

**The Role of Disease Advocacy Groups in Establishing Common Infrastructure:
Excellent Science Driven by Novel Collaborations to Create Shared Resources**

Sharon F. Terry, MA
Genetic Alliance & PXE International

The goal of this workshop is to understand what it means to develop an internet-based platform with common data elements utilizing a federated rare disease registry. Technically, this is challenging, but the standards compliant (LOINC, SNOMED CT, etc.) interchange of information (e.g., HL7) is simple, compared to the challenge of disassembling the competitive nature of science, and the quest for funding and recognition. Participants in this endeavor must be bold and be willing to assume the risk of disturbing the status quo.

Establishing, and continually improving, evidence-based registries and biorepositories that capitalize on new advances in information technology, repository science and social networking, requires a collaborative effort¹. Further, disease specific organizations are uniquely positioned to establish common infrastructure, though there are both opportunities and challenges in this activity.

True transformation to the point of shared infrastructure requires radical transformation of advocacy organizations as we know them today. Technologies abound that make rare common at this point in the evolution of web 2.0 and social network phenomenon. The leap that must be undertaken requires culture change including: open space, shared commons, dynamic networks and permeable boundaries between organizations and systems. These changes must occur in the research & advocacy community².

Using long tail and open innovation technology, it is possible to assemble a system that creates virtual and actual registries and biorepositories. A common platform, shared infrastructure and trainings is attainable in an open environment. As a first generation product, a group of disease specific organizations established a cooperative called the Genetic Alliance BioBank in 2003. The common elements are applicable to the goals of this workshop. A common infrastructure would include:

- Training and mentoring to disease advocacy organizations by experts in biobanking and registries, and providing templates for all necessary documents and protocols.
- Tools for disease advocacy organizations to recruit participants in their registries in the federated structure, using state-of-the art methods that emphasize trust, privacy protections, data security, empowerment of participants and the member advocacy groups, and ongoing education.³
- A robust and dynamic web 2.0/3.0 process for informed donor decision-making, leading to truly informed consent, tailored to specific uses of the samples and related information.⁴
- Multiple options for state-of-the-art storage and systems for collection, processing, archiving and distributing biological samples. Intelligent and intentional distribution of samples is critical.
- Clinical data collection system with customizable, web-based interface for participant data entry. Controlled vocabulary and minimum data sets provide enhanced data mining opportunities.
- Shared and networked web-applications for ease of sample and data management by the disease organization, including on the fly data analysis.
- Facilitated collaboration of disease organizations, academic, government and industry partnerships.

The result of a virtual federated network approach would allow professional advisory boards of disease and cross-disease entities to drive research in a focused and hypothesis driven way, while protecting these valuable resources.

References:

1. Terry, S.F., Terry, P.F., Rauert, K.A., Uitto, J., and Bercovitch, L.G. (2007). Advocacy groups as research organizations: the PXE International example. *Nat Rev Genet* 8, 157-164.
2. Terry, S.F., and Boyd, C.D. (2001). Researching the biology of PXE: partnering in the process. *Am J Med Genet* 106, 177-184.
3. Terry, S.F., and Terry, P.F. (2001). A consumer perspective on informed consent and third-party issues. *J Contin Educ Health Prof* 21, 256-264.
4. Beskow, L.M., Botkin, J.R., Daly, M., Juengst, E.T., Lehmann, L.S., Merz, J.F., Pentz, R., Press, N.A., Ross, L.F., Sugarman, J., et al. (2004). Ethical issues in identifying and recruiting participants for familial genetic research. *Am J Med Genet A* 130, 424-431.

Uniting Rare Diseases



**Advancing Rare Disease Research:
The Intersection of Patient Registries, Biospecimen
Repositories and Clinical Data**

Session IV
**Patient Participation & Outreach Activities/Patient Advocacy:
The role of disease advocacy groups in establishing common
infrastructure**



Sharon F. Terry, MA
President & CEO, Genetic Alliance
Founding Executive Director, PXE International



