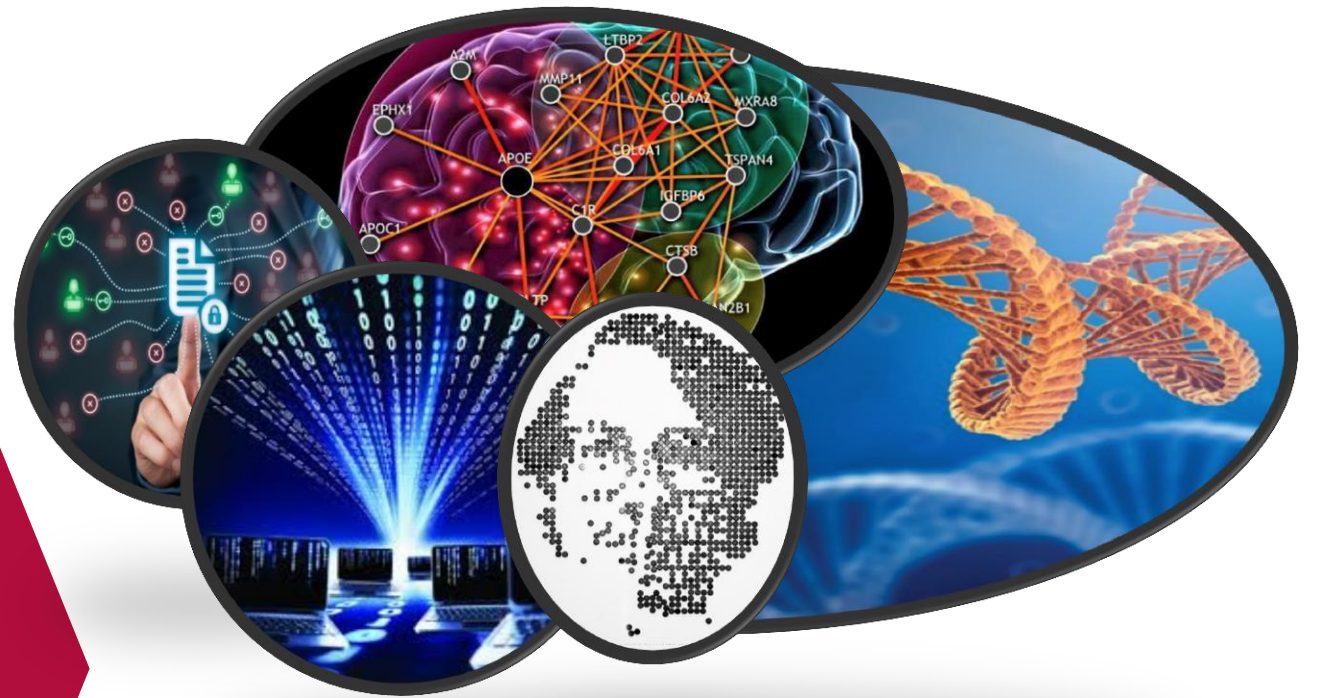


YOU and Big Data in Data Wonderland: It's NOT a Data Jabberwocky



Vivian OTA WANG, Ph.D., CGC, FACMG

***Deputy Director, Office of Data Sharing
Center for Biomedical Informatics &
Information Technology (CBIIT)
National Cancer Institute-NIH***

***Professional Development Workshop and Mock Review
03 June 2019***

Agenda

Data Wonderland

*Big Data
Human Rights
Open Science*



Jabberwocky

*The Policies
Office of Data Sharing*



Science

*The Data
The People
The Ethics*





Data Wonderland

Big Data
Open Science
Human Rights



Big Data: Human Rights and the Democratization of Knowledge



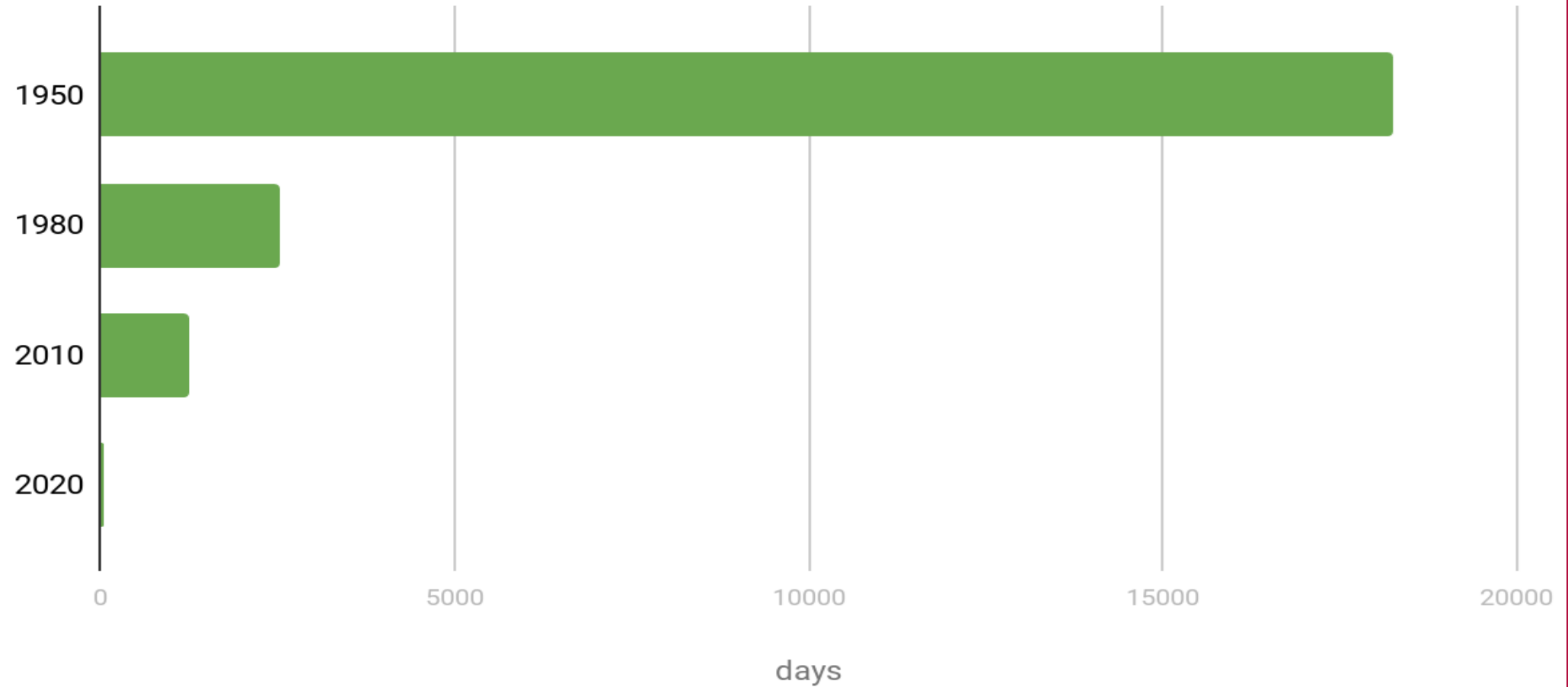
Big Data



- Large amounts of data and data types
 - *Mobile devices, tracking systems, RFID, sensor networks, social networks, Internet searches, automated record keeping, video archives, e-commerce*
- Secondary analyses of primary and derived data
- Identify trends
- Improve research quality

Big Data

Doubling Time of Health Knowledge



Open Science



DATA SHARING AND INNOVATION

- Open access
 - Accessible research & data to all levels of society (*e.g., amateurs, citizen scientists, and professionals*)
- Open data
- Open sources

Open Science and Data Sharing

Facilitates innovation of
research tools and
methods



Open Science and Data Sharing

Facilitates innovation of
research tools and
methods

- Increases statistical
power



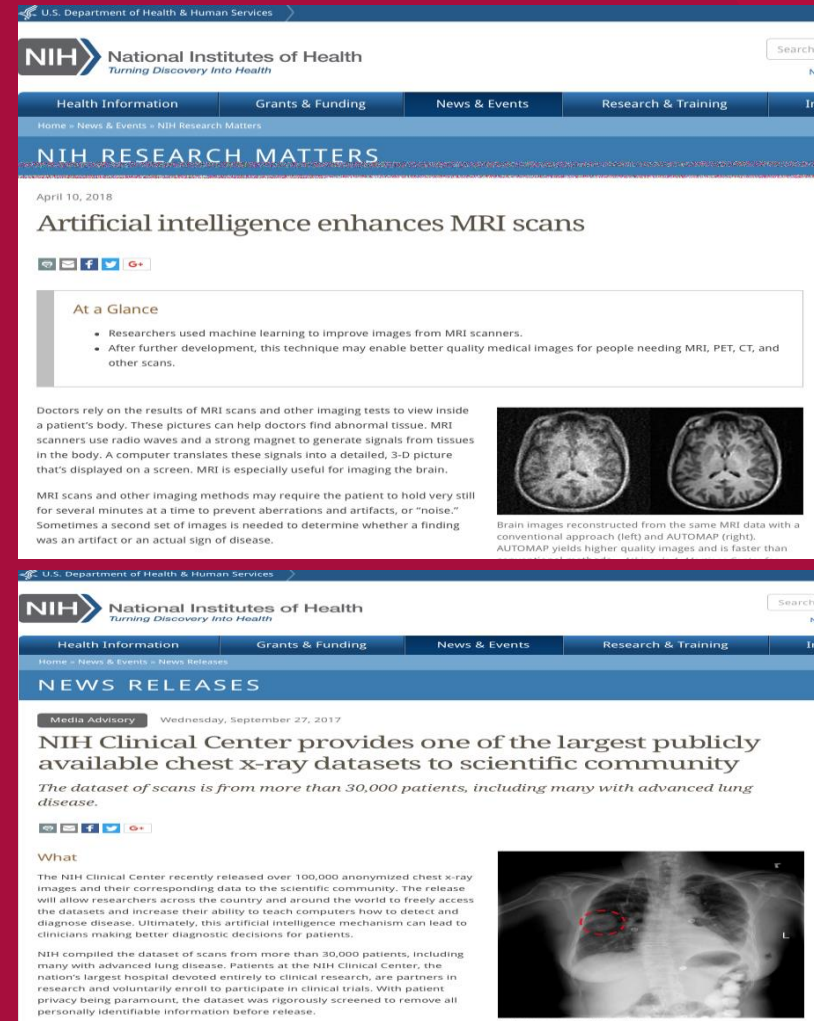
Open Science and Data Sharing

- Facilitates innovation of research tools and methods
- Increases statistical power
- Improves research quality through validation and replication



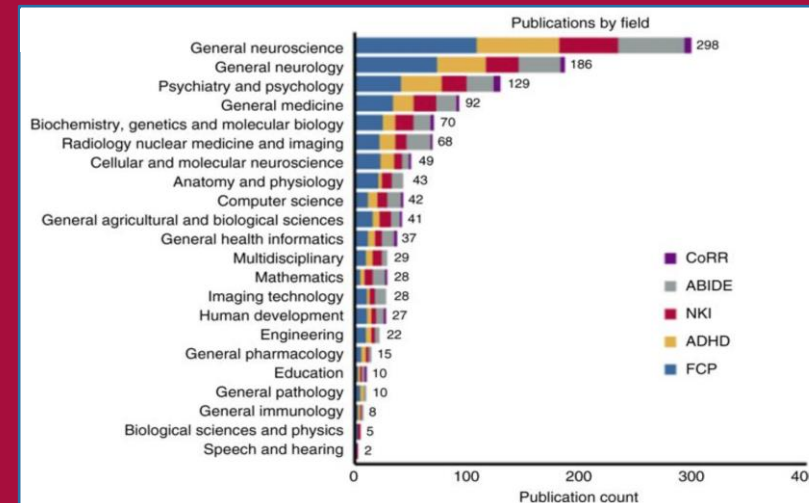
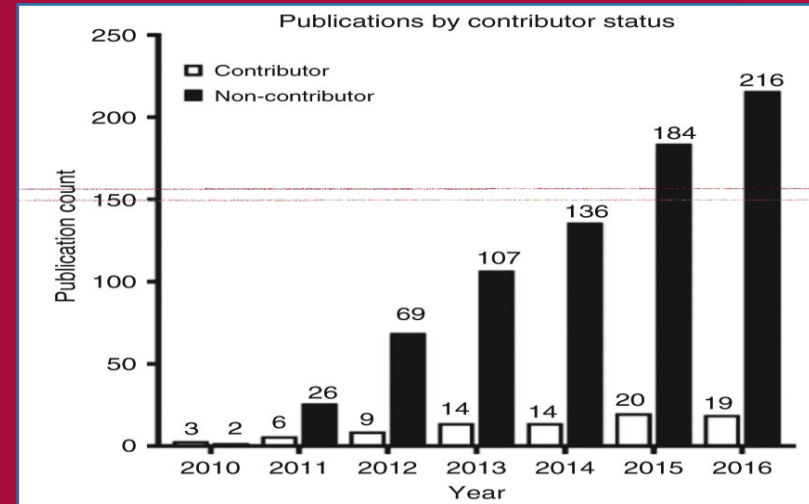
Open Science and Data Sharing

