Role of the Advocacy Organization in Rare Disease Research

International Rare Disease Research Consortium
07 November 2014

Sharon F. Terry
President & CEO, Genetic Alliance
Founding CEO, PXE International
President, EspeRare
Principal Investigator, CENA

Elizabeth and Ian diagnosed with pseudoxanthoma elasticum (PXE)

1994

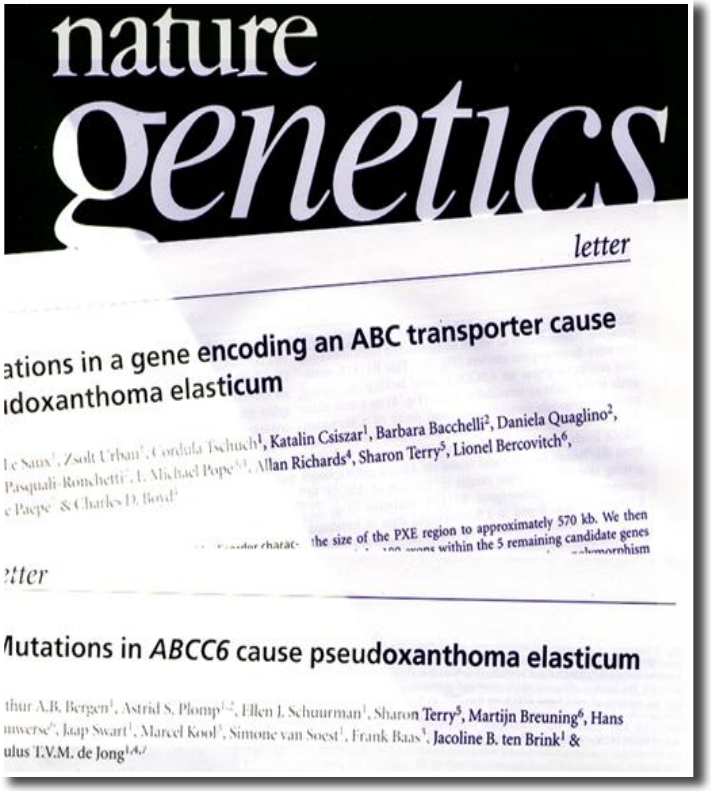
2014

Elizabeth:
Teach for America

Ian:
Organic Farmer

PXE international

Gene Discovery



nature genetics

letter

ations in a gene encoding an ABC transporter cause
ıdoxanthoma elasticum

Le Saux[1], Zsolt Urban[2], Cordula Tschuch[1], Katalin Csiszar[1], Barbara Bacchelli[3], Daniela Quaglino[3], Pasquali-Ronchetti[3], J. Michael Pope[5,6], Allan Richards[4], Sharon Terry[5], Lionel Bercovitch[6], e Paepe[7] & Charles D. Bond[1]

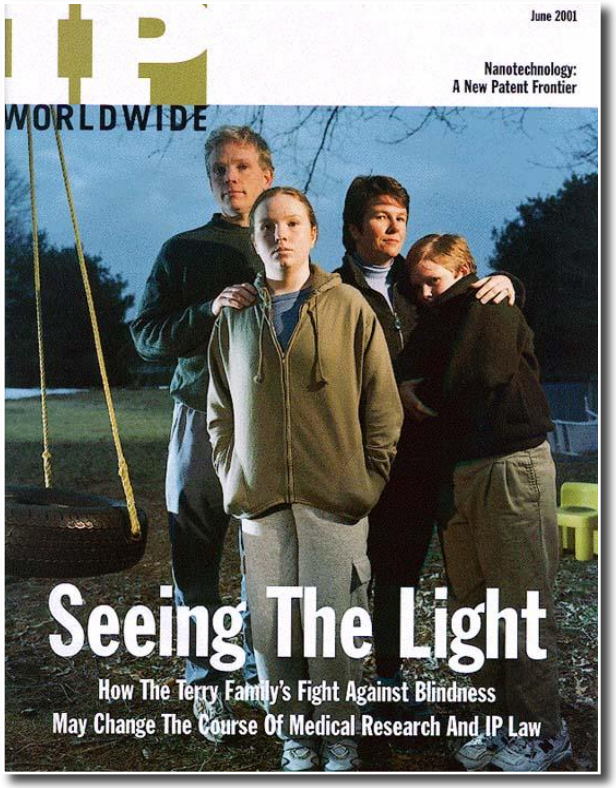
the size of the PXE region to approximately 570 kb. We then within the 5 remaining candidate genes

etter

Mutations in *ABCC6* cause pseudoxanthoma elasticum

thur A.B. Bergen[1], Astrid S. Plomp[1,2], Ellen J. Schuurman[1], Sharon Terry[5], Martijn Breuning[6], Hans unverse[6], Jaap Swart[1], Marcel Kool[1], Simone van Soest[1], Frank Baas[1], Jacoline B. ten Brink[1] & ulus T.V.M. de Jong[1,4,7]

