Advocacy Organizations and Emerging Technologies

International Summit on Gene Editing
Washington, DC
1 December 2015

Sharon F. Terry, MA
President & CEO

Genetic Alliance

Elizabeth and Ian diagnosed with pseudoxanthoma elasticum (PXE)

(rare genetic condition: vision loss, cardiovascular, gastro…)

1994

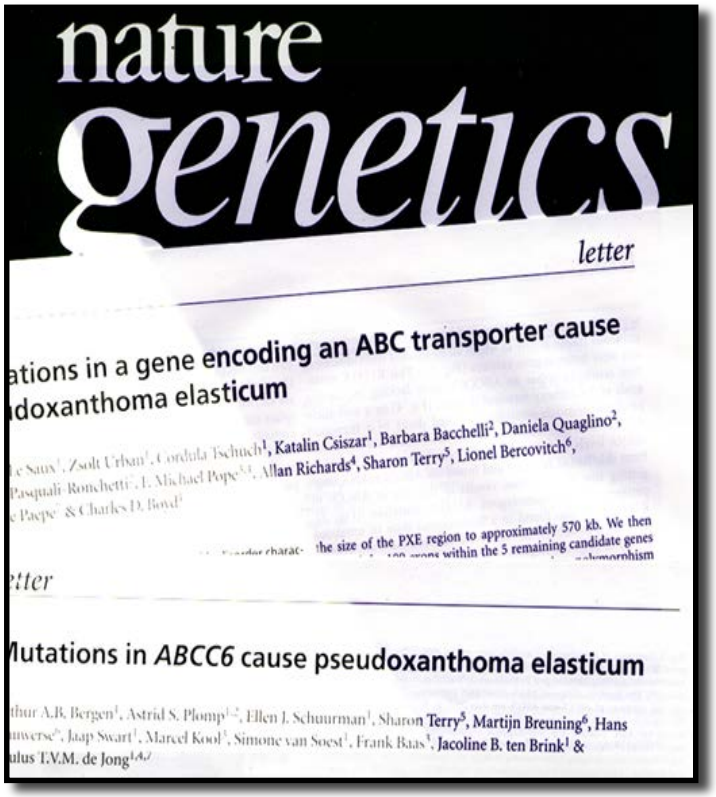
2015

Elizabeth
Teacher

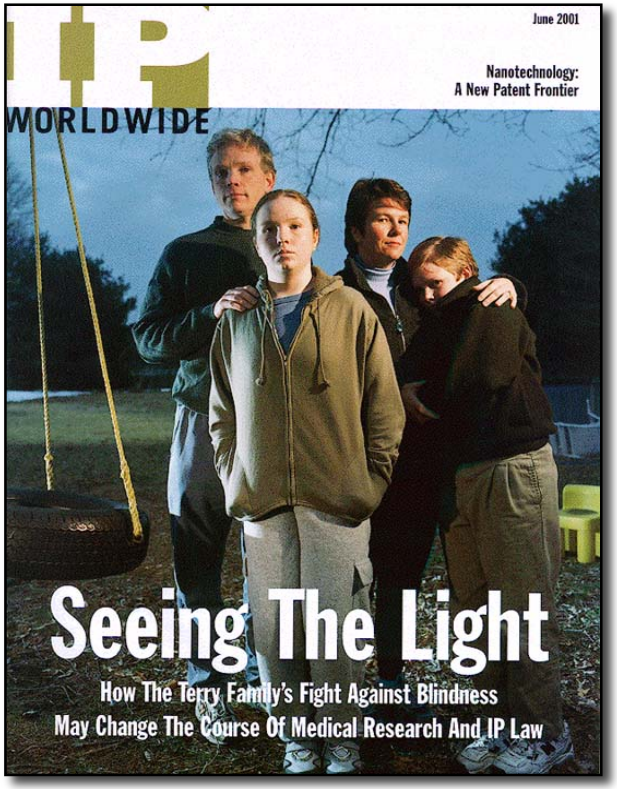
Ian
Organic Farmer

PXE international

Gene Discovery



BioBank



Testing

Clinical Diagnostic Test Development via FDA & CLIA Regulatory Strategies

Human Clinical Trials

Drug Screening & Development Approaches

Therapeutics

--Small Molecules

--Nonsense mutants

Patenting

Licensing & Intellectual Property Management

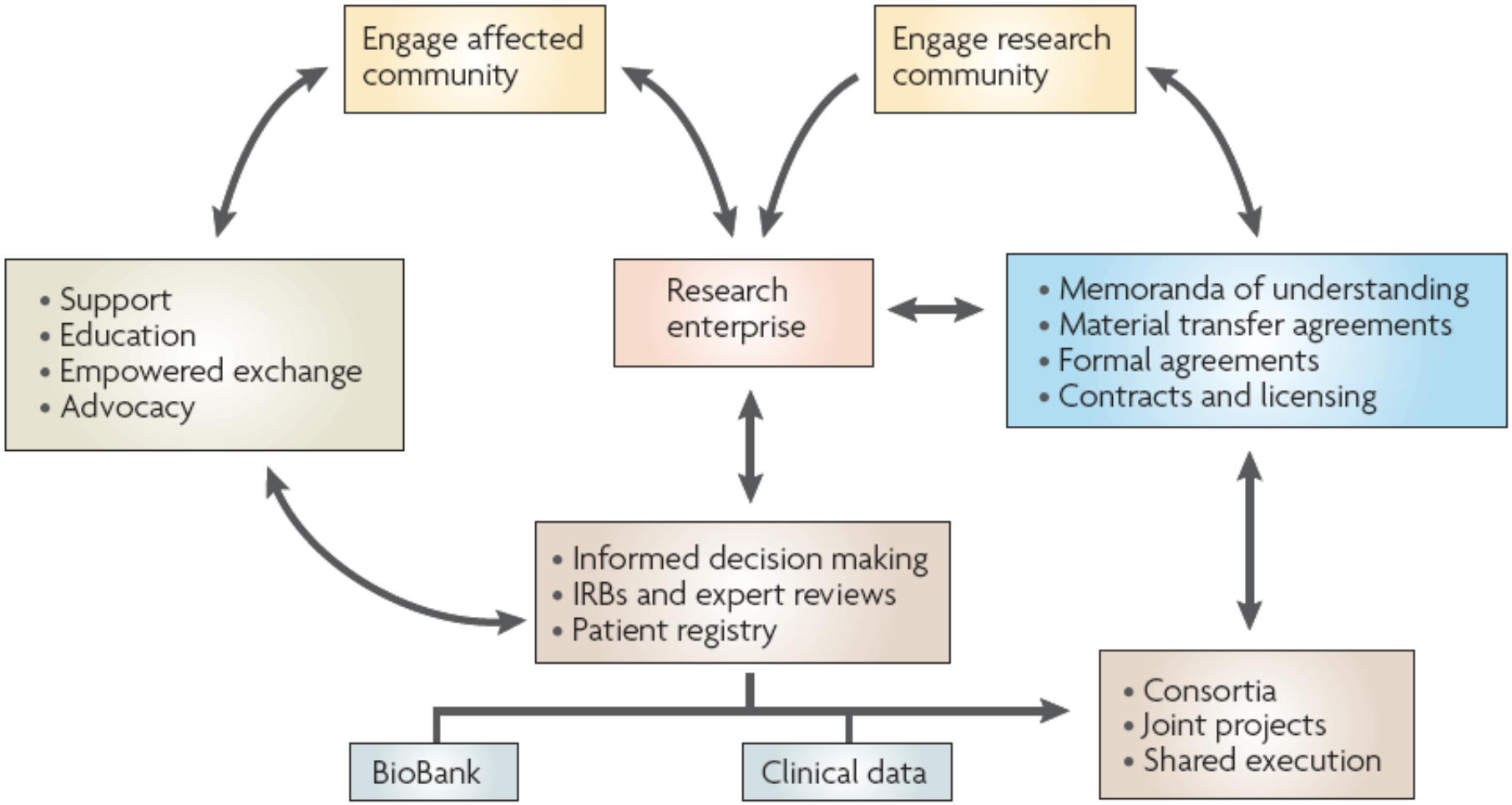


Figure 1 | **The PXE International strategy.** PXE International uses a variety of approaches to bring the PXE community together with research scientists to accelerate translational research. IRB, institutional review board.

Terry SF, Terry PF, Rauen K, Uitto J, Bercovitch L. Advocacy Organizations as Research Organizations: the PXE International example. Nature Reviews Genetics. 2007 Feb; Vol. 8, No. 2

Network of thousands of organizations around the world, 1200 of which are disease advocacy organizations.

Working to accelerate development and access to interventions for all conditions driven by patients/participants/consumers

Advocacy Orgs

As heterozygous as the diseases

- Kitchen table – Mini-pharma companies