Increases scientific value
and analyses by enabling
data from multiple
studies to be combined
and explored



Open Science and Data Sharing

- Increases scientific value and analyses by enabling data from multiple studies to be combined and explored
- Increases scale of studies, # publications, and types of scientists from a broader range of disciplines



Open Science and Data Sharing



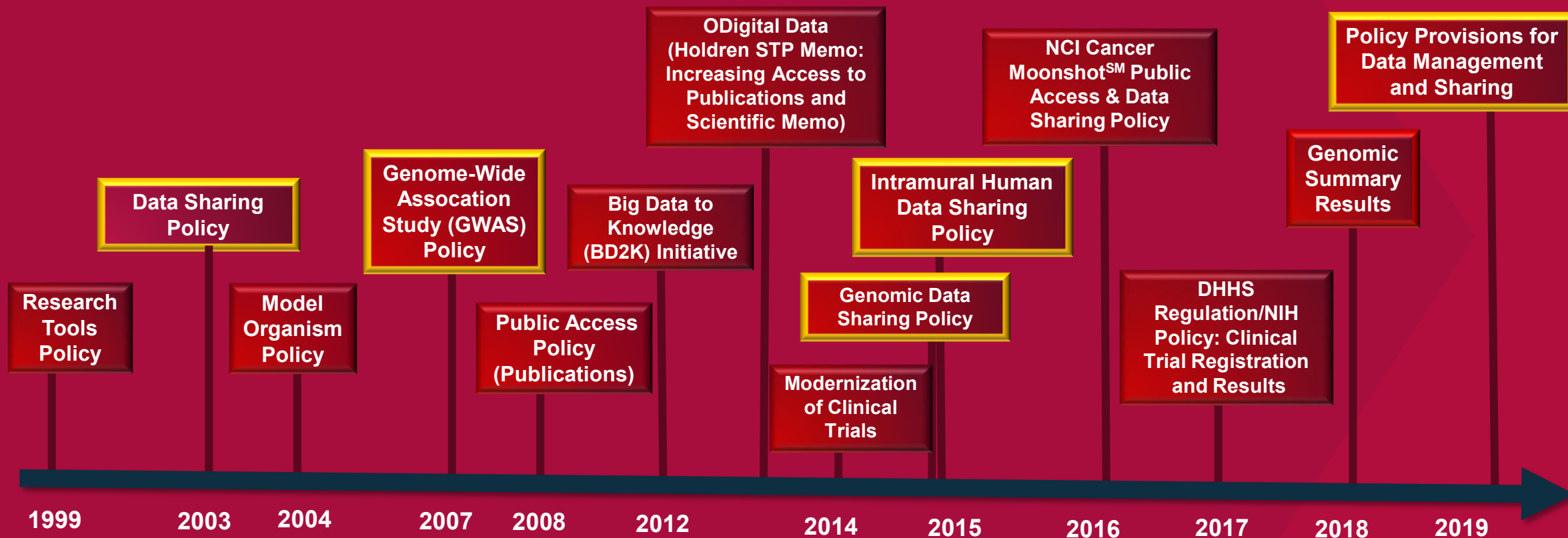
- Biology and Medicine are now *data intensive enterprises*
- *Rapidly changing* scale
- Technology, *data computing* and *information technology* (IT) are *pervasive* in the *lab*, *clinic*, and *home*



The Jabberwocky

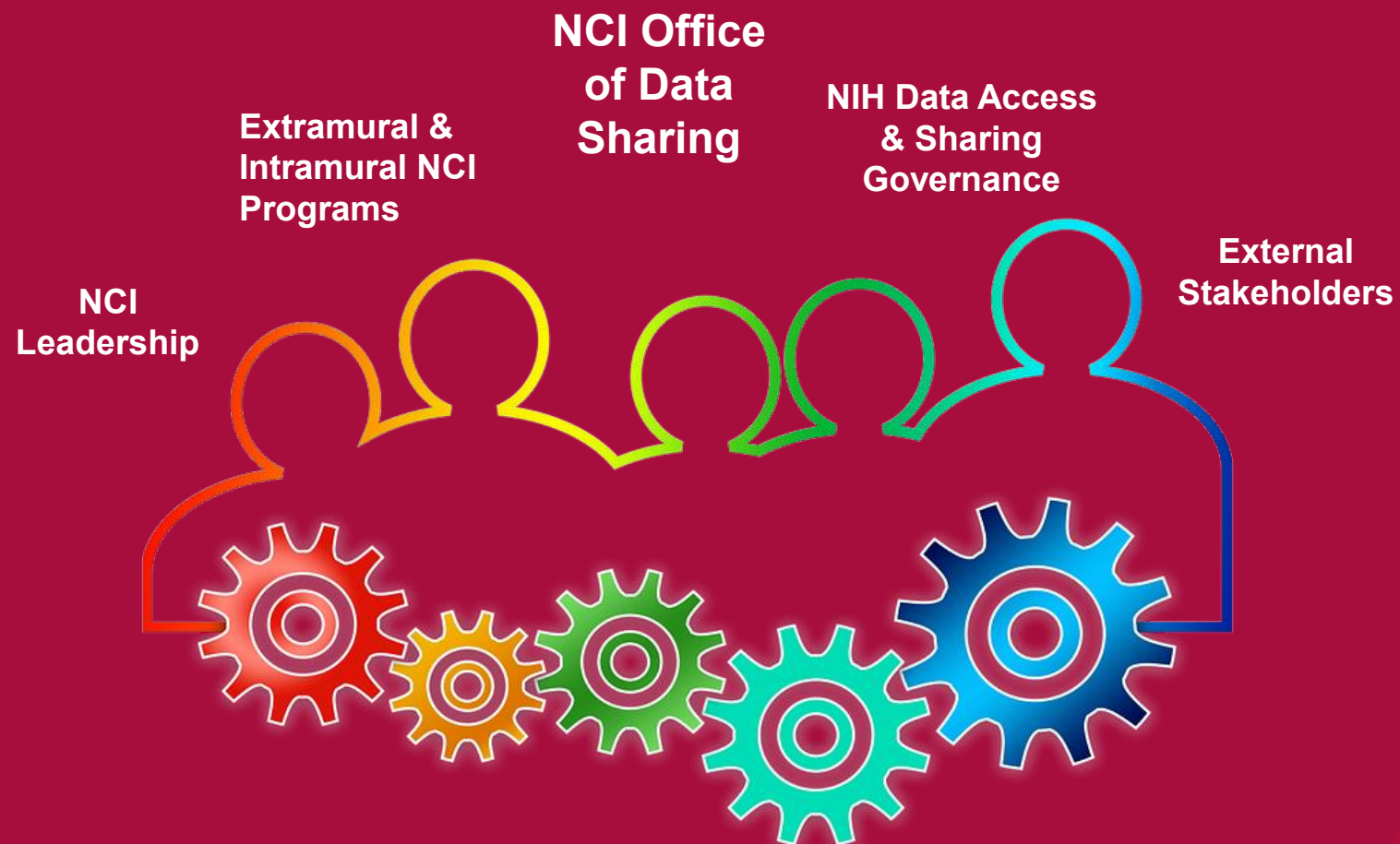
Policies
NCI Office of Data Sharing

NIH Data Sharing Policies



NCI Office of Data Sharing

nciofficeofdatasharing@nih.gov



NCI Office of Data Sharing

nciofficeofdatasharing@nih.gov



Provide **leadership and guidance** to enhance data sharing for NCI and the cancer research community.



Guide NCI approach to **implementation and interpretation** of NIH and NCI data management and sharing policies.



Coordinate registration, submission, and access **procedures** for NCI datasets/repositories.



Advise on considerations for **ethical and minority and health disparity issues** related to data access and sharing for the cancer community.

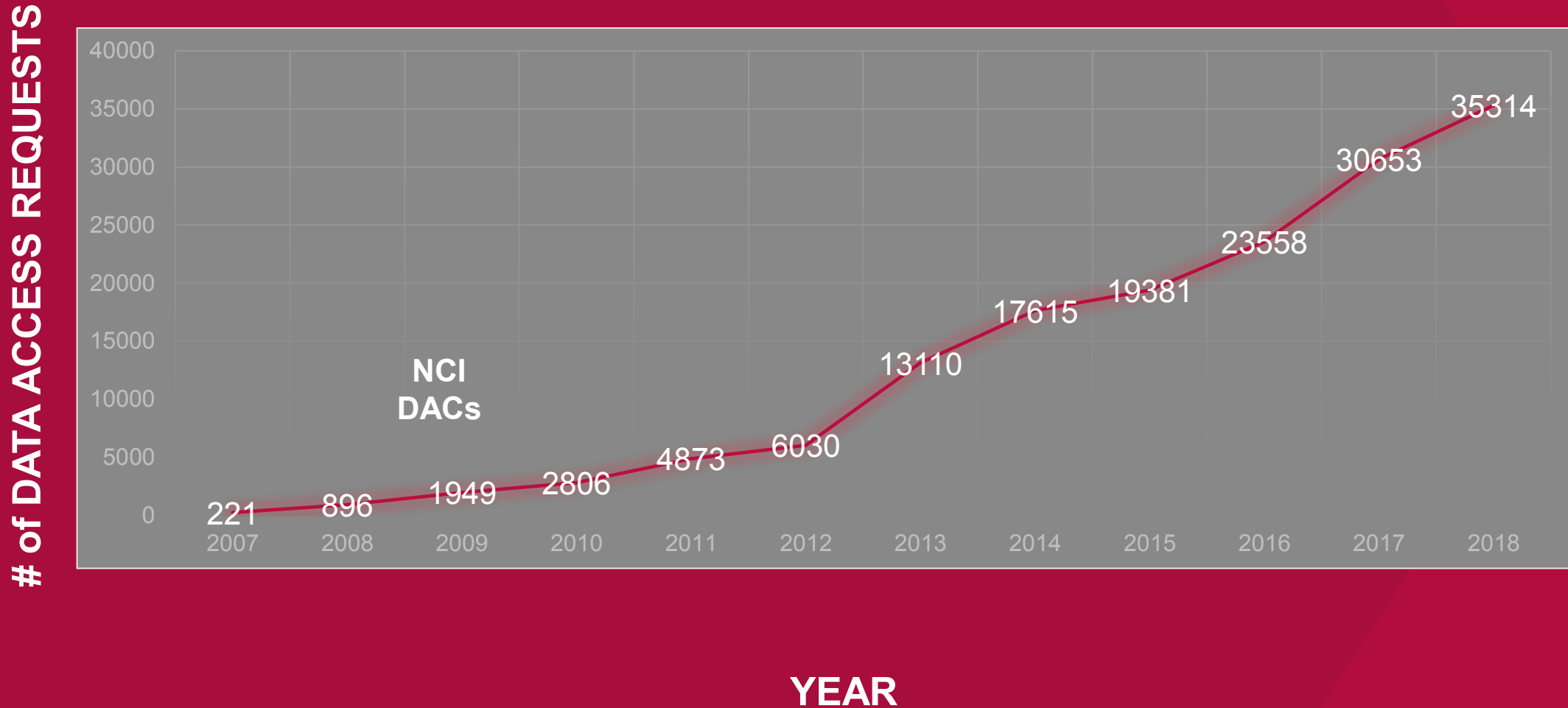


Encourage **participation** in major data sharing initiatives.



