

# House of Lords Inquiry on Genomic Medicine

Public-Private Partnership

Revolutionizing Access

6 June 2008

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



Elizabeth and Ian  
diagnosed with  
a rare genetic  
condition:  
pseudoxanthoma  
elasticum (PXE)



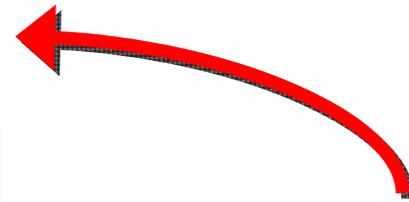
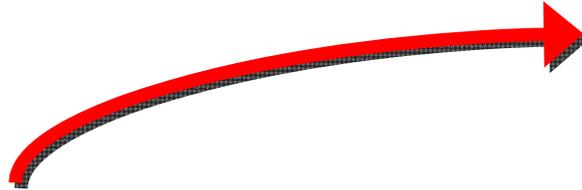
2008



**Surveyed Literature**  
**Interviewed Researchers**  
**Investigated issues with**  
**the consumer community**  
**[www.geneticalliance.org](http://www.geneticalliance.org)**



# PXE International, Inc.

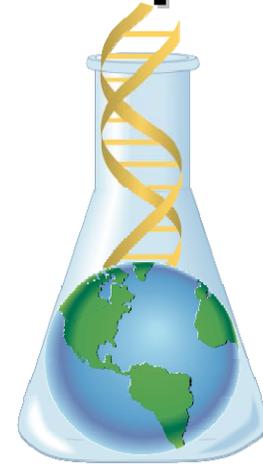


## Participants

- Centralization & Coordination
- IRBs & Study Management
- Informed Consent Process
- Blood & Tissue Bank
- Geno/pheno database
- Coded Patient IDs
- Reporting & Re-contacting
- Longitudinal Follow-up
- Patient Education & Updates