- Rare – common
- Various inheritance: AR, AD, mtDNA
- Prenatal, newborn, childhood, adult
- Variable morbidity
- Support – research focus
- Grasstops – grassroots

Informal Poll

- Hell, yes!
- The only way we will make any progress to have a better outcome for our community is with gene editing.
- Fully explore this – scientifically, ethically.
- Gene what?
- Let’s not think about it until the technology is perfected.

One example: Batten's Disease

Antisense Oligonucleotides for the Treatment of Juvenile Neuronal Ceroid Lipofuscinosis (CLN3)
Principal Investigator: Michelle Hastings, Ph.D., Rosalind Franklin School of Medicine and Science
$50,000, BDSRA

- CLN3 is most commonly caused by a 1.02kb deletion that encompass 2 exons—7 and 8. The loss of these exons causes 'misreading' of the mRNA and ultimately not allowing whole bodily systems to work correctly. This research seeks to correct the way the mRNA functions fixing the string of proteins that affect CLN3.

To edit or not?

What is a condition/disease/problem?

- Intersex
- Deafness
- Klinefelter’s syndrome
- Turner’s syndrome
- Achondroplasia
- Weight
- Height
- Intelligence
- Social interaction

Tempered Urgency

- Needs are enormous and immediate
- Technical and ethical issues must be resolved satisfactorily
- Accessible technology
- Data sharing and transparency to accelerate progress
- Regulatory dialogue concurrent with technology development

Hope, not hype.

Sharon F. Terry, MA

sterry@geneticalliance.org