

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**





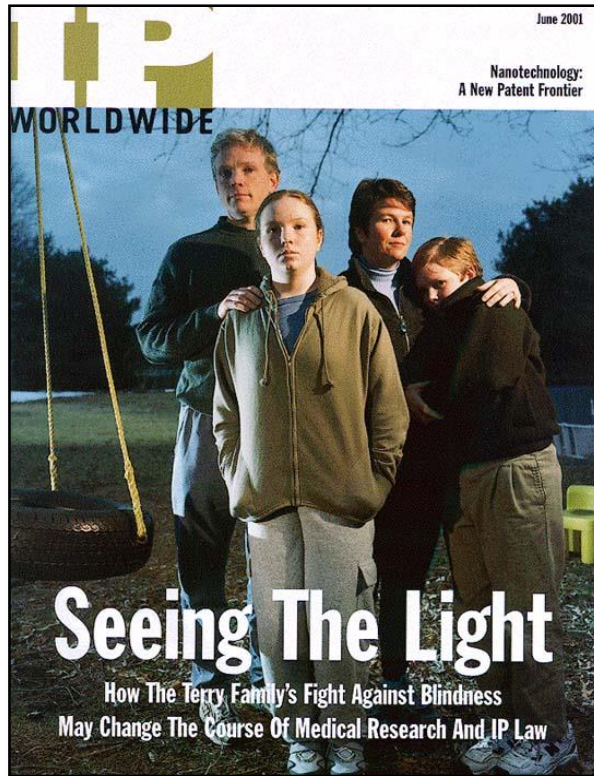
PXE
international

Gene
1999
Discovery

BioBank

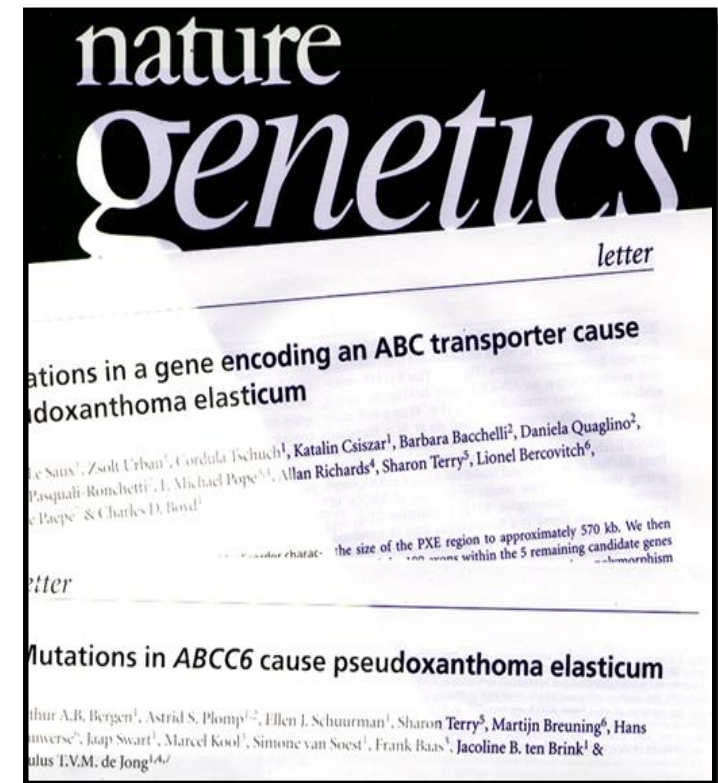
Testing

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

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 CERN efficiently, by creating a large, highly representative national patient-centered clinical research network for conducting clinical outcomes research.

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.