**Firewall**

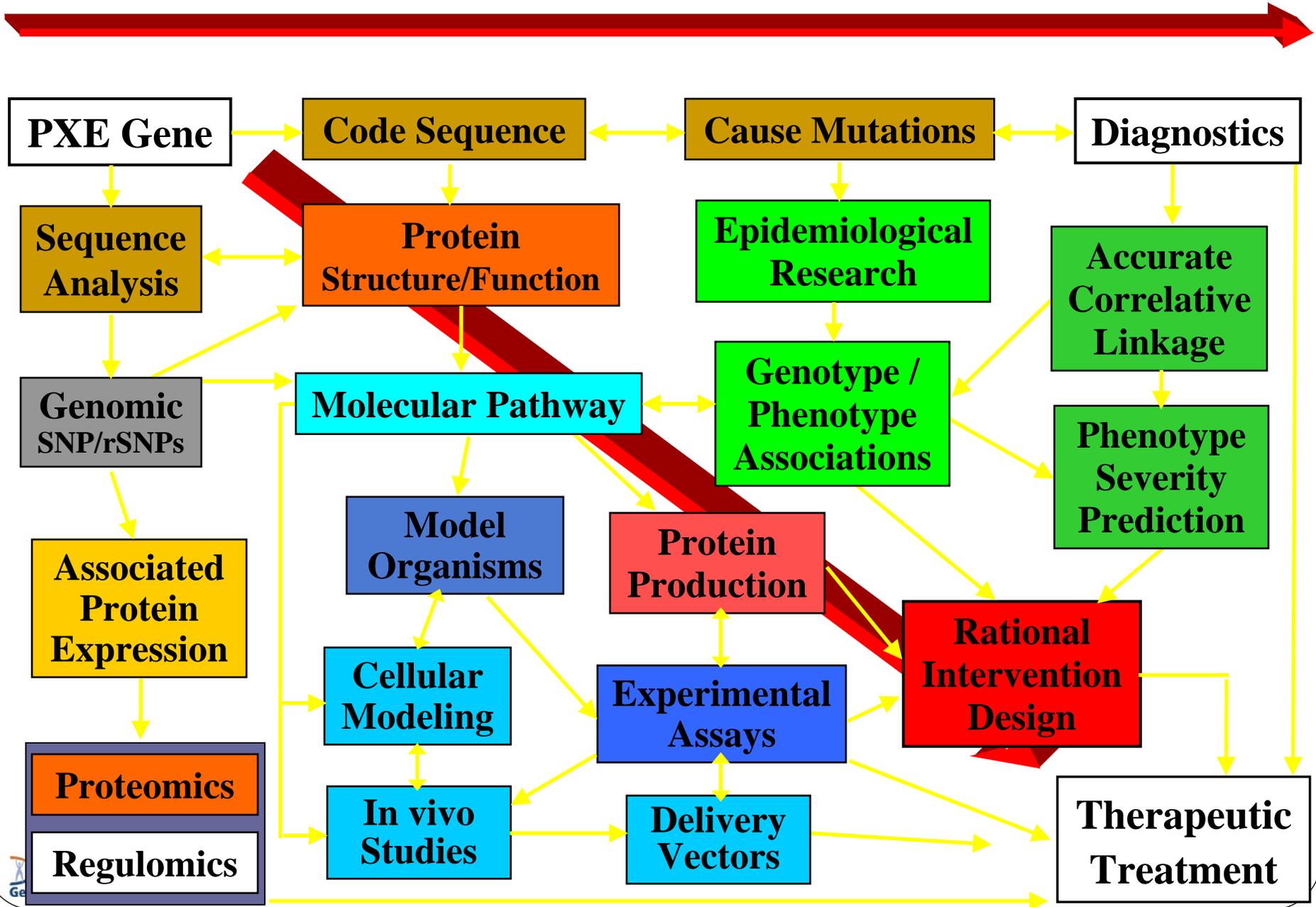
## Research Enterprise



- Coordination of Labs
- Shared Info & Tech Trans



**LOGIC: Data Collection + Data Integration + Time/Serendipity + Knowledge**



# nature Genetics

letter

## Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum

de Saix<sup>1</sup>, Zsolt Urban<sup>1</sup>, Cordula Tschuch<sup>1</sup>, Katalin Csiszar<sup>1</sup>, Barbara Danzelli<sup>2</sup>, Daniela Quaglino<sup>2</sup>,  
Pasquali-Ronchetti<sup>3</sup>, E. Michael Pope<sup>3,4</sup>, Allan Richards<sup>5</sup>, Sharon Terry<sup>5</sup>, Ronel Bercovitch<sup>6</sup>,  
de Paepe<sup>7</sup> & Charles D. Boyd<sup>1</sup>

... the size of the PXE region to approximately 570 kb. We then  
... within the 5 remaining candidate genes