BioBank



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

Testing

Clinical Diagnostic Test Development via FDA & CLIA Regulatory Strategies

Human Clinical Trials

Drug Screening & Development Approaches

Patenting

Licensing & Intellectual Property Management

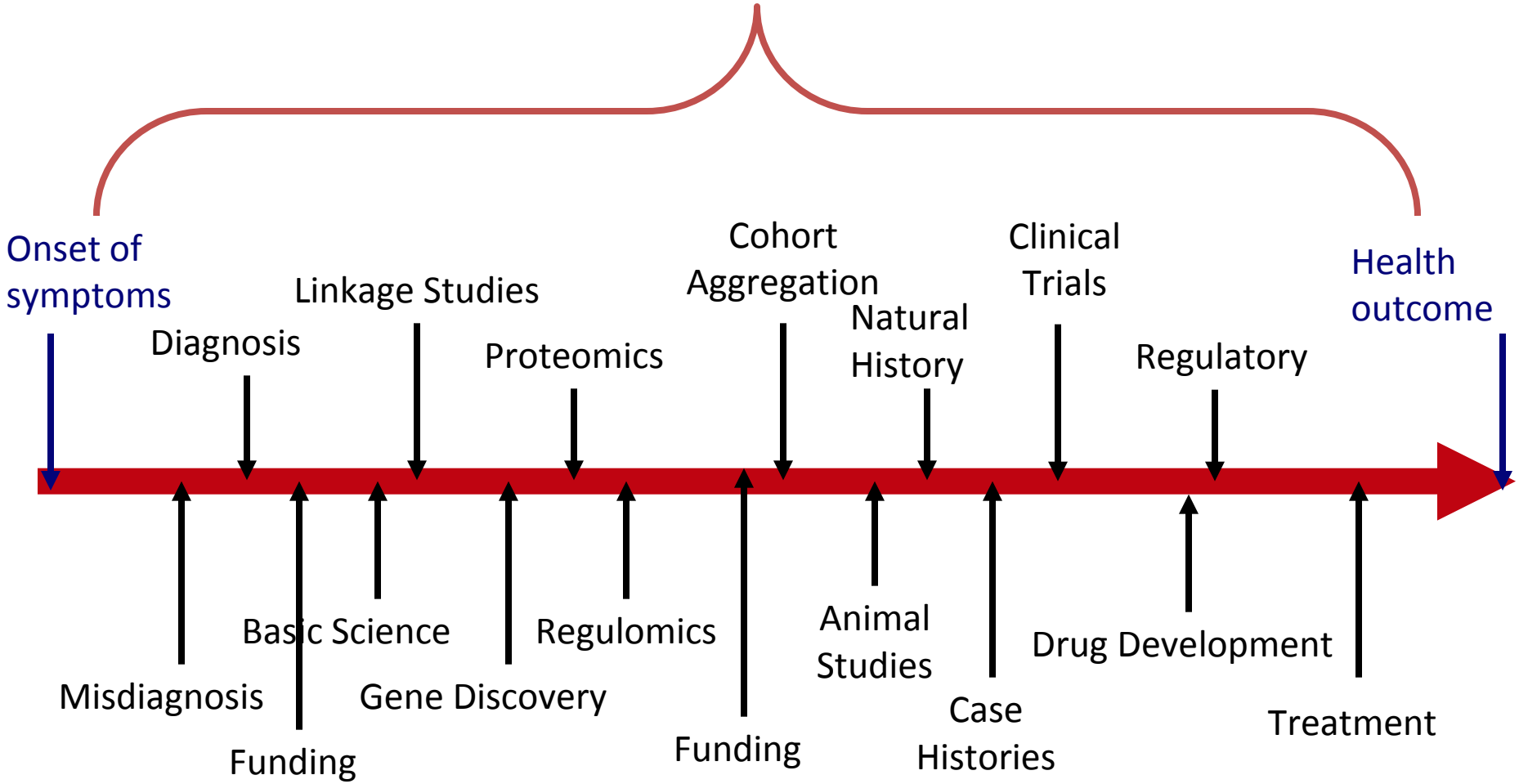
Therapeutics

--Small Molecules

--Nonsense mutants

Traversing the ‘diagnosed line’, individuals and families journey---- to seek answers.

Perspective → Vision



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** – **Networks in the Commons** – ***Translation & Delivery***

Culture Shift in Information Age

Industrial Age (old)	Information Age (new)
Control means of production	**Open** means of production
Based on **scarcity**	Based on **abundance**
Hierarchical / Command & Control	**Network** / Collaboration
Linear / Sequential	**Organic**
Win / Lose	Win / Win
Materials	**Information**

Needles in Haystacks

But for the advocacy organization, the haystack is made of needles.

DISEASE INFOSEARCH
Powered by Genetic Alliance

Enter a condition name...

Share 1 | Tweet 1 | Recommend 0 | print

Home | About | List an Organization | Learn More | Contact

Cystic Fibrosis

To find out how your experience compares with others about this condition Click Here

Common Name(s)

Cystic Fibrosis

Cystic fibrosis (CF) is an inherited condition that causes mucus to build up and clog some of the organs in the body, particularly in the lungs and pancreas. When mucus clogs the lungs, it can make breathing very difficult. The thick mucus also causes bacteria to get stuck in the airways, which causes inflammation and infections. These infections can cause chronic coughing, and
infections can lead to permanent lung
(fibrosis) and cysts in the lungs. Mucu
pancreas, leading to digestive proble
and is inherited in an autosomal rece

Source: Genetic and Rare Diseases Informa
NHGRI.

Jump to Topic | Advocacy and Support Organizations | Health Questions | Reco
Scientific Literature | Symptoms, Diagnosis, and Treatment | Clin

Advocacy and Support Organizations

How do you compare with others?

Login to Reg4ALL

Your answers: 0

Cystic fibrosis (also known as CF) is a common hereditary disease which affects the entire body, causing progressive disability.

I have been diagnosed with cystic fibrosis...

- no
- yes

feedback on question (0)

Don't know | Skip

Advocacy and Support Organizations

Condition Specific Organizations

Following organizations serve the condition "Cystic Fibrosis" for support, advocacy or research.