\$1.26 Billion

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

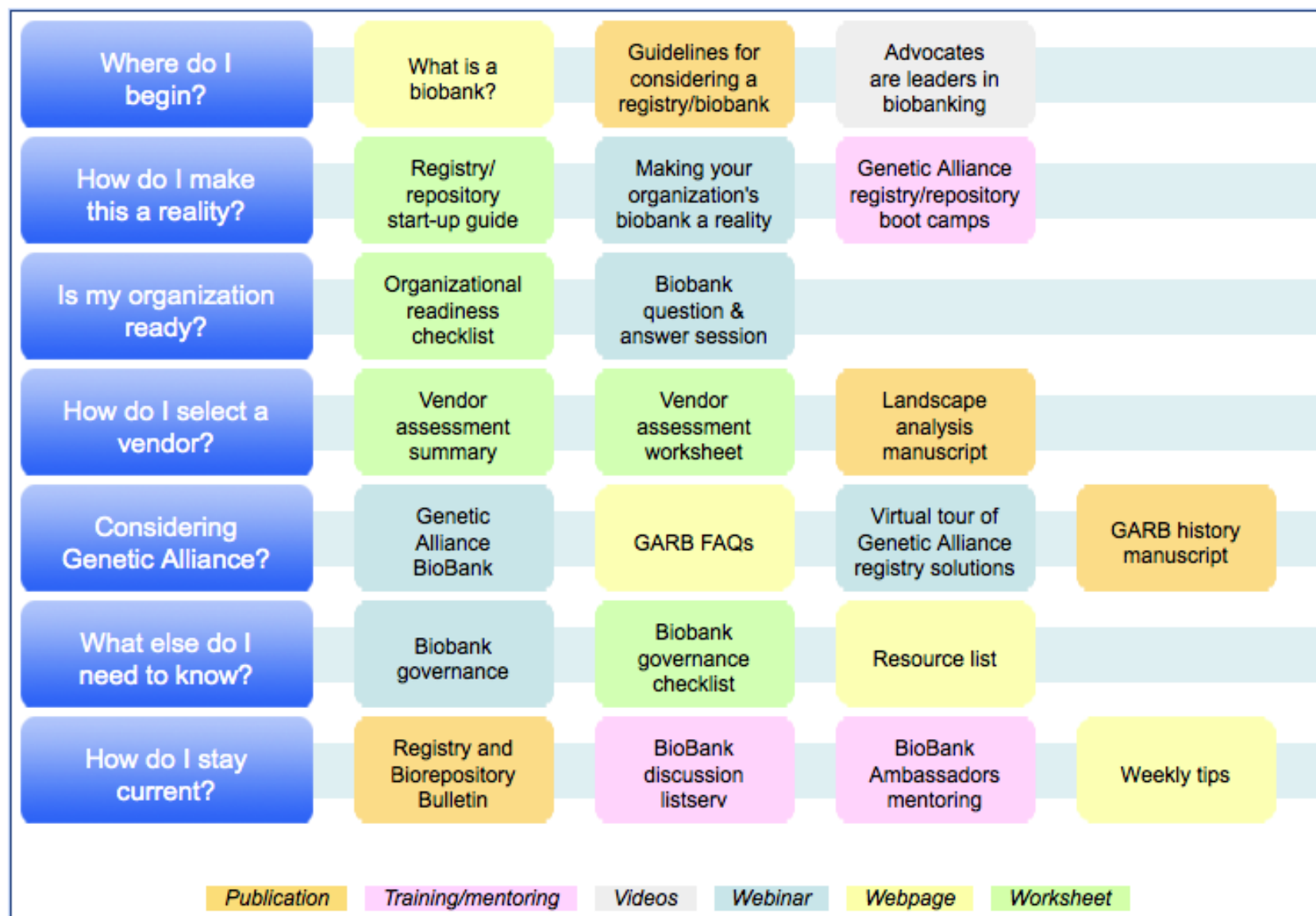
....in both rare and common disorders

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 10

‘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

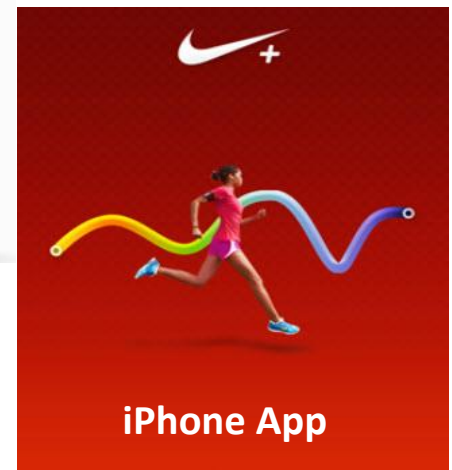
Can we connect without adding friction to people's lives?



FitBit®



Twitter
Facebook



iMapMyRUN+



iMapMyRIDE+



iMapMyWALK



iMapMyFITNESS



iMapMyHIKE

Needles in Haystacks



The haystack is made of needles...



Research Enterprise



Consent

...current methods of informed consent are challenged...

