US Perspective on Advances for Rare Diseases

ICORD
Ede, Netherlands
9 October 2014

Sharon F. Terry, President & CEO

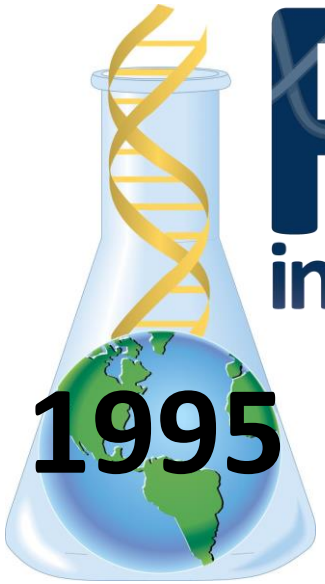
Elizabeth and Ian diagnosed with pseudoxanthoma elasticum (PXE)

1994

2014

Elizabeth:
Teach for America

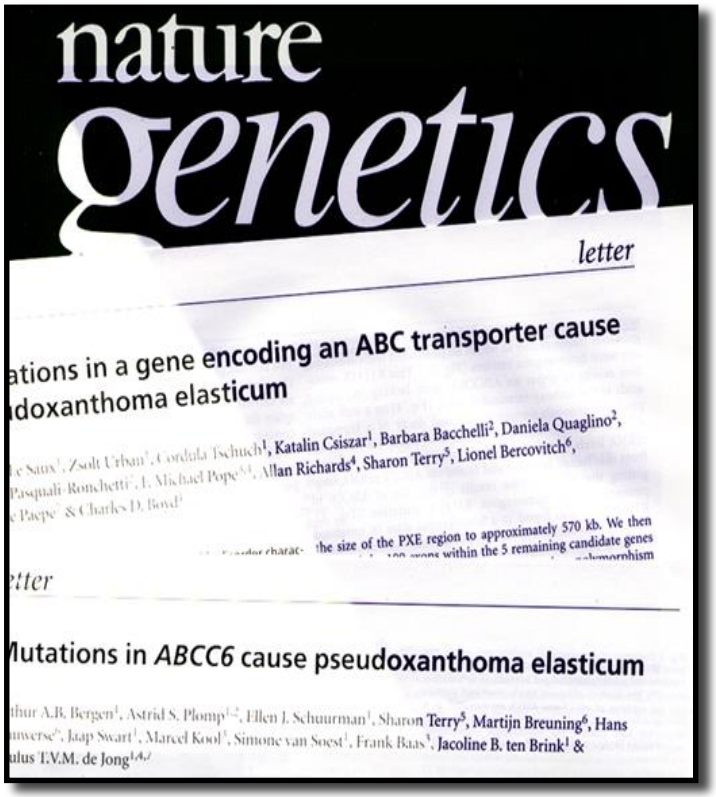
Ian:
Organic Farmer



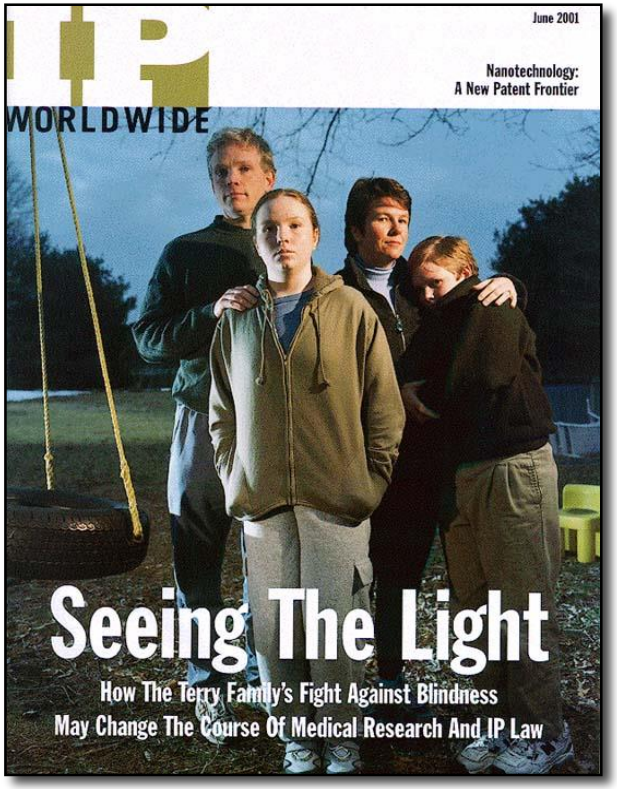
1995

PXE international

Gene Discovery 1999



BioBank



Testing

Clinical Diagnostic Test Development via FDA & CLIA Regulatory Strategies

2000

Patenting

Licensing & Intellectual Property Management

Human Clinical Trials

2012

Drug Screening & Development Approaches

Therapeutics

--Small Molecules

--Nonsense mutants

Maturing Disease Advocacy

- **1950s-1960s** – **Medical Models**
  - Voluntary Health Agencies
- **1970s** – **Nascent Patient Movement** – ***Missing Services***
  - Self-organized Disease Specific Organizations
- **1980s** – **Maturing Patient Movement** – ***IS & IT Technology***
  - New Alliances and New Strategies Emerge, industry alliances
- **1990s** – **Powerful Momentum "Patient Power"** – ***Websites & Email***
  - Institutionalized Advocacy Coalitions
  - Patient Organized Networked Research Organizations
  - Effecting Broad Change of Public Policy
- **2000s** – **Successful Models "Research Advocacy"** – ***BioBanks***
  - Active Engagement in the Research Enterprise
  - Breaking Conventional Boundaries of the Medical Model
  - Demand for Quality, Services, Choice, & Personalized Delivery
  - Patient Rights Public Policy – Changing the Status-Quo
- **2010s** – **Networks in the Commons** – ***Translation & Delivery***

Genetic Alliance engages individuals, families, and communities to transform health.

2003: Registry and BioBank founded

PCORnet: the National Patient-Centered Clinical Research Network

The **goal** of PCORI's National Patient-Centered Clinical Research Network Program is to **improve the nation's capacity to conduct CER** efficiently, by **creating a large, highly representative, national patient-centered clinical research network** for conducting clinical outcomes research.