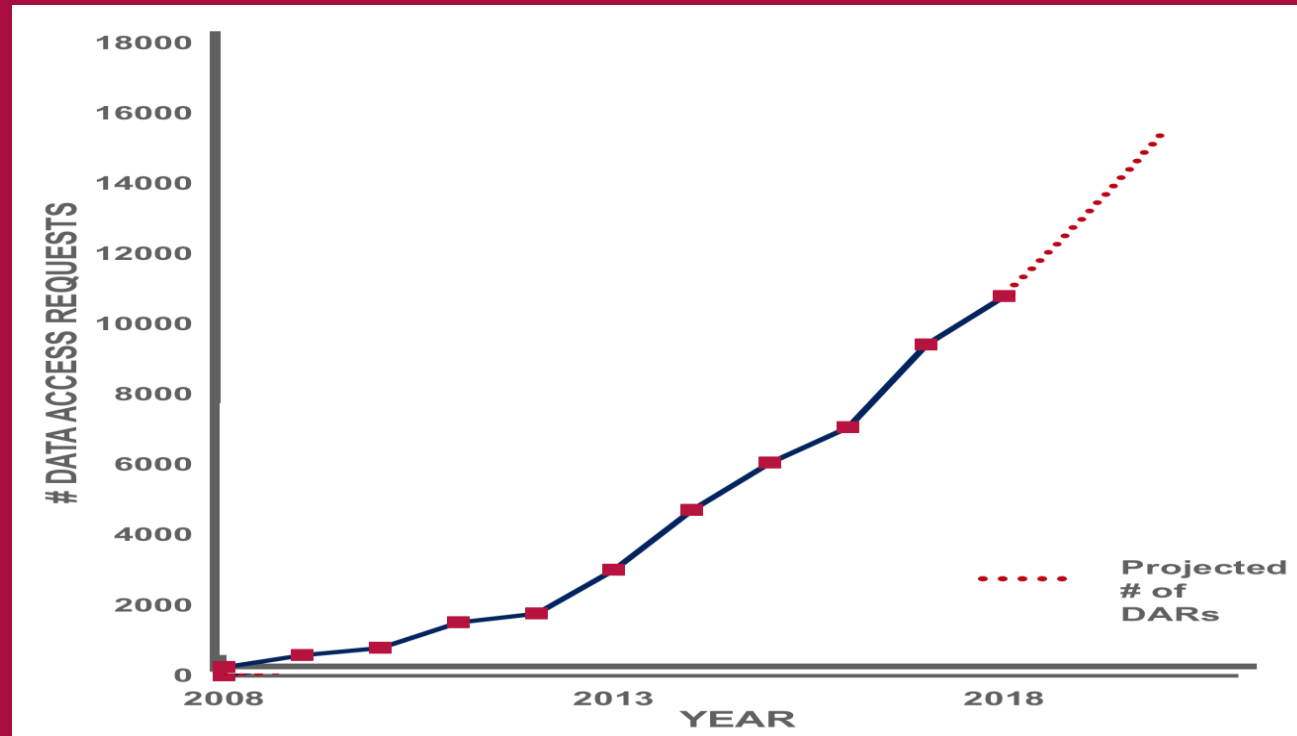
Create **data sharing resources** to inform and guide the cancer communities.

Total Number of Actual and Projected NCI dbGaP Data Access Requests By Year



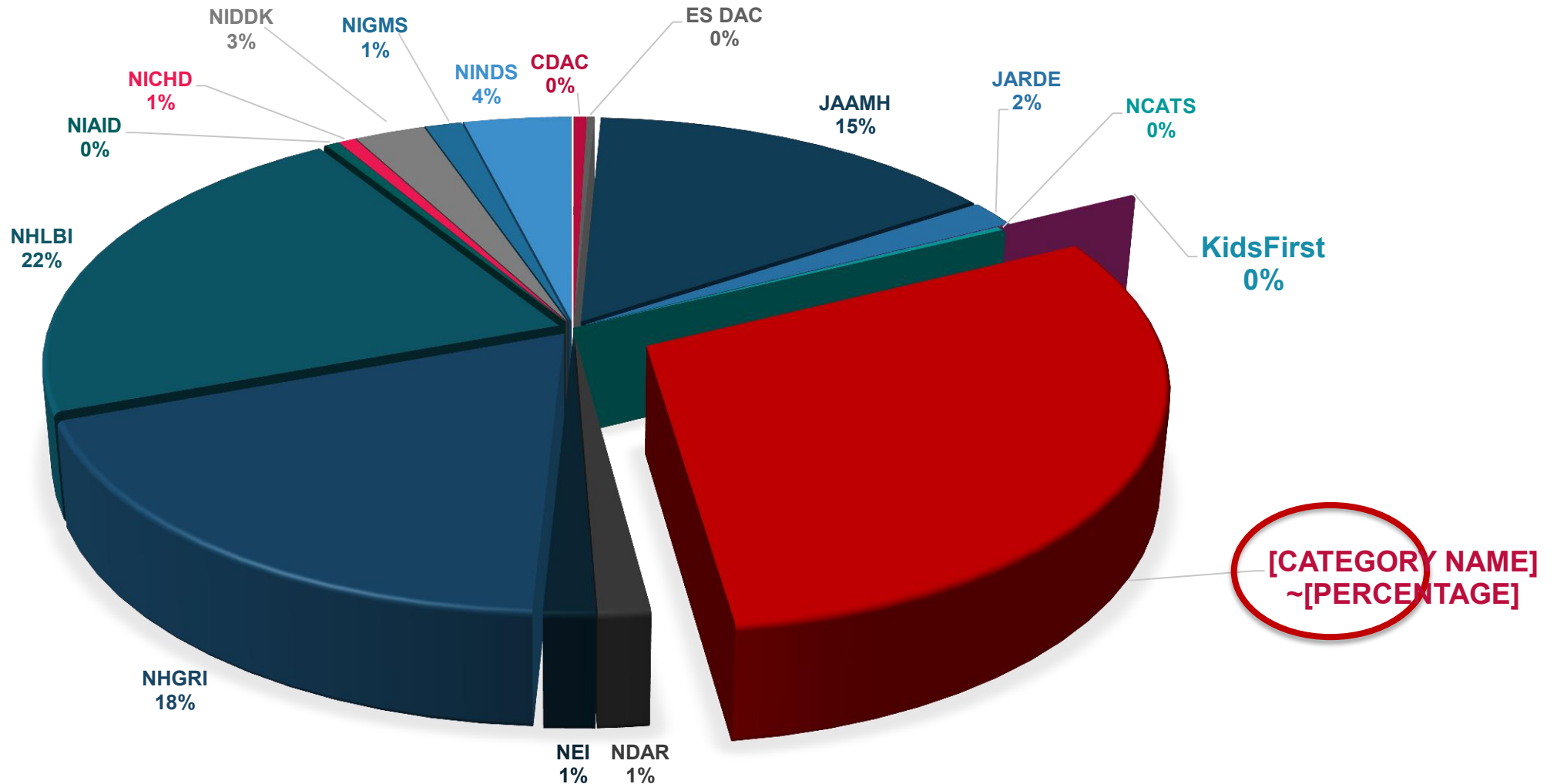
Total Number of Actual and Projected NCI dbGaP Data Access Requests By Year

of DATA ACCESS REQUESTS

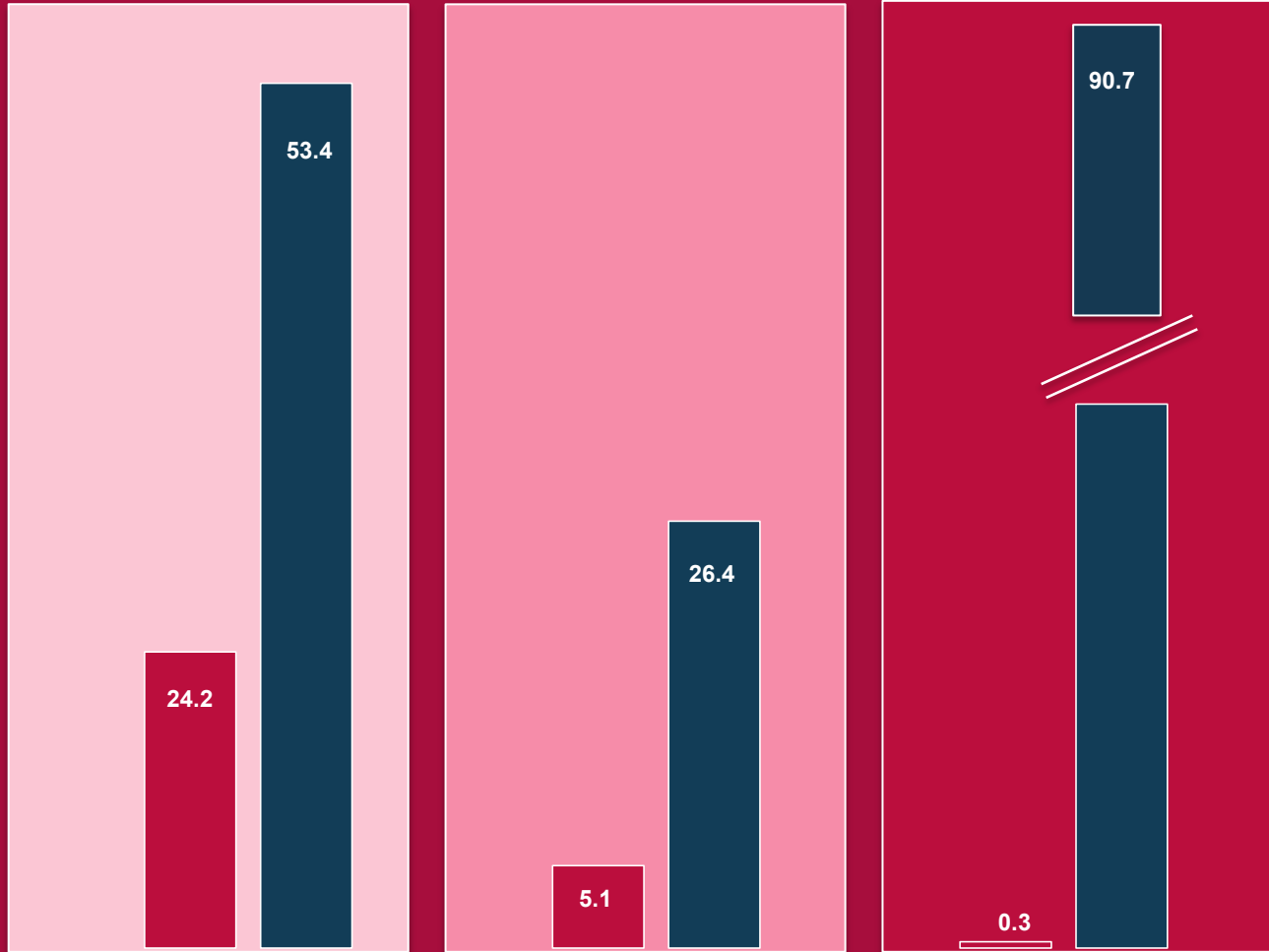


YEAR

Percentage of Data Access Requests By NIH I/Cs 01/01/2018 - 05/31/2019



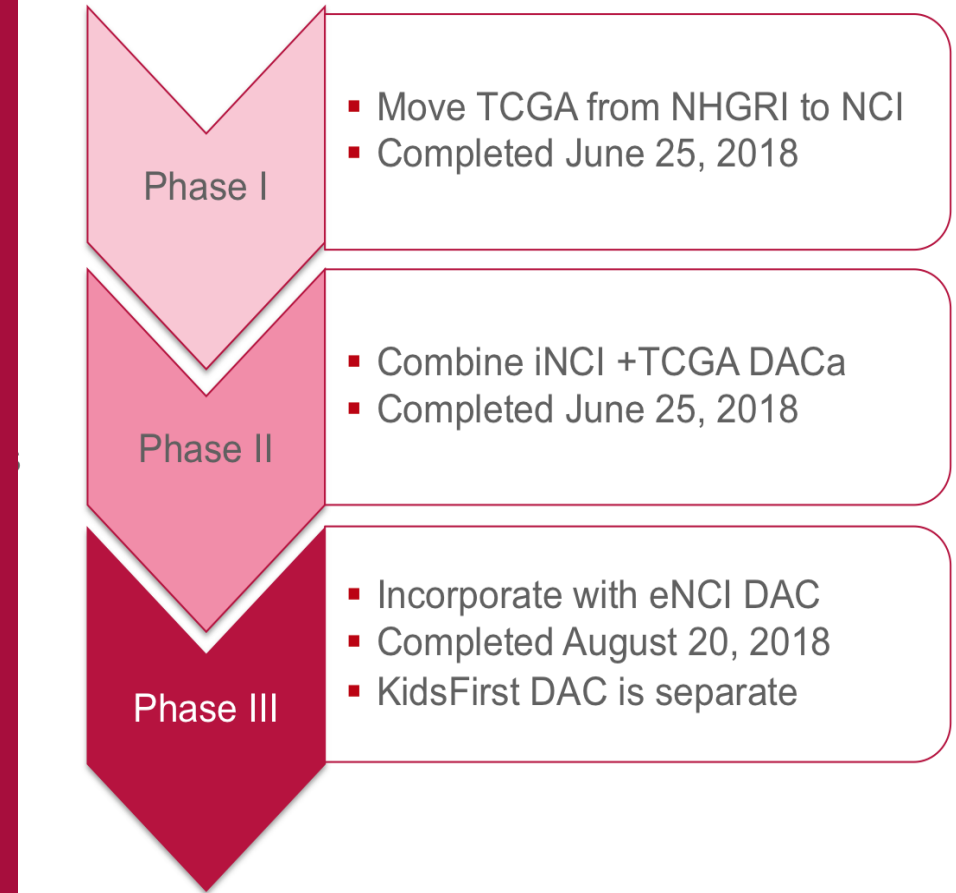
NCI Office of Data Sharing*



Pre-centralization to Phase I
(6/25/17 - 6/22/18)