letter

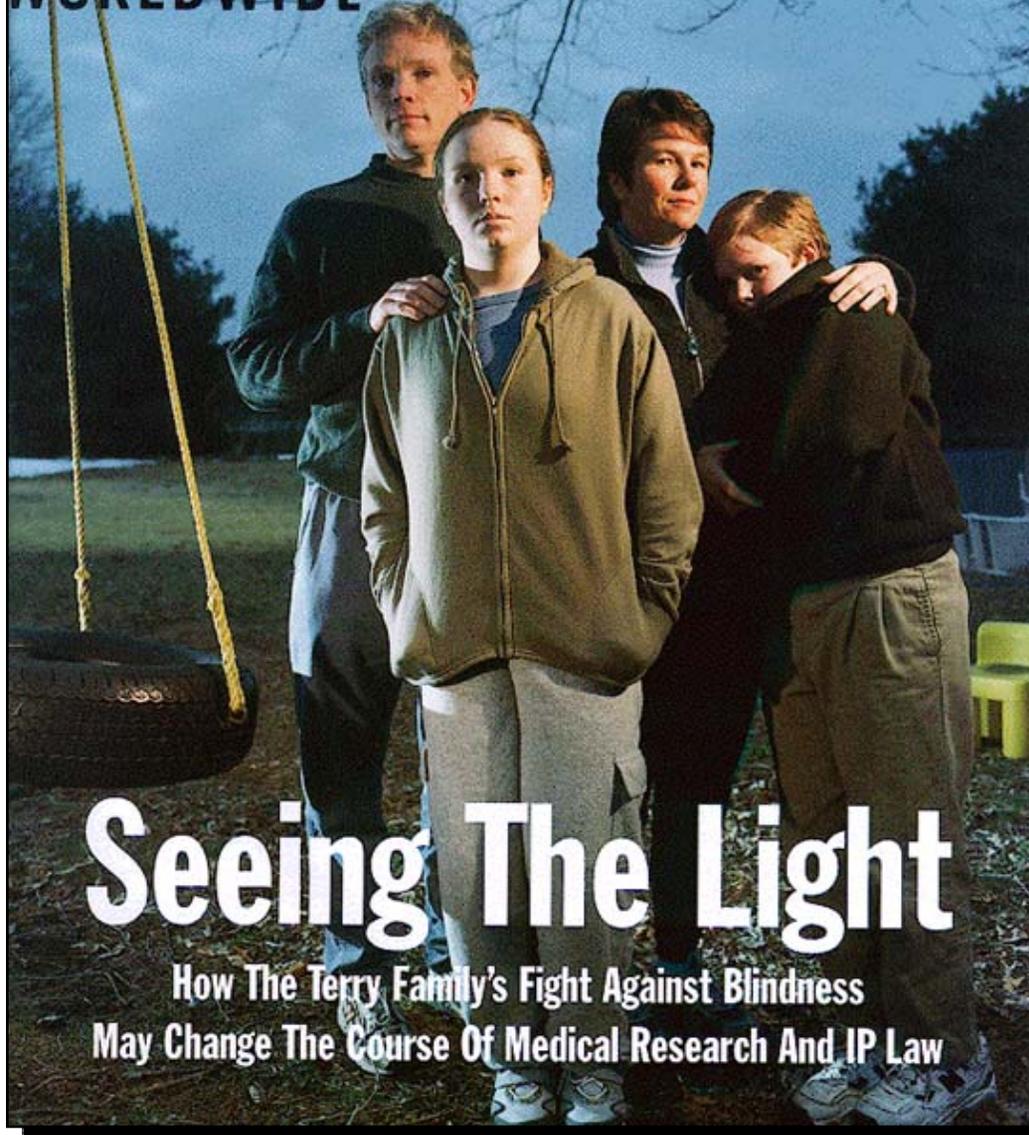
## Mutations in *ABCC6* cause pseudoxanthoma elasticum

thir A.B. Bergen<sup>1</sup>, Astrid S. Plomp<sup>1,2</sup>, Ellen J. Schuurman<sup>1</sup>, Sharon Terry<sup>5</sup>, Martijn Breuning<sup>6</sup>, Hans  
niverse<sup>6</sup>, Jaap Swart<sup>1</sup>, Marcel Kool<sup>3</sup>, Simone van Soest<sup>1</sup>, Fran...  
ulus T.V.M. de Jong<sup>1,4,7</sup>