granular and dynamic engagement

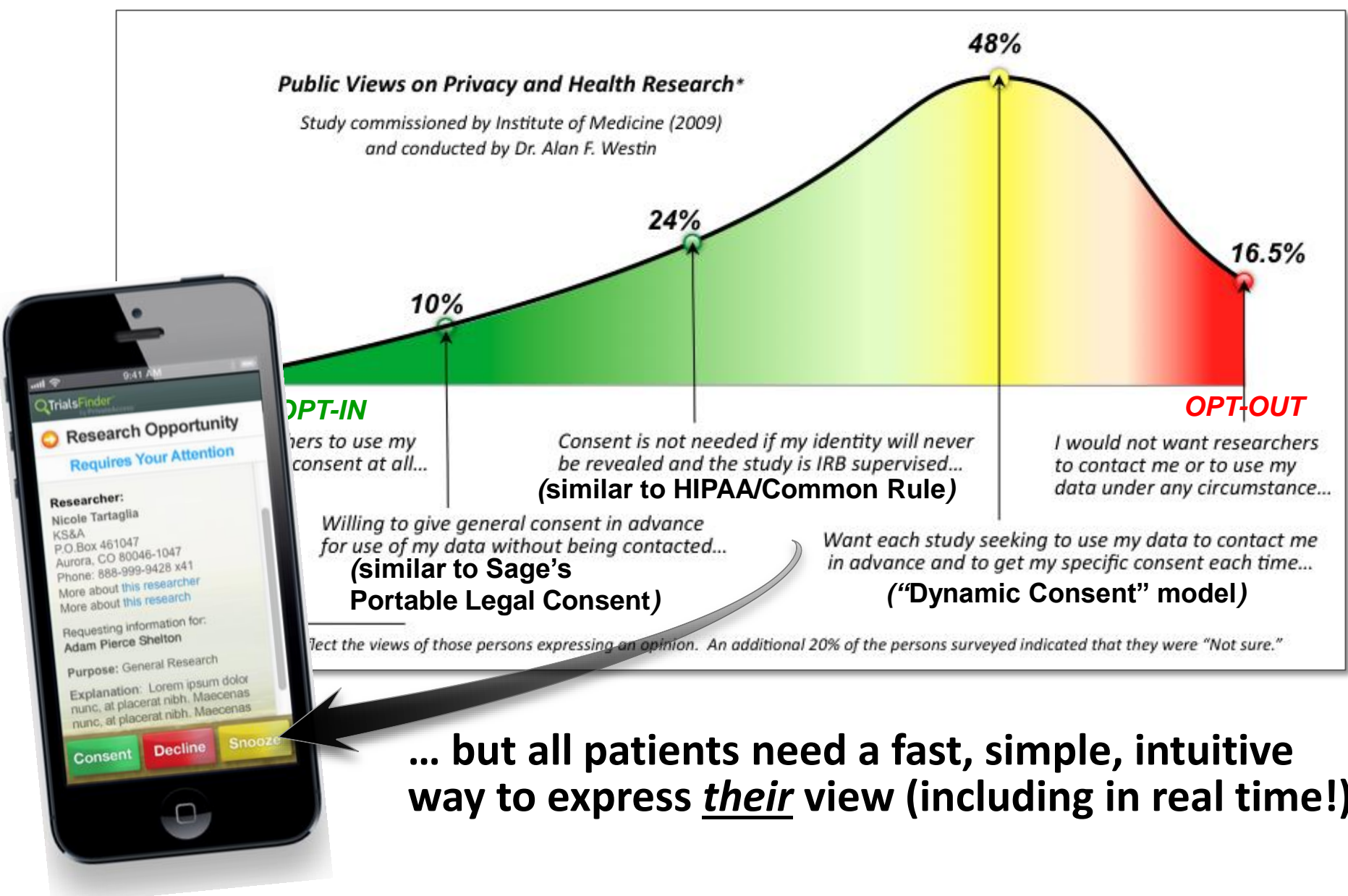
not consent

not a transaction

not binary