Phase I to Phase II
(6/25/18 - 8/17/18)

Post Phase III
(8/18/18 - 5/31/19)



* NCI DAC receives ~30% of DARs to dbGaP; results include significant efficiency in DAR processing times and eliminating 1000+ DAR backlog

The World's Cheapest Car | 23 Hot Summer Gadgets

Get Ready for the
Google Phone

WIRED

TIME | JUL 2008



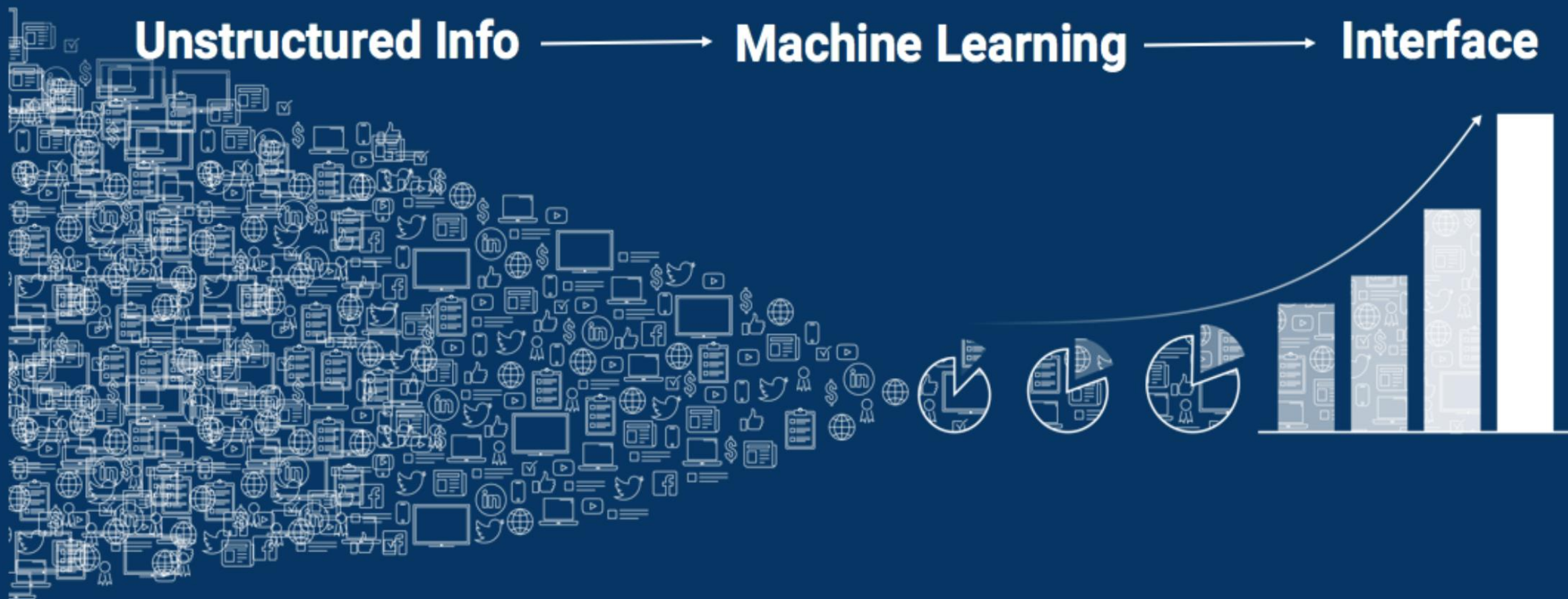
The End of Science

The quest for knowledge used to begin with grand theories.
Now it begins with massive amounts of data. Welcome to the Petabyte Age.

The Science

The Data
The People
The Ethics

Unstructured Info → Machine Learning → Interface



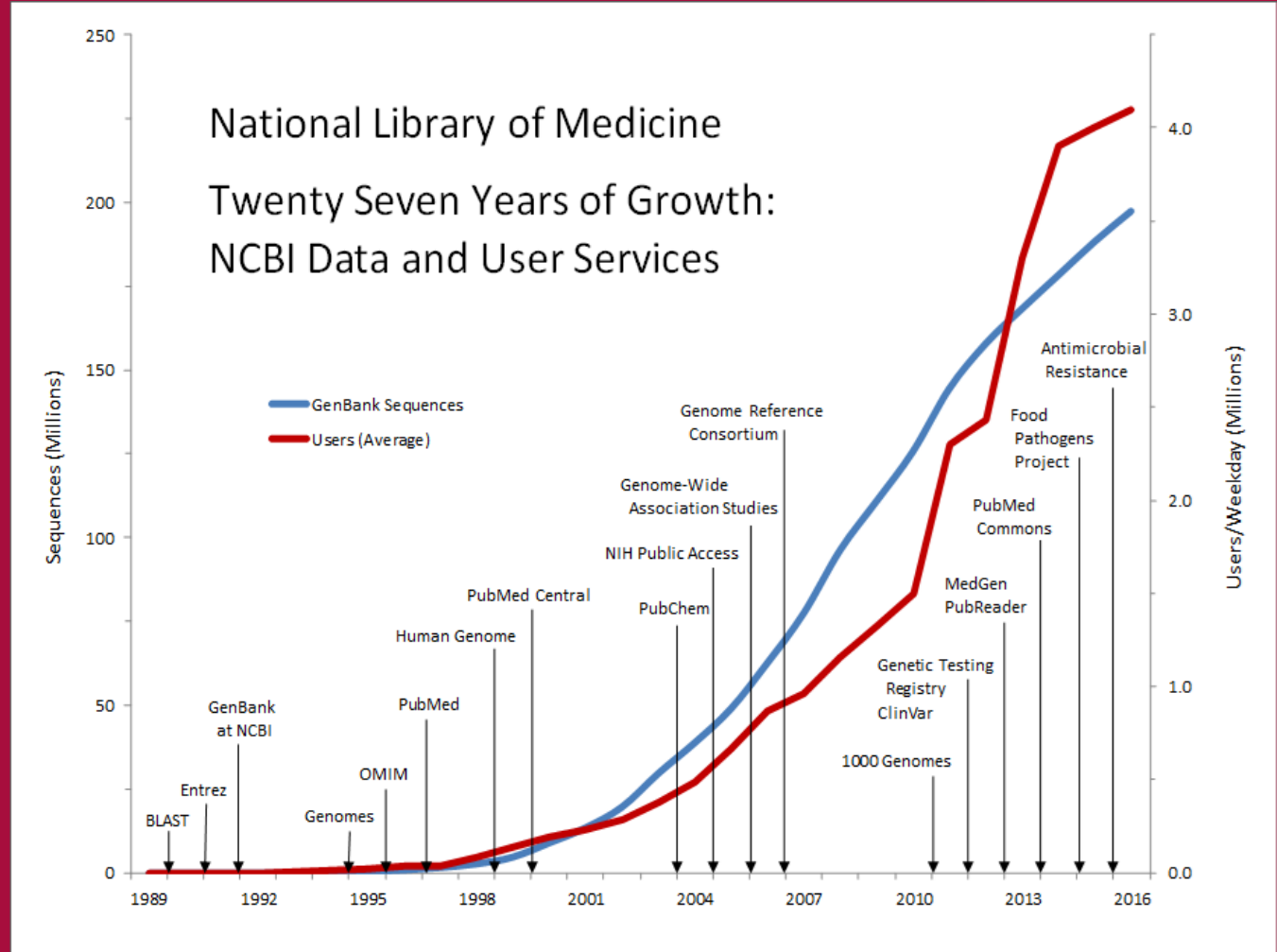
Machine learning technology
analyzes millions of unstructured
sources in real-time and...

selects and synthesizes
that knowledge
so you can...

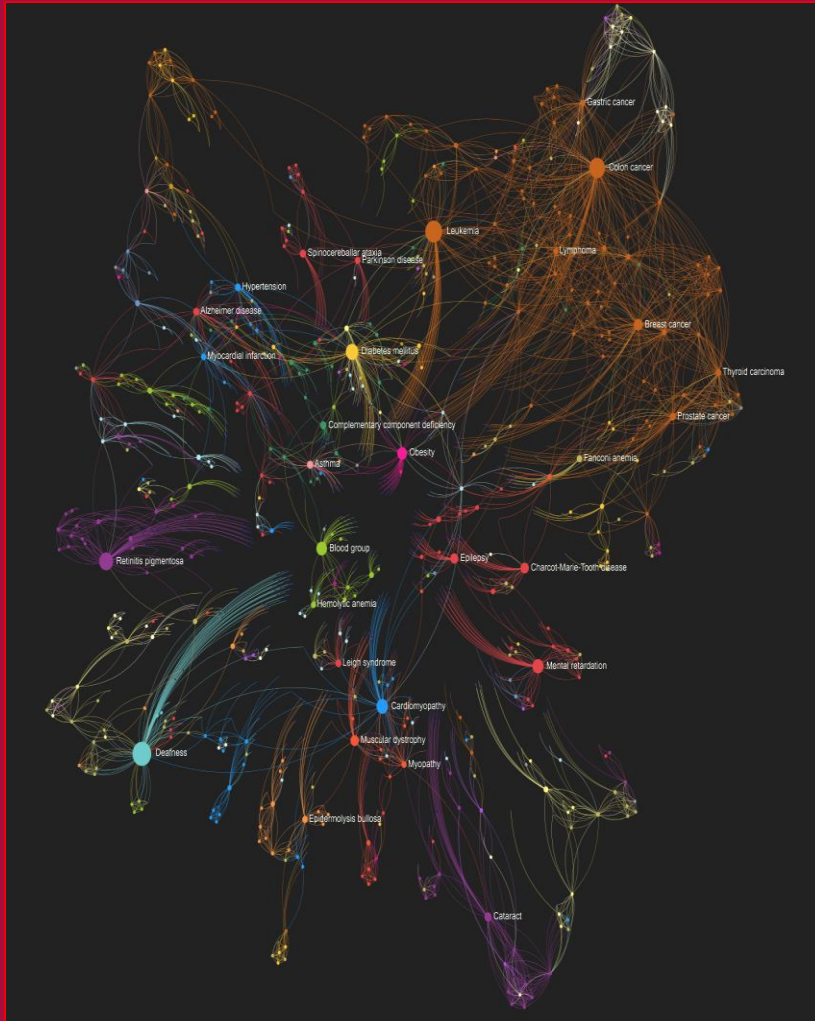
see trends easily &
quickly.