Boomer Esiason Foundation ›

The Boomer Esiason Foundation is a dynamic partnership of leaders in the medical and business communities joining with a committed core of volunteers to heighten awareness, education and quality of life for those affected by cystic fibrosis, while providing financial support to research aimed at finding a cure.

http://www.esiason.org

Last Updated: 15 Feb 2013

View Details

Center for Jewish Genetics ›

The Center is dedicated to gathering and disseminating knowledge about Jewish genetic disorders and hereditary cancers. Its mission is to educate and serve health care professionals, clergy and the Jewish community.

http://www.jewishgenetics.org

Last Updated: 26 Dec 2012

View Details

Cystic Fibrosis Foundation ›

Our mission is to assure the development of the means to cure and control Cystic Fibrosis and to improve the quality of life for those with the disease.

http://www.cff.org

Last Updated: 19 Mar 2013

View Details

Scientific Literature

Top

Articles from the PubMed Database

Research articles describe the outcome of a single study. They are the published results of original research.
The terms "Cystic Fibrosis" returned 4176 free, full-text research articles on human participants. First 3 results:

Ivacaftor in a G551D homozygote with cystic fibrosis. ›

Author(s): Michael J Harrison, Desmond M Murphy, Barry J Plant
Journal: N. Engl. J. Med.. 2013 Sep;369(13):1280-2.

Last Updated: 26 Sep 2013

Go To URL

Genetic interaction of GSH metabolic pathway genes in cystic fibrosis. ›

Author(s): Fernando Augusto de Lima Marson, Carmen Sílvia Bertuzzo, Rodrigo Secolin, Antônio Fernando Ribeiro, José Dirceu Ribeiro
Journal:

Last Updated: 19 Jun 2013

Go To URL

Cystic fibrosis (CF) is a monogenic disease caused by CFTR gene mutations, with clinical expression similar to complex disease, influenced by genetic and environmental factors. Among the possible modifier genes, those associated to metabolic pathways of glutathione (GSH) have been ...

The role of serum Pseudomonas aeruginosa antibodies in the diagnosis and follow-up of cystic fibrosis. ›

Last Updated: 22 May 2013

Reviews from the PubMed Database

Review articles summarize what is currently known about a disease. They discuss research previously published by others.
The terms "Cystic Fibrosis" returned 418 free, full-text review articles on human participants. First 3 results:

Transepithelial nasal potential difference (NPD) measurements in cystic fibrosis (CF). ›

Last Updated: 10 Jun 2013

Author(s): Dorota Sands
Journal: Med Wieku Rozwoj. ;17(1):13-7.

Go To URL

The main underlying physiologic abnormality in cystic fibrosis (CF) is dysfunction of the CF transmembrane conductance regulator (CFTR), which results in abnormal transport of sodium and chloride across epithelial surfaces. CFTR function could be tested in vivo using measurements ...

Ivacaftor treatment in patients with cystic fibrosis and the G551D-CFTR mutation. ›

Last Updated: 4 Mar 2013

Author(s): Isabelle Sermet-Gaudelus
Journal: Eur Respir Rev. 2013 Mar;22(127):66-71.

Go To URL

Cystic fibrosis (CF) is an autosomal recessive lethal disease caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene that encodes for CFTR, an epithelial cell-surface expressed protein responsible for the transport of chloride (Cl(-)). Gating mutations ...

Targeting a genetic defect: cystic fibrosis transmembrane conductance regulator modulators in cystic fibrosis. ›

Last Updated: 4 Mar 2013

Author(s): Nico Derichs
Journal: Eur Respir Rev. 2013 Mar;22(127):58-65.

Go To URL

Clinical Trial Information

Top

This information is provided by ClinicalTrials.gov

Metabolic Efficiency of Combined Pancreatic Islet and Lung Transplant for the Treatment of End-Stage Cystic Fibrosis ›

Status: Not yet recruiting
Condition Summary: Cystic Fibrosis; Diabetes Related Cystic Fibrosis

Last Updated: 5 Mar 2012

Go to URL

Exocrine Pancreatic Function Testing in Cystic Fibrosis ›

Status: Recruiting
Condition Summary: Cystic Fibrosis

Last Updated: 8 Jul 2013

Go to URL

Circadian Rhythm In Tobramycin Elimination In Cystic Fibrosis ›

Status: Recruiting
Condition Summary: Cystic Fibrosis

Last Updated: 4 Oct 2012

Go to URL

According to ClinicalTrials.gov there are currently 204 additional "open" studies for "Cystic Fibrosis" (open studies are recruiting volunteers) and 604 "Cystic Fibrosis" studies with "all" status. Visit ClinicalTrials.gov now to view them. Or alternatively, consider TrialsFinder for assistance:

TrialsFinder®

Relief is when you and the right researcher find each other

Finding the right clinical trial for Cystic Fibrosis can be challenging.

PRIVACY ASSURED

The Advocacy ATLAS

Accessible Tools for Leadership and Advocacy Success

www.geneticalliance.org/advocacy-atlas

WikiAdvocacy.org

page | discussion | view source | history

navigation
- Main page
- Community portal
- Current events
- Recent changes
- Random page
- Help

search

Search

Go | Search

toolbox
- What links here
- Related changes
- Special pages
- Printable version
- Permanent link

Main Page

Welcome to WikiAdvocacy

WikiAdvocacy is a free, reader-built guide, as well as a community for advocacy. It covers every aspect of founding and growing an advocacy organization from fundraising to detailed explanations of issues, skills, and the elements of creating a registry and samples repository.

If this is your first time visiting WikiAdvocacy, we recommend reading **About WikiAdvocacy** and **How to Get Started**. We also recommend checking out the **community portal** and **help** pages.

| About WikiAdvocacy | How to Get Started | Community Portal | Help | How to: Create a New Wiki Page | How to post a file on WikiAdvocacy