$1.26 Billion

The **vision** is to support a learning US healthcare system, which would allow for **large-scale research** to be conducted with **enhanced accuracy and efficiency**.

PCORnet: "The National Patient-Centered Clinical Research Network"

- 1000 researchers, traditional and lay
- 29 funded entities covering all 50 states
- Focus on patient-centered outcomes research
- No "one size fits all"

- Community Engaged Network for All (CENA)
- 9 disease-specific advocacy organizations, UCSF, UCD
- From hepatitis (affects millions) to Alström syndrome (affects a several hundred)

Clinical Data Research Network's Disease Cohorts

Organization	Common Cohort	Rare Cohort
ADVANCE	Diabetes	Co-infection with HIV and hepatitis C virus
CAPriCORN	Anemia; Asthma	Sickle cell disease; Recurrent C. Difficile colitis
Great Plains Collaborative	Breast Cancer	Amyotrophic Lateral Sclerosis (ALS)
Louisiana Clinical Data Research Network	Diabetes	Sickle Cell Disease, Rare Cancers
NYC-CDRN	Diabetes	Cystic fibrosis
Mid-South CDRN	Coronary Heart Disease (CHD)	Sickle Cell Disease (SCD)
PEDSNet	Inflammatory bowel disease	Hypoplastic left heart syndrome
PORTAL	Colorectal Cancer	Severe Congenital Heart Disease
pSCANNER	Congestive Heart Failure	Kawasaki Disease
P2ATH	Atrial Fibrillation	Idiopathic Pulmonary Fibrosis
SCIHLS	Osteoarthritis	Pulmonary arterial hypertension

Patient Powered Research Networks span a range of conditions

Organization	PI	Condition	Proposed PPRN Population Size
Accelerated Cure Project for Multiple Sclerosis	Robert McBurney	Multiple Sclerosis	20,000
American Sleep Apnea Association	Susan Redline	Sleep Apnea	50,000
Cincinnati Children's Hospital Medical Center	Peter Margolis	Pediatric Crohn's Disease and Ulcerative Colitis	15,000
COPD Foundation	Richard Mularski	Chronic Obstructive Pulmonary Disease	50,000
Crohn's and Colitis Foundation of America	R. Balfour Sartor	Inflammatory Bowel Disease (Crohn's disease and ulcerative colitis)	30,000
Global Healthy Living Foundation	Seth Ginsberg	Arthritis (rheumatoid arthritis, spondyloarthritis), musculoskeletal disorders (osteoporosis), and inflammatory conditions (psoriasis)	50,000
Massachusetts General Hospital	Andrew Nierenberg	Major Depressive Disorder (MDD) and Bipolar Disorder (BP)	50,000
Univ of California, San Francisco	Mark Pletcher	Cardiovascular health	100,000
University of South Florida	Rebecca Sutphen	Hereditary Breast and Ovarian Cancer (HBOC)	17,000