not tiered

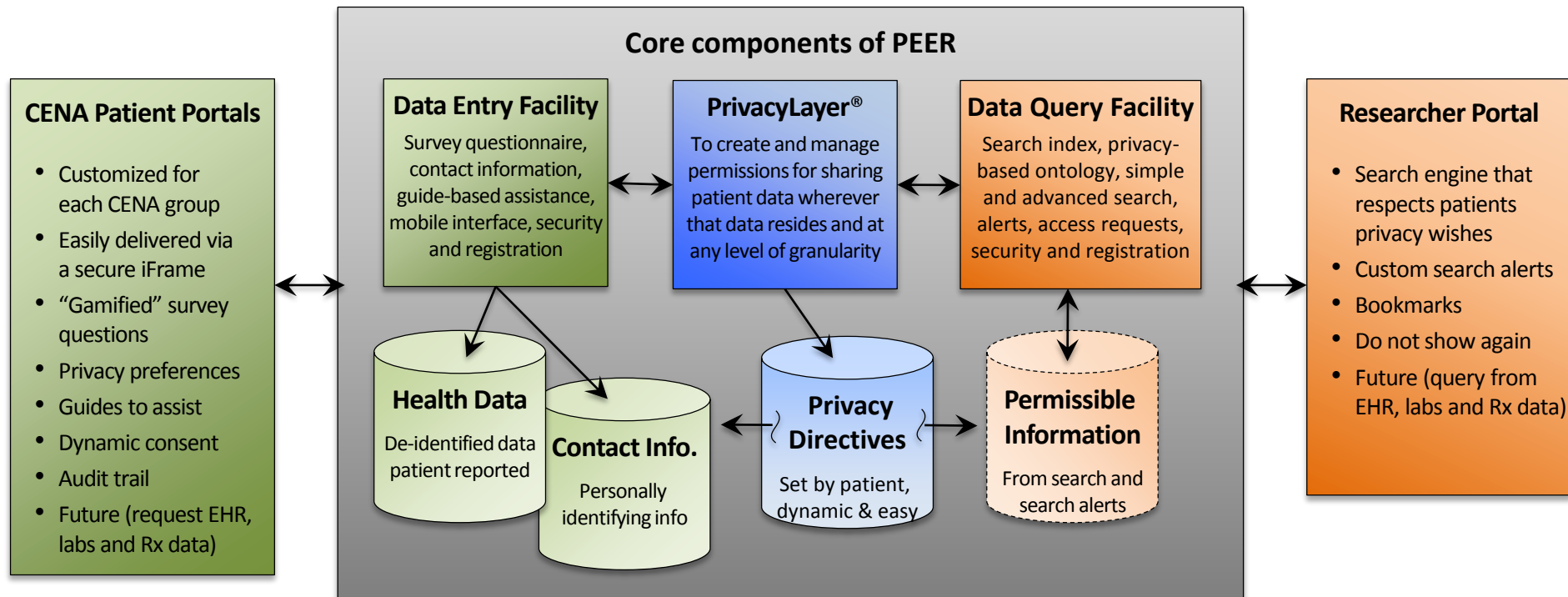
Because people have vastly different views about privacy and sharing