Data: Variety, Volume, Velocity, and Veracity

- More scientific data domains are emerging with capacities to capture real time health information
 - Proteomics
 - Metabolomics
 - Microscopy
 - Medical imaging
 - Other various technologies



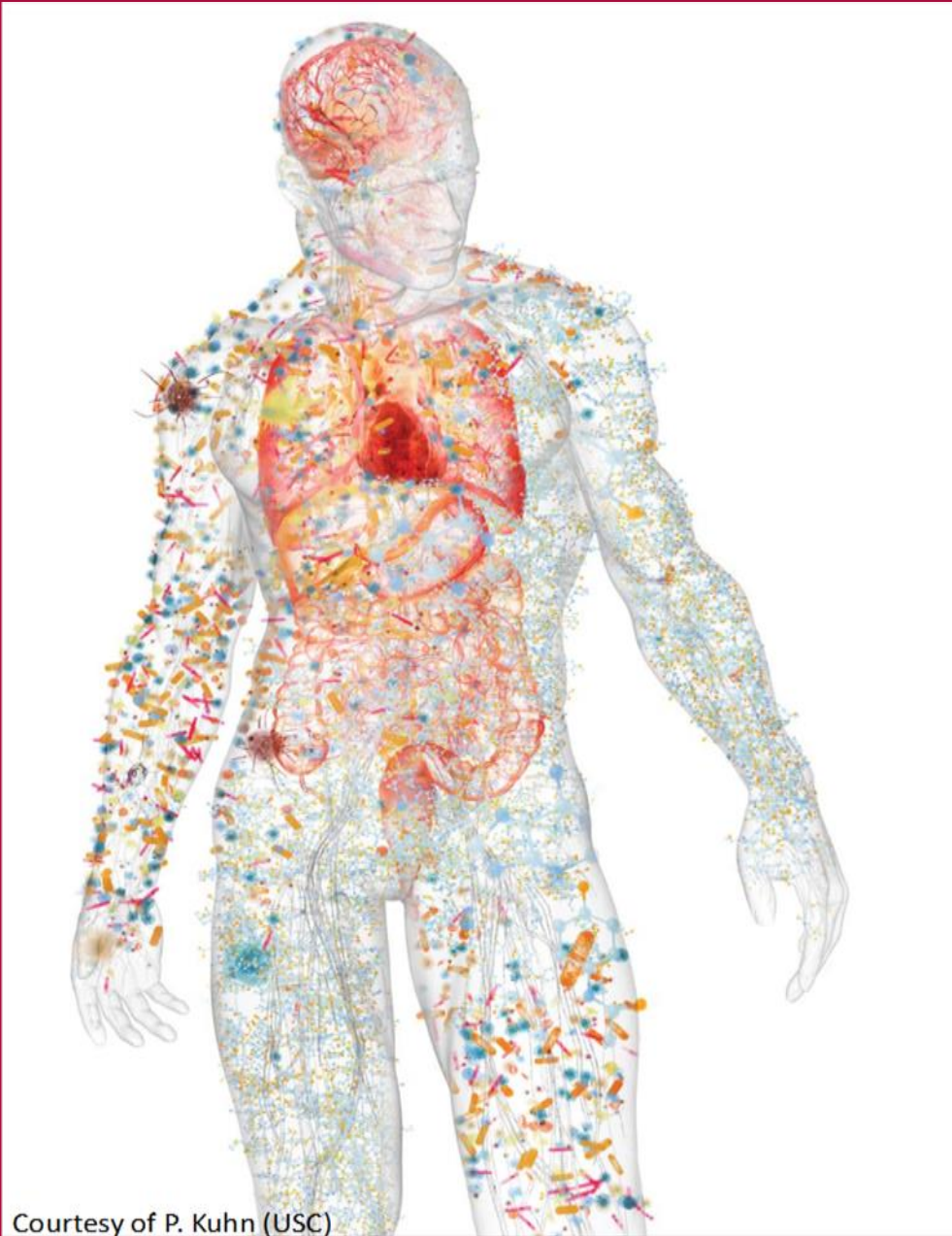
Human Disease Networks (2015)



- Identify disease gene-phenotype associations at higher cellular and organismal levels

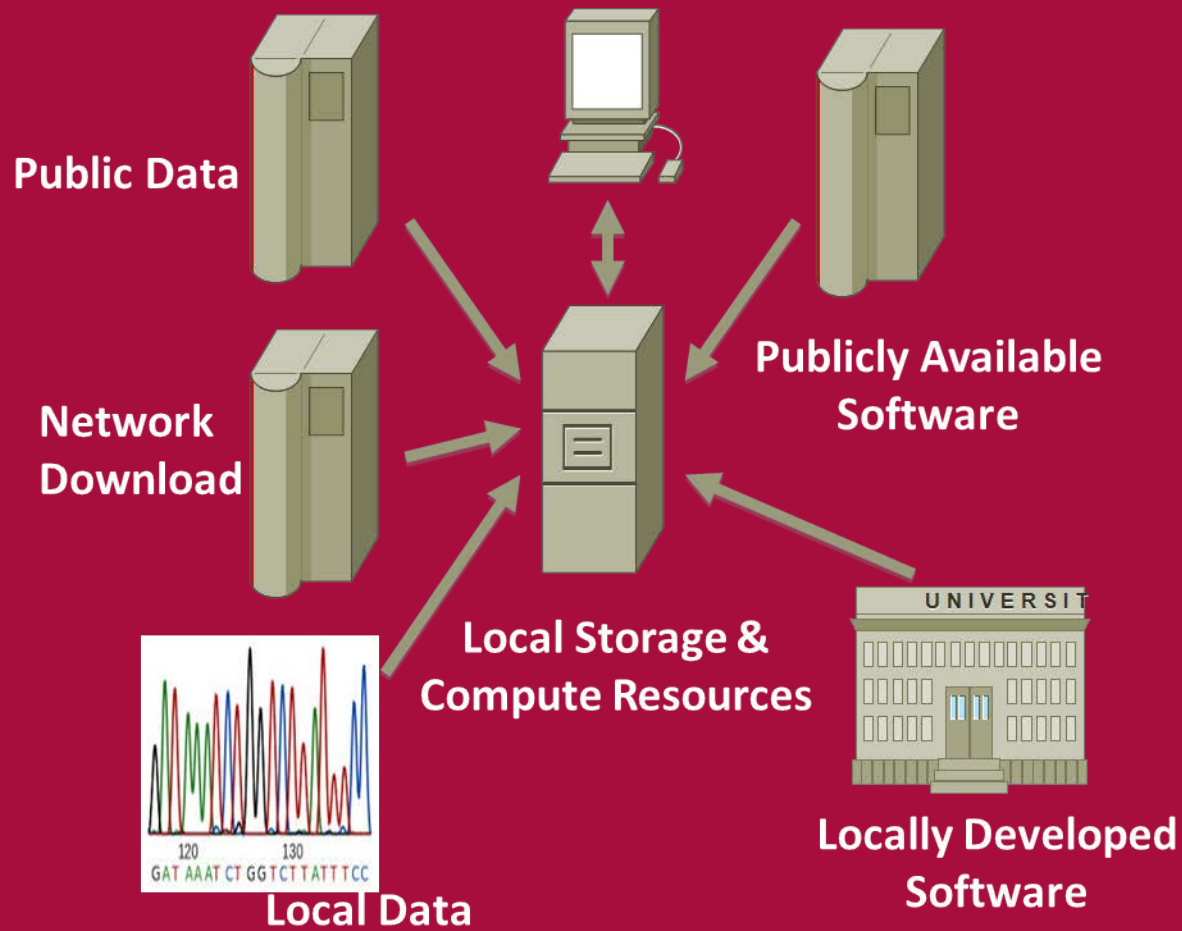
Precision Medicine

- Learning system that accounts for complexity of underlying biology
- Requires
 - Deep biological understanding
 - Advances in scientific methods
 - Advances in instrumentation
 - Advances in technology
 - Advances in data management and computation
- *Can change* disease classifications
- Genomic, imaging, clinical, and laboratory data



Courtesy of P. Kuhn (USC)

Standard Model of Computational Analyses (circa 2014)



NATIONAL CANCER INSTITUTE THE CANCER GENOME ATLAS

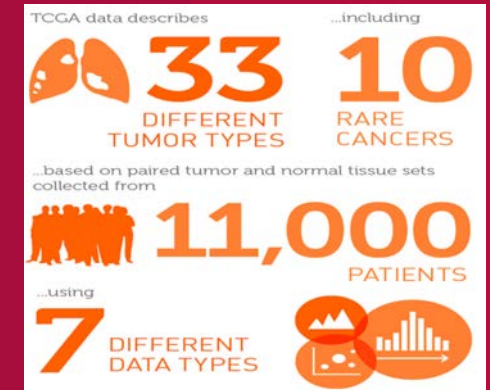
TCGA BY THE NUMBERS

TCGA produced over

2.5

PETABYTES

of data



TCGA RESULTS & FINDINGS



MOLECULAR
BASIS OF
CANCER

Improved our understanding of the genomic underpinnings of cancer



TUMOR
SUBTYPES

Revolutionized how cancer is classified

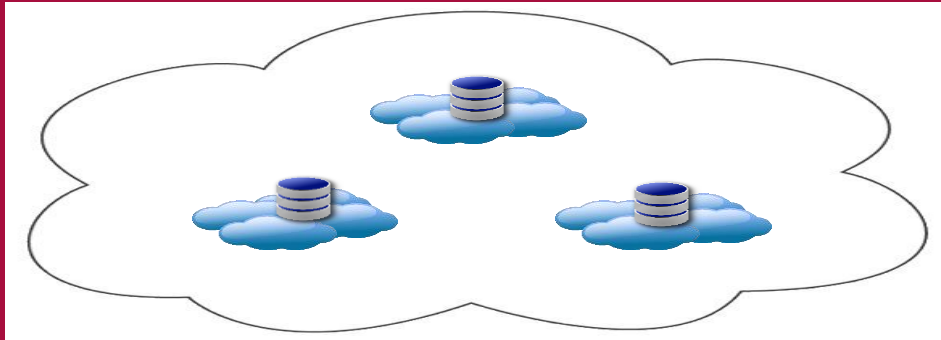


THERAPEUTIC
TARGETS

Identified genomic characteristics of tumors that can be targeted with currently available therapies or used to help with drug development

NCI Cloud Resources

- Democratize access to NCI-generated data
- Create cost-effective scalable computational capacity



- Access and analyze 11,000 TCGA samples without having to download data
- Upload your own data for analysis

Data



- Perform large scale analysis using the elastic compute power of commercial cloud platforms

Compute



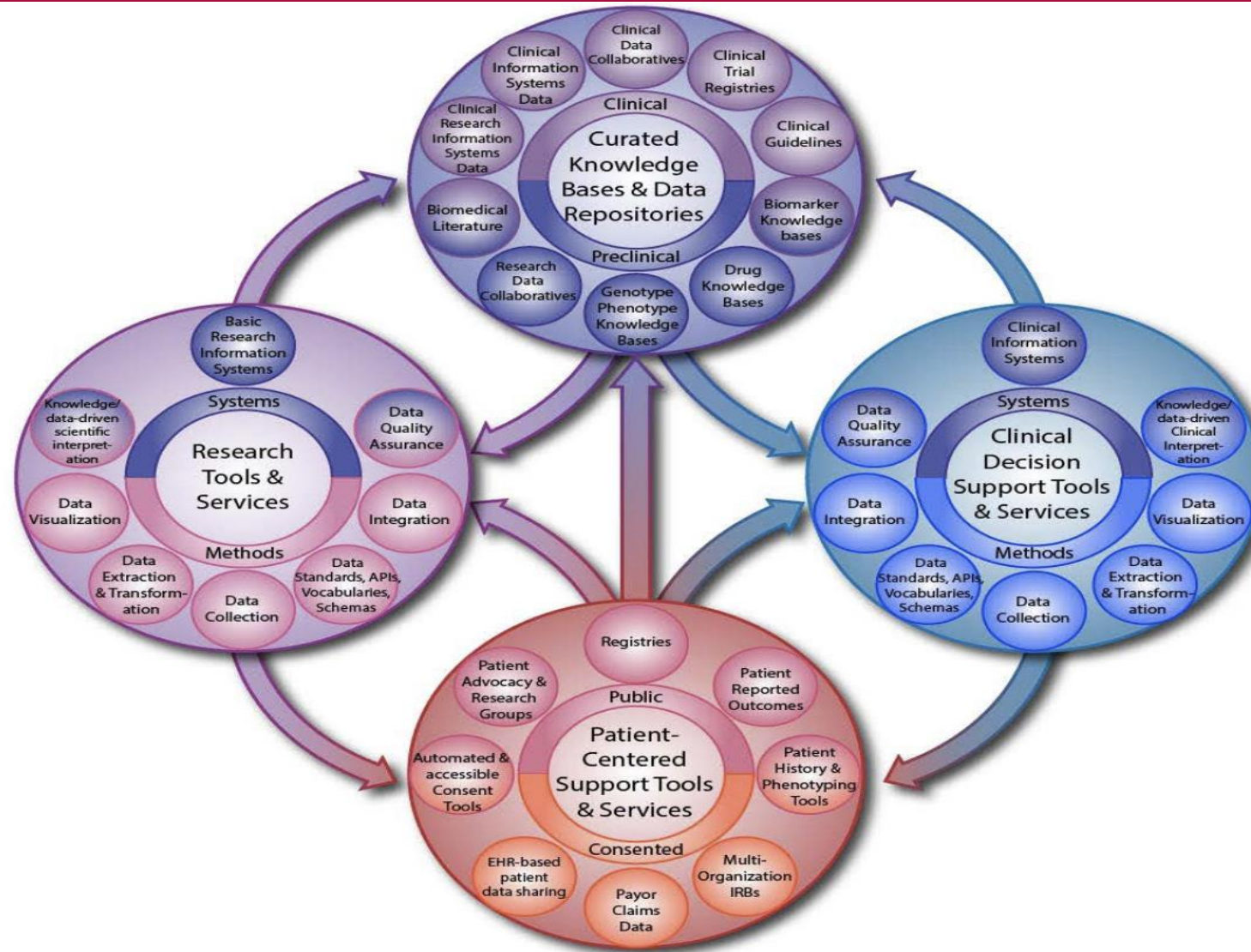
- dbGaP-authorized users can access controlled TCGA data
- Systems meet strict Federal security guidelines

Security



- Access large data sets without downloading data
- Bring tools and pipelines to the data
- Bring and combine own data and analyze with existing data
- Workspace to save and share data and results of analyses