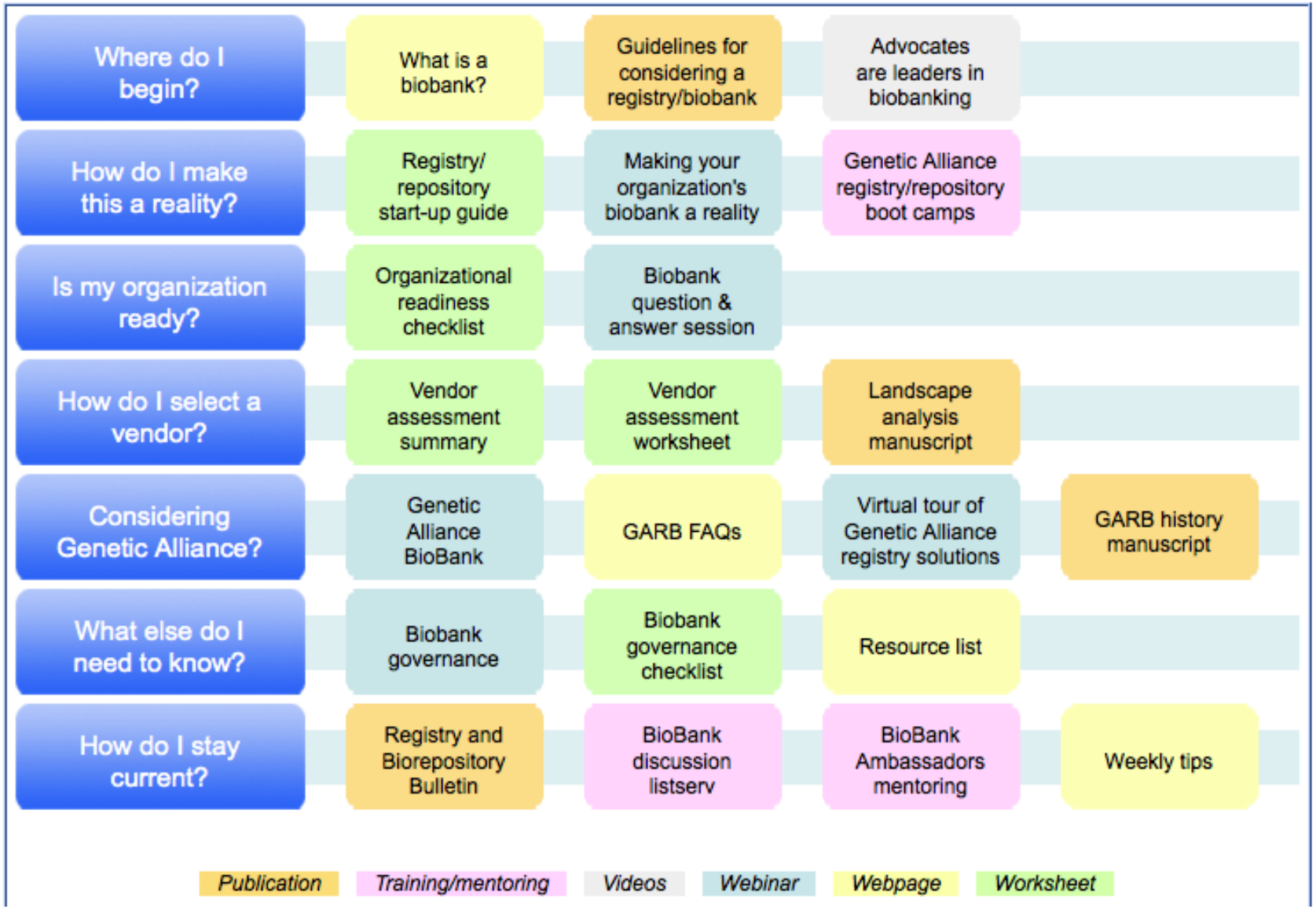
….in both rare and common disorders

Organization	PI	Condition	Proposed PPRN Population Size
ALD Connect, Inc	Florian Eichler	Adrenoleukodystrophy	3,000
Arbor Research Collaborative for Health	Bruce Robinson	Primary Nephrotic Syndrome (Focal Segmental Glomerulosclerosis [FSGS], Minimal Change Disease [MCD], and Membranous Nephropathy [MN] Multiple Sclerosis	1,250
Duke University	Laura Schanberg	Juvenile Rheumatic Disease	9,000
Epilepsy Foundation	Janice Beulow	Aicardi Syndrome, Lennox-Gastaut Syndrome, Phelan-McDermid Syndrome, Hypothalamic Hamartoma, Dravet Syndrome, and Tuberous Sclerosis	1,500
Genetic Alliance, Inc	Sharon Terry	Alström syndrome , Dyskeratosis congenital, Gaucher disease, Hepatitis, Inflammatory breast cancer, Joubert syndrome, Klinefelter syndrome and associated conditions, Metachromatic leukodystrophy, Pseudoxanthoma elasticum (PXE)	50- 50,000
Immune Deficiency Foundation	Kathleen Sullivan	Primary Immunodeficiency Diseases	1,250
Parent Project Muscular Dystrophy	Holly Peay	Duchenne and Becker muscular dystrophy	4,000
Phelan-McDermid Syndrome Foundation	Megan O'Boyle	Phelan-McDermid Syndrome	737
University of Pennsylvania	Peter Merkel	Vasculitis	500

‘Registry and BioBank in a Box’

- Add water and serve
- Deliver ‘white label product’
- Deliver technical assistance
- Cooperative – learn from each other
- Low cost and driving lower
- Community based and local trusted entities’
- Global standards and rigor

Genetic Alliance Registry and BioBank Toolbox



CENA DAO Partners

- Alström Syndrome International
- Dyskeratosis Congenita Outreach
- Inflammatory Breast Cancer Research Foundation
- Hepatitis Foundation International
- Joubert Syndrome Foundation
- KS&A
- MLD Foundation
- National Gaucher Foundation
- PXE International

Can we connect without adding friction to people's lives?