Creative Commons Attribution 3.0 License 1

Transforming Health Through Genetics

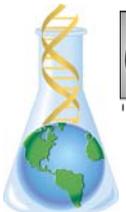


Network of 10,000 organizations, universities and companies

Openess is our product and process

Shared infrastructure to transform health
is our goal

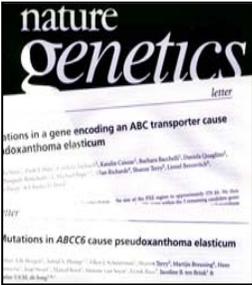
Creative Commons Attribution 3.0 License 2



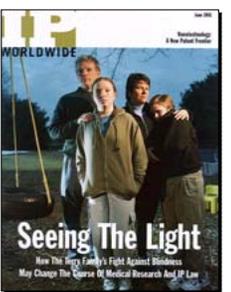
BioBank



**Gene
Discovery**



**Human
Clinical
Trials**



**Drug
Screening &
Development
Approaches**

Testing

Clinical
Diagnostic Test
Development
via FDA & CLIA
Regulatory
Strategies

Patenting

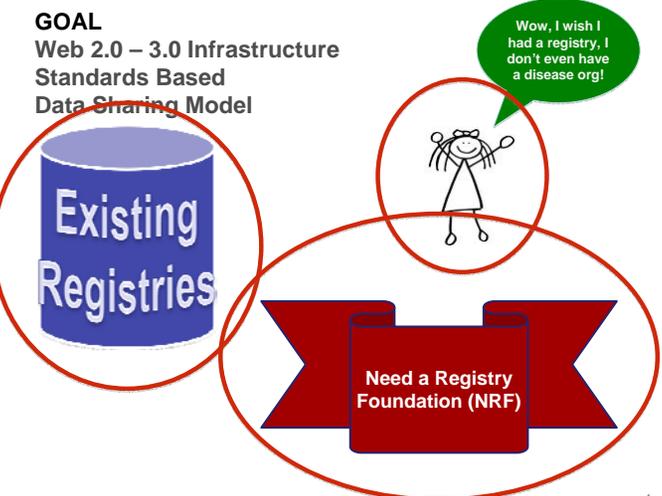
Licensing & Intellectual Property Management

Therapeutics

--Small Molecules
--Nonsense mutants

Creative Commons Attribution 3.0 License 3

GOAL
Web 2.0 – 3.0 Infrastructure
Standards Based
Data Sharing Model



Creative Commons Attribution 3.0 License 4

Challenges for Disease Advocacy Organizations

- We never wanted to be in this anyway
- We worry about our funding and branding (our IP)
- We worry our inadequacies will be exposed
- We want someone to do it for us
- We can't see beyond our own problems
- We fall in the huge culture gaps between advocates, academia, industry
- We can't see the evidence that creating common infrastructure will make a BETTER difference.

Creative Commons Attribution 3.0 License

5

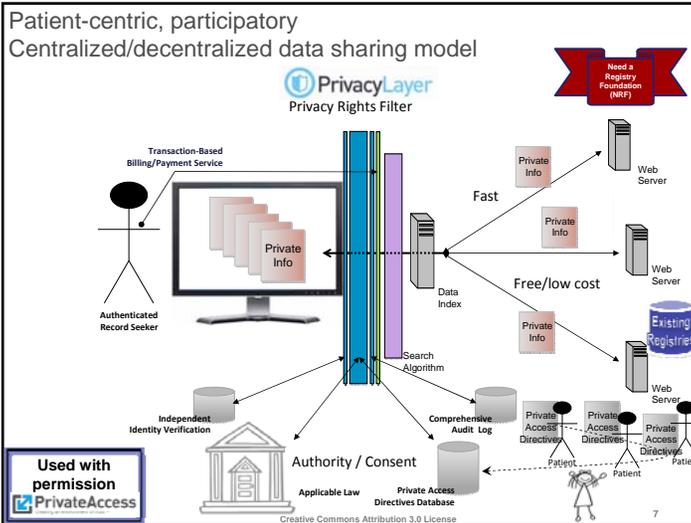
- It appears harder to work for the common good
- We have difficulty building on a systems level, it is easier to work one-offs