June 2001

**Nanotechnology:  
A New Patent Frontier**

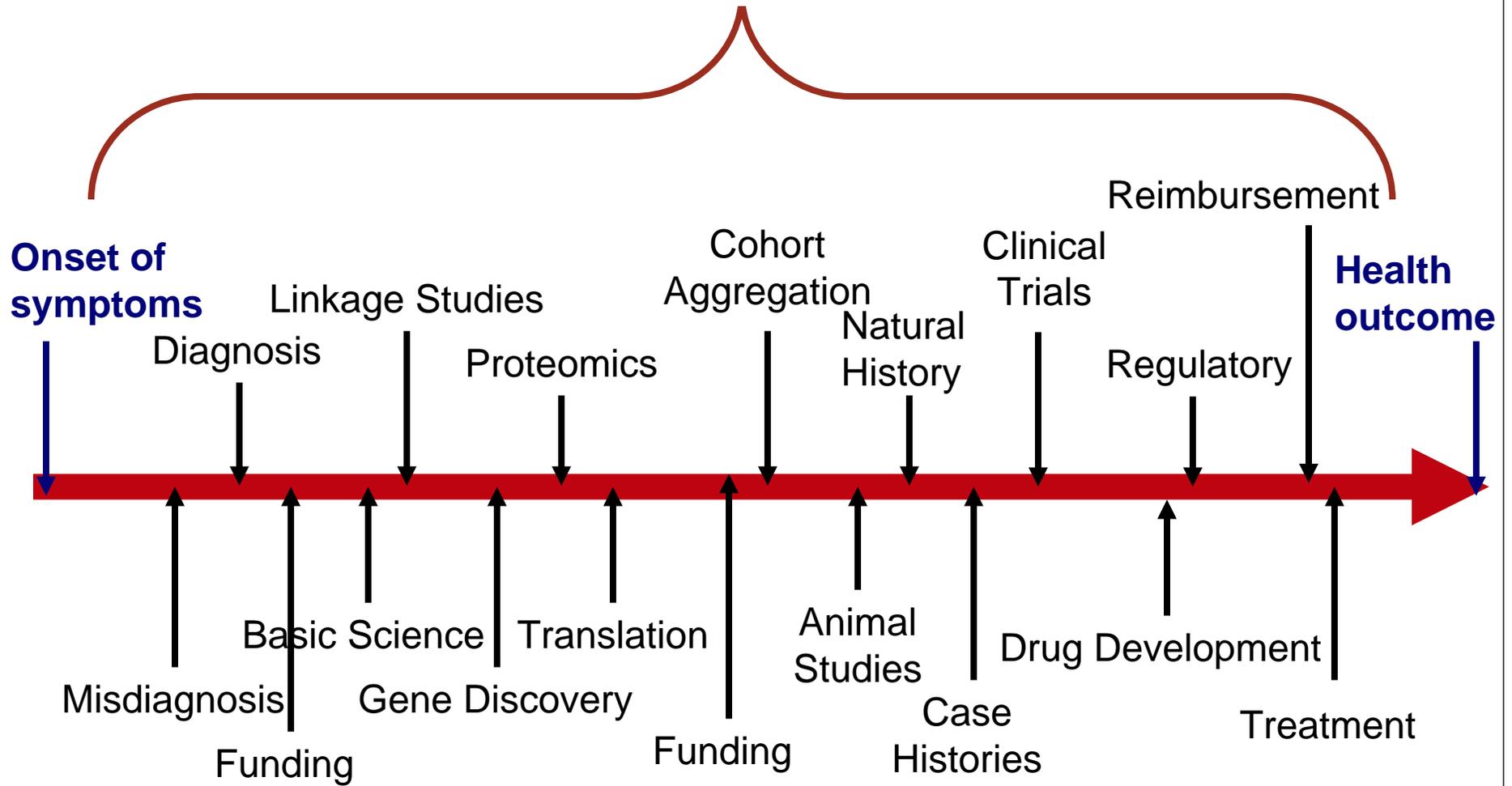
**WORLDWIDE**



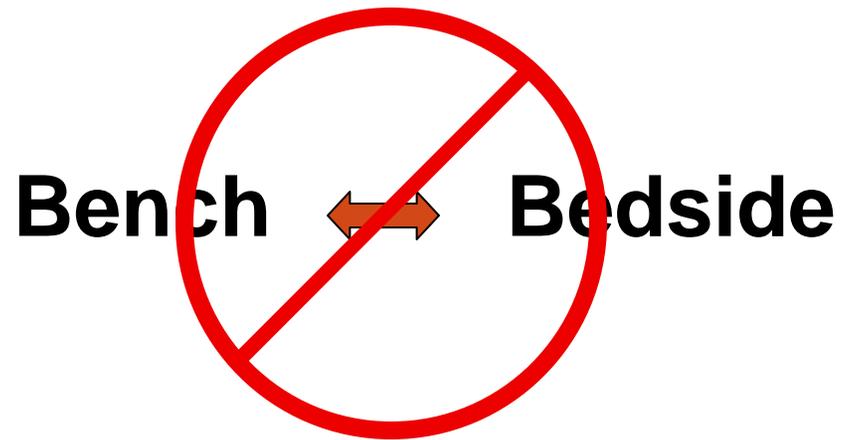
# Seeing The Light

**How The Terry Family's Fight Against Blindness  
May Change The Course Of Medical Research And IP Law**

# Perspective → Vision



# What Matters?



# What Matters?

Improved Health Outcome

Diagnostics

Medicines

Prevention  
Strategy

Clinical  
Trials

Applied  
Research &  
Development

Translation  
Research

New  
Knowledge  
&  
Technology

BASIC BIOMEDICAL RESEARCH BASE

# Maturing Patient 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
- **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 – Information Sharing in the Commons – *Translation & Delivery!!!***