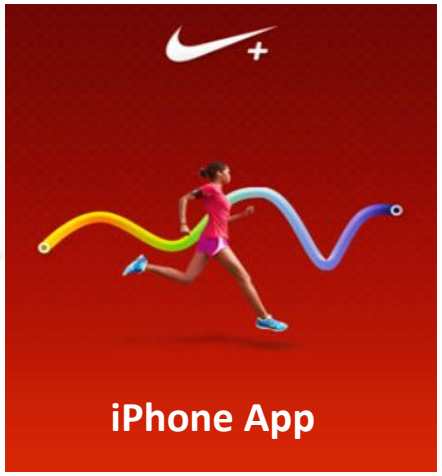
23andMe

FitBit ®

iPhone App
daily burn

Microsoft HealthVault

Twitter
Facebook



Wi Fi

iMapMyRUN+ iMapMyRIDE+ iMapMyWALK iMapMyFITNESS iMapMyHIKE

Needles in Haystacks

The haystack is made of needles…

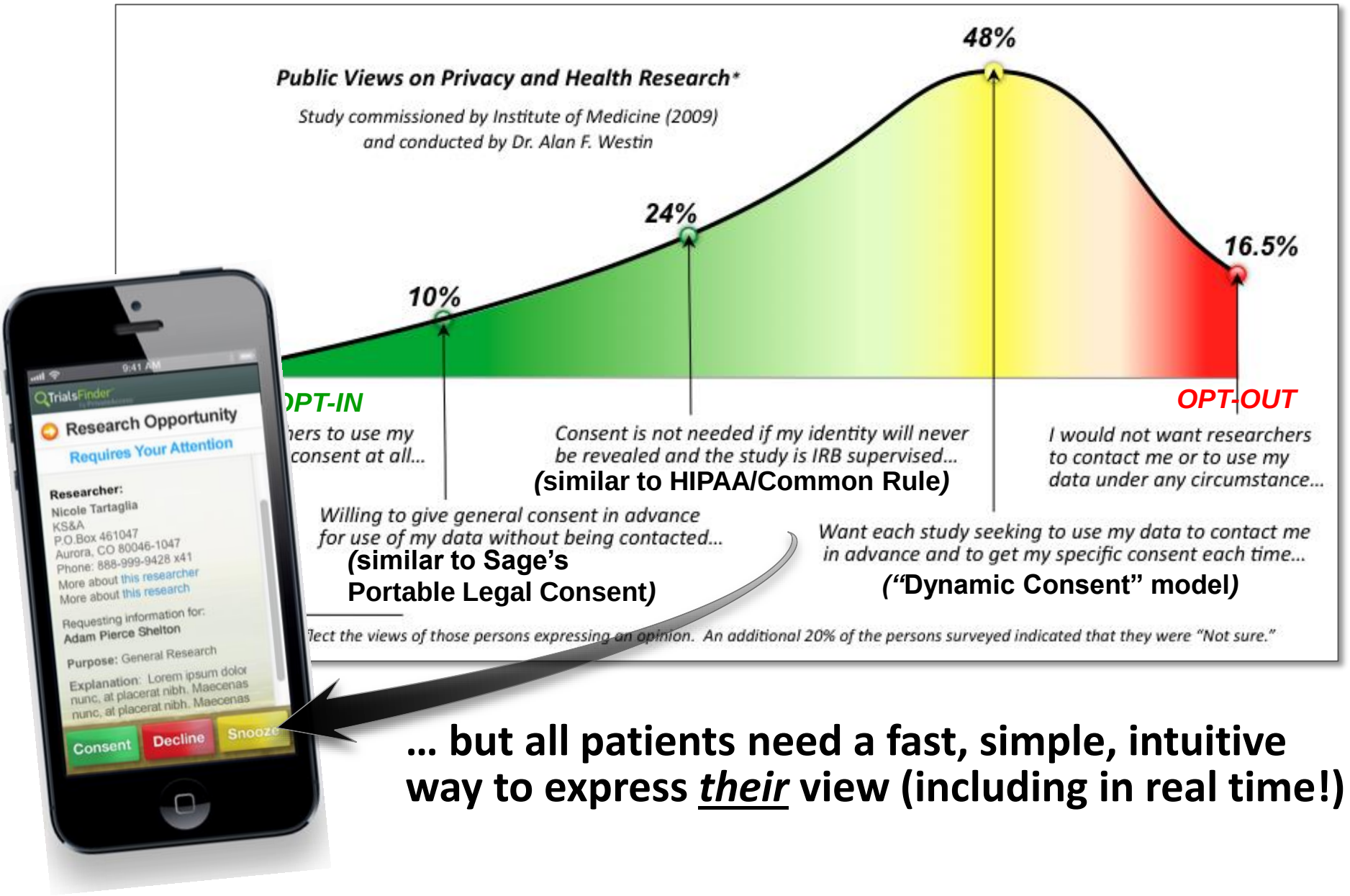
Research Enterprise

…current methods of informed consent are challenged...

granular and dynamic engagement

not consent
not a transaction
not binary
not tiered

Because people have vastly different views about privacy and sharing

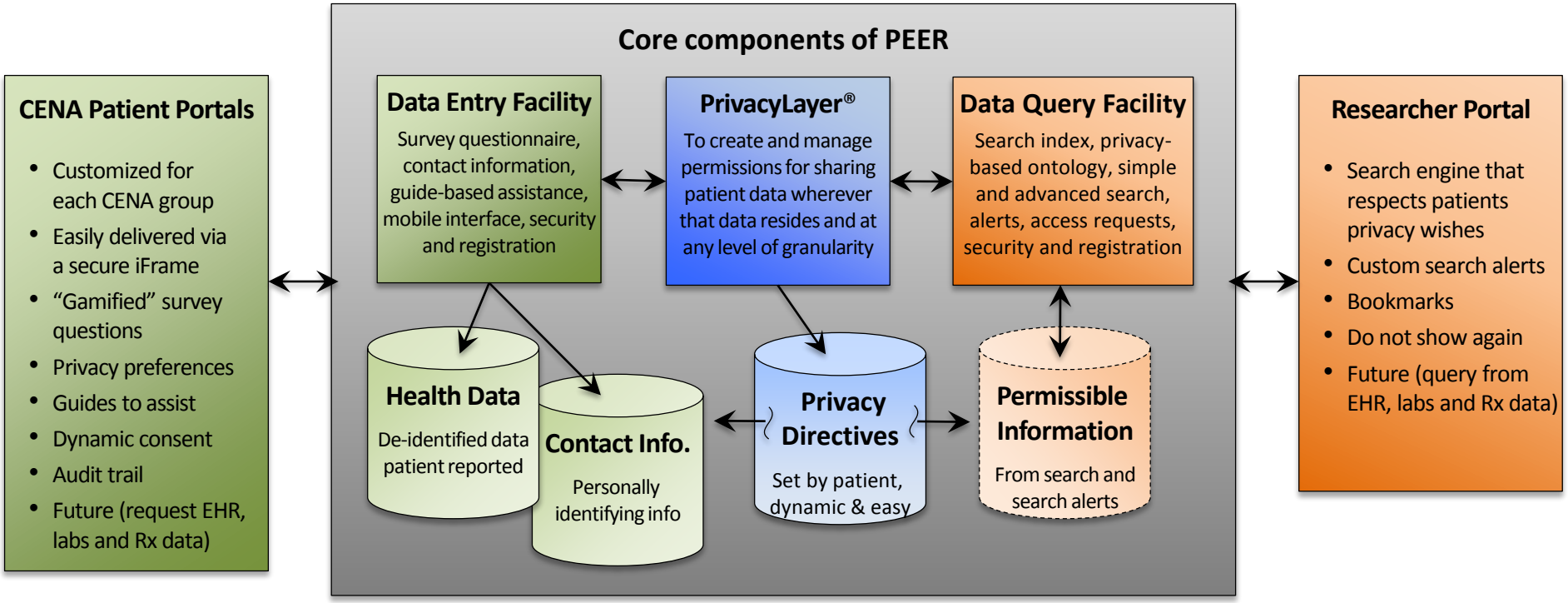


… but all patients need a fast, simple, intuitive way to express *their* view (including in real time!)