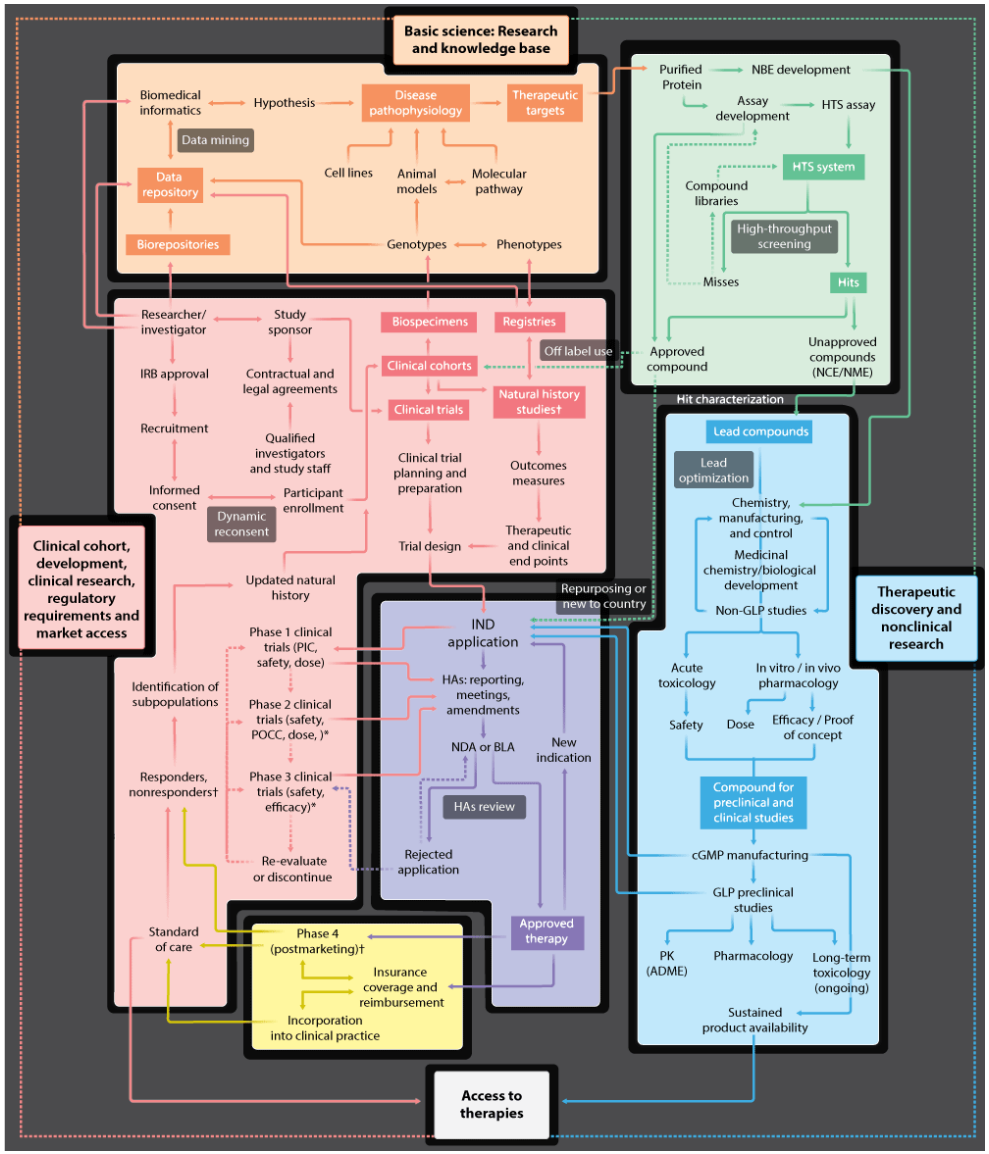
Introduction
- Defining Our Terms
- The Power of Advocacy Organizations
- Why Go There?
- How to Use This Guide
- History of Advocacy Organizations and Genetic Alliance
- The Kitchen Table Is a Good Place to Start
- Genetic Alliance

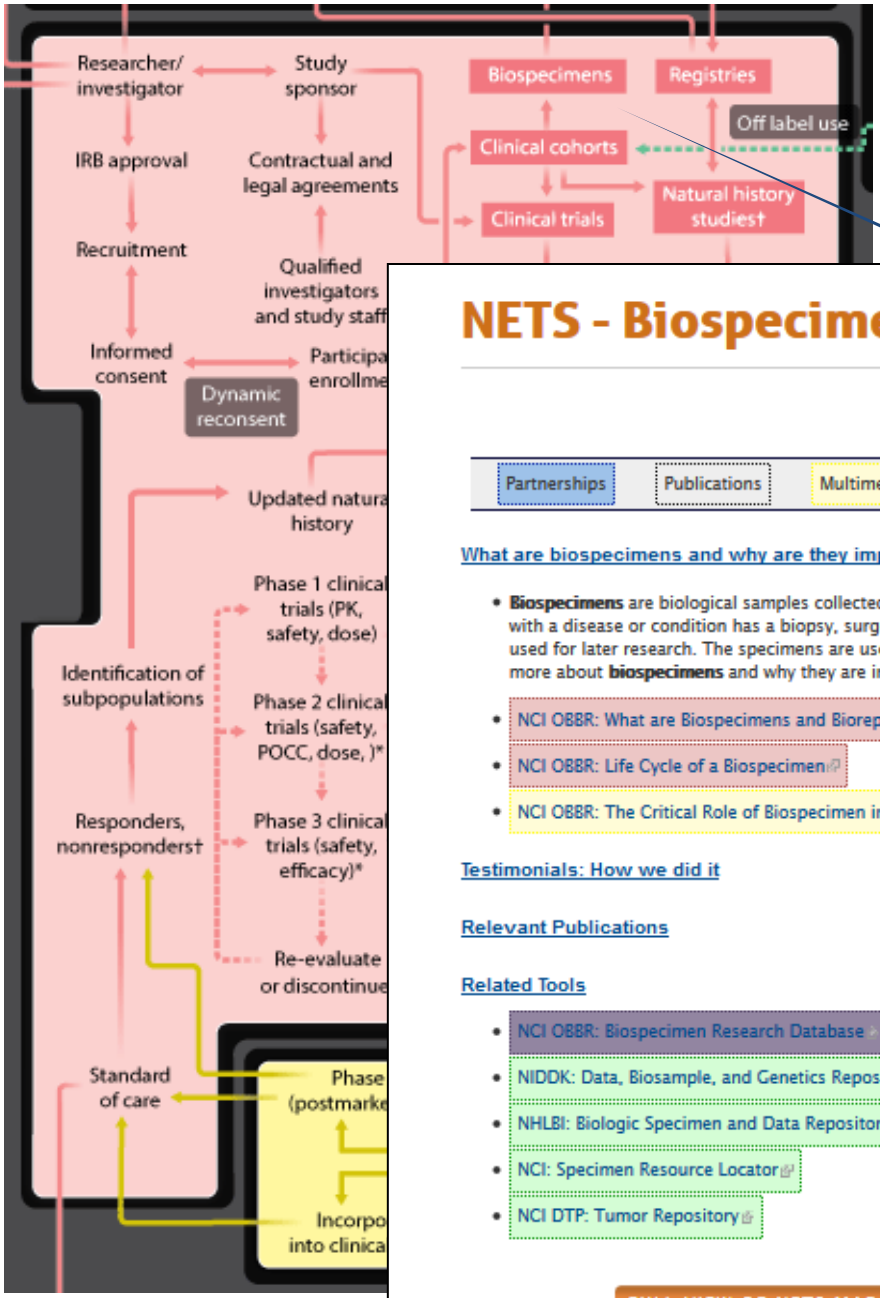
Organizational Assessment
- Determine Goals
- Characterize Condition
- Characterize Resources
- Compare Goals and Resources, with the Characteristics of the Condition
- Considering Starting a Support Group for a Condition that Already Has One?

