

Participant perspectives on data sharing: What is important and why?

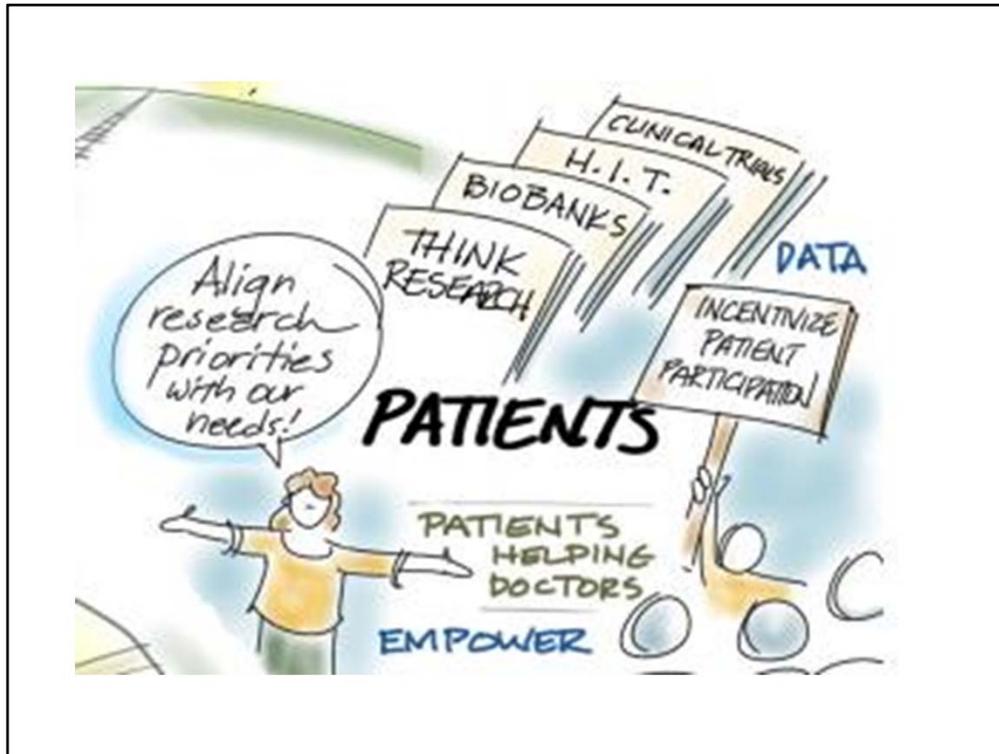
There is a small but growing number of people in this country who understand the power sharing their own data to improve their health. I am one of them.

Today I'm going to speak to you about Participant Perspectives in Data Sharing: What's Important and Why.

But first, my disclosures. I'm not a geneticist or a scientist. I'm not a cancer survivor.



What I am is a Previsor. This is me in 2012 after my prophylactic bilateral mastectomy. I carry a BRCA1 mutation called C61G, and we're lucky enough to have enough data on that mutation to know it's extremely harmful.



This is why data sharing is important to me. But my perspective isn't the only voice in all of this, and I don't speak for all patients.

In preparing for this workshop, I reached out to dozens of patient advocates in multiple communities, and we thought of all sorts of tangible next steps. I have a print-out of recommendations from these patient advocates, which is only a first step in what we hope is a much longer conversation.

I'll talk about 3 key themes emerged when talking to patients:

1. Let patients / participants lead.
2. Give us equal access to data.
3. Practical, useful tools designed by patients for patients.

Let patients lead.

Let's create a Patient advisory board should be part of permanent governance of this Cohort, with voting power guide decisions and funding.

A patient advisory board is nothing new. Who gets to speak for patients?
And questions we have including:

How do we use an advisory board to work through a thoughtful design process?

How do we LET participants ASK RESEARCH QUESTIONS?

How do we work with genetic counselors and doctors to share what we learn?

Take look at great working models where patients are doing great things – like the Stanford Medicine X program, and the Department of Defense Breast Cancer Research Program.

It's clear that patients are ready and willing to lead.



<<Patients are an UNDERUTILIZED RESOURCE!!>>

Emily Kramer-Golinkoff – who is creating an innovative network of Cystic Fibrosis families who are building a drug discovery roadmap for rare CF mutations. She is working with scientists like Dr. Allan Berkman UCSF to accelerate the pace of drug discovery.