National Cancer Data Ecosystem



**The
Economist**

JANUARY 14TH–20TH 2017

Trump v the spooks

The stain of Guantánamo

Pop stars and patronage in Congo

Inflation's welcome return

Lifelong learning

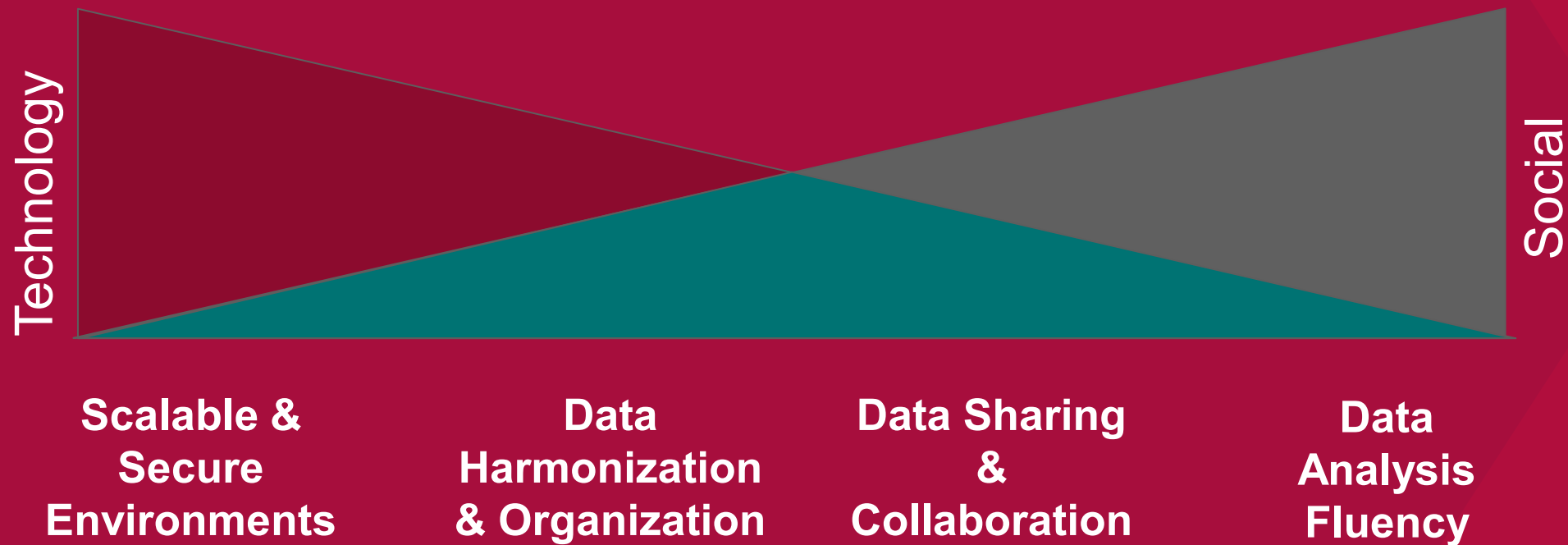
**How to survive in the age
of automation**

A SPECIAL REPORT



The People

Data Access and Sharing: Isn't Only a Technology Challenge



Data Access and Sharing: Isn't Only a Technology Challenge

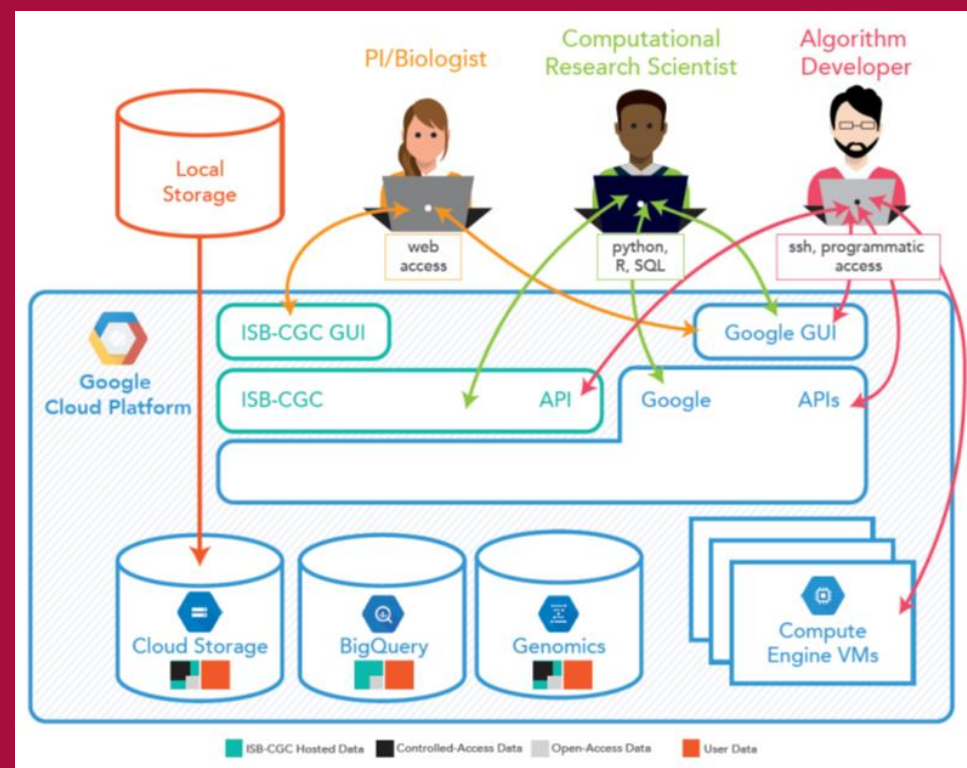
Multidisciplinary Teams with Diverse Expertise and Resources

Biology/Social/Psychology Researcher

Computational Scientist

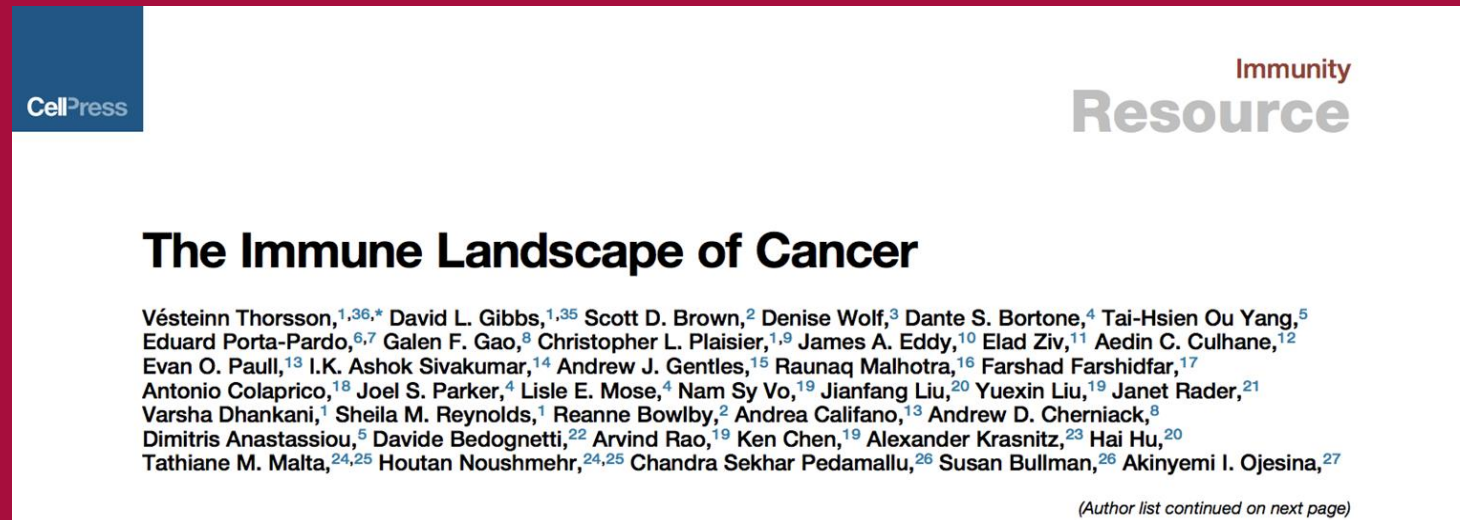
Algorithm Developer

- test new algorithm on hundreds or thousands of BAM or FASTQ files
- run novel image segmentation method across whole-slide images



Data Access and Sharing: Isn't Only a Technology Challenge

Multidisciplinary Teams with Diverse Expertise and Resources

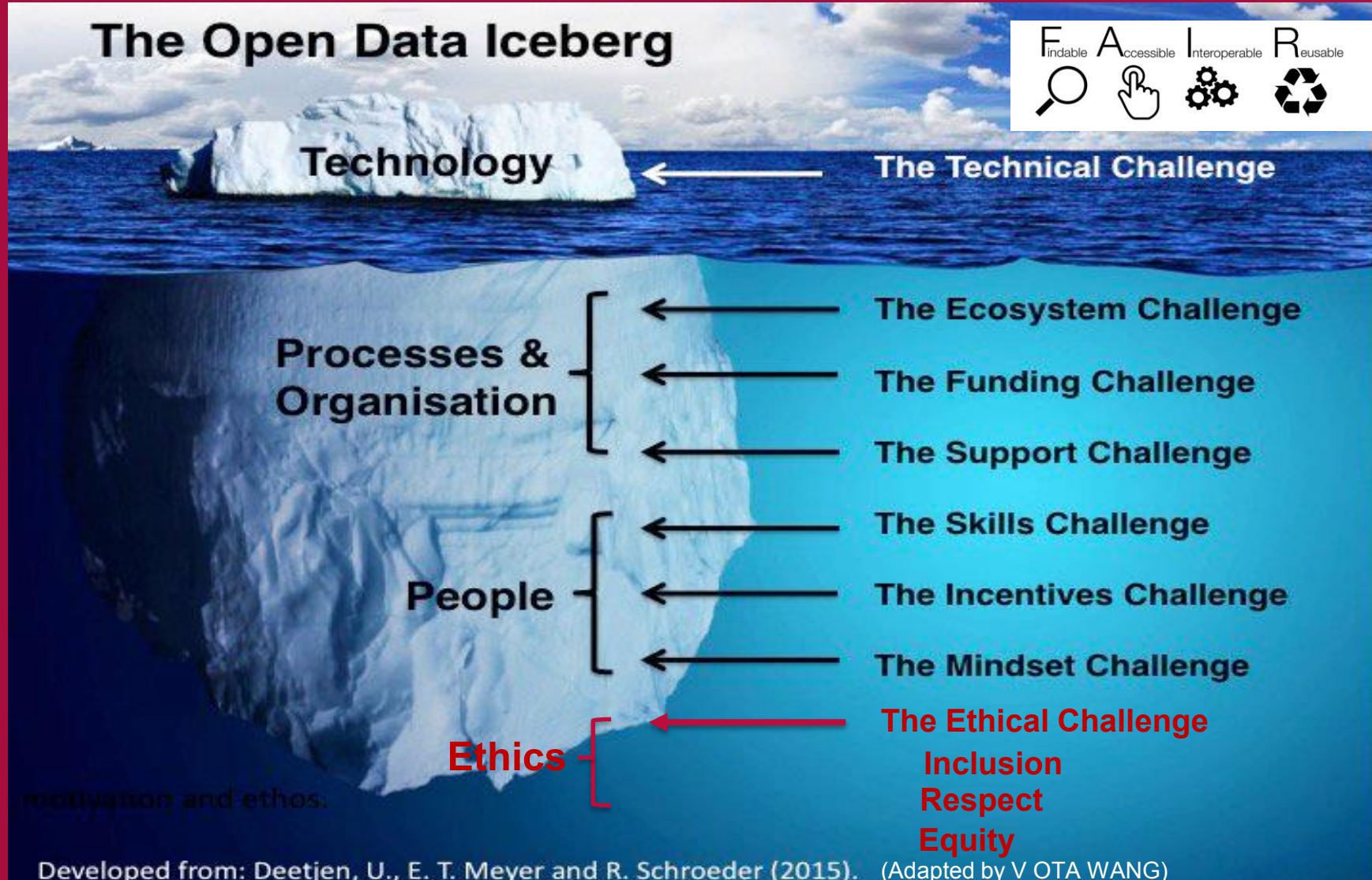
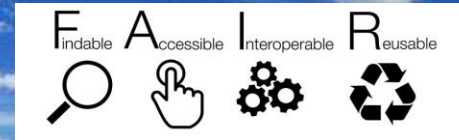


55 authors (+ TCGA network)