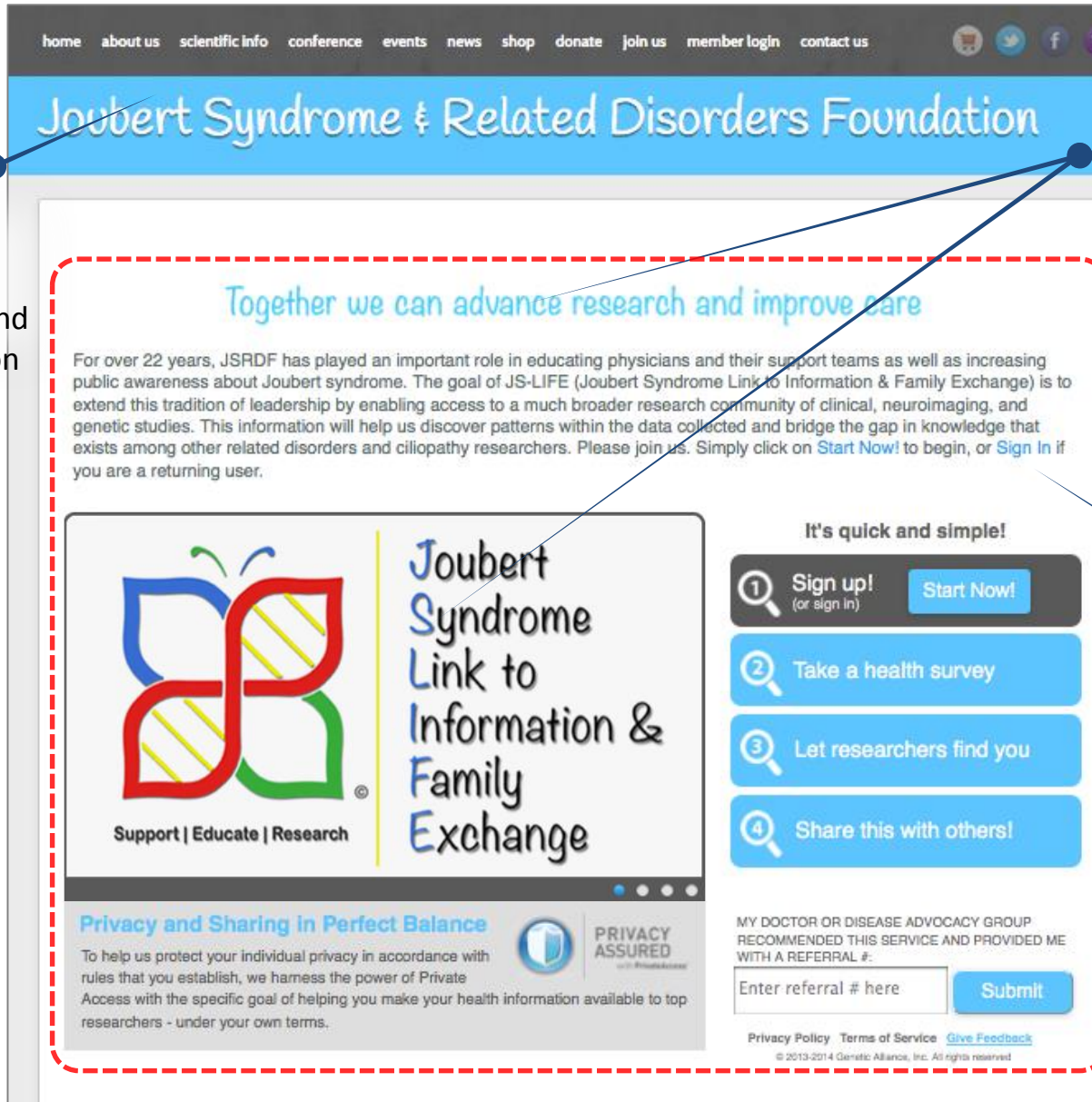
High level architecture of the PEER Platform