# Advocates in the Information Age:

Business models based on new economic constructs

## Industrial Age (old)

**Control** means of production

Based on **scarcity**

**Hierarchical** / Command & Control

**Linear** / Sequential

Win / **Lose**

**Material**

## Information Age (new)

**Open** means of production

Based on **abundance**

**Network** / Collaboration

**Organic**

Win / Win

**Information**

# Disease Organization Gears



# State of the Art Today

Lack of community



Lack of cooperation



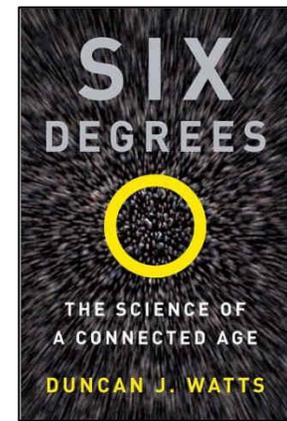
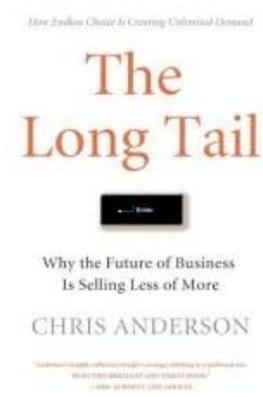
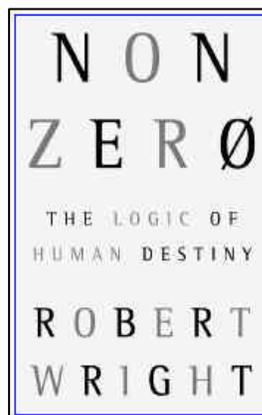
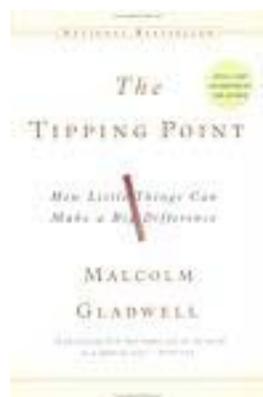
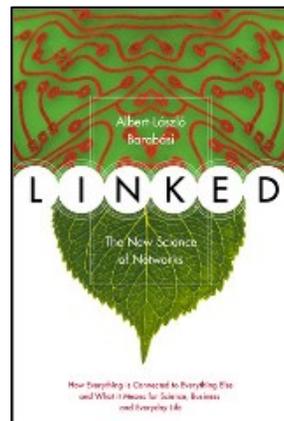
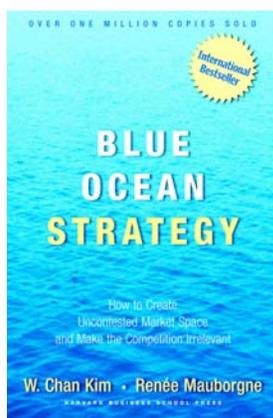
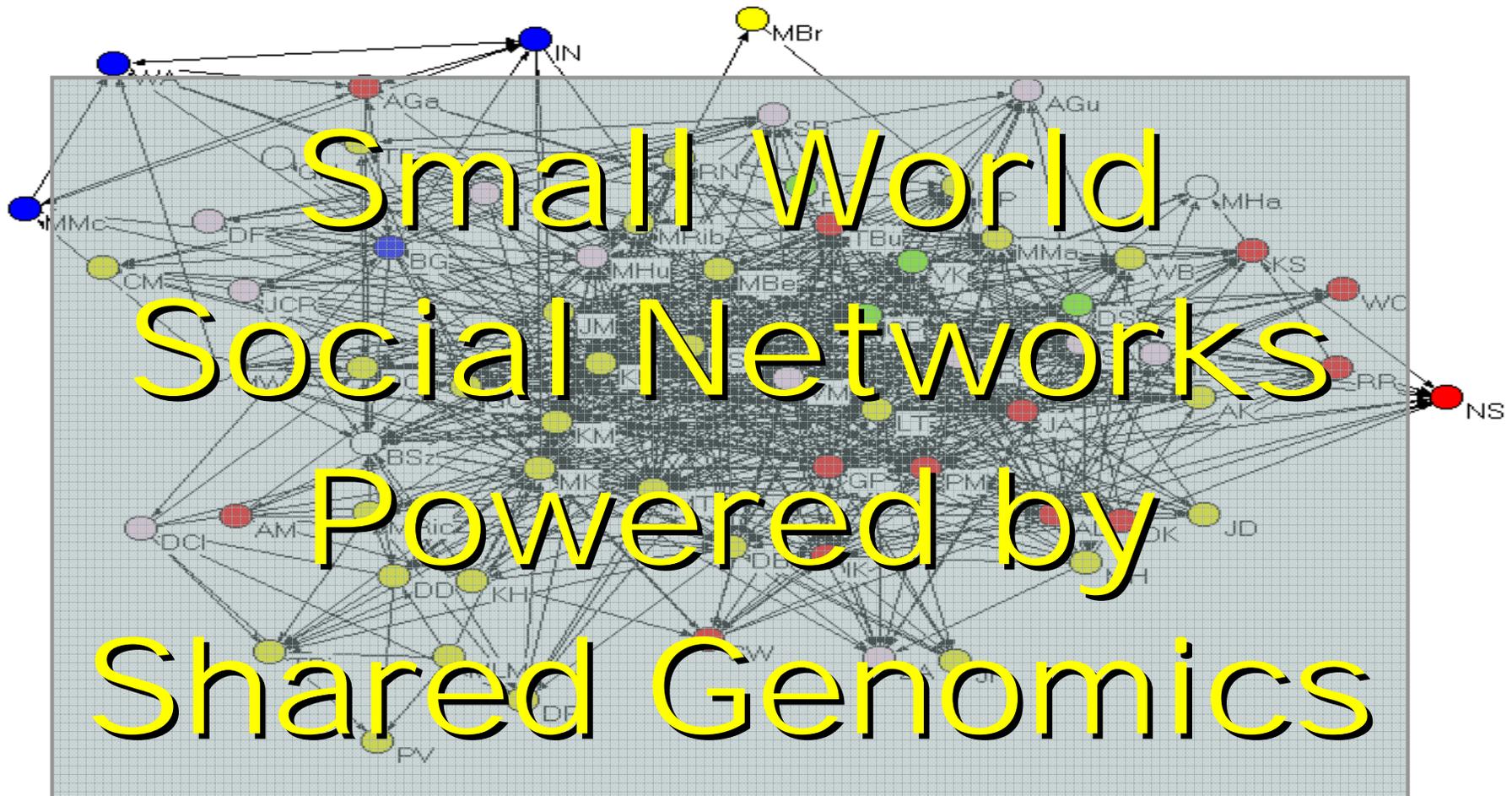
Privacy fears