Organization Development: Organizational Structure
- Advisory Boards
- Bylaws and Articles
- Governance Board
- Ensuring Cultural Competence
- Finding a Lawyer
- Insurance and Policies
- International Offices
- NAC Best Practices
- Organization's Name
- Staff
- Tax and Finance

Organization Development: Resources for Development
- Becoming the Organization You Imagine
- Building a Website
- Charity Rating Listings
- Conference Call Services
- Donations
- Events
- Financial Assets
- Getting Grants
- Harnessing the Resources That Are Hard to Measure
- Helping Your Membership Help Your Group
- How to Obtain Donated Office Space

Navigating the Ecosystem of Translational Science





NETS - Biospecimens

Partnerships | Publications | Multimedia | Worksheets | Guides | Toolkits | Databases | Services

What are biospecimens and why are they important in drug development?

- **Biospecimens** are biological samples collected from humans such as blood samples, tumor samples, or other tissues. When someone with a disease or condition has a biopsy, surgery, or other procedure, often a small amount of the specimen taken can be stored and used for later research. The specimens are used in biomedical research to learn about how disease affects the human body. To learn more about **biospecimens** and why they are important for drug development, click on the links below.
- NCI OBBR: What are Biospecimens and Biorepositories?
- NCI OBBR: Life Cycle of a Biospecimen
- NCI OBBR: The Critical Role of Biospecimen in Cancer Research

Testimonials: How we did it

Relevant Publications

Related Tools

- NCI OBBR: Biospecimen Research Database
- NIDDK: Data, Biosample, and Genetics Repository
- NHLBI: Biologic Specimen and Data Repository Information Coordinating Center
- NCI: Specimen Resource Locator
- NCI DTP: Tumor Repository

FULL VIEW OF NETS MAP | NEIGHBORHOOD VIEW OF NETS MAP | BACK TO NETS

‘Registry and BioBank in a Box’

- 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

Where do I begin?	What is a biobank?	Guidelines for considering a registry/biobank	Advocates are leaders in biobanking	
How do I make this a reality?	Registry/ repository start-up guide	Making your organization's biobank a reality	Genetic Alliance registry/repository boot camps	
Is my organization ready?	Organizational readiness checklist	Biobank question & answer session		
How do I select a vendor?	Vendor assessment summary	Vendor assessment worksheet	Landscape analysis manuscript	
Considering Genetic Alliance?	Genetic Alliance BioBank	GARB FAQs	Virtual tour of Genetic Alliance registry solutions	GARB history manuscript
What else do I need to know?	Biobank governance	Biobank governance checklist	Resource list	
How do I stay current?	Registry and Biorepository Bulletin	BioBank discussion listserv	BioBank Ambassadors mentoring	Weekly tips

Publication *Training/mentoring* *Videos* *Webinar* *Webpage* *Worksheet*

Platform for Engaging Everyone Responsibily – branded according to the advocacy organizations wishes

The portal fits directly onto any web page and retains the host site's 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.

home about us scientific info conference events news shop donate join us member login contact us

Joubert Syndrome & Related Disorders Foundation

Together we can advance research and improve care

For over 22 years, JSRDF has played an important role in educating physicians and their support teams as well as increasing public awareness about Joubert syndrome. The goal of JS-LIFE (Joubert Syndrome Link to Information & Family Exchange) is to extend this tradition of leadership by enabling access to a much broader research community of clinical, neuroimaging, and genetic studies. This information will help us discover patterns within the data collected and bridge the gap in knowledge that exists among other related disorders and ciliopathy researchers. Please join us. Simply click on Start Now! to begin, or Sign In if you are a returning user.

Support | Educate | Research

Joubert Syndrome Link to Information & Family Exchange

It's quick and simple!

1. Sign up! (or sign in) Start Now!
2. Take a health survey
3. Let researchers find you
4. Share this with others!

Privacy and Sharing in Perfect Balance

To help us protect your individual privacy in accordance with rules that you establish, we harness the power of Private Access with the specific goal of helping you make your health information available to top researchers - under your own terms.

PRIVACY ASSURED

MY DOCTOR OR DISEASE ADVOCACY GROUP RECOMMENDED THIS SERVICE AND PROVIDED ME WITH A REFERRAL #:

Enter referral # here Submit

Privacy Policy Terms of Service Give Feedback

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Everything is ready and because it is patient-centric. 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 trusted, local, "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?