High level architecture of the PEER Platform

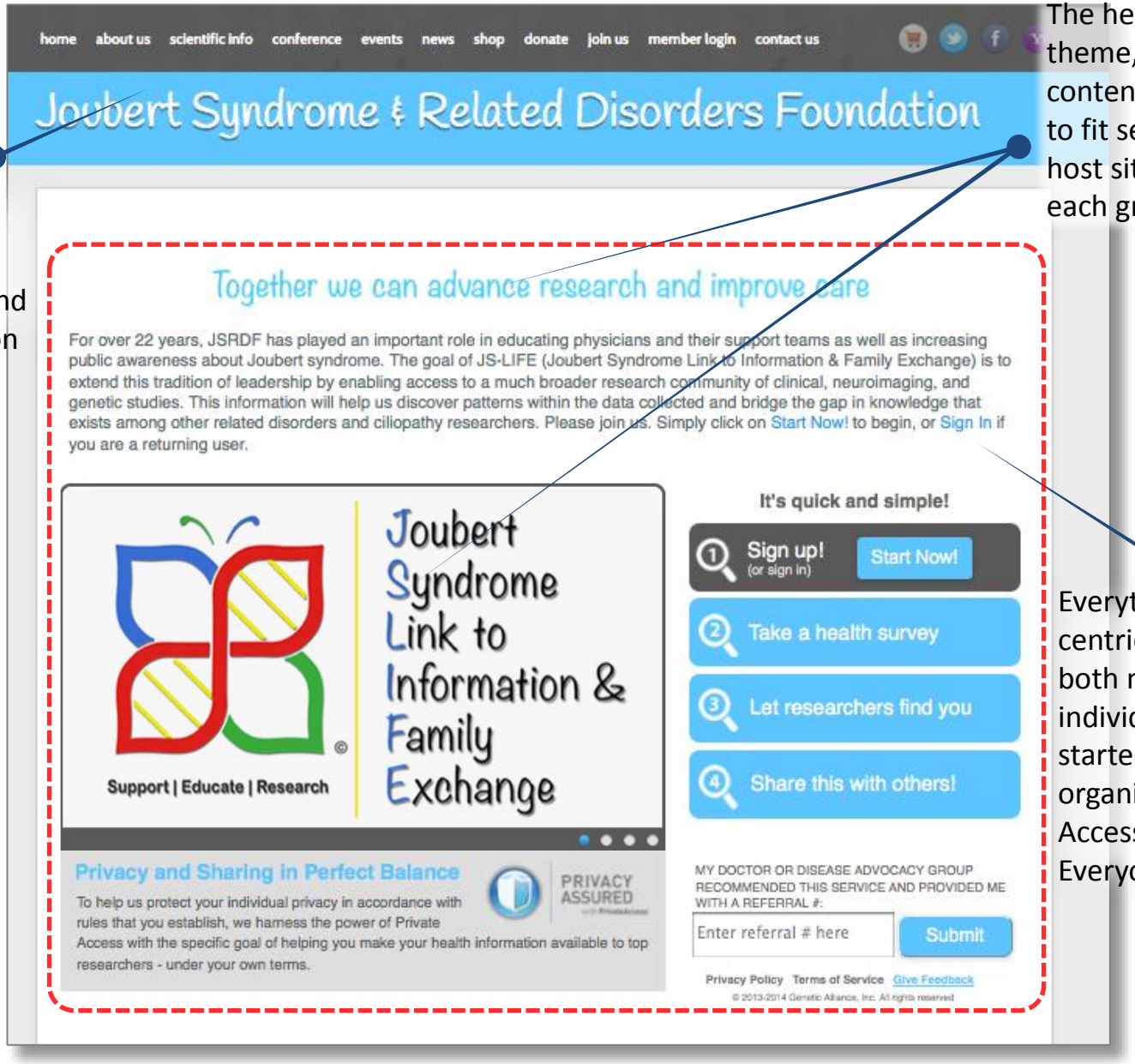
CENA is powered by the PEER (**P**latform for **E**ngaging **E**veryone **R**esponsibly) Network for medical research, developed by Genetic Alliance, a 28-year old consumer health advocacy non-profit, and Private Access, Inc.

The **P**latform for **E**ngaging **E**veryone **R**esponsibly
in Medical Research for all Disease Areas



How **the PEER portal looks is entirely up to** ***where*** **it's located**

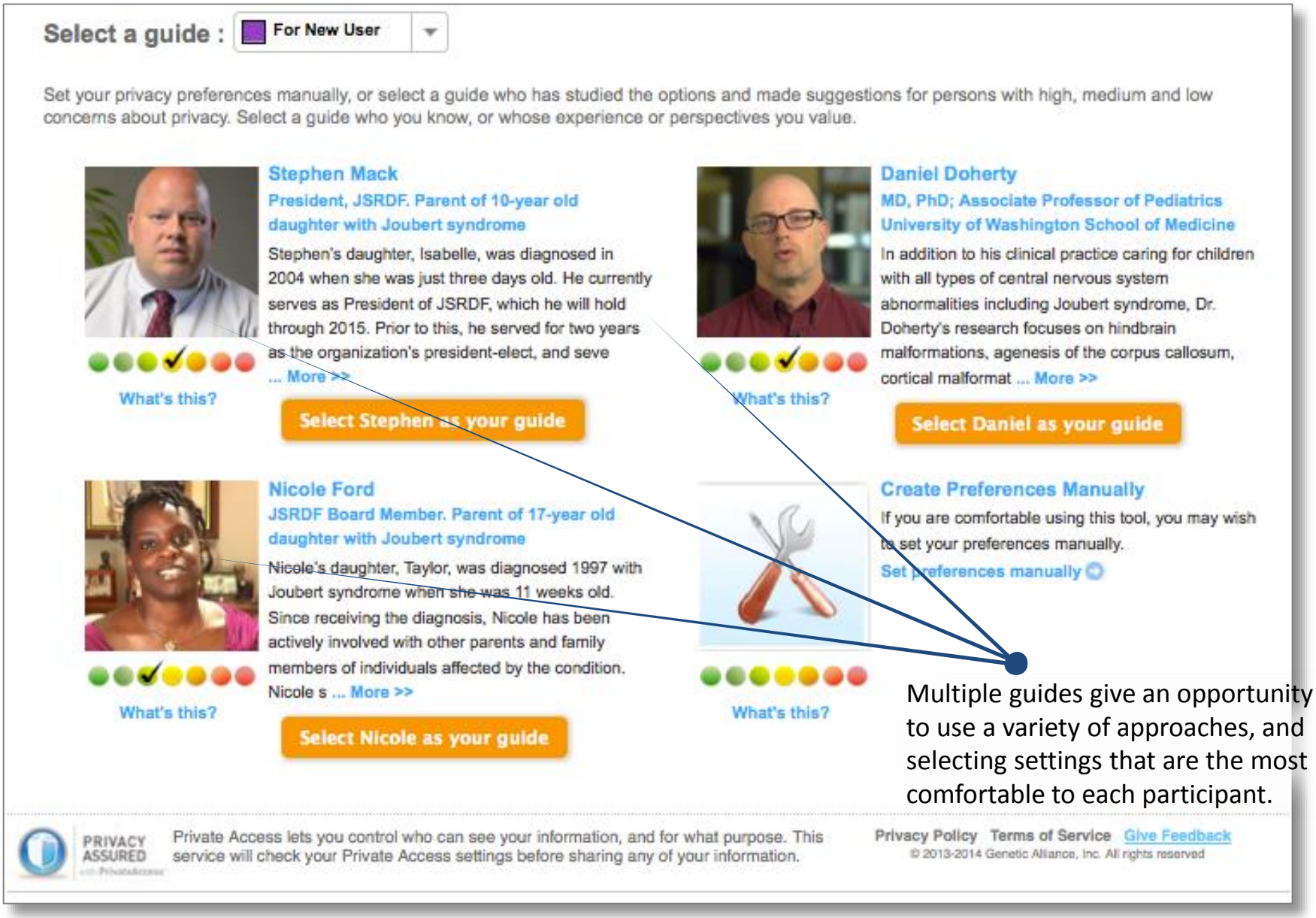
The portal fits directly onto any web page and retains the top and bottom navigation



The headline, colors, theme, video and other content can be tailored to fit seamlessly into the host site and support each group's message.

