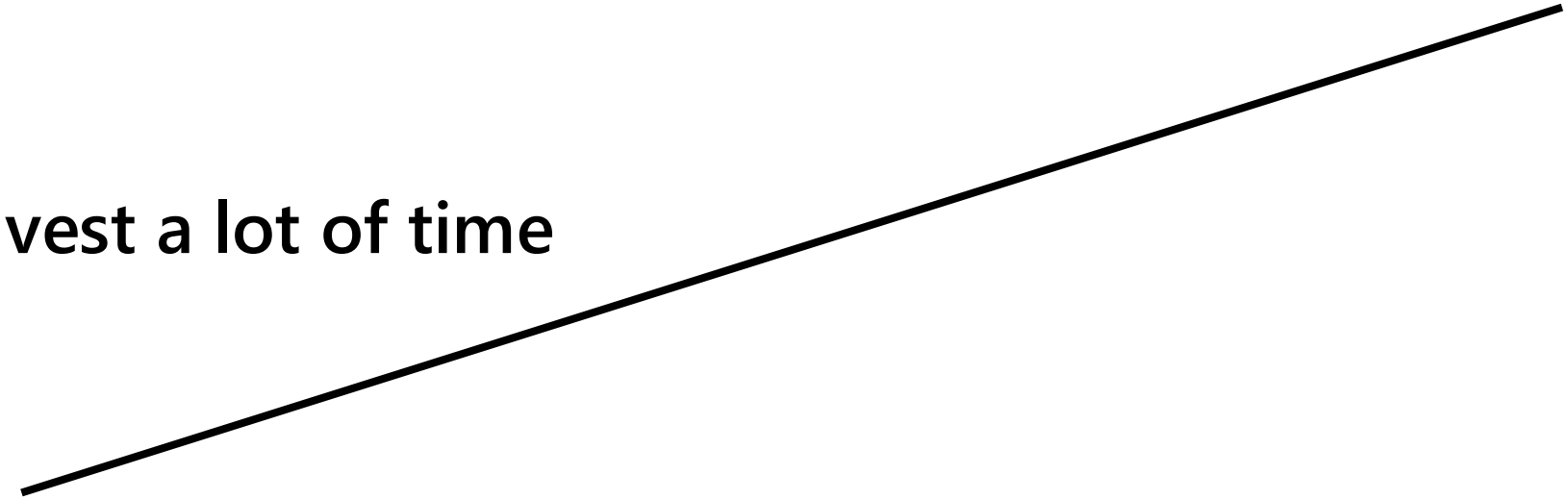
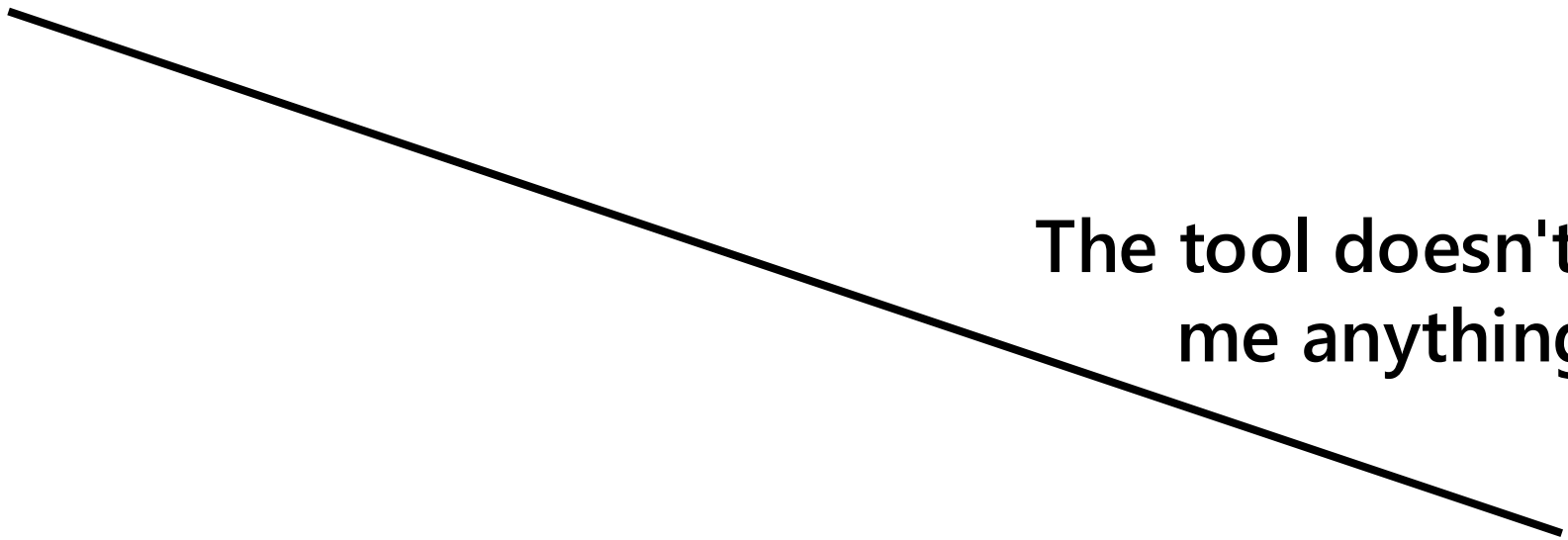
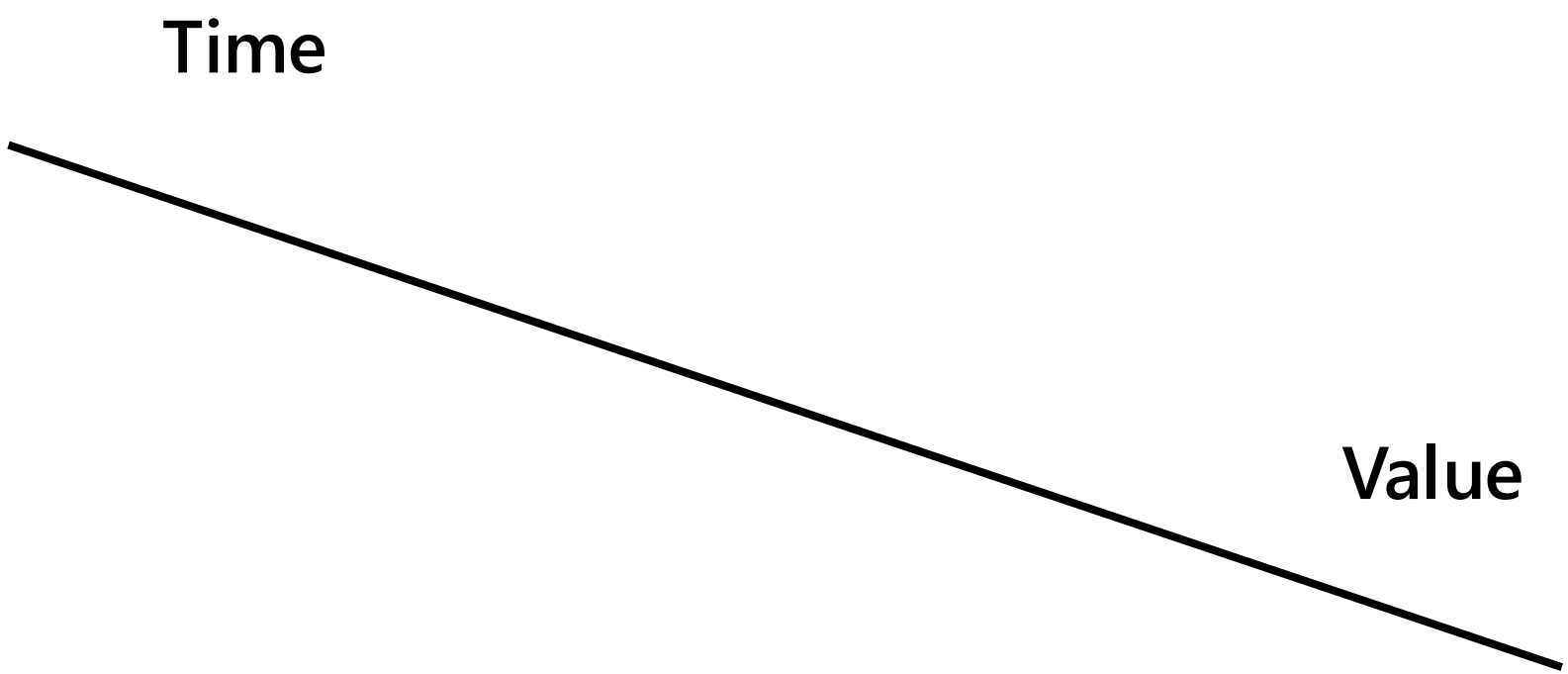


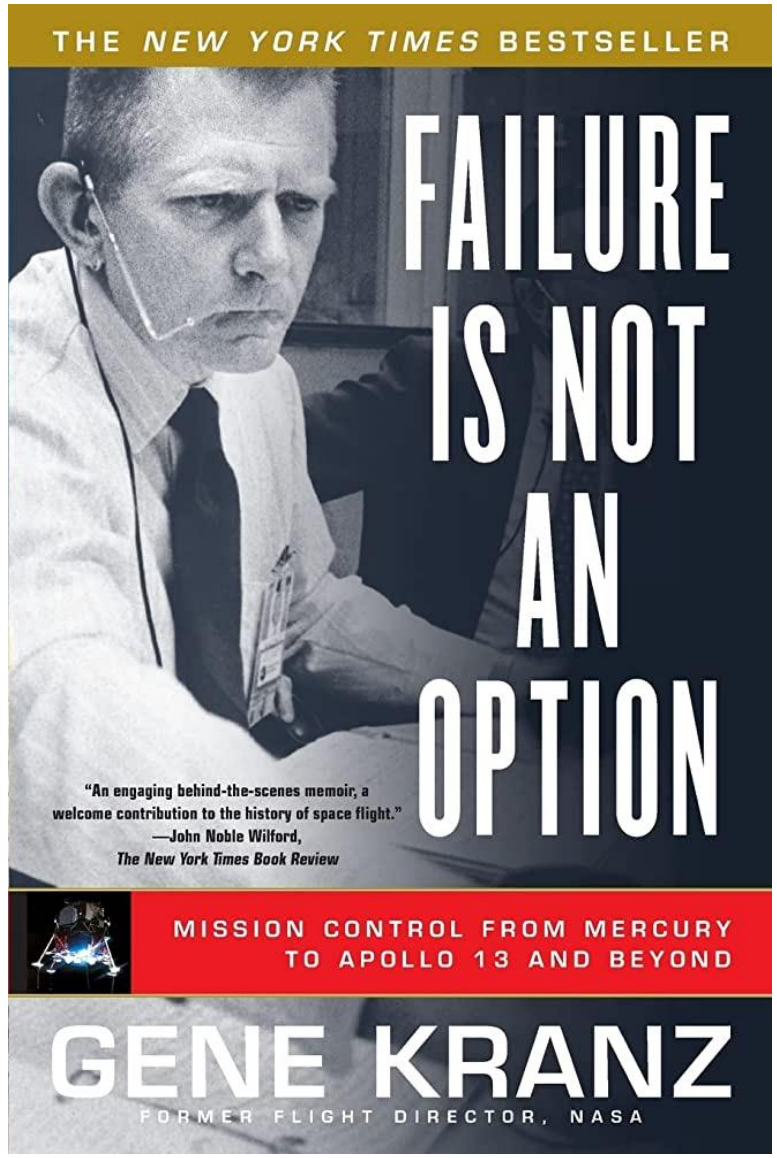
I invest a lot of time

I invest a lot of time



The tool doesn't give me anything


Time
Value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GENE KRANZ
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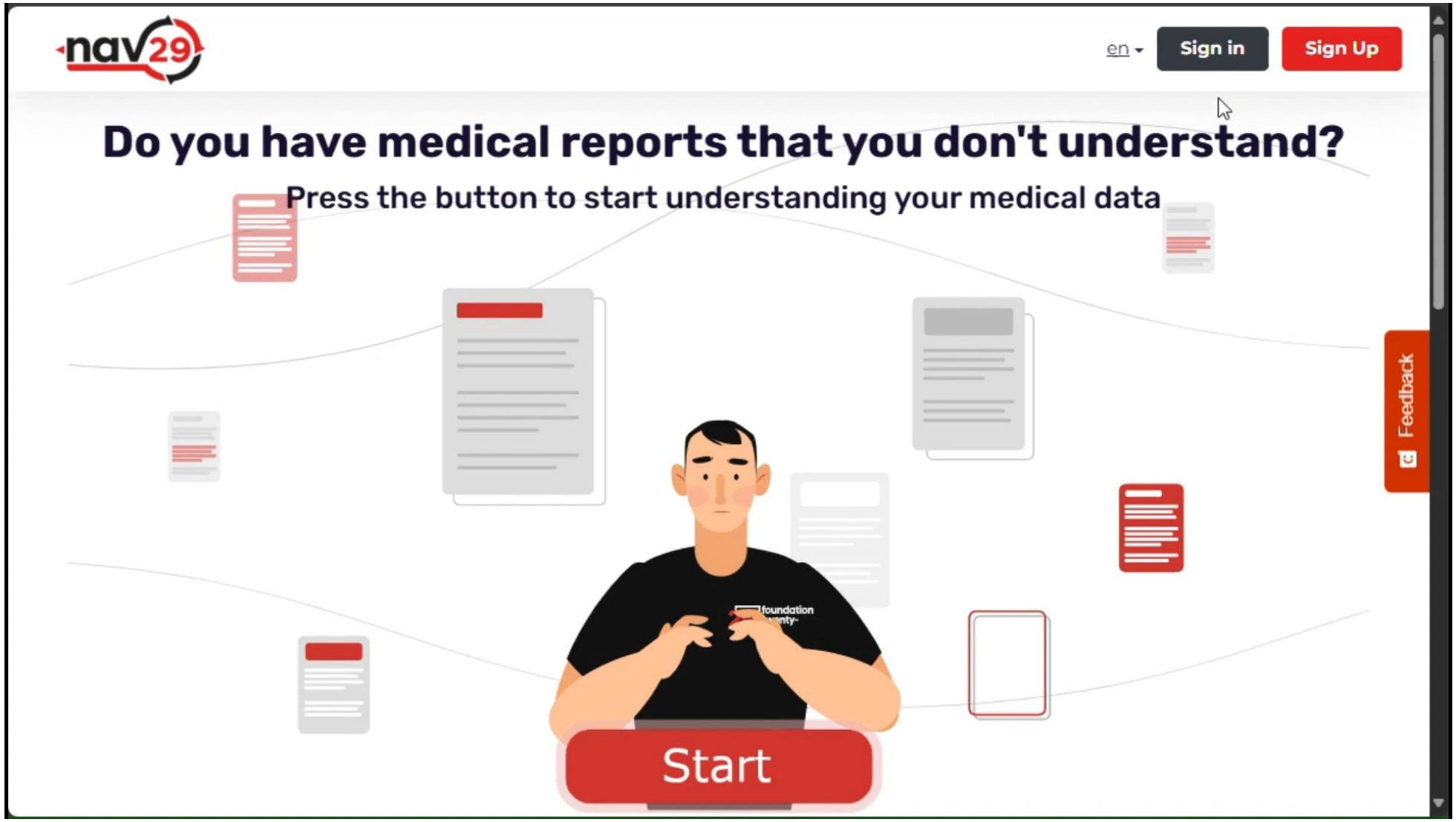

www.nav29.org

Our learnings collecting data

- Patient data is the new gold
- Physicians and researchers don't have a strong motivator to share data
- Physicians don't have time to collect data
- Patients don't have time to collect data
- Everything is about time/value deal
- Competition for data is there
- Shit happens

Ask for your data

Fight for your data

julian.isla@foundation29.org

Thanks

dravet europe
Dravet Syndrome European Federation

29 foundation twenty-nine