**Ultimately
WE ARE AFRAID**

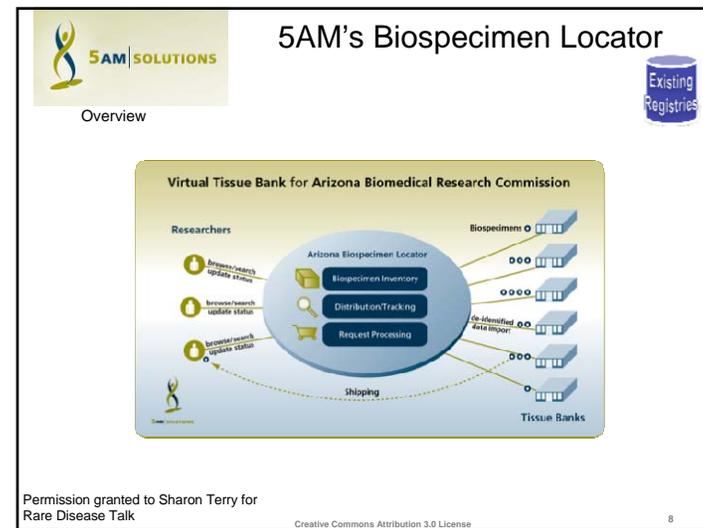
Let's fearlessly do the thought-experiment
in this meeting

Creative Commons Attribution 3.0 License

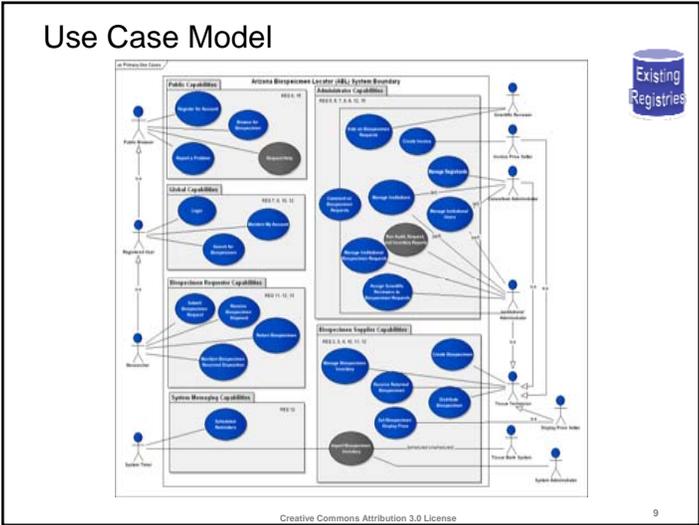
6



7



8



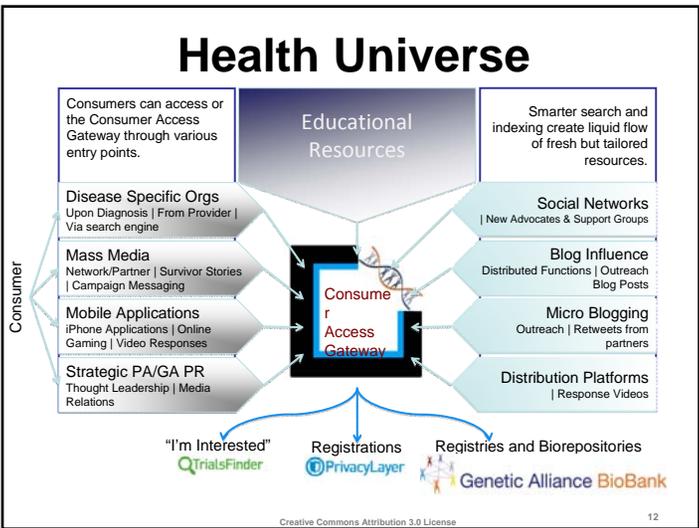
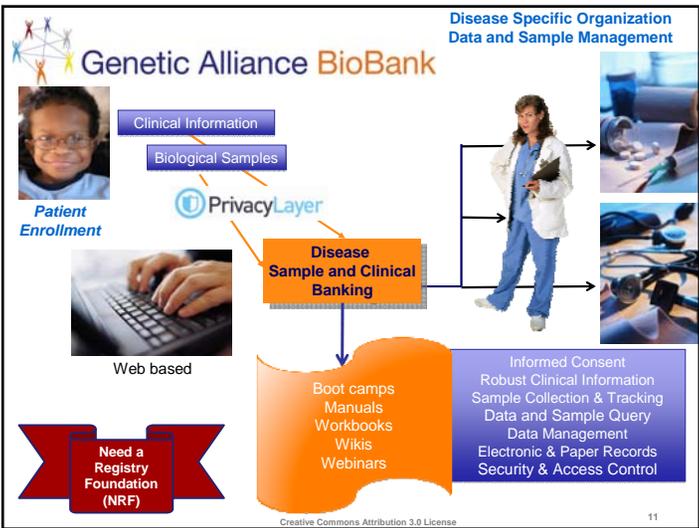
Genetic Alliance BioBank