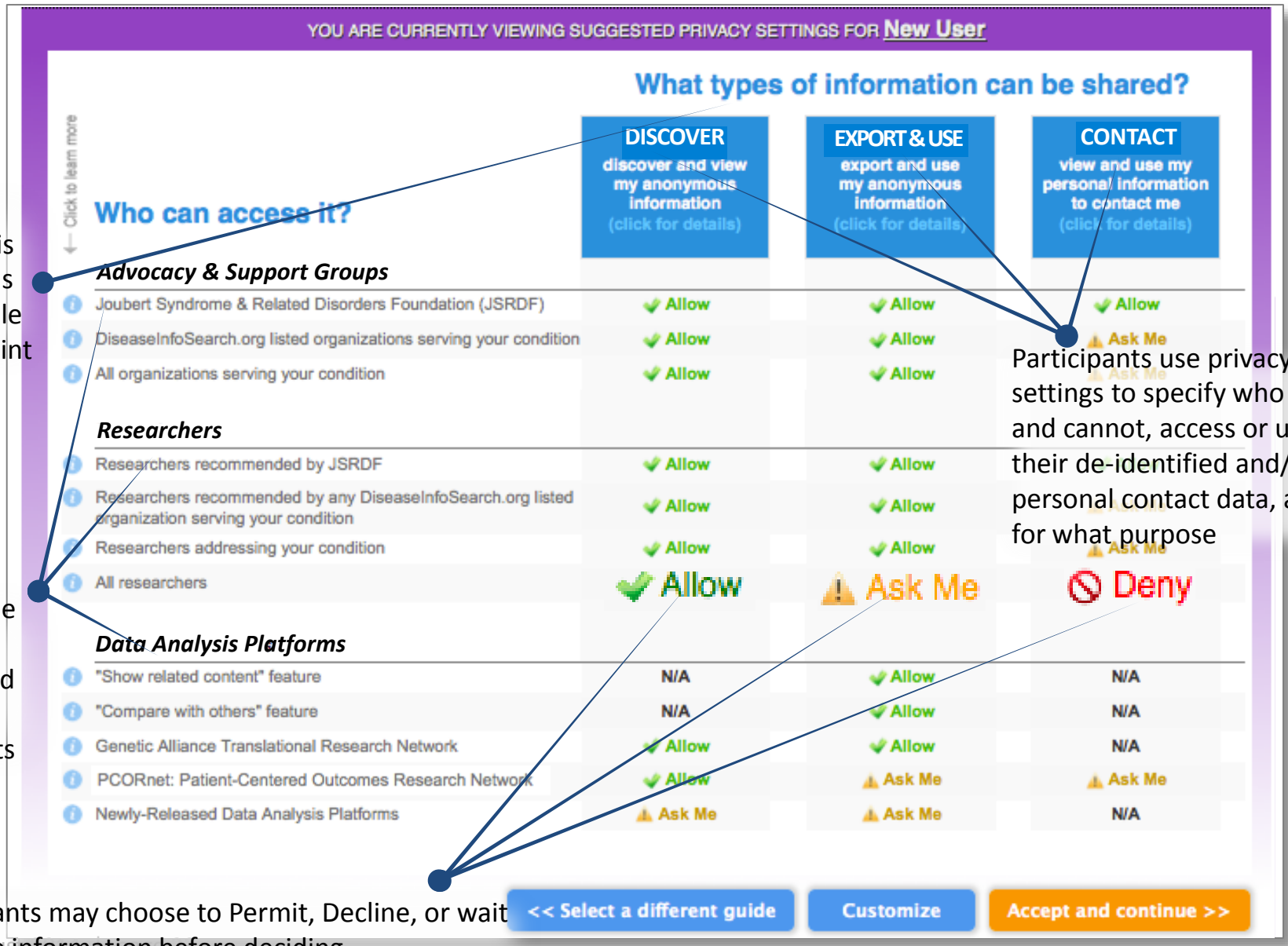
Everything is patient-centric, and it supports both new users as well as individuals who may have started on a different organization's Private Access-enabled site. Everyone benefits!

Users are assisted by highly intuitive, non-coercive “guides”



Multiple guides give an opportunity to use a variety of approaches, and selecting settings that are the most comfortable to each participant.