34 author affiliations

2 cloud resources + local infrastructures

The Open Data Iceberg



Wilkinson, M. D. *et al.* (2016). The FAIR Guiding Principles for scientific data management and stewardship. *Sci. Data*3:160018 doi: 10.1038/sdata.2016.18

datasharing Ethical, Economic, Legal, Social Implications (dEELSI)

- Data and Information are not Neutral



datasharing Ethical, Economic, Legal, Social Implications (dEELSI)

- Data and Information are not Neutral
 - Identity, Phenotypes, and Bias
 - Identifiability and Privacy
 - Bias – Machine Learning and Artificial Intelligence
 - Incidental Findings and Return of Results
 - Informed Consent and Broad Data Uses
- Governance, and Trustworthiness

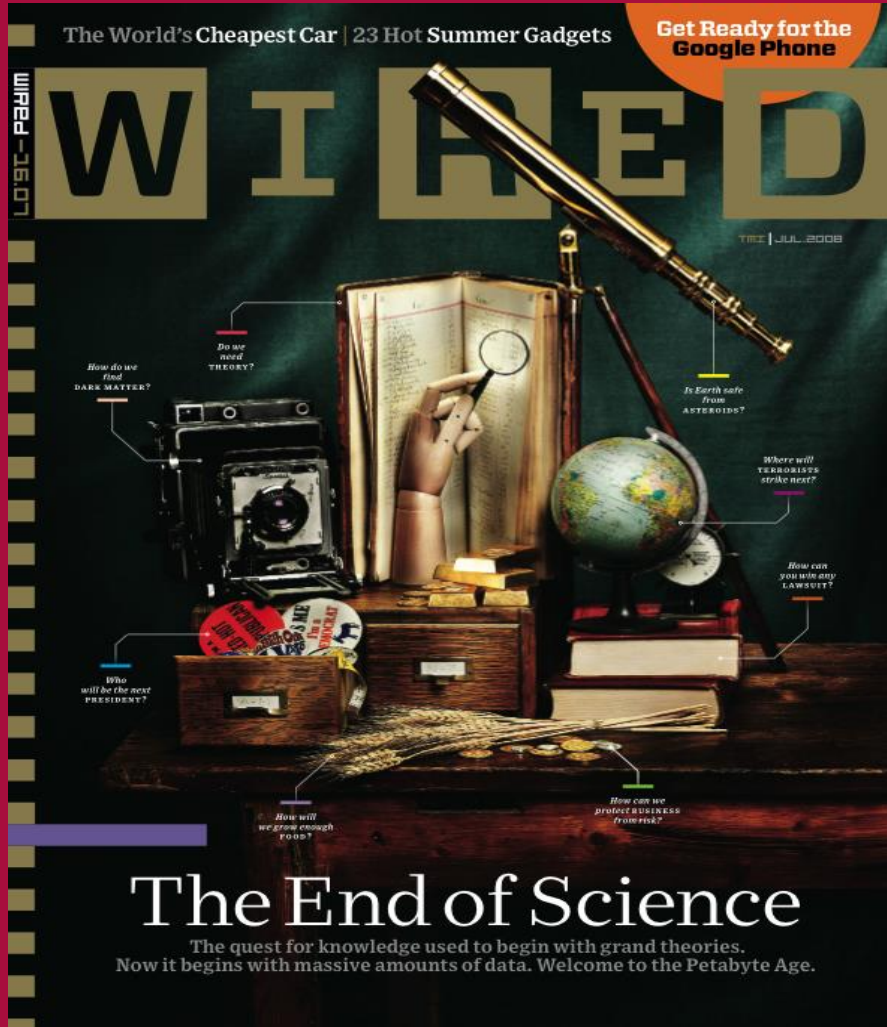


Minority and Health Disparity Issues

- **Data and Information are not Neutral**
 - **Stigma:** People/groups/communities/diagnoses/phenotypes
 - **Inclusion:** Data collected in basic/applied/clinical trial research
 - **Diversity and Workforce** issues
- **Citizen Science** and *community and patient engagement*
- **Inclusion, Equity,** and Data Access – The Haves and Have Nots



Challenges

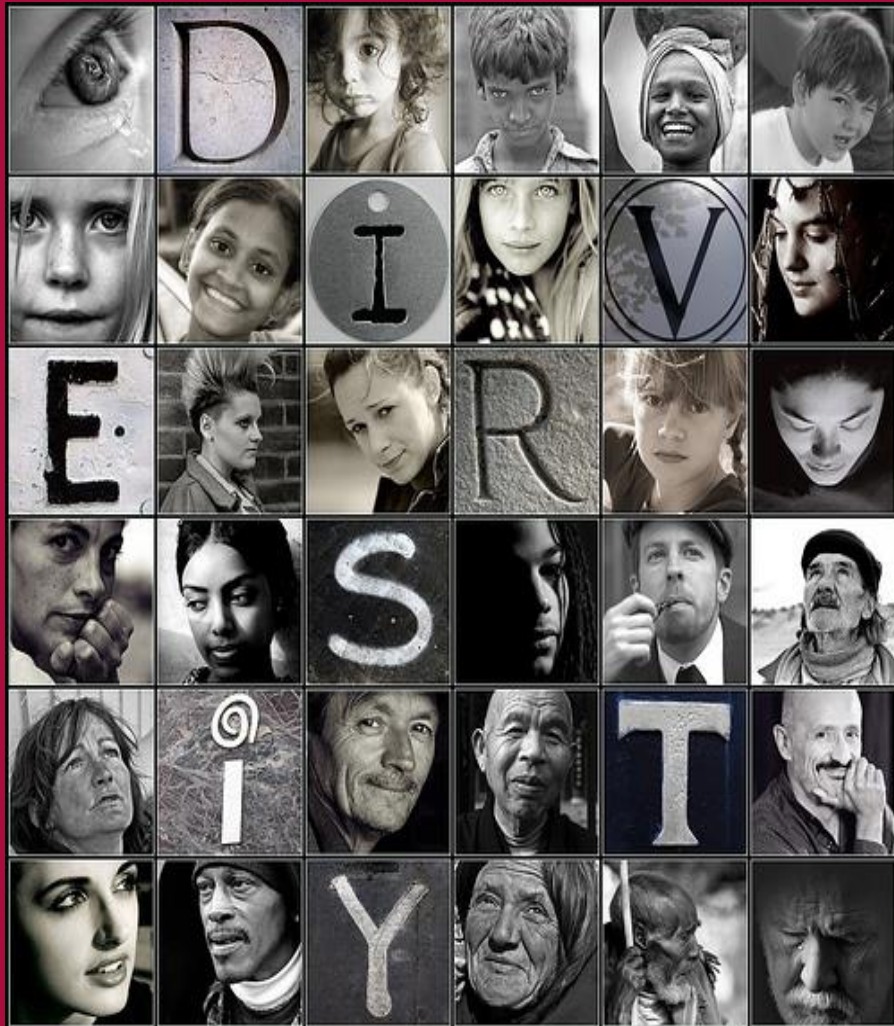


Data are difficult to

- collect
- store
- delete
- search
- share
- visualize
- curate
- process
- analyze

*with current available
databases and tools*

Challenges



Genomics is failing on diversity

An analysis by Alice B. Popejoy and Stephanie M. Fullerton indicates that some populations are still being left behind on the road to precision medicine.

A 2009 analysis revealed that 96% of participants in genome-wide association studies (GWAS) were of European descent¹. Such studies scan the genomes of thousands of people to find variants associated with disease traits. The finding prompted warnings that a much broader range of populations should be investigated² to avoid genomic medicine being of benefit merely to “a privileged few”.

Seven years on, we’ve updated that

analysis. Our findings indicate that the proportion of individuals included in GWAS who are not of European descent has increased to nearly 20%. Much of this rise, however, is a result of more studies being done in Asia on populations of Asian ancestry. The degree to which people of African and Latin American ancestry, Hispanic people and indigenous peoples are represented in GWAS has barely shifted.

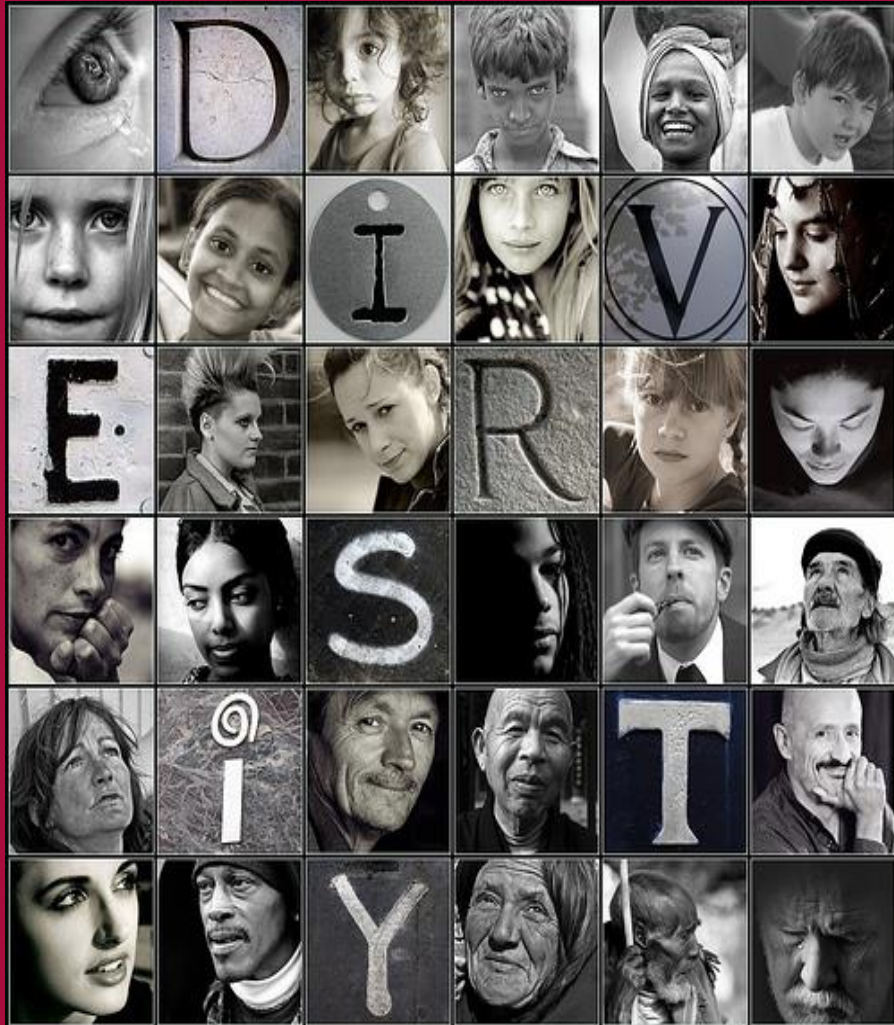
Thus, more than 20 years after the

US National Institutes of Health (NIH) mandated the inclusion of diverse participants in the biomedical research it funds, GWAS funded by the NIH and other sources are continuing to miss a vast portion of the world’s genetic variation.

Over the past decade, GWAS have been the preferred tool for discovering the genetic factors involved in common diseases. Tens of thousands of significant associations between genetic variants and biological traits have ▶

13 OCTOBER 2016 | VOL 538 | NATURE | 161

Challenges



How to have integrity
in [data
and data sharing]
in a world
that does not
affirm everyone's
humanity

- Adapted from Thomas A. Parham

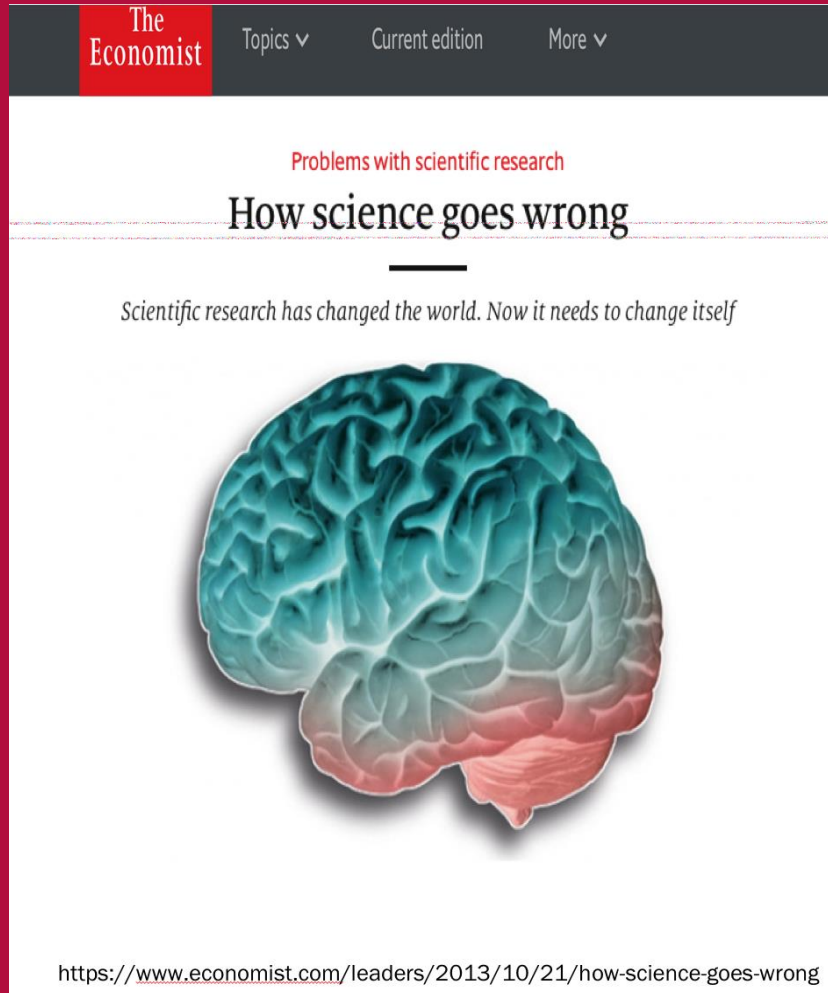
Challenges



Science Isn't Broken

“...it’s just...a lot harder than we give it credit for...”