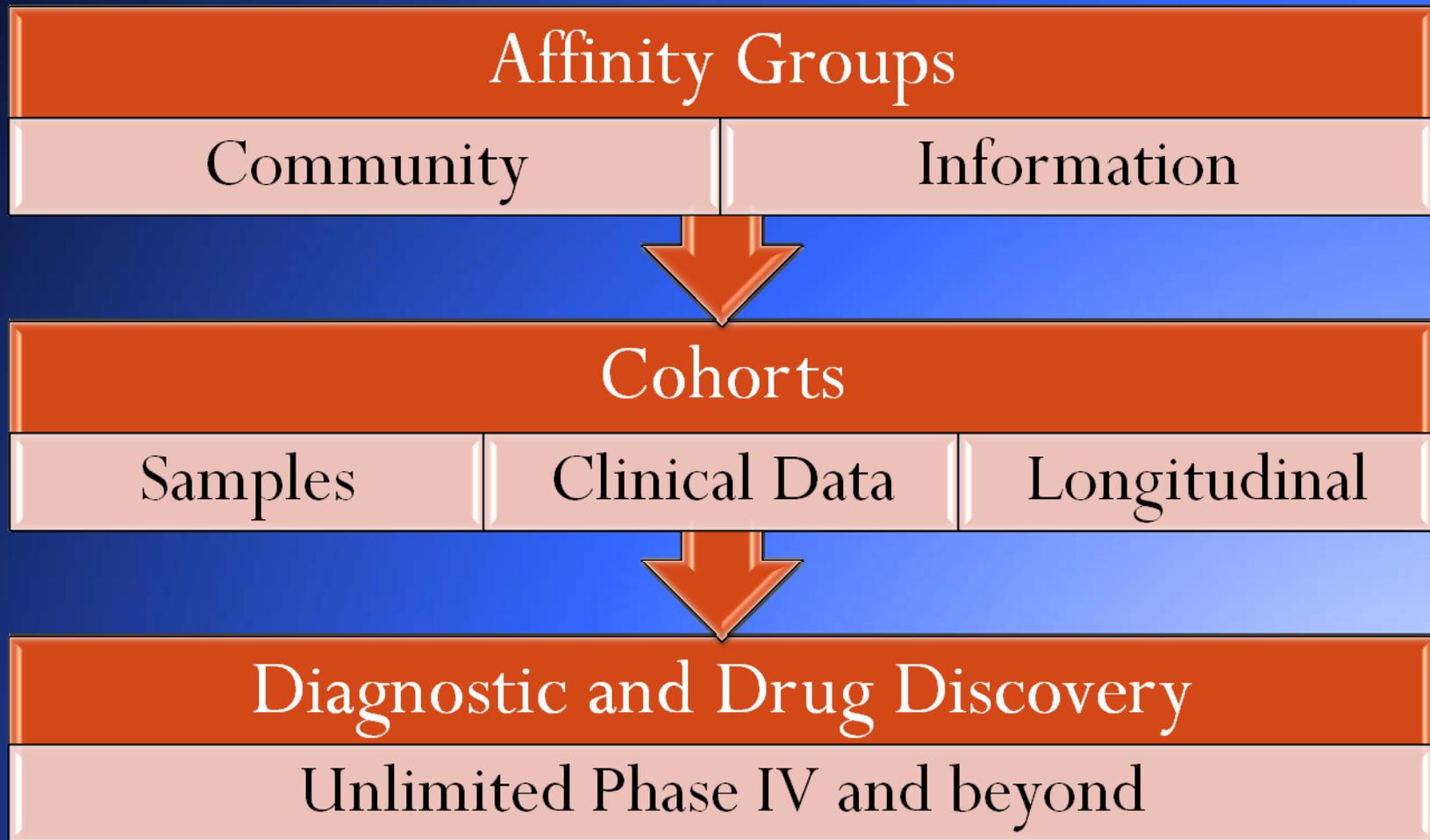
Substandard information



**There is no deli number...**



# Disruptive Innovation



# Affinity Groups

Community

Information



# Cohorts

Samples

Clinical Data

Longitudinal



Genetic Alliance BioBank



# Diagnostic and Drug Discovery

## Unlimited Phase IV and beyond

Advocacy organizations maintain adverse drug reaction registries

**BILL & MELINDA  
GATES foundation**

*Bringing innovations in health and  
learning to the global community*

**FOUNDATION**  
FOR THE  
National Institutes of Health

**ff** Cystic  
Fibrosis  
Foundation

**The C**  
**PATH**  
INSTITUTE

NATIONAL INSTITUTES  
OF HEALTH

**American  
Cancer  
Society®**

# Policy Issues



- **Innovation Access**
- **FDA Regulations**
- **CLIA Oversight**
- **Reimbursement**
- **Biomarker Funding**
- **Biobanking**
- **Healthcare Reform**
- **Adaptive Trials**
- **IP Rights**
- **Combinations & Hybrid products**
- **Info Medicine**