To enable ease *and* an extraordinary range of granularity



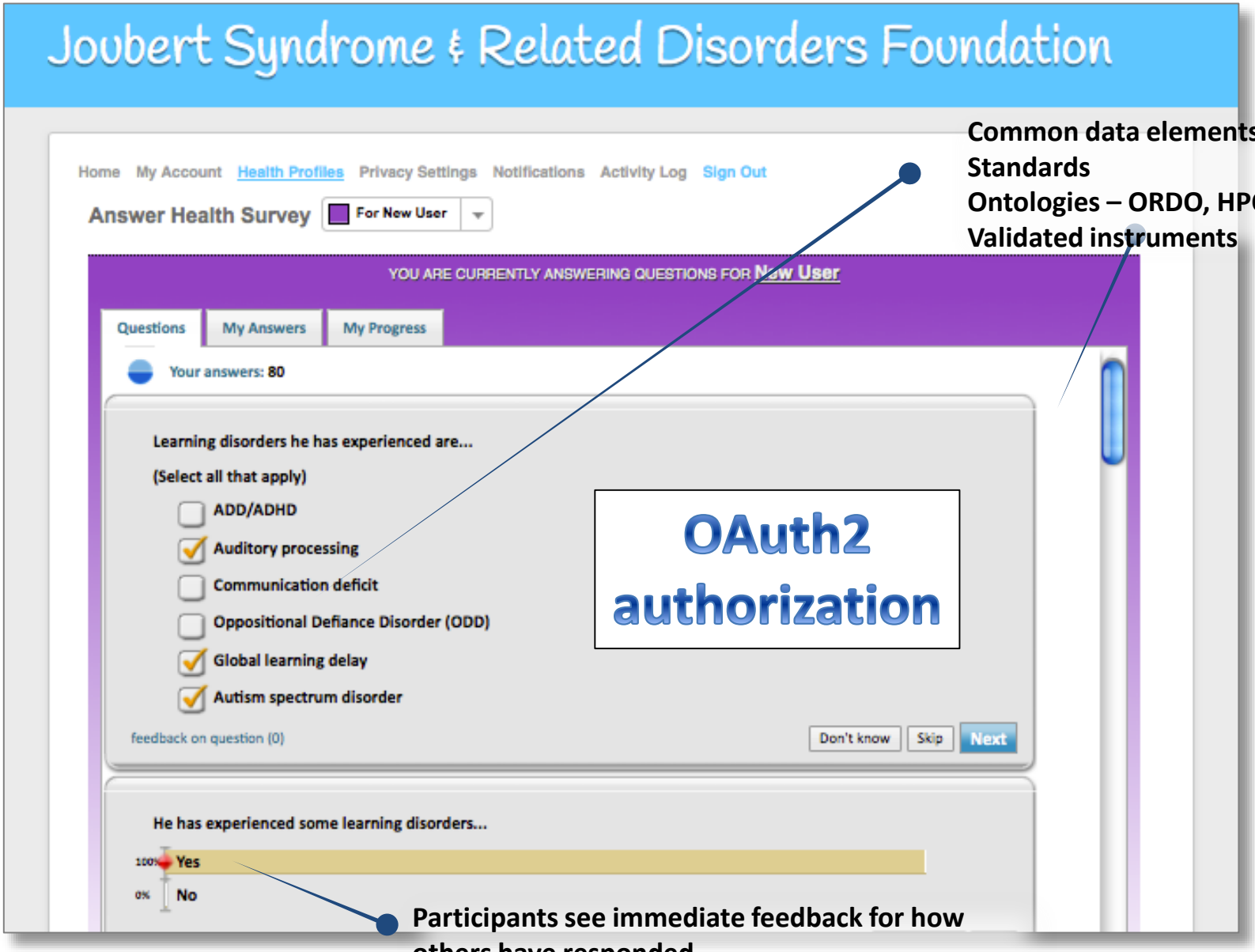
Each guide suggests his or her ideas as a possible starting point

For multiple categories of uses, and specified usage rights

Participants use privacy settings to specify who can, and cannot, access or use their de-identified and/or personal contact data, and for what purpose

Participants may choose to Permit, Decline, or wait for more information before deciding

Gamified questionnaire with easily customized topics



Beginning in October 2014… Longitudinal data and automated reminders

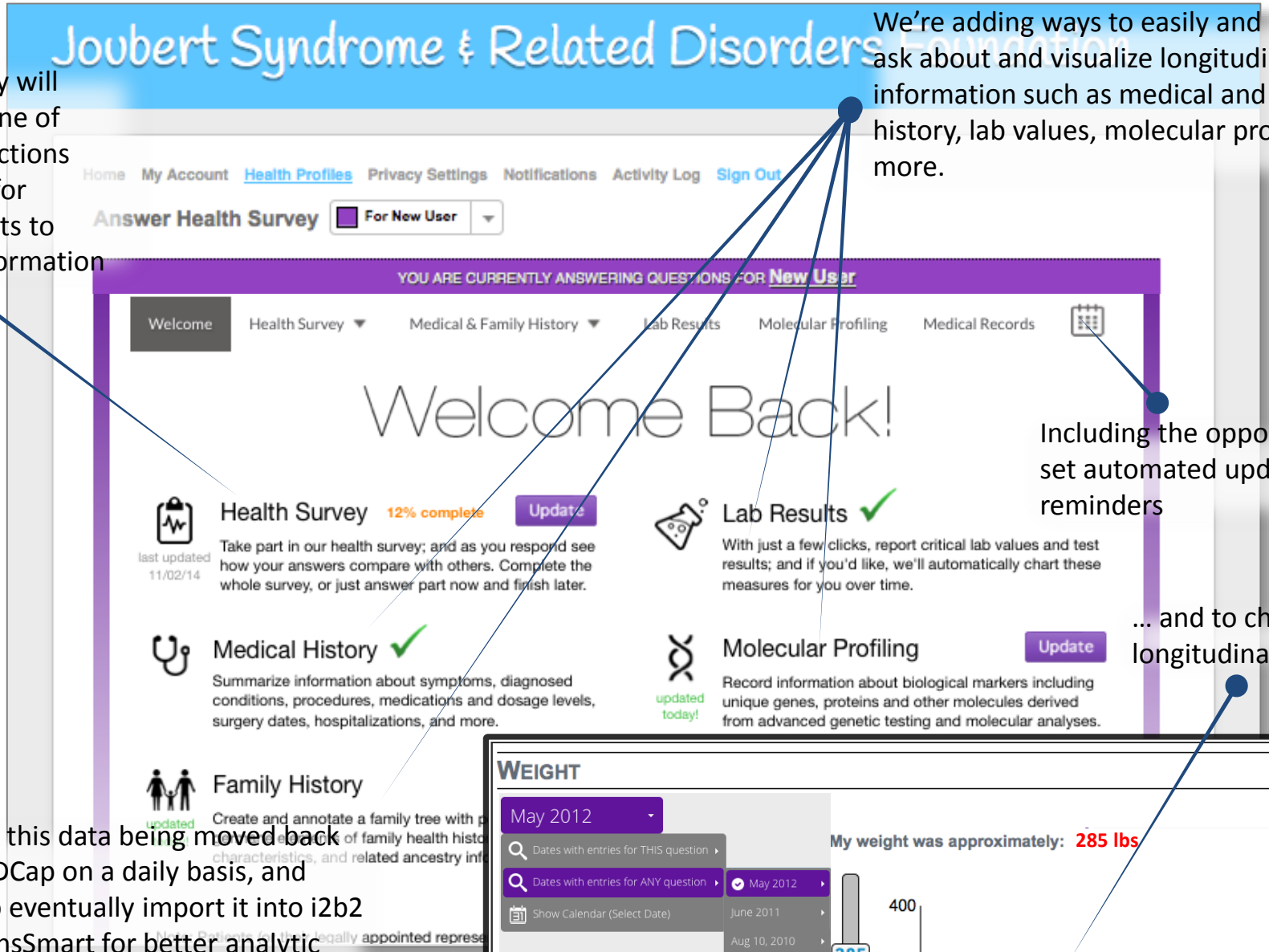
The survey will become one of several sections available for participants to report information

We're adding ways to easily and intuitively ask about and visualize longitudinal information such as medical and family history, lab values, molecular profiles, and more.