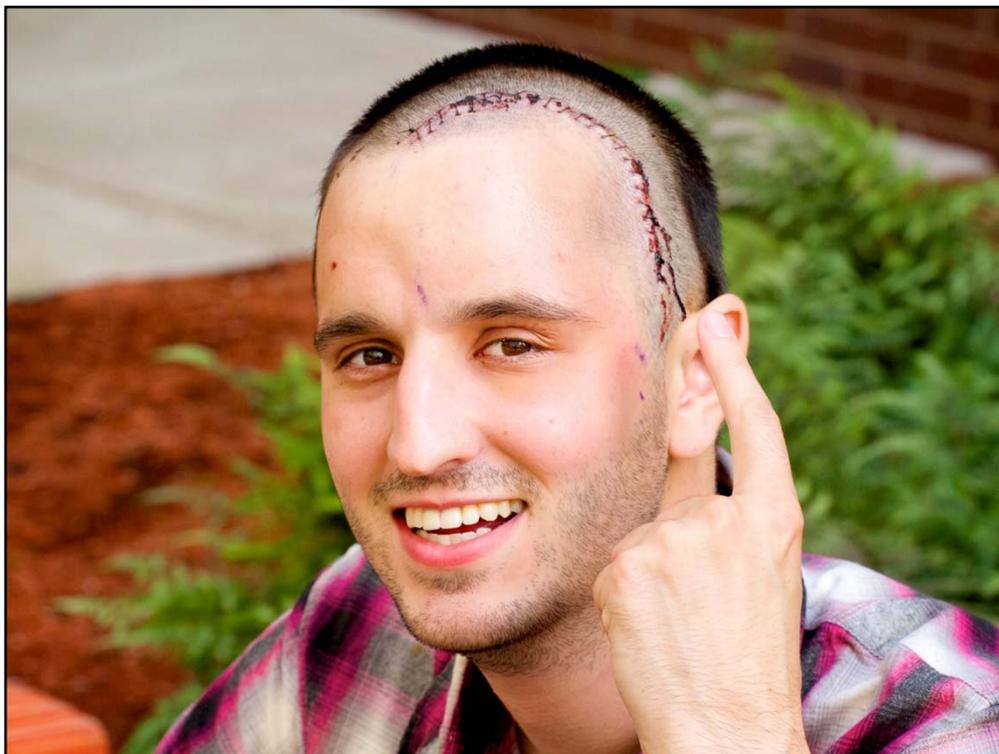
Participants need equal access to
the data.

Transparency/Accountability, Equal Access & Autonomy for Patients.

How can this Cohort ensure that grantees are following ethical data sharing practices? How do we define those practices?

How can you give participants raw data to share with whom we choose.

But here is an example of why we need equal access to the data.



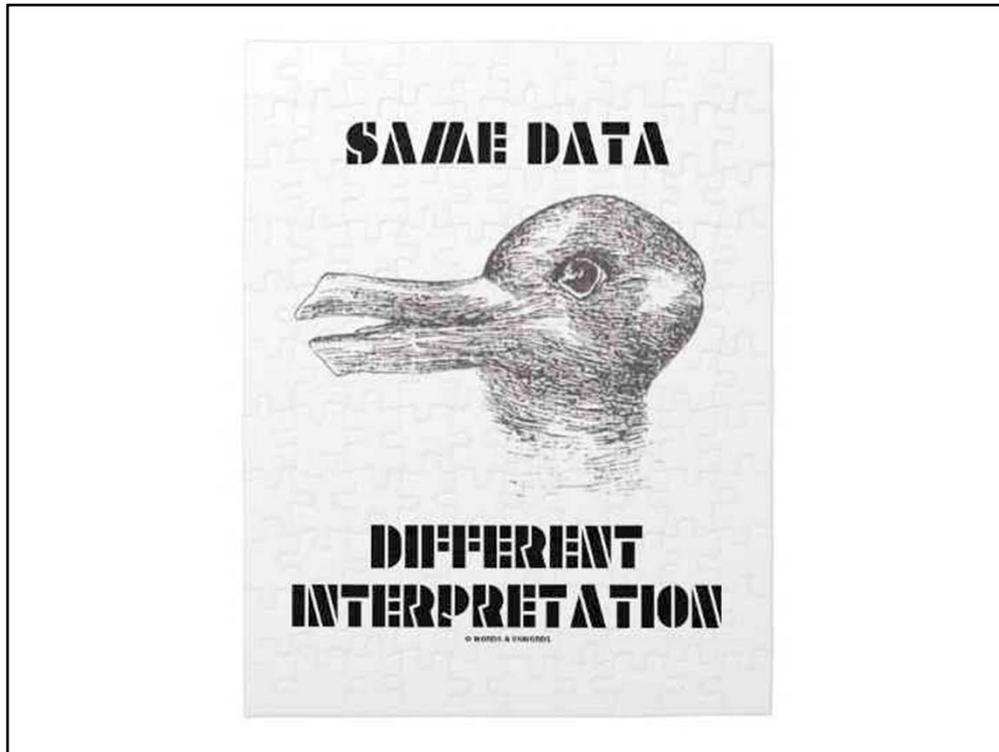
Let me give you the example of Steven Keating, a phd candidate at MIT and an astrocytoma patient, whose access to his own data ultimately saved his life when he discovered that he had a 12cm brain tumor.