CENA is powered by the PEER (Platform for Engaging Everyone Responsibly) 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 headline, colors, theme, video and other content can be tailored to fit seamlessly into the host site and support each group's message.


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

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”

Select a guide : For New User

Set your privacy preferences manually, or select a guide who has studied the options and made suggestions for persons with high, medium and low concerns about privacy. Select a guide who you know, or whose experience or perspectives you value.




Stephen Mack
President, JSRDF. Parent of 10-year old daughter with Joubert syndrome

Stephen's daughter, Isabelle, was diagnosed in 2004 when she was just three days old. He currently serves as President of JSRDF, which he will hold through 2015. Prior to this, he served for two years as the organization's president-elect, and seve

... [More >>](#)

What's this?

Select Stephen as your guide




Daniel Doherty
MD, PhD; Associate Professor of Pediatrics University of Washington School of Medicine

In addition to his clinical practice caring for children with all types of central nervous system abnormalities including Joubert syndrome, Dr. Doherty's research focuses on hindbrain malformations, agenesis of the corpus callosum, cortical malformat ... [More >>](#)

What's this?

Select Daniel as your guide




Nicole Ford
JSRDF Board Member. Parent of 17-year old daughter with Joubert syndrome

Nicole's daughter, Taylor, was diagnosed 1997 with Joubert syndrome when she was 11 weeks old. Since receiving the diagnosis, Nicole has been actively involved with other parents and family members of individuals affected by the condition. Nicole s ... [More >>](#)

What's this?

Select Nicole as your guide




Create Preferences Manually

If you are comfortable using this tool, you may wish to set your preferences manually.

[Set preferences manually](#)

What's this?

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



PRIVACY ASSURED
Private Access lets you control who can see your information, and for what purpose. This service will check your Private Access settings before sharing any of your information.

[Privacy Policy](#) [Terms of Service](#) [Give Feedback](#)

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

YOU ARE CURRENTLY VIEWING SUGGESTED PRIVACY SETTINGS FOR New User

What types of information can be shared?

	DISCOVER discover and view my anonymous information (click for details)	EXPORT & USE export and use my anonymous information (click for details)	CONTACT view and use my personal information to contact me (click for details)
Who can access it?			
Advocacy & Support Groups			
ⓘ Joubert Syndrome & Related Disorders Foundation (JSRDF)	✓ Allow	✓ Allow	✓ Allow
ⓘ DiseaseInfoSearch.org listed organizations serving your condition	✓ Allow	✓ Allow	⚠ Ask Me
ⓘ All organizations serving your condition	✓ Allow	✓ Allow	⚠ Ask Me
Researchers			
ⓘ Researchers recommended by JSRDF	✓ Allow	✓ Allow	⚠ Ask Me
ⓘ Researchers recommended by any DiseaseInfoSearch.org listed organization serving your condition	✓ Allow	✓ Allow	⚠ Ask Me
ⓘ Researchers addressing your condition	✓ Allow	✓ Allow	⚠ Ask Me
ⓘ All researchers	✓ Allow	⚠ Ask Me	🚫 Deny
Data Analysis Platforms			
ⓘ "Show related content" feature	N/A	✓ Allow	N/A
ⓘ "Compare with others" feature	N/A	✓ Allow	N/A
ⓘ Genetic Alliance Translational Research Network	✓ Allow	✓ Allow	N/A
ⓘ PCORnet: Patient-Centered Outcomes Research Network	✓ Allow	⚠ Ask Me	⚠ Ask Me
ⓘ Newly-Released Data Analysis Platforms	⚠ Ask Me	⚠ Ask Me	N/A