PRIVACY ASSURED

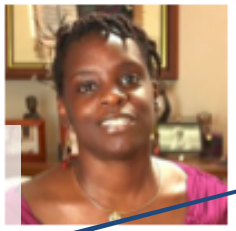
Private Access lets you control who can see your information, and for what purpose. This service will check your Private Access settings before sharing any of your information.

Multiple guides give an opportunity to use a variety of approaches, and selecting settings that are the most comfortable to each participant. Well known guides can be used across an entire condition, or be specific to an individual disease organization

To enable ease *and* an extraordinary range of granularity

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

Set Privacy Settings : For New User



Nicole Ford's suggested privacy settings for persons with : Moderate concerns about privacy

1. Use the above selected level or choose a level of concern about privacy that more closely reflects your views.
2. If you are satisfied with Nicole's suggested privacy settings shown below, click "Accept and continue".
3. If not, either click "Customize" to refine these settings, or choose "Select a different guide".

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

YOU ARE CURRENTLY VIEWING SUGGESTED PRIVACY SETTINGS FOR New User

← Click to learn more

Who can access it?

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)
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	Allow
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

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

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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

For multiple categories of uses, and specified usage rights

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

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.

Joubert Syndrome & Related Disorders Foundation

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
updated today!
Record information about biological markers including unique genes, proteins and other molecules derived from advanced genetic testing and molecular analyses.

Family History — Update
updated today!
Create and annotate a family tree with potentially germane elements of family health history, heritable characteristics, and related ancestry information.

Medical Records — *Coming Soon!*
Create, send, check on the status of requests to access, and view records from medical professionals* so this information can be included in the registry.

* Note: Patients (or their legally appointed representatives) have a federally guaranteed right to see and get a copy of the patient's medical

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

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updated today!
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Family History
Create and annotate a family tree with p… family health histo… characteristics, and related ancestry inf…

WEIGHT

May 2012
Dates with entries for THIS question
Dates with entries for ANY question
Show Calendar (Select Date)
May 2012
June 2011
Aug 10, 2010
May 12, 2010
Feb 2009

My weight was approximately: 285 lbs

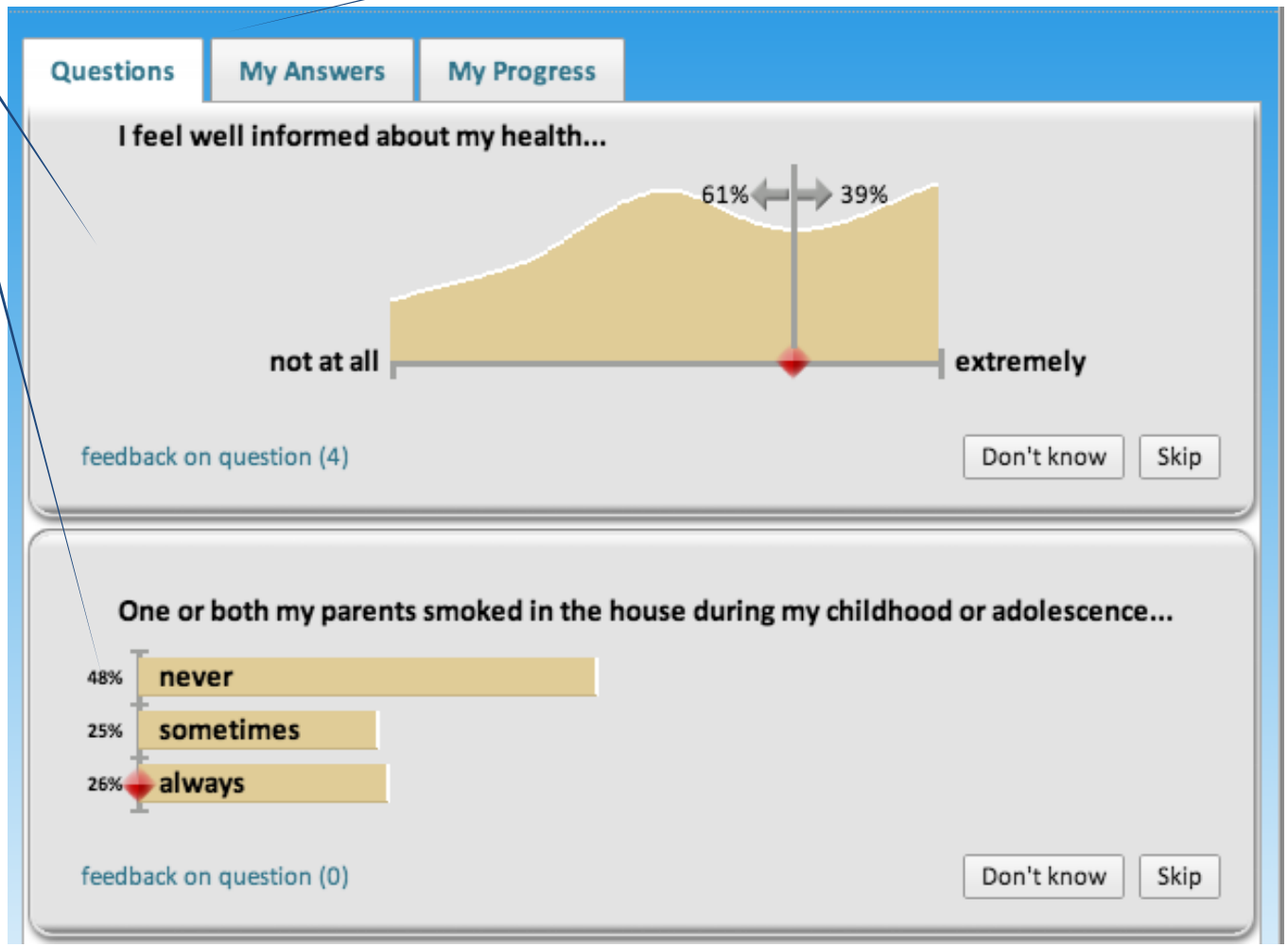
285

400
300
200

“Gamified” Interface for Questions and Answers

Questions appear in a dynamic user interface, and provide immediate feedback on how others responded to the same question…

Participants can review their prior answers, make updates and/or remove the data at any time.



