

Understanding Rare Disease Registries

> Part 1 July 31, 2013

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KEY CONSIDERATIONS FOR PLANNING AND BUILDING A ROBUST PATIENT REGISTRY

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Overview

- **Dr. Shira Kramer**, President, GlobalEpi Research. *Key Considerations for Planning and Building a Robust Patient Registry*
- Dr. Stephen Groft, Director, NIH Office of Rare Diseases Research (ORDR). Global Rare Diseases Patient Registry Data Repository Pilot Project Overview
- Dr. Marshall Summar, Division Chief, Children's Research Institute, Center for Genetic Medicine Research, Children's National Medical Center. Lessons Learned from the Urea Cycle Disorders Consortium and the Rare Disease Clinical Research Networks
- **Megan O'Boyle**, Board of Directors, Phelan-McDermid Syndrome Foundation. The Phelan-McDermid Syndrome International Registry: Lessons Learned in Building a Patient Registry







What is a patient registry?

- Collection of data on patients with a specific condition (or related conditions)
- <u>Purpose</u> defines the registry
 - Locate and list patients ("rolodex")
 - Facilitate patient/family networking
 - Clinical trial recruitment
 - Evaluate patient-reported outcomes
 - Collect natural history of disease
 - Drug development
 - Research tool
 - Improve clinical care and establish evidence-based medical practices





What is a Natural History Study?

- Collection of data <u>over time</u> on patients with a specific condition to determine the "typical" progression of the disease <u>in the absence</u> <u>of treatment</u>
- Term is sometimes used interchangeably with "patient registry"
 - Some subtle differences, but concepts are similar
 - Most of the rest of the webinar will use the term "patient registry," but concepts apply to natural history studies as well





Data sources for a patient registry

Patient-reported data

- Patients live with diseases every day and can provide very accurate information about their experiences, patient-reported outcomes, quality of life, etc.
- Clinician- or health professional-reported data
 - The addition of some clinician-entered data and/or data from medical records and laboratory tests is optimal, but not always feasible

Voluntary, opt-in

- Patients in registries are unlikely to be representative of <u>all</u> patients with the condition
- Except in rare circumstances, it is not optimal for estimating incidence or prevalence of the condition





- A. Identify key stakeholders and create a Registry Advisory Committee
- B. Establish registry goals (short- and long-term)
- C. Determine the target population and inclusion/exclusion criteria
- D. Establish registry protocol, policies and data management plan
- E. Determine the budget and consider mechanisms for sustainability
- F. Determine registry platform
- G. Design data collection tools





A. Identify key stakeholders and create a registry Advisory Committee

- Advisory Committee should include experts on the condition and experts in registry design/analysis
 - Disease experts, KOLs, and/or treating physicians
 - Representatives of patients, patient advocacy group(s), disease foundations
 - Registry management staff
 - Epidemiologists
 - BioPharma (when possible)
 - Other registry stakeholders
- One person/entity must be responsible for final decisions and managing the Advisory Committee





B. Establish registry goals (short- and long-term)

- Advisory Committee/stakeholders should define up-front the goals for the registry
 - Independent of budgetary considerations
 - Ensure that the registry goals map to the issues of greatest relevance to the patient community, researchers/drug developers, the FDA (or other regulatory bodies), or other relevant stakeholders
- Registry design, registry platform selection, and data collection tools should reflect these goals
- To the degree possible, design for future value (long-term goals), even if initial efforts can only accomplish short-term goals
 - Example: Can take many years to collect complete data on natural history, but you have to start somewhere





C. Determine the target population and inclusion/exclusion criteria

- Identify target population
 - All patients with the disorder, regardless of etiology?
 - Patient subgroups
 - Geographies
 - Likelihood of identification
- Inclusion/exclusion criteria
 - Case definition
 - Data requirements and clinical validation
 - Case classification criteria





D. Establish registry protocol, policies, and data management plan

- Develop protocols/policies/plans for:
 - Registry management and coordinating data collection and recruitment/retention
 - Data quality control and/or curation of medical records (when applicable)
 - Addressing issues of privacy and data ownership
 - Addressing IRB, HIPPA, and other requirements
 - How to evaluate data requests and how data will be shared
- Speaker in webinar #2 will address these issues and considerations in greater detail





E. Determine budget and consider mechanisms for sustainability

- Budget for registry design, implementation, and maintenance
- Explore funding mechanisms for sustainability
 - Grants (private, government)
 - Donors, fundraising
 - Crowd-sourcing
 - Partnering with BioPharma and/or academia
 - Fees for access to registry data
- A long-term funding stream is critical to maintaining (and growing) a registry and maximizing the investments made
 - But not necessary to get started





F. Determine registry platform

- Numerous platforms available, including (but not limited to):
 - Patient Crossroads CONNECT
 - Reg4All (Genetic Alliance)
 - PatientsLikeMe
 - CoRDS Registry (Sanford)
 - The Informatics Marketplace (TIM^e) (Remedy Informatics)
 - ORDR/GRDR and NORD patient registry and natural history resources (forthcoming)
 - Custom-designed platforms
- Different strengths, limitations, and costs associated with each
- Webinar #2 will include speakers from several of these platforms to address these issues and considerations in greater detail





G. Design data collection tools

- Obtain guidance from Advisory Committee
- Recommended to be done by professionals trained in proper questionnaire design and survey research methods (e.g., Epidemiologists, Psychometricians)
- Balance ease of registration and completeness of data collection with goals of registry
 - What data are "must know now" versus "like to know later"?
- Leverage existing tools and resources available for standardizing data collection





Dr. Stephen Groft



- Director, Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS) at the National Institutes of Health (NIH)
- Recent recipient of National Organization for Rare Disorders (NORD) award for Vision and Pioneering Guidance on behalf of rare disease patients









Global Rare Diseases Patient Registry Data Repository Pilot Project Overview

Stephen C. Groft, Pharm.D. Yaffa Rubinstein, Ph.D. Office of Rare Diseases Research (ORDR) National Center for Advancing Translational Sciences (NCATS) National Institutes of Health (NIH) Department of Health and Human Services Global Genes RARE /Project July 31, 2013





Global Rare Diseases Patient Registry Data Repository (GRDR) Pilot Project Overview

- New Patient Registries + Existing Registries (Registries retain ownership & control of data)
- Collect And Aggregate Patients Data From Multiple Registries In A Standardized Manner
- Share De-identified Patient Data
- Ability To Conduct Across-Disease Analyses And Recruitment
- Develop And Use Additional Rare Disease Common Data Elements (CDE)
- Utilize A Global Unique Identifier (GUID) to Link Patient Data with Bio-Specimen Samples
- Explore Integration Of Electronic Health Records Into GRDR
- Evaluate Data Mapping, Data Export/Import Processes, And Data Mining Capabilities
- Develop An Accessible Web/Open Source Software-based Patient Registry Template
- Collaborations With NIH Institutes and Centers and Patient Advocacy Groups To Develop Disease -Specific standard questions and CDEs
- Developing Data Contribution And Data Access Agreements



The Need for Patient Registries and Natural History Studies



Benefits Of Patient Registries to Stakeholders

Stakeholder	Benefits
Patients & Foundations	 Ability to organize patient populations for clinical trials & studies Patients can learn from others through survey results Raise visibility to patients and researchers Complete questionnaires in local language
Industry	 Ability to share de-identified pan-disease patient information Link proprietary information to shared patient record Ability to share information with patients based on specific profile Multi-lingual capabilities collect international patient data
Researchers and Academia	 Learn directly from patients and families Ability to recruit for clinical studies & trials pan-disorder Gain access to broader pool of clinical candidates Interactive maps enable clinical trial site planning
Government	 Access to patient reported outcomes Self-sustaining business model frees funding for other research





Submitting Your Registry Data to GRDR





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Home

Home

http://cde.nih.gov

NIH encourages the use of common data elements (CDEs) in clinical research, patient registries, and other human subject research in order to improve data quality and opportunities for comparison and combination of data from multiple studies and with electronic health records. This portal provides access to NIH-supported CDE initiatives and other tools and resources that can assist investigators developing protocols for data collection. <u>What is a CDE?</u>

NIH CDE Initiatives

Collections of CDEs that have been identified for use in particular NIH-supported research projects or registries after a formal evaluation and selection processes.

Summary Table Areas

NIH CDE Tools and Resources

Databases and repositories of data elements and case report forms that may assist investigators in identifying and selecting data elements for use in their projects.



The CDE Resource Portal also includes Other CDE Resources and Relevant Standards. Descriptions of all four groups can be found in the Glossary.

The CDE Working Group of the <u>Trans-NIH BioMedical Informatics Coordinating Committee</u> (BMIC) developed this Portal to improve the coordination of CDEs. BMIC encourages researchers to use CDEs from the Resources in this Portal where applicable, and to consider existing CDE initiatives before starting additional initiatives.

Are we missing a CDE Resource? Contact us.



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Scientific and Clinical Value of GRDR

- Integrating Patient-reported & Clinical Data From Multiple Sources Into Single Repository
- Stimulating New Research On The Causes, Treatments, & Consequences Of Disorders
- Accelerating Knowledge Discovery & Health Of Patients With Rare Diseases
- Enhancing Creative Data Mining Within & Across Disorders
- Leading New Scientific Insights Into Rare Diseases
- Developed CDEs To Be Used By Any Patient Registry & For GRDR
- Developing Library Of Disease-Specific Questions For Patient Reporting
- Developed Informed Consent Template For Participation In Patient Registries
- Developing Open Source Software Patient Registry Template For The Rare Disease
 Community





- Implementing NIH and Office Of Rare Diseases Research Common Data Elements (CDEs)
- Integrating Specific Questions For Individual Diseases Into Patient Registries
- Contributing De-identified Patient Clinical Data To The GRDR From Registries Developed And Curated By Patients' Organizations, Academic Researchers and the Biopharmaceutical Industry
- Registry Will Enable People With Rare Diseases, Their Clinicians, And Researchers To Actively Collaborate In The Research Process.
- For Registry Developers, There Is No Established Forum For Sharing Experiences. Each Time A New Registry Is Developed, It Is Started Using A Different Platform.
- Training Webinars.
- ICD 11 Nomenclature Beta Phase Website For Rare Diseases

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<u>http://www.who.int/classifications/icd/revision/en/index.html</u>





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Patient Registries – Future Needs

- Development and Acceptance of Common Data Elements by Developers, National Library of Medicine, Institutes and Centers, Researchers, Industry, Patient Advocacy Groups
- Data Security and Patient Privacy Guarantees
- Integration of Patient Clinical Data with Biospecimen Samples Information and Use of A Global Unique Identifier (GUID)
- Controlled Sources and Tools of Data Collection, Storage, and Access Across
 Common Platforms
- Data Aggregation and Analyses of De-Identified Data
- AHRQ Registry of Patient Registries (RoPR)
- Patient Centered Outcomes Research Institute (PCORI)
- GRDR 2







- An Integrated Platform Connecting, Registries, Biobanks, and Clinical Bioinformatics for Rare Diseases Research
- PI Hanns Lochmuller, University of Newcastle upon Tyne (UK)
- Common Data Elements
 - Collect All Existing Definitions And Analyses
 - Identification Of Gaps; Non- Harmonized Fields;
 - Specific Necessities Of Projects And Biobanks
 - Final Report List CDEs
- Implement Standardized (Coding, Classifications, Ontologies)
 - Analyses Of The Different Possibilities
 - RD Specificities
 - Resources Accessible Through The Ontology Systems
- Operating Procedures (Collecting; Storing; Retrieving Data)
 - Procedures Manual
 - Quality Standards Criteria
 - Rare Diseases Research Repository
- <u>http://www.irdirc.org/</u>



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Global Rare Diseases Patient Registry Data Repository (GRDR)





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Dr. Marshall Summar



- Children's National Medical Center (CNMC):
 - Chief, Division of Genetics and Metabolism
 - Margaret O'Malley Chair of Molecular Genetics
 - Director, Clinical Research Center
- Founding Investigator, Urea Cycle Disorders Consortium





The Utility of Academics in the Rare Disease Space "you are sort of stuck with us" Lessons Learned from the Urea Cycle Disorders Consortium and the Rare Disease Clinical Research Networks

> Marshall Summar, MD Chief: Division of Genetics and Metabolism Children's National Medical Center Washington D.C.





Standardized longitudinal natural history study data sets and infrastructure can be key in the regulatory process for Rare Disease Drug Approval

- Identify key outcome measures
- Quantify their effects for clinical trial power calculations
- Organize the community of patients
- Organize the community of clinicians/scientists
- Organize the community of pharmas and investors
- Provide standards for data collection, format, etc.
- Works for post-marketing too!





Points to Consider

- Rare Diseases are by their very nature.....rare
- Most expertise is in an academic medical setting usually amongst a handful of physicians
- Large sets of natural history information typically don't exist.
- Most data is from expert opinion or small case report series.





Definitions are Important

The **WHO** definition of "patient registry" is "a file of documents containing uniform information about individual persons, collected in a systematic and comprehensive way, in order to serve a pre-determined scientific, clinical or policy purpose." It does not pre-judge the amount of collected data which can be minimal or extensive, but implies continuity, as distinct from a cross-sectional survey.

The **US National Committee on Vital and Health Statistics** defines registries as "an organized system for the collection, storage, retrieval, analysis, and dissemination of information on individual persons who have either a particular disease, a condition (e.g., a risk factor) that predisposes (them) to the occurrence of a health-related event, or prior exposure to substances (or circumstances) known or suspected to cause adverse health effects."

Terms:

- Natural History Study
- Registry (with longitudinal data)
- Contact Registry (often called just registry in US which is confusing)





Some Points to Consider

- Over 7300 NIH listed rare diseases
- Redesigning and recollecting a database and info system for each one is stupid and wasteful.
- Parent/Family Organizations exist for many of these
- Many of the key outcomes are not known or quantified for these diseases.





Are Such Efforts Underway?

- The NIH Rare Disease Research Network
 - 17 rare disease groups disease specific
 - Investigator initiated
- NIH standard Registries project
- The NORD Rare Disease Database Project
- Others and Other Organizations





Key Issues For Pharma

- FDA pre-acceptance of data for use
- Ownership of data (joint custody, solo, etc.)
- Housing
- Legacy
- Cost
- IRB
- Administration
- Buy in by clinical and patient stakeholders





Case Study

- National Urea Cycle Disorders Consortium
- NIH-Philanthropy sponsored RDCRