

Treating Data as an Asset: Data Entrepreneurship in the Service of Patients

Eric D Perakslis PhD

Rubenstein Fellow, Duke University

Lecturer, Department of Biomedical Informatics, Harvard Medical School

Strategic Advisor on Innovation

Manson Unit

Médecins Sans Frontières

@eperakslis

Data & Digital Transformation: How are patients changing?

They are active, connected, informed and savvy.

Ill Literates or Illiterates? Investigating the eHealth Literacy of Users of Online Health Communities.

[Petrič G¹](#), [Atanasova S¹](#), [Kamin T²](#)

The reliability of patient reported outcomes is still highly variable.

The Impact of Participation in Online Cancer Communities on Patient Reported Outcomes: Systematic Review.

[van Eenbergen MC^{#1}](#), [van de Poll-Franse LV^{1,2}](#), [Heine P³](#), [Mols F⁴](#).

They desire price transparency but do not yet understand how to shop.

Patients' views on price shopping and price transparency.

[Semigran HL](#), [Gourevitch R](#), [Sinaiko AD](#), [Cowling D](#), [Mehrotra A¹](#).

The younger & next generation is very different.

Exploring the digital technology preferences of teenagers and young adults (TYA) with cancer and survivors: a cross-sectional service evaluation questionnaire.

[Abrol E¹](#), [Groszmann M²](#), [Pitman A^{3,4}](#), [Hough R⁵](#), [Taylor RM⁶](#), [Aref-Adib G^{4,7}](#).

Direct-to-consumer R&D is becoming mainstream.

The RUDY study: using digital technologies to enable a research partnership

[Harriet J A Teare^{1,*}](#) et al

The #1 reason patients use pharmacies is information.

<http://vhpharmacyrx.com/compounding/top-5-reasons-people-go-to-the-pharmacy/>

How is product selection and delivery changing?



80 Million Prime Subscribers Ready for Amazon's Pharmacy?
Moyet Moken, Jun. 9, 2017

What will that look like?

Bayer Aspirin Regimen, Low Dose (81 mg), Enteric Coated, 300 Count

[Bayer](#) 4.7 out of 5 stars | [1,170 customer reviews](#), | [67 answered questions](#)

[#1 Best Seller in Aspirin](#)

Top customer reviews

[4.0 out of 5 stars](#) [It's aspirin!](#) By [Serge Mikhaylov](#) on December 9, 2016

[1.0 out of 5 stars](#) [The Bayer product is great, but I received the bottle with the security ...](#) By [SILVIA DE BARRAZA](#) on August 29, 2016







[1.0 out of 5 stars](#) [Short expiration date](#) By [Joyce Koppel](#) on January 25, 2014

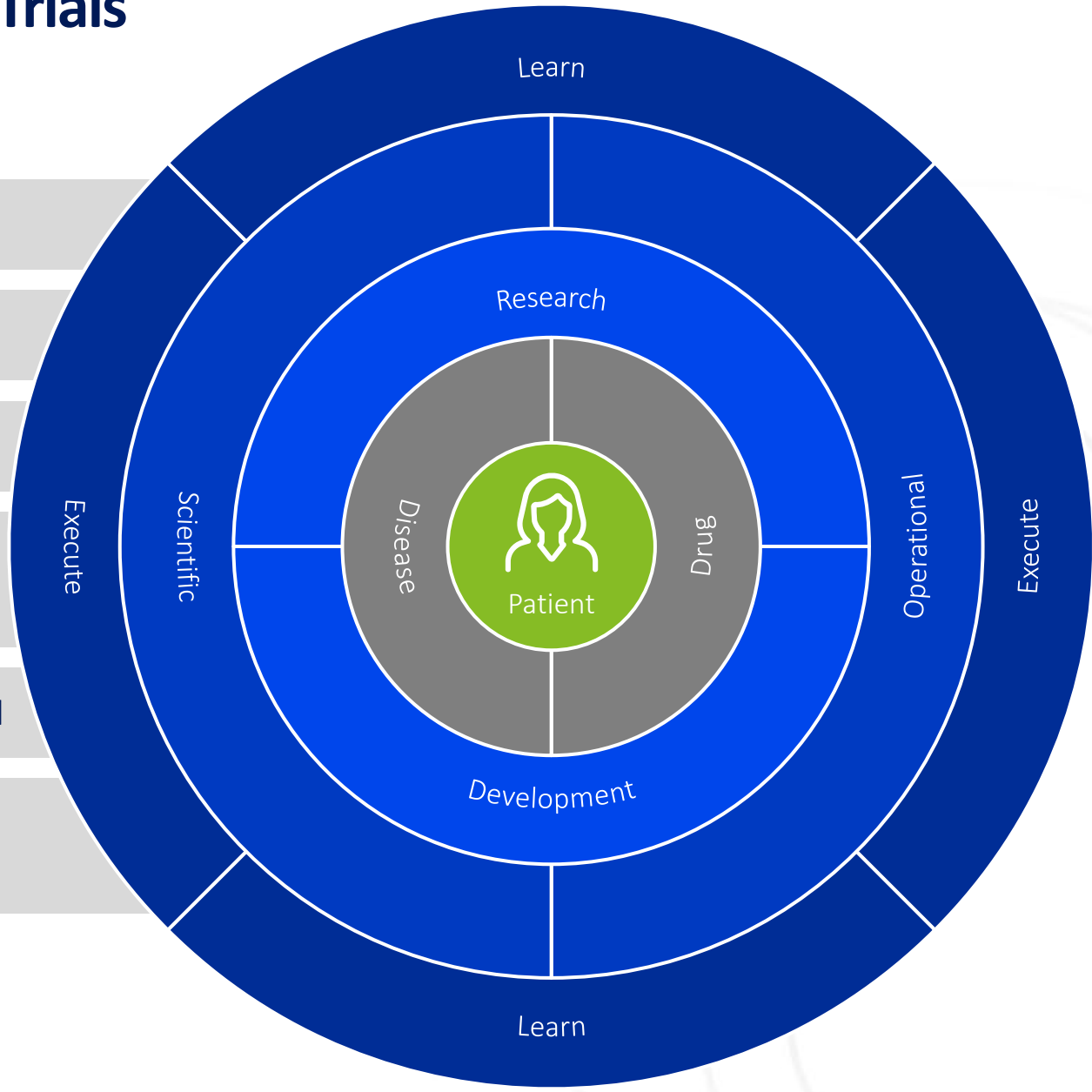


Patients are sharing their data and collaborating enthusiastically in open science and open data

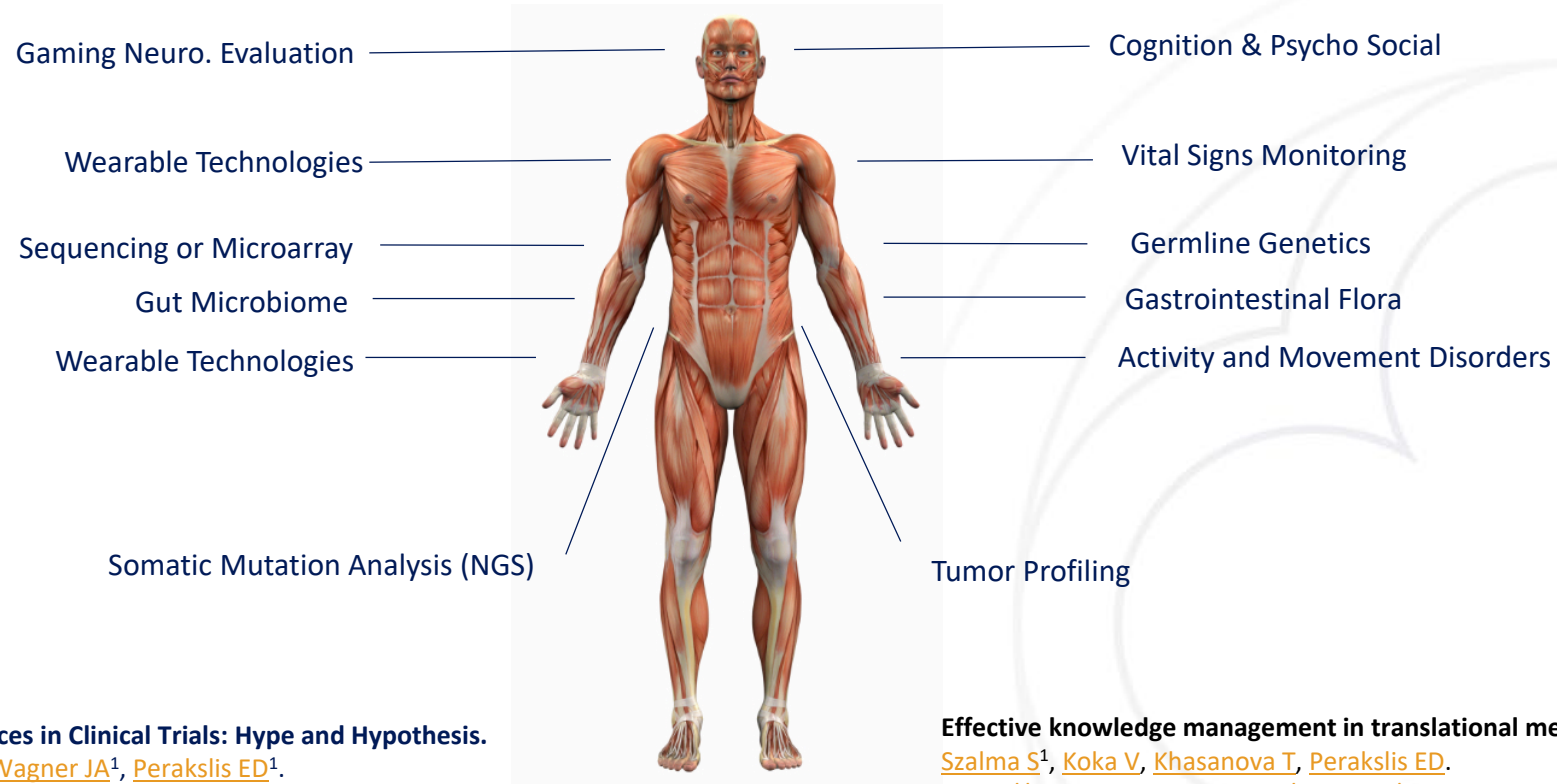
The screenshot shows the Undiagnosed Diseases Network website. At the top left is the logo with the text 'Undiagnosed Diseases Network'. To the right are social media icons for Facebook and Twitter with the text 'Follow Us!'. Below the logo is a navigation menu with links: 'About Us', 'How to Apply', 'Sites', 'Participants', 'Research', 'News', and 'Contact Us'. A search bar is located on the right side of the navigation bar. The main content area features a blue background with a grid of participant photos. A callout box on the right says: 'If any of these participants sound like you or someone you know, please contact us!' with a 'CONTACT US' button. The profile for 'Participant 052' is highlighted, showing a photo of a young girl with glasses. The text next to the photo reads: 'On this page, you will find information about a UDN participant. Sharing information on this website is not a requirement of UDN participation. Only descriptions about participants who give explicit consent will appear here. Female, age 5 with global developmental delay, rhythmic and repetitive movements (stereotypic movements), and difficulty feeding'. Below the profile is a table with two rows: 'Date of Report' (Feb 01, 2018) and 'Description' (Shortly after birth, the patient was noticed to have decreased tone and difficulty breathing. She struggled to drink from a bottle and would breathe liquids into her airway (aspirate). At 6 months, a G tube was placed. Currently she struggles with chewing and swallowing and is unable to drink out of a cup or straw. The patient said her first words at 13 months and began walking at 22 months. She gets tired easily (easy fatigability) and makes rhythmic and repetitive movements with her upper and lower limbs (stereotypic movements).).

Data Transformation of Clinical Trials

-  Predictive Trial Modeling & Simulation
-  Targeted Patient Enrollment
-  Clinical Trial Oversight & Monitoring
-  Data Flow & Learning Health (functional & organizational)
-  Ability to Scale Up and Down to Meet Demand
-  Collaboration with Partners and Industry



Deep human data phenotyping and molecular profiling strategies are becoming ubiquitous



Wearable Devices in Clinical Trials: Hype and Hypothesis.
[Izmailova ES¹](#), [Wagner JA¹](#), [Perakslis ED¹](#).
<https://www.ncbi.nlm.nih.gov/pubmed/29205294>

Effective knowledge management in translational medicine.
[Szalma S¹](#), [Koka V](#), [Khasanova T](#), [Perakslis ED](#).
<https://www.ncbi.nlm.nih.gov/pubmed/20642836>

The only 100% common element of digital transformation across all industries is data

Harvard
Business
Review

What the Companies on the Right Side of the Digital Business Divide Have in Common

[Robert Bock](#), [Marco Iansiti](#), [Karim R. Lakhani](#)... **“differentiate themselves from competitors based on their their data platform.”**

Forbes

The Power Of Digital Transformation In A Data-Driven World

[Peter Bendor-Samuel](#). **“digital transformation journey moves an organization from a process-defined world to a data-driven world”**

information age

Data science needs to be a fundamental component of any digital transformation effort.

[Ben Rossi](#)

HIT
CONSULTANT

Why Data Block Is the Leading Cause of Death for Digital Health Startups.

by Our Thought Leaders


mobihealthnews

Five digital health trends investors are watching in 2017

By [Skip Fleshman](#) February 16, 2017. **“#3 Data Integration and Analytics”**

Duke

The Press Towards Open Health Data Continues

In Stunning Win For Open Science, Johnson & Johnson Decides To Release Its Clinical Trial Data To Researchers

[Matthew Herper, Forbes Staff](#)

I cover science and medicine, and believe this is biology's century.

[PHARMA & HEALTHCARE | 1/30/2014 @ 7:09AM | 17,254 views](#) **Forbes**

The NEW ENGLAND JOURNAL of MEDICINE

Perspective

October 24, 2013

Access to Patient-Level Trial Data — A Boon to Drug Developers

Hans-Georg Eichler, M.D., Frank Pétavy, M.Sc., Francesco Pignatti, M.D., and Guido Rasi, M.D.

The provision of access to clinical trial results that include patient-level data is generating much debate. A growing chorus of transparency advocates is pushing for open access to these data...



Highly Turbulent Regulatory Environment

Text Resize **A A A** Print Share

FOR IMMEDIATE RELEASE
February 11, 2019

Contact: HHS Press Office
202-690-6343
media@hhs.gov

HHS Proposes New Rules to Improve the Interoperability of Electronic Health Information

New innovations in technology promote patient access and could make no-cost health data exchange a reality for millions

The U.S. Department of Health and Human Services (HHS) today proposed new rules to support seamless and secure access, exchange, and use of electronic health information. The rules, issued by the Centers for Medicare & Medicaid Services (CMS) and the Office of the National Coordinator for Health Information Technology (ONC), would increase choice and competition while fostering innovation that promotes patient access to and control over their health information. The proposed ONC rule would require that patient electronic access to this electronic health information (EHI) be made available at no cost.

NBCNEWS.com

Is privacy possible in the digital age?

If it isn't dead, then it's hanging on by a thread

Jump to discuss
Loading comments...

Recommend 0
 Tweet
 Share 220

Below: Discuss

FORTUNE In the Wake of GDPR, Will the U.S. Embrace Data Privacy?

privacy rights of Europeans can be upheld in the context of the age of "big data."



Fury awaited Facebook CEO Mark Zuckerberg ahead of his May visit to Brussels. Dario Pigantelli—

Opinion

The Privacy Project

Latest

April 28, 2019 **Losing Our Fourth Amendment Data Protection** 
The courts have shielded information when we have a "reasonable expectation" it will stay private. What happens when we stop believing?
By Josephine Wolff

April 27, 2019 **Your Privacy Is Our Business** 
Let us reassure you: You're worried only because you don't understand anything about anything.
By Jessica Powell

THE LANCET Digital Health

COMMENT | [VOLUME 1, ISSUE 1, PE8-E9, MAY 01, 2019](#)

Is health-care data the new blood?

[Eric Perakslis](#) ✉ • [Andrea Coravos](#)

[Open Access](#) • Published: May, 2019 • DOI: [https://doi.org/10.1016/S2589-7500\(19\)30001-9](https://doi.org/10.1016/S2589-7500(19)30001-9)

THE LANCET Digital Health



Editorial

A digital (r)evolution: introducing
The Lancet Digital Health
See page e1

Comment

The challenges of cybersecurity in health
care
See page e10

Articles

An artificial intelligence approach for
diabetic retinopathy screening in Zambia
See page e35

Most Data about our Patients, and Us, are Not in our Medical Records

Under-Used Data



*Bibliometric/
Scientometrics*



Social Media



*Competitive
Intelligence*



*Consumer
Behavior*



*Siloed
Data*

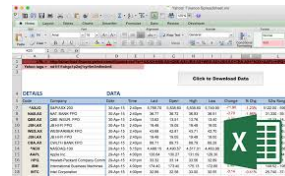
Over-Used Data



Single Papers



Single Trials



Under Translated Data



*Genetics/
Genomics*



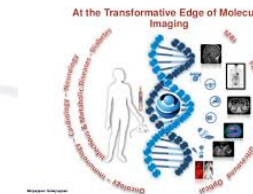
Phenotyping



Digital Sensors



Geospatial

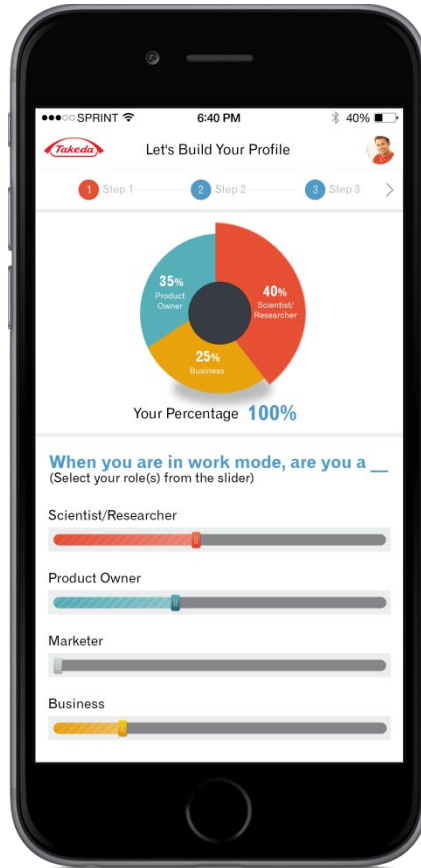


Molecular Imaging



Anecdotal Learning

Data Architecture and Knowledge Management

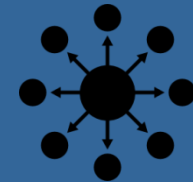


Architecture:

- Simplified technology stack
- Enterprise R&D Data
- Cloud/SaaS based
- Scalable demand based model
- Built for purpose solutions
- Search, NLP, Machine Learning



Data and Analytics



Collaboration



Workflow Automation and Integration



Compliance

Standards and Governance:

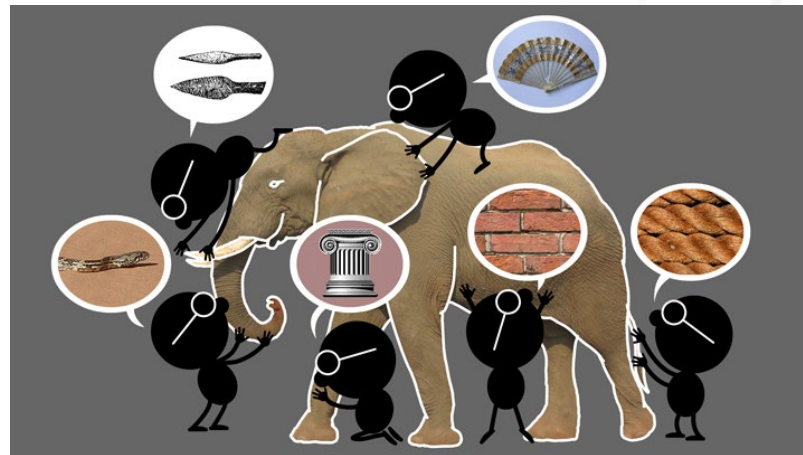
- Leverage industry data standards and formats for internal use and exchange with partners
- Ontology for data concepts and metadata
- Master data management
- Single source of truth
- Role-based access

Delivery:

- Agile Methodology
- Technology and skill aligned partners
- Common infrastructure and platforms
- Road map and prioritized portfolio
- Security and Compliance built-in
- User experience



Data Governance

1. What roles and committees are there for implementing data governance?
2. How is data stored and maintained, and what constitutes the truth in the organization?
3. How to guarantee data is accurate and suitable for analysis? Who owns the data?
4. How do users get access to the data they need & no more than that, & how do they use it in modelling & reporting?
5. How to guarantee data remains private, secure and compliant with regulations?



Eric D Perakslis: It's time to address the medical benefits and risks of the internet of things

January 29, 2018

It's time for us to seriously consider which patient care and monitoring devices should be connected to the internet  



In the first *Jurassic Park* movie, Jeff Goldblum's character made this profound (for Hollywood) statement about humans manufacturing dinosaurs: "You were so busy proving you could do something that you never stopped to consider if you should." There's a lesson here for today's technology innovators, and it is time for us all to seriously consider which patient care and monitoring devices should be connected to the internet.

We have no clear or common frameworks for cyber threats in healthcare. Yet with US hospitals currently having on average 10-15 connected devices per bed, we desperately need them.

Cyber Time in 2017

Technology Time	<p><i>7 years or older: age of systems for 61% of daily MS security updates</i></p> <p><i>5 years or older: age of 69.82% of current MS Operating instances</i></p> <p><i>100-120 days: average lag time from receipt-to-installation of a security patch</i></p> <p><i>Daily: an average of 15 security patches are received</i></p> <p><i>Daily: 44% of security operations managers see at least 5000 security updates</i></p>
Cyber-Threat Time	<p><i>8 months-3 years: duration of average zero-day attack</i></p> <p><i>310 days: how long cybercriminals know about a hacker-discovered-vulnerability before the public or vendor</i></p> <p><i>Weekly: 1-2 zero-day vulnerabilities are exploited</i></p> <p><i>2-3 days: a new Adobe vulnerability is discovered</i></p> <p><i>Daily: 200,000 new malware samples are captured</i></p> <p><i>Daily: 4000 new ransom ware attacks</i></p> <p><i>Minute: 400 new threats (of all types) are launched</i></p>
Regulatory & Standards Time	<p><i>3.8 years: average time between federal cyber law updates since HIPAA (1996)</i></p> <p><i>2 years: time between significant updates to NIST cyber /guides standards</i></p> <p><i>1 year: time from commission-to-report of the HHS Healthcare Cyber Task Force</i></p>

Healthcare IT News

Cloud computing reduces HIPAA compliance risk in managing genomic data

September 4, 2013

“...a review of data on HIPAA breaches published by the US Department of Health and Human Services (HHS) shows that these concerns are Misplaced.”

In fact, by using a cloud-based service with an appropriate security and compliance infrastructure, an organization can significantly reduce its compliance risk...”

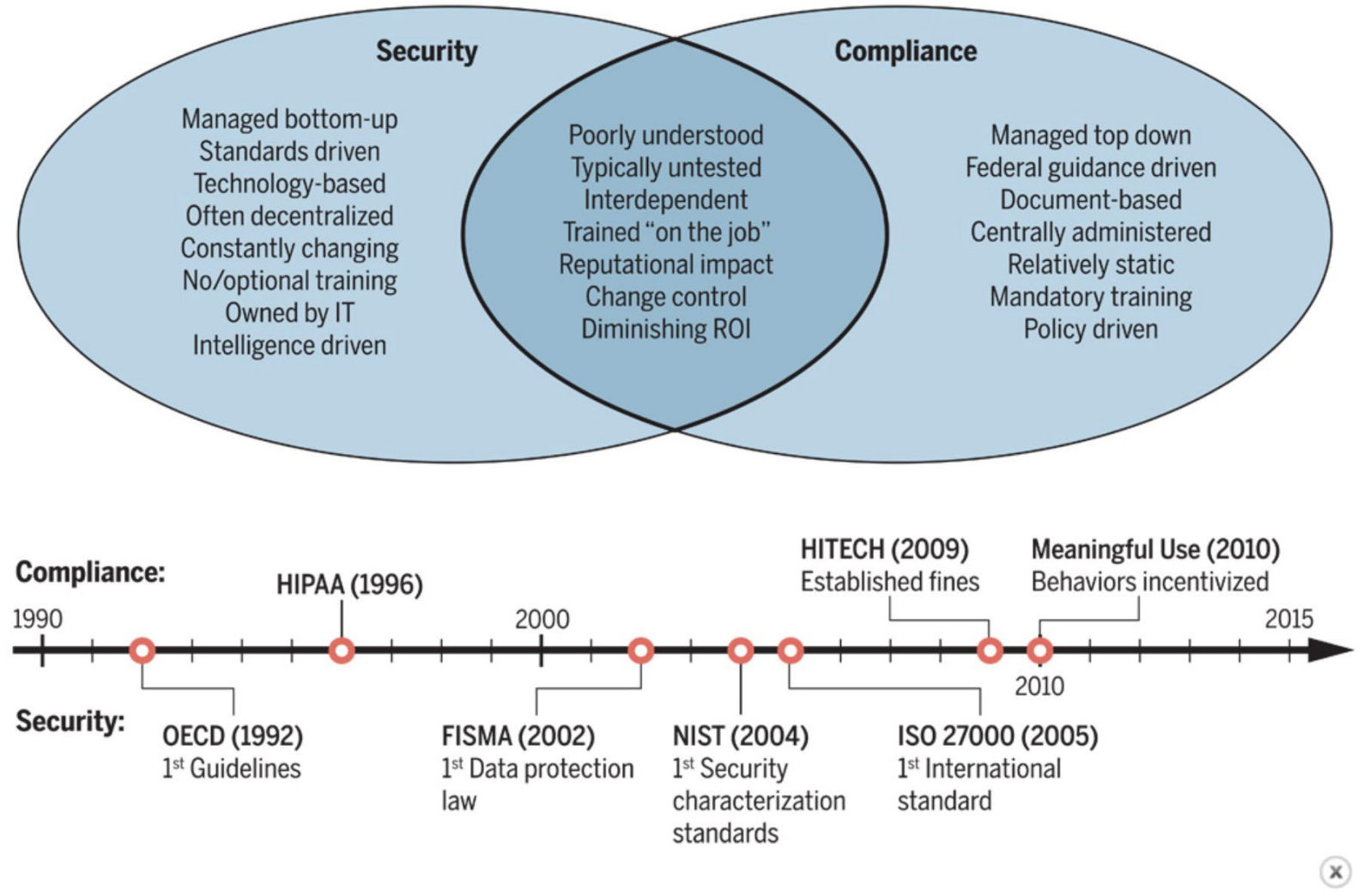


[Sci Transl Med.](#) 2016 Jan 20;8(322):322ps2. doi: 10.1126/scitranslmed.aaa4493.

A cybersecurity primer for translational research.

[Perakslis ED](#)¹, [Stanley M](#)².

Compliance and security: History, gaps and overlaps



Data are Assets

Technology is an Expense

Treating Data as an Asset: Data Entrepreneurship in the Service of Patients

1. Break down the macro and micro silos of healthcare data
2. Keep patients safer
3. Crowdfund the most difficult problems
4. Aggregate, align and donate to friends and freaks
5. Catch a thief
6. Out-shout medical misinformation
7. Shrink space and time to improve research access
8. Nourish your family but read the label
9. Disable the surveillance state and stand up for patient/consumer rights
10. Discover new math

Entrepreneurial Idea #1: Break Down the Macro and Micro Silos of Healthcare

We characterize **macro silos** as data technology platforms and/or knowledge structures that are completely non-overlapping or intentionally isolated and/or competitive and de-incentivized by design. Examples:

- IOS v. Android
- Medical practice and biomedical product development
- Healthcare and non-healthcare

We characterize **micro silos** as data that can be utilized across fragmented but somewhat similar and Overlapping constructs. Examples:

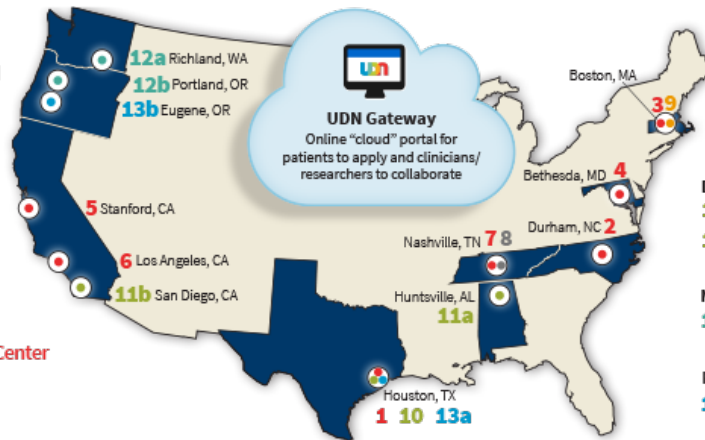
- Local MRI center that is not connected to major local hospitals
- Clinical trials data from different sponsors on similar drugs
- Competitive software sub-ecosystems like clinical trials management and HER
- Independent grants and clinical studies

Example – PCRF Linking Initiative

UDN Undiagnosed Diseases Network

Seven clinical sites

- 1 Baylor College of Medicine and Texas Children's Hospital
- 2 Duke Medicine with Columbia University Medical Center
- 3 Harvard Teaching Hospitals (BCH, BWH, MGH)
- 4 National Institutes of Health
- 5 Stanford Medicine
- 6 UCLA School of Medicine
- 7 Vanderbilt University Medical Center



Six additional research sites

- Central Biorepository
- 8 Vanderbilt University Medical Center
- Coordinating Center
- 9 Harvard Medical School
- DNA Sequencing Core Facilities
- 10 Baylor College of Medicine
- 11 a HudsonAlpha Institute for Biotechnology with b Illumina
- Metabolomics Core Facility
- 12 a Pacific Northwest National Laboratories with b Oregon Health & Science University
- Model Organisms Screening Center
- 13 a Baylor College of Medicine with b University of Oregon

*Applications

Applications received: 2346
Applications under review: 306
Participants accepted: 1030

Evaluations

Participants evaluated: 735
Participants diagnosed: 222
Participants with exome or genome sequencing complete: 682
Participants with metabolomic analyses complete: 113
Participants with variants studied by the Model Organisms Screening Center: 96
Participants with samples in the Central Biorepository: 516

Data Sharing

Manuscripts published: 27
Participants with data submitted to [dbGaP](#): 150
Participants with data submitted to [PhenomeCentral](#): 491
Participants with variants in [ClinVar](#): 28*

Last updated: April 23, 2018

*Note: only proband data is reported here


<https://undiagnosed.hms.harvard.edu/>

E2: Keep Patients Safer

World Health Organization

Health Topics ▾ Countries ▾ News ▾ Emergencies ▾

10 / 10



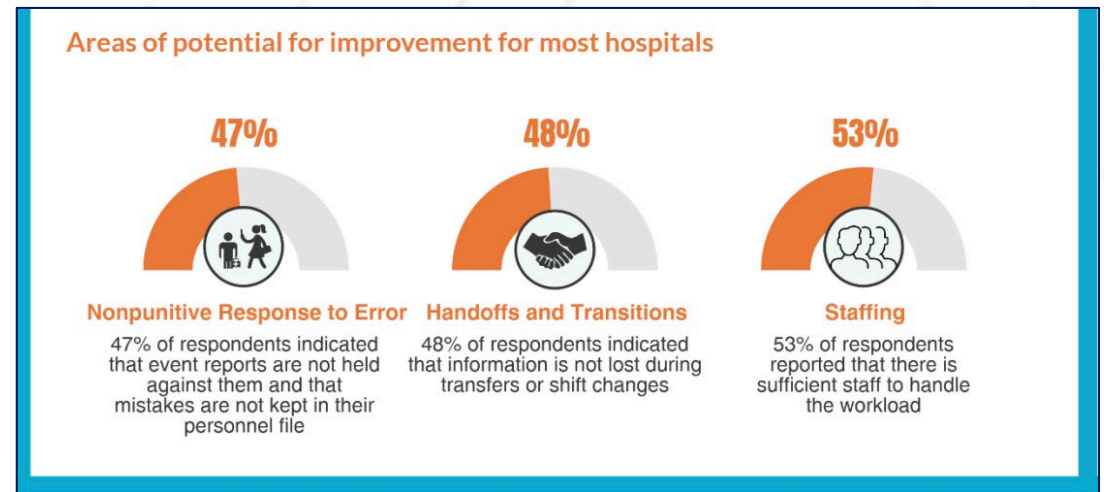
WHO/H. Ruiz

Fact 10: Administrative errors account for up to half of all medical errors in primary care

Recent literature reviews have revealed that medical errors in primary care occur between 5 and 80 times per 100 000 consultations. Administrative errors - those associated with the systems and processes of delivering care - are the most frequently reported type of errors in primary care. It is estimated that from 5 to 50% of all medical errors in primary care are administrative errors.

https://www.who.int/features/factfiles/patient_safety/en/

Hospital Survey on Patient Safety Culture: 2018 User Database Report Prepared for: Agency for Healthcare Research and Quality U.S. Department of Health and Human Services 5600 Fishers Lane Rockville, MD 20857



<https://www.ahrq.gov/sites/default/files/wysiwyg/sops/quality-patient-safety/patientsafetyculture/2018hospitalreport.pdf>

E3: Crowdfund the Most Difficult Problems

The screenshot shows the Undiagnosed Diseases Network website. At the top left is the logo "UDN Undiagnosed Diseases Network". To the right are social media icons for Facebook and Twitter with the text "Follow Us!". Below the logo is a navigation menu with links: "About Us", "How to Apply", "Sites", "Participants", "Research", "News", and "Contact Us". A search bar is located on the right side of the navigation menu.

The main content area features a profile for "Participant 052". On the left is a photo of a young girl with dark hair and pink glasses. To the right of the photo is text: "On this page, you will find information about a UDN participant." followed by "Sharing information on this website is not a requirement of UDN participation. Only descriptions about participants who give explicit consent will appear here." Below this is a description: "Female, age 5 with global developmental delay, rhythmic and repetitive movements (stereotypic movements), and difficulty feeding".

On the right side of the main content area, there is a yellow and orange call-to-action box with the text: "If any of these participants sound like you or someone you know, please contact us!" and a blue button labeled "CONTACT US".

Below the main content area is a table with two rows:

Date of Report	Feb 01, 2018
Description	<p>Shortly after birth, the patient was noticed to have decreased tone and difficulty breathing. She struggled to drink from a bottle and would breathe liquids into her airway (aspirate). At 6 months, a G tube was placed. Currently she struggles with chewing and swallowing and is unable to drink out of a cup or straw.</p> <p>The patient said her first words at 13 months and began walking at 22 months. She gets tired easily (easy fatigability) and makes rhythmic and repetitive movements with her upper and lower limbs (stereotypic movements).</p>

E4: Aggregate, Align and Donate to Friends & Freaks

MMRF CoMMpass Study Data Featured at AACR 2019

The 2019 American Association for Cancer Research (AACR) meeting took place March 29 -April 3 in Atlanta. This gathering of over 20,000 cancer clinicians and researchers featured four presentations made possible by researchers using data from our CoMMpass Study, which is the largest genomic dataset in all of cancer. CoMMpass continues to be an invaluable resource for investigators to use to test new hypotheses and accelerate precision medicine treatments into the clinic for all myeloma patients. Below are summaries of the four CoMMpass Study presentations:

Analysis of CoMMpass genomic data has revealed 12 subtypes of myeloma. Myeloma also occurs more frequently in males and in African Americans. A group from the Translational Genomics Research Institute (TGen) analyzed the data from 607 CoMMpass patients to look at the contribution of variations in normal DNA sequences in this increase in incidence. They found

Cystic Fibrosis Foundation

Home / Research / Researcher Resources / Patient Registry /

Care Center Data

The CF Foundation's Patient Registry tracks the health outcomes of people with cystic fibrosis who receive care at Foundation-accredited care centers and makes this information easily available to the CF community.

Share [f](#) [t](#) [e](#) [r](#) | Print [p](#)

Freakonomics

Book by Stephen J. Dubner and Steven Levitt

Book preview
51/393 pages available

PREVIEW

4/5 · [Goodreads](#)

90% liked this book
Google users

Freakonomics: A Rogue Economist Explores the Hidden Side of Everything is the debut non-fiction book by University of Chicago economist Steven Levitt and New York Times journalist Stephen J. Dubner. It was published on April 12, 2005, by William Morrow. [Wikipedia](#)

E5: Catch a Thief



Security Center > IoT > What is a honeypot? How it can lure cyberattackers

What is a honeypot? How it can lure cyberattackers

f t in

Authored by a Symantec employee

A **honeypot** is a computer or computer system intended to mimic likely targets of cyberattacks. It can be used to detect attacks or deflect them from a legitimate target. It can also be used to gain information about how cybercriminals operate.

Like mice to cheese-baited mousetraps, cybercriminals are attracted to honeypots — not because they're honeypots. The bad guys think the honeypot is a legitimate target, something worthy of their time. That's because the bait includes applications and data that simulate a real computer system.