National Gaucher Foundation, Inc.
EDUCATION • SUPPORT • HOPE

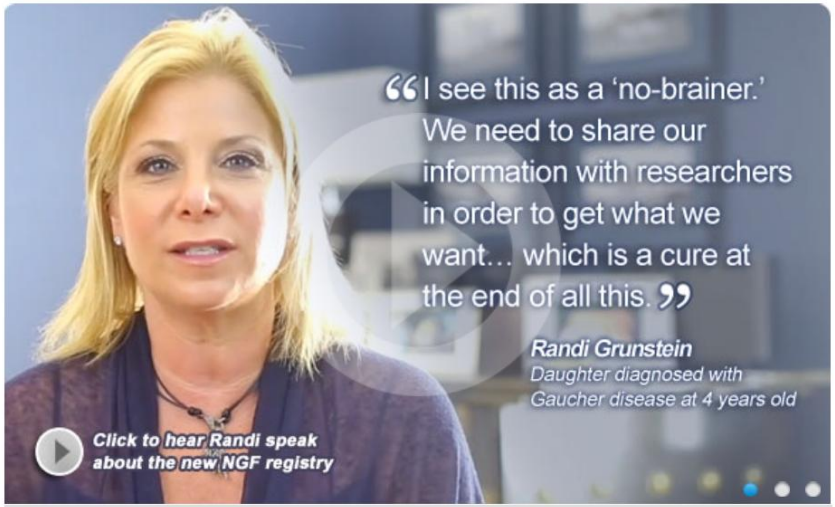
You're not alone.

The NGF provides support through financial, educational and research programs

Gaucher Disease | Treatments/Pharmas | Resources | News | Events | Programs | Media Library | Support NGF

The Gaucher Network Registry will help advance research and improve care!

By sharing our stories in a way that safeguards each individual's respective privacy concerns, the information we provide can be a vital force in helping to advance research. That's why the NGF is sponsoring the new Gaucher Network Registry (GNR), the first ever participant powered Gaucher registry. Through learning more about how Gaucher disease affects each individual and -- with express permission – making it easier for top researchers to contact us about new studies, we can accelerate our quest to find a cure for this debilitating and often fatal disease. Simply click on Start Now! to begin, or Sign In if you are a returning user.



Respecting Your Privacy is a Priority

PRIVACY ASSURED with PrivateAccess

To help us protect your individual privacy in accordance with rules that you establish, we harness the power of Private Access with the specific goal of helping you make your health information available to top researchers - under your own terms.

It's quick and simple!

1. Sign Up! (or sign in) — Start Now!
2. Take health survey
3. Let researchers find you
4. Share with others!

MY DOCTOR OR DISEASE ADVOCACY GROUP RECOMMENDED THIS SERVICE AND PROVIDED ME WITH A REFERRAL #:

Enter referral # here | Submit

Privacy Policy | Terms of Service | Give Feedback

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Citizens For Quality Sickle Cell Care

Now We Have A Voice!

0

Let's tell the FDA how important it is to find a cure for Sickle Cell Disease

April 8th is the last day for the Food & Drug Administration (FDA) to hear public comments about sickle cell disease, and our voices need to be heard. This is our opportunity to tell the government about the needs and concerns of millions of Americans affected by sickle cell trait and disease. You can help just by answering a few questions; and if you want, you can also allow researchers to alert you about new studies for which you may qualify...

"This is our chance to tell the FDA about the need for more drugs and therapies of people with Sickle Cell Disease"

Katrina Rice
44 year old mother of a child with Sickle Cell Disease

We Respect Your Right To Privacy

PRIVACY ASSURED with PrivateAccess

We understand that many people in our community have concerns about privacy. We are committed to protecting your right to decide how active and open to be. And that's why we're using Private Access to let you establish the rules for who can see your information, when, and for what purpose.

It's Easy as 1, 2, 3, 4

1. Protect your privacy — Start Now!
2. Tell the FDA your opinion
3. Provide More Health Info
4. Help Sprade The Word!

MY DOCTOR OR DISEASE ADVOCACY GROUP RECOMMENDED THIS SERVICE AND PROVIDED ME WITH A REFERRAL CODE:

Enter referral code here | Submit

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Let's tell the FDA how important it is to find a cure!

By April 8, the FDA needs to hear from us about sickle cell disease, and it's critical your voice is heard. This is our opportunity to tell the government about the needs and concerns of millions of Americans affected by sickle cell trait and disease. You can help just by answering a few questions; and if you want, you can also allow researchers to alert you about new studies for which you may qualify...

"Priorities for research and advocacy should align with real-world perspectives of people who are affected by Sickle Cell Disease."

Mattie Robinson
Project Manager
William E. Proudford Sickle Cell Fund, Inc.