<< Select a different guide Customize Accept and continue >>

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

Gamified questionnaire with easily customized topics

The screenshot displays the 'Joubert Syndrome & Related Disorders Foundation' website. The navigation bar includes links for Home, My Account, Health Profiles, Privacy Settings, Notifications, Activity Log, and Sign Out. The main heading is 'Answer Health Survey' with a dropdown menu set to 'For New User'. A purple banner indicates 'YOU ARE CURRENTLY ANSWERING QUESTIONS FOR New User'. The interface has three tabs: Questions, My Answers, and My Progress. A progress indicator shows 'Your answers: 80'. The current question is 'Learning disorders he has experienced are...' with the instruction '(Select all that apply)'. The options are: ADD/ADHD, Auditory processing (checked), Communication deficit, Oppositional Defiance Disorder (ODD), Global learning delay (checked), and Autism spectrum disorder (checked). A central box contains the text 'OAuth2 authorization'. At the bottom of the question box are buttons for 'Don't know', 'Skip', and 'Next'. Below the question box is a horizontal bar chart titled 'He has experienced some learning disorders...'. The chart shows a red dot at the 'Yes' end (100%) and a yellow bar extending to the 'No' end (0%).

Common data elements
Standards
Ontologies – ORDO, HPO
Validated instruments

OAuth2
authorization

Participants see immediate feedback for how others have responded

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.

Joubert Syndrome & Related Disorders Foundation

Home My Account Health Profiles Privacy Settings Notifications Activity Log Sign Out

Answer Health Survey ☐ For New User

YOU ARE CURRENTLY ANSWERING QUESTIONS FOR New User

Welcome Health Survey Medical & Family History Lab Results Molecular Profiling Medical Records

Welcome Back!

Health Survey 12% complete **Update**
last updated 11/02/14
Take part in our health survey; and as you respond see how your answers compare with others. Complete the whole survey, or just answer part now and finish later.

Lab Results ✓
With just a few clicks, report critical lab values and test results; and if you'd like, we'll automatically chart these measures for you over time.

Medical History ✓
Summarize information about symptoms, diagnosed conditions, procedures, medications and dosage levels, surgery dates, hospitalizations, and more.

Molecular Profiling **Update**
Record information about biological markers including unique genes, proteins and other molecules derived from advanced genetic testing and molecular analyses.

Family History **Update**
Create and annotate a family tree with p... of family health histo... characteristics, and related ancestry info...

WEIGHT

May 2012

My weight was approximately: **285 lbs**

400
300
200

285

June 2011
Aug 10, 2010
May 12, 2010
Feb 2009

Dates with entries for THIS question
Dates with entries for ANY question
Show Calendar (Select Date)

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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

Also create custom badges to display on other websites

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

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

The screenshot displays the 'PEER Partner Admin' interface for a user named 'Rally4Liver'. The interface includes a top navigation bar with buttons for 'Create Widget', 'Create Badges', 'Referral Codes', 'View Stats', and 'View Data'. Below this is a progress bar with four steps: 'STEP 1: Create Theme >>', 'STEP 2: Edit Content >>', 'STEP 3: Select Questions >>', and 'Get Code'. The main content area is divided into several sections: 'Reset Default Colors' and 'Stretch to browser' (checked) at the top; 'Headline Logo (or no)' with an 'Include 255x55 JPG logo' checkbox; 'Select Featured Content' with a dropdown menu showing 'Show Host', 'Show Video', and 'Show Custom 585x353 Image'; 'Select button functions' with a list of steps and corresponding dropdowns; and a color selection section at the bottom with various color swatches and labels like 'Theme 1 Color', 'Link Color', 'Button Color', etc. A 'Live Preview' button is located at the bottom left of the interface.

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/JS LIFE-demo>