Our Product
Cooperative – extensible,
interoperative, cross-disease, cost-sharing platform

Need a Registry Foundation (NRF)

Existing Registries

Creative Commons Attribution 3.0 License



patientslikeme Patients helping patients live better every day.

Share Your Experience »
Find Patients Like You »
Learn From Others »
Join Now (It's Free!)

Wow, there is an affinity groups for anything, I am not alone!

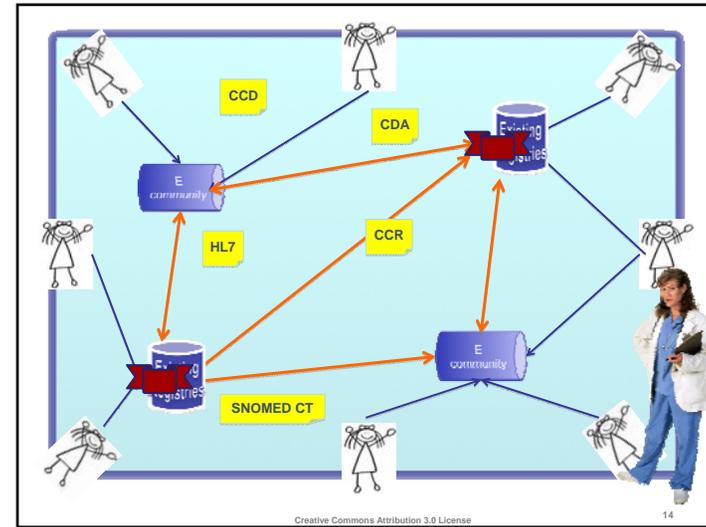
CarePages™

facebook

Inspire together we're better™

23andMe

Creative Commons Attribution 3.0 License 13



Genetic Alliance Biobank Competition

Win \$20,000 Towards the Initiation Fee for the Genetic Alliance Biobank

Who is Eligible?

- Non-Profit Organizations
- Non-Profit Collaborations focusing on the same Condition, Gene, or Disease Pathway

What's Included?

- \$20,000 towards the initiation fee for the Genetic Alliance Biobank.
- Training and mentoring to disease advocacy organizations by experts in BioBanking.
- Tools for disease advocacy organizations to recruit participants to the BioBank.

Applications available February 1, 2010
www.geneticalliance.org and www.biobank.org
 Contact: jbialick@geneticalliance.org

Creative Commons Attribution 3.0 License 15

The Solution

- Web-based, open source, interoperable, global
- Linking all types of registries and repositories
- Create 'add water and serve' solutions for disease orgs with no existing registry/biorepository
- Create registry-lite for diseases with no registry enabling infrastructure
- Outreach to nontraditional Web 2.0 affinity groups
- Outreach to undiagnosed, and un-affiliated
- Switch (not store), servers reside local to community, standards-based
- Mining tools, assuring confidentiality and high access
- Electronic sample management and data distribution
- Cross-disease analysis tools
- Iterative feedback loops, for researchers to deposit findings, and for system to improve (learning system)

Creative Commons Attribution 3.0 License 16

Steps to the Solution

Let go – become transparent and boundriless, without concern for 'property' of any kind
Multi-stakeholder workgroup established in part by community consensus and visionary leadership
Workgroup focuses on "What Matters"

System architecture design: iterative, learning, open access
Use cases to determine all scenarios
Metrics of success agreed upon
Existing resources are brought to the table
Gaps are identified
Contractors engaged to construct system
Training teams developed
Measurement methods to test against metrics deployed
System tested
Iterate again

Training and re-training (technical assistance and culture shifts)

It's not enough to rage against the lie...
you've got to replace it with the truth.

Bono