E6: Out-Shout Medical Misinformation

Fri, May 10, 2019

Newsweek

U.S. | World | Business | Tech & Science | Culture | Sports | Health

OPINION

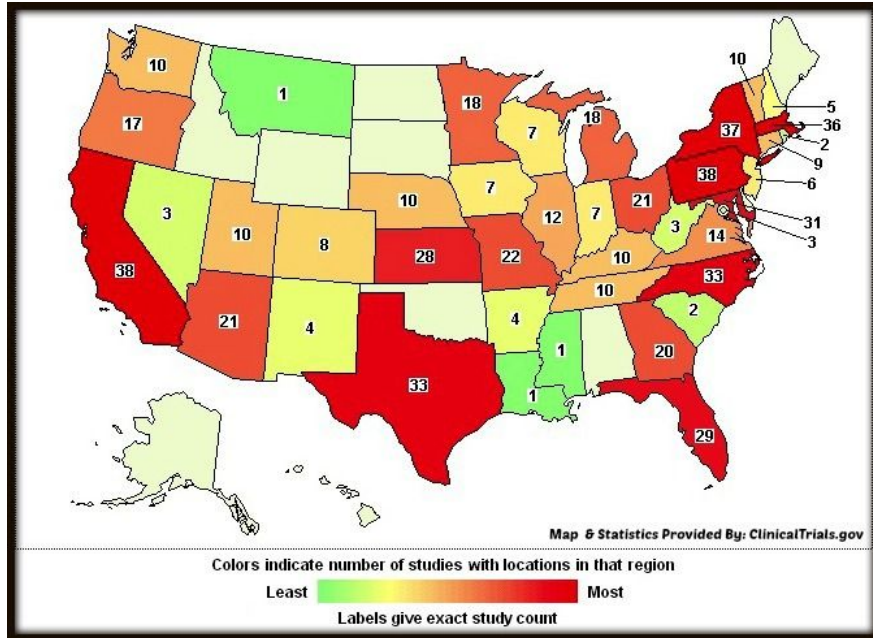
AS MEASLES CASES CLIMB, OUR MISSION IS CLEAR: TAKE DOWN THE THREE-HEADED ANTI-VAX MONSTER | OPINION

DR. PETER HOTEZ
ON 5/9/19 AT 12:32 PM EDT

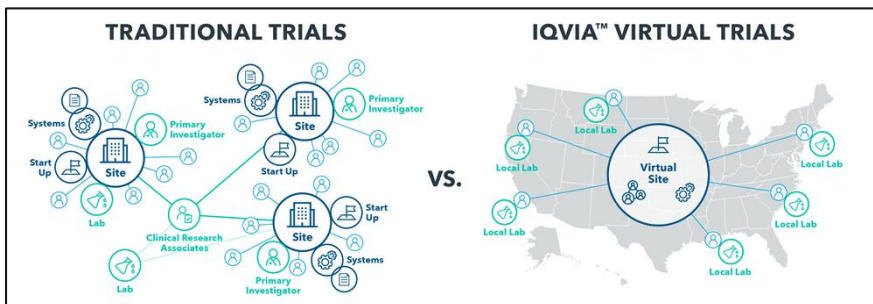


- Positive Imagery
- Education
- Political Action
- Big Tech Activism
- Social Voice
- Cyber Warfare Techniques against nation-state actors
- ...

E7: Shrink Space and Time to Improve Research Access



- Minimize the Regulatory Regime Chasm between Medical Practice and Biomedical Products R&D
- Disrupt the Clinical Trials Industrial Complex
- Virtual teleporter from work or the soccer field to the clinical trial and back.



E8: Nourish Your Family but Read the Label

Medicine

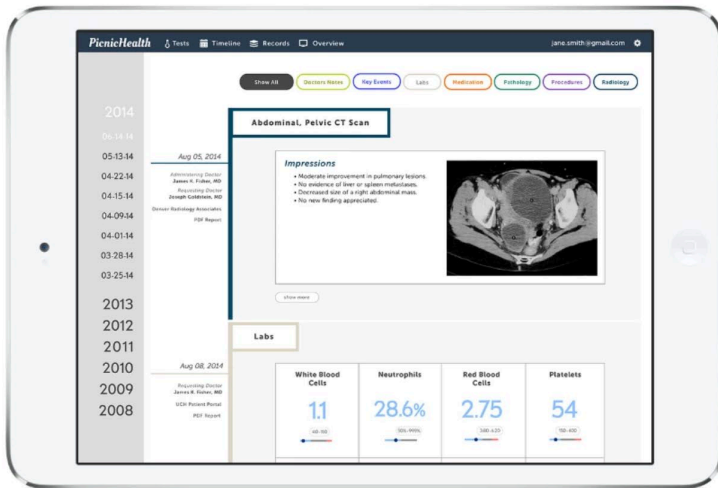
What is a personal health record?

A personal health record, or [PHR](#), is an electronic application through which patients can maintain and manage their health information (and that of others for whom they are authorized) in a private, secure, and confidential environment.

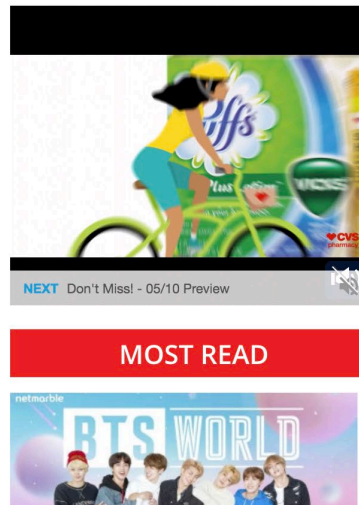
For information about [PHRs](#) and the Health Insurance Portability and Accountability Act ([HIPAA](#)) Privacy Rule, please [view the U.S. Department of Health and Human Services \(HHS\) Office for Civil Rights' guidance on this topic](#).

Picnic Health narrows its focus to helping cancer patients with their patient records

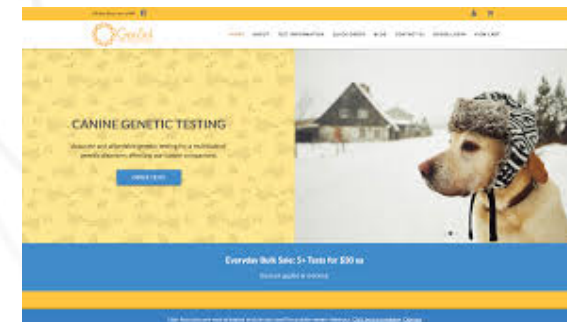
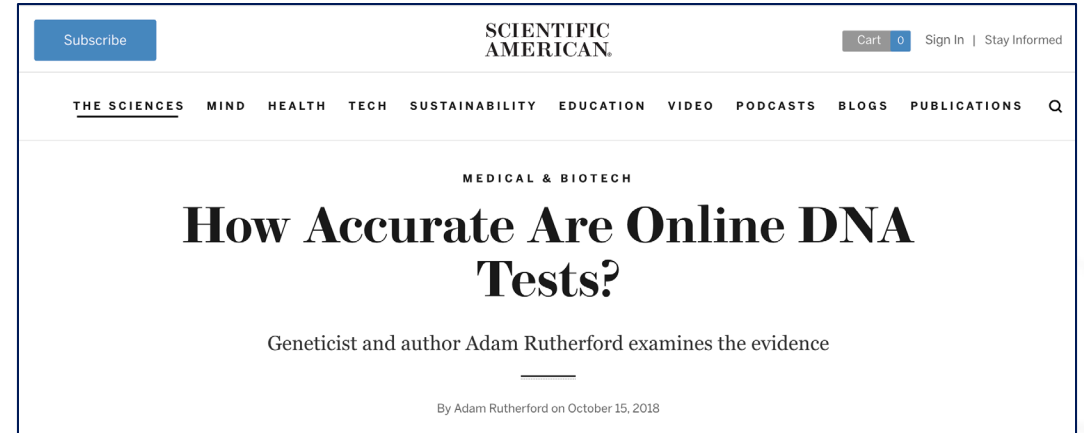
MARK SULLIVAN @THESULLIVAN AUGUST 20, 2014 7:30 PM



Above: Picnic Health's personal health record



Recreation



E9: Disable the Surveillance State & Stand Up for Patient/Consumer Rights

You Retweeted



Privacy Project @PrivacyProject · 11h

From @DKThomp for @TheAtlantic: "If you have a hard time understanding the meaning of privacy and the scale of digital surveillance in the modern age—and let's face it, who doesn't?—consider a toy named Cayla"



Why Surveillance Is the Climate Change of the Internet

The Atlantic podcast Crazy/Genius returns to explain how privacy became the most important idea on the Internet—and why it's still so confusing.

theatlantic.com

You Retweeted



Brave Bosom @BraveBosom · 13h

Wowza: FTC Members Unanimously Press Congress for Tough National Privacy Protections gizmodo.com/ftc-members-un... via @gizmodo



FTC Members Unanimously Press Congress for Tough National Priva...

All five members of the Federal Trade Commission appeared Wednesday before the House Communications and Technology Subcommittee. One ...

gizmodo.com

E10: Discover New Math

EDITORIAL



Eric D. Perakis is Senior Vice President, Data Science, Takeda Oncology, and Visiting Faculty, Department of Biomedical Informatics, Harvard Medical School, Boston, MA, 02115, USA. Email: eric.perakis@takeda.com



Isaac S. Kohane is Chair and Marion V. Nelson Professor of the Department of Biomedical Informatics, Harvard Medical School, Boston, MA, 02115, USA. Email: isaac.kohane@harvard.edu

Citation:
E. D. Perakis, I. S. Kohane, Treating the enigmatic "exceptional responders" as patients with undiagnosed diseases. *Sci Transl Med* 8, 340e08 (2016).

10.1126/scitranslmed.aaf0146

DEEP PHENOTYPING

Treating the enigmatic "exceptional responders" as patients with undiagnosed diseases

EXCEPTIONAL RESPONDERS ARE PATIENTS WHO RESPOND TO THERAPIES IN WAYS THAT are both dramatically and unexpectedly positive but also statistically insignificant within the current context of most biopharmaceutical drug trials. Recent studies have demonstrated the genetic bases of some exceptional responders and unearthed secondary resistance mechanisms (1). These insights have stimulated great interest and a thirst for more knowledge. Organizations such as the U.S. National Cancer Institute are launching exceptional-programs to learn, from these unusual patients, how to improve therapies for traditional patients (2).

Occupying a similarly tiny but more unfortunate end of the statistical spectrum are adults and children who are afflicted with rare and undiagnosed diseases. Many rare diseases are genetic in origin, and the study of such disorders can yield insights into fundamental biology, which can, in turn, illuminate mechanisms that underlie more common diseases. This has proven to be true in next-generation sequencing studies of rare tumor samples, which have led to more intricate characterization of tumor pathologies, the establishment of new cancer classifications and subtypes, and new mechanistic insights.

The U.S. National Institutes of Health (NIH) Office of Rare Diseases Research (ORDR) defines rare and undiagnosed diseases as those that affect fewer than 200,000 individuals; diagnosis takes 1 to 5 years in 33% of cases studied thus far. In operation since 2008 and recently expanded to a full network of clinical and research sites nationwide, NIH's Undiagnosed Diseases Program (UDP) offers key insights into the management of exceptional responders in clinical trials regarding the efficiency and cost-effectiveness of focused and time-bound clinical assessment; the utility of agnostic whole-genome genetic studies, including patient pedigrees, to produce candidates for causal variants; the utility of rare patient populations in new disease discoveries; and the importance of deep phenotyping and careful review of the entire medical record of each patient (3). Each UDP patient is evaluated at the NIH clinical center in an intensive manner that is consolidated into a single 4- to 5-day visit. For patients with an apparent neurological phenotype this week could include consultations in 10 areas of medical specialties, 20 various laboratory investigations, and 25 diagnostic procedures, some of which require sedation. All of these data are then considered in the context of the complete medical history of the patient, which is compiled in the 2 to 3 months prior to the clinical visit.

Would a cancer-specific, deep-phenotyping process similar to that of UDP patients be useful for exceptional responders? Considering the complexity and possible contributions of environmental and genetic factors, such a study could surely lead to insights or at least hypotheses that would enable a deeper understanding of such patients. This knowledge could then inform more targeted patient selection for future clinical trials.

Deep molecular phenotyping provides a richer understanding of population-wide, disease-course divergences, intratumor cellular heterogeneity, and histological variations and might pinpoint physiological alterations that can serve as therapeutic targets; at the very least, deep

To Your Health

Cancer researchers: It's time to pay more attention to 'miracle' patients. By Erin Blakemore



Emily Whitehead, right, beat cancer through immunotherapy. She's pictured onstage with Lady Gaga at the launch of the Parker Institute for Cancer Immunotherapy. (Jonathan Leibson/Getty Images for Parker Media)



06:36
0 </>

Harvard Project Seeks 'Exceptional Responder' Cancer Patients To Figure Out What Went Right

June 21, 2018 By Carey Goldberg



Treating Data as an Asset: Data Entrepreneurship in the Service of Patients

1. Break down the macro and micro silos of healthcare data
2. Keep patients safer
3. Crowdfund the most difficult problems
4. Aggregate, align and donate to friends and freaks
5. Catch a thief
6. Out-shout medical misinformation
7. Shrink space and time to improve research access
8. Nourish your family but read the label
9. Disable the surveillance state and stand up for patient/consumer rights
10. Discover new math