Including the opportunity to set automated update reminders

… and to chart longitudinal results

With all this data being moved back into REDCap on a daily basis, and plans to eventually import it into i2b2 and TransSmart for better analytic tools.



Each DAO creates & manages their customized PEER

Select and edit each element in the PEER portal to appear in the theme of its current website

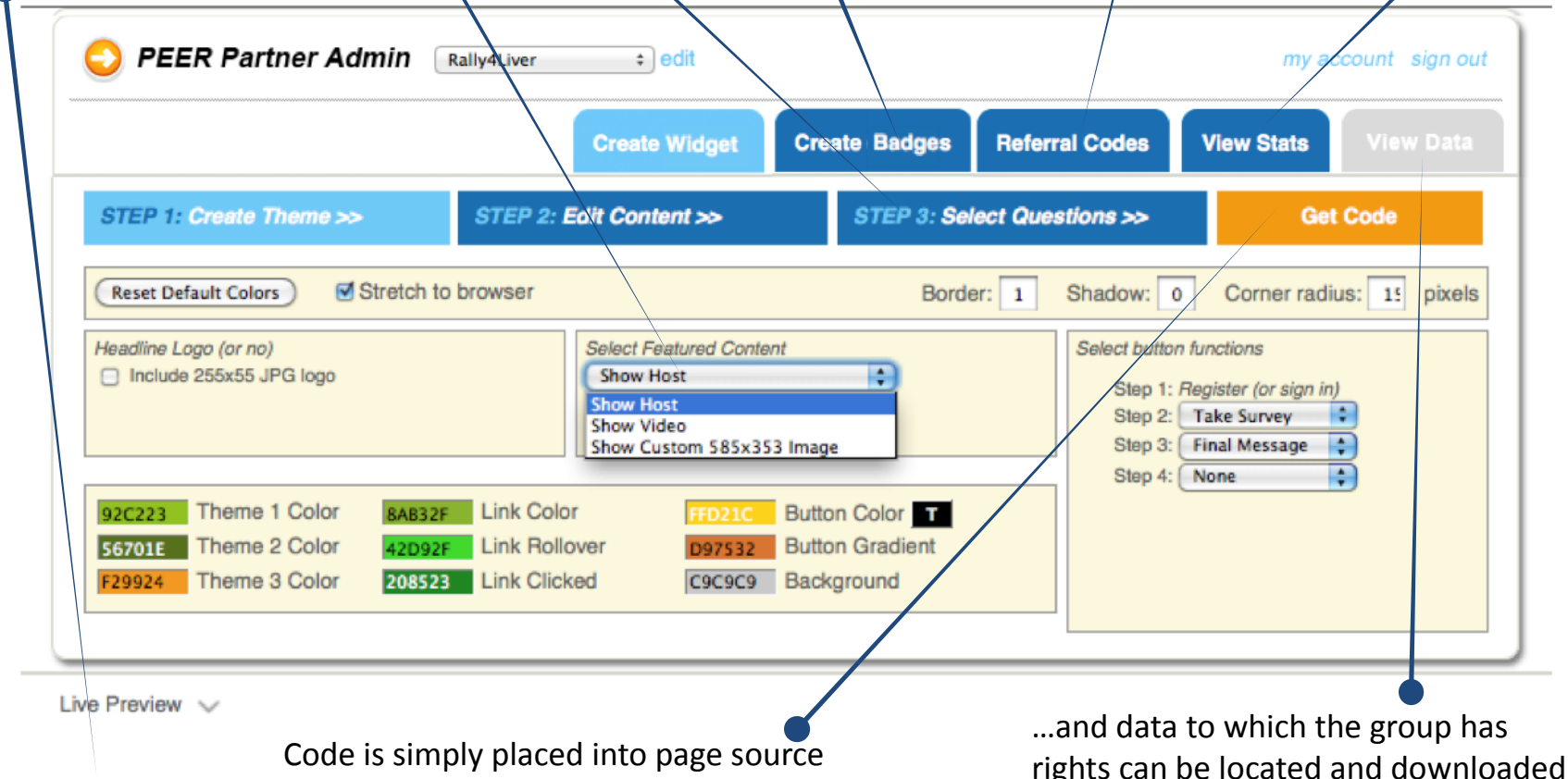
…and view a live preview of the page as it is modified

Curate inquiries (content, sequence, dependencies, etc) from a starting point of over 22,000 questions

Create referral codes to use on any printed communications like letters and posters

Also create custom badges to display on other websites

…and view statistics for how all of these assets perform and why



Code is simply placed into page source and it begins working instantly

…and data to which the group has rights can be located and downloaded in CSV format for analysis

EspeRare: Merging patients & commercial interests

Mission

In partnership with patient groups, academia and medical reference centers, EspeRare uncovers the potential of existing drugs to address severe therapeutic unmet needs in neurological and immunological rare diseases.

Strategic Goals

- **Gives a chance to unexplored therapies in rare diseases:**
  - Identify & drive translational validation of "dormant" opportunities
  - Leverage established patient groups & biomedical networks
  - Invest foundation's R&D revenues and grants in rare disease programs
- **De-risk early development of rare diseases programs:**
  - Combine not-for profit & public grants with commercial funds/assets
  - Bring Biopharma expertise to academia & patient groups collaborations
  - Translate patient engagement into scientific and regulatory efforts
- **Hand-over programs to commercial partners for late development:**
  - PhII/PhIII ready programs with strong network of patient groups & experts
  - Flexible partnering model, tailored to the asset & disease

“You never change things by fighting existing reality. To change something, build a new model that makes the existing model obsolete.”

Buckminster Fuller

Build the WE

Contact Information

For more information:

Sharon Terry
CEO and President, Genetic Alliance, Inc.
(202) 966-5557, Ext. 202
sterry@geneticalliance.org

General Information: *http://www.geneticalliance.org/programs/biotrust/cena*

Online demo (for JSRDF shown here): *http://jsrdf.org/JSLIFE-demo*