Steven shared his own genetic data and research imaging to make informed decision about his own treatment options, to find the right clinical trials, and to guide decisions about surgery.

Not only did access to data fight the tumor, it enabled steven to create a 3D print of his brain tumor so that he could advocate for research in important new ways.

Education & well-designed tools.



We need better ways to bridge the literacy gap for these data. Better tools and training – for patients, for doctors, and for genetic counselors.

How do we visualize data in a way that's accessible and useful to doctors and patients?
How do we minimize mistakes in interpretation through these tools?

How do we train medical professionals to use these tools in a way that scale?

Two examples...

NCBI Resources How To Sign in to NCBI

ClinVar Search ClinVar for gene symbols, HGVS expressions, conditions, and more Search Help

Home About Data use and maintenance Using the website How to submit Statistics FTP site

NM_007294.3(BRCA1):c.181T>G (p.Cys61Gly)

NM_007294.3(BRCA1):c.181T>G (p.Cys61Gly) Go to: Help

Clinical significance
 NM_007294.3(BRCA1):c.181T>G (p.Cys61Gly)
 Clinical significance: Pathogenic/Likely pathogenic
 Review status: ★ ★ ★ ★
 Number of submission(s): 5

Condition(s)
 Breast-ovarian cancer, familial 1 [MedGen - Orphanet - OMIM]
 BRCA1 and BRCA2 Hereditary Breast and Ovarian Cancer [MedGen]
 Neoplastic Syndromes, Hereditary [MedGen]

See supporting ClinVar records

1 Affected Gene

breast cancer 1, early onset (BRCA1) [Gene - OMIM - Variation viewer]
 Search ClinVar for variants within BRCA1
 Search ClinVar for variants including BRCA1

Browser views

RefSeqGene
 Variation viewer [GRCh38 - GRCh37]
 UCSC [GRCh38/hg38 - GRCh37/hg19]

Variant type: single nucleotide variant

Cytogenetic location: 17q21.3

Genomic location: Chr17:41258504 (on Assembly GRCh37)
 Chr17:43106487 (on Assembly GRCh38)

Other names: 300T>G

Protein change: C61G

HGVS: NG_005905.2:g.111497T>G
 NM_007294.3:c.181T>G
 U14680.1:n.300T>G [...more](#)

Links: OMIM: 113705.0002
 dbSNP: 28897672

NCBI 1000 Genomes Browser: [rs28897672](#)

Molecular consequence: NM_007294.3:c.181T>G: missense variant
 SO:0001583
 NR_027676.1:n.342T>G: non-coding transcript variant
 SO:0001619

This is my mutation in ClinVar. Most patients and doctors don't know what Clinvar does, and most doctors have no idea how to use this tool. Oh and the data is incomplete because companies don't share their variants.



My world has certainly changed a lot in these few short years. This is me at the steps of the Supreme Court in 2013 telling my story about why human genes shouldn't be patented by the company that offered me genetic testing. The world has already changed by leaps and bounds since this day.

Now that the patents are overturned, I want smarter options to screen for, treat, and prevent cancer. I want a roadmap to get there.

Data can heal. Patients should be given the choice and tools to easily access/share/contribute data.

My data are not an abstraction to me. These data represent generations of suffering within my family, and I want to be a part of the solution to alleviate that suffering for my children.

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