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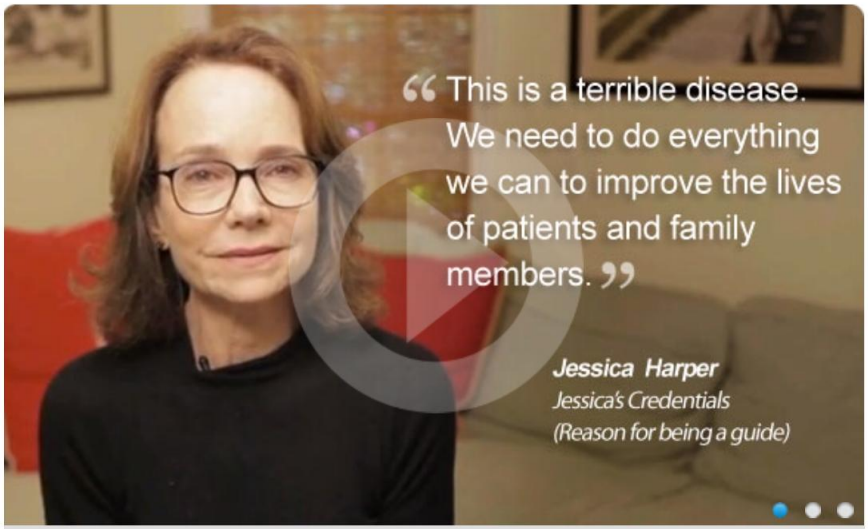
PXE international

About Us | About PXE | Research | Get Involved | News | Events

Your piece of the puzzle is a critical one!

We do not understand how PXE progresses, or how to slow it down. It is time to change that! Please join our effort to complete the puzzle! Simply click on Start Now! to begin, or Sign In if you are a returning user.



Respecting Your Wishes is Our Priority

PRIVACY ASSURED with PrivateAccess

To help us protect your individual privacy in accordance with rules that you yourself establish, we harness the power of Private Access with the specific goal of helping you make your health information available to top researchers for your condition - under your own terms.

It's quick and simple!

1. Sign Up! (or sign in) — Start Now!
2. Take health survey...
3. Let researchers find you...
4. Share this with others!

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Privacy Policy Terms of Service Give Feedback

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PEER Deployed

- Joubert Syndrome LIFE Registry: https://www.jsrdf.org/JSLIFE
- Gaucher Registry: https://www.gaucherdisease.org/gaucher-network-registry.php
- Pseudoxanthoma Elasticum Registry: https://www.pxe.org
- Registries for All: https://www.reg4all.org
- TrialsFinder: https://www.trialsfinder.org
- Free the Data: https://www.free-the-data.org
- United Mitochondrial Disease Foundation: http://www.umdf.org/site/c.8qKOJ0MvF7LUG/b.9135169/k.D604/Registry.htm
- North Alabama Sickle Cell Foundation:http://sicklecellna.org/peer.php
- Sickle Cell Disease Association of America, Southern Connecticut: http://www.scdaaofsouthernct.org/tellthefda.html

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.

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**.

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$1.26 Billion

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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)

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

EspeRare: Merging patients & commercial interests

Mission

In partnership with advocacy 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

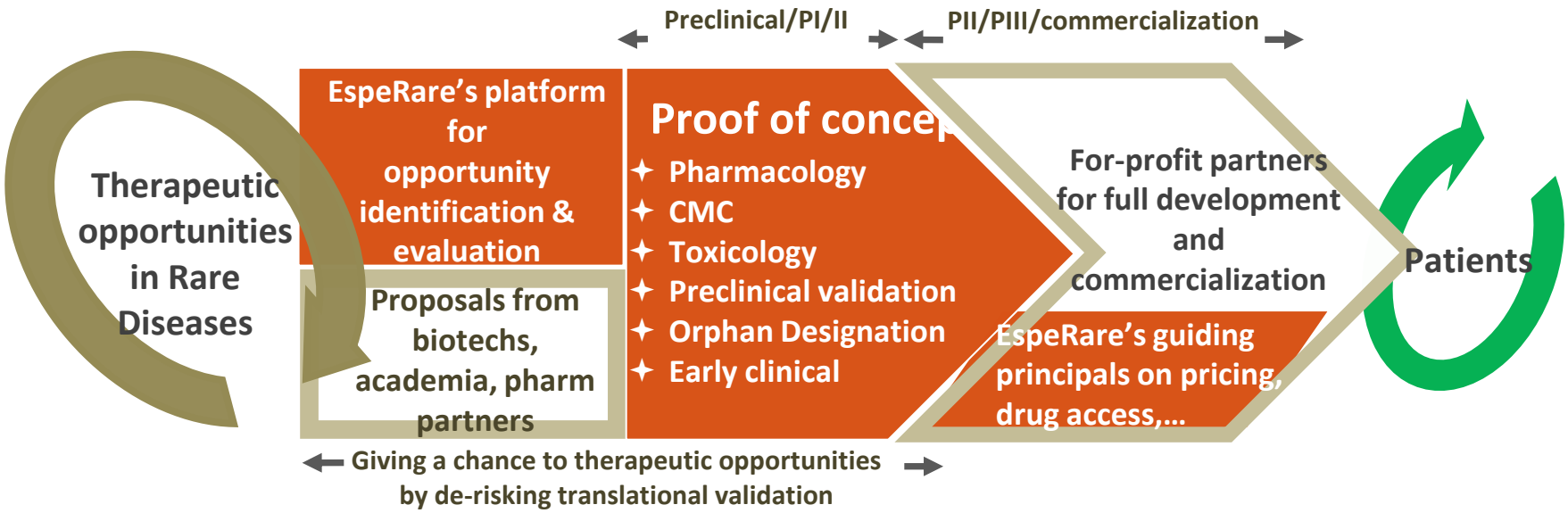
✦ **De-risk early development of rare diseases programs:**

- 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

EspeRare uncovers therapeutic opportunities and aims to insure treatment access



Include Informed Public in All Stages of Clinical Research

- Participant perspective to study design
  - Protocol Review
  - IRB service
  - Data safety monitoring boards
- Informed decision making
- Reasonable compliance - AIDS Community examples
- Enhanced community education
- Better cohort accrual - Herceptin example
- Improve participant retention
- Advance public trust in research

Advocacy Organizations

- Willing partners
- Ready to work hard
- Have exceptional access
- Are excellent allies
- Add value throughout the system
- Technical assistance for them

Contact Information

For more information:

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(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

What do we have to lose?

Everything.