Challenges



Science Isn't Broken

“...it's just...a lot harder than we give it credit for...”

“Now it needs to change itself...”

The Challenge

PARADIGM
SHIFT

Hypothesis
Confirmation
TO
Hypothesis
Generation

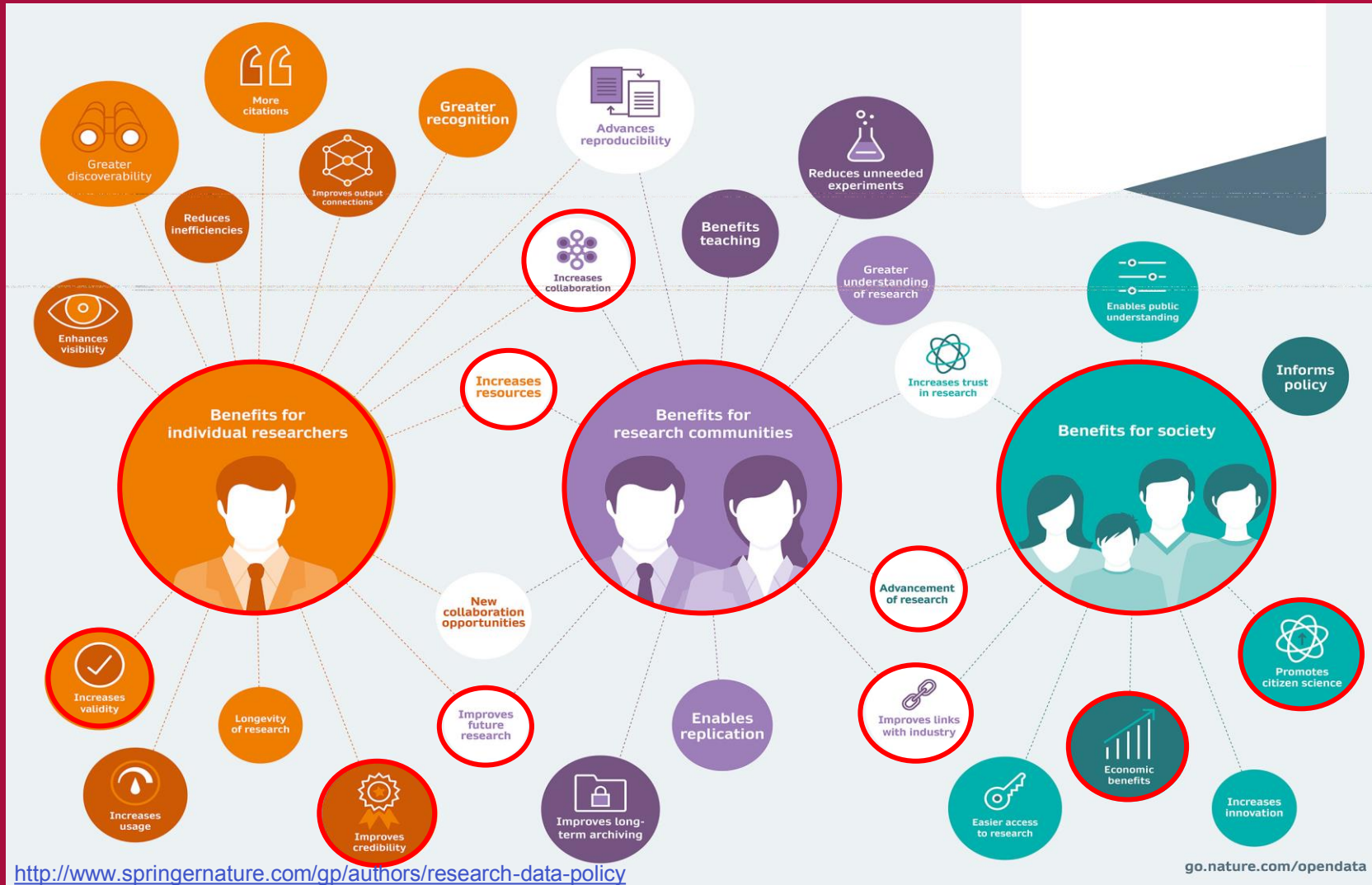
50TH ANNIVERSARY EDITION

THE STRUCTURE OF SCIENTIFIC
REVOLUTIONS

THOMAS S. KUHN

WITH AN INTRODUCTORY ESSAY BY IAN HACKING

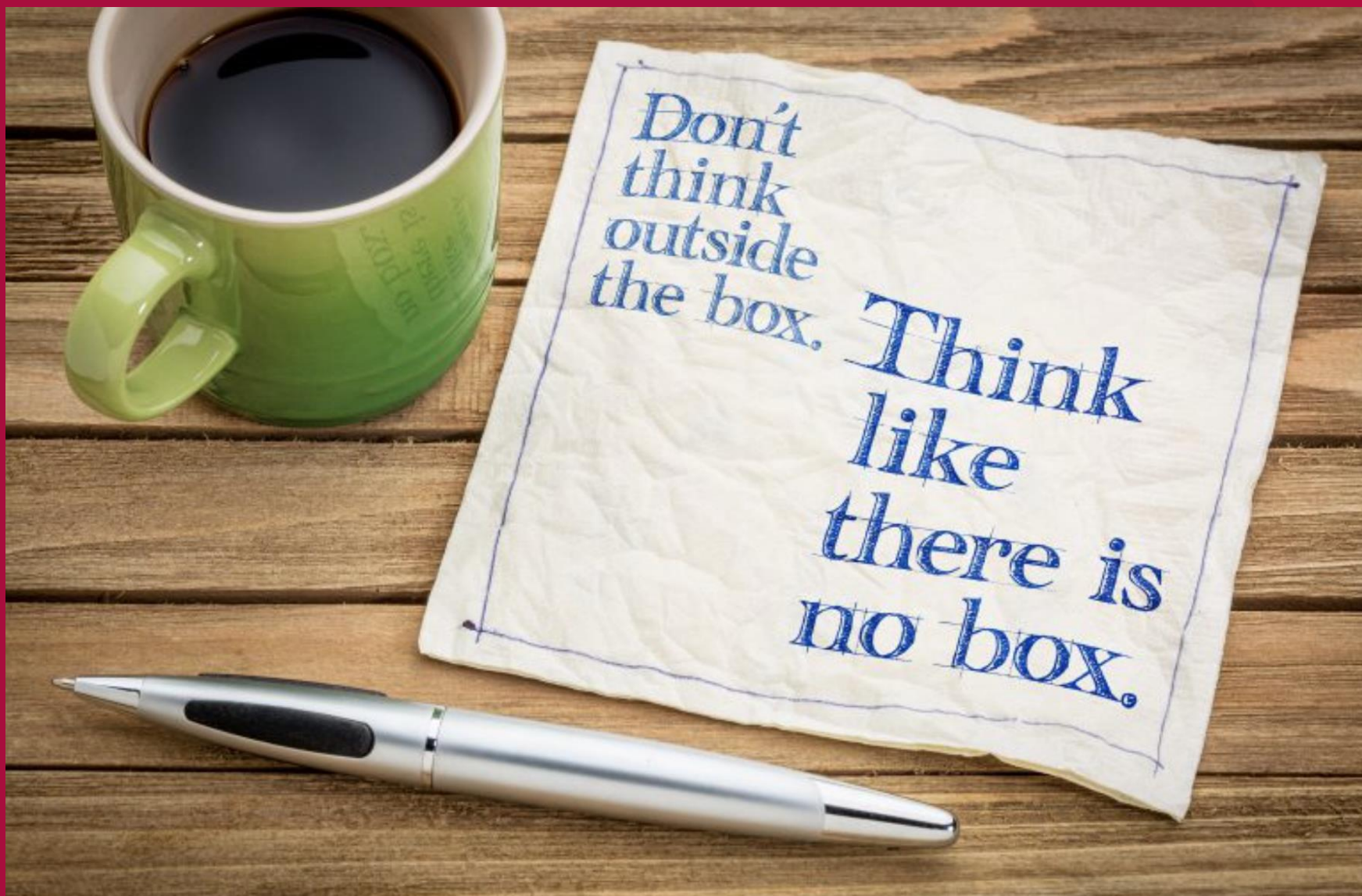
The Challenge



Setting You and your Data Free

- Catalyze new collaborations
- Increase confidence in results
- Larger projects (scope & sample size)
- Local vs global (>external validity)
- Generate greater recognition
- Credit
 - Digital Object Identifier (DOI) enables independent discoverable citability for researcher credit
 - Data tracking on the impact of research
 - Journal data availability statements
 - Open data badges

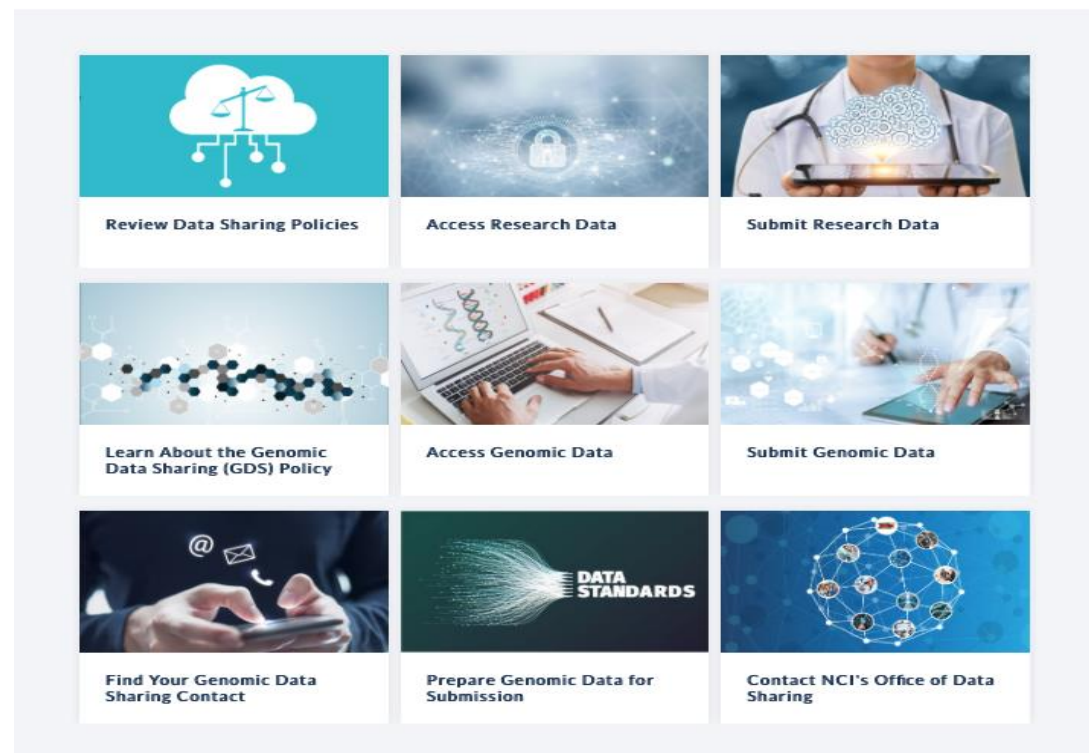




<https://datascience.cancer.gov/data-sharing>



NCI is dedicated to building upon the critical impact sharing data has on accelerating treatment for cancer.





**NATIONAL
CANCER
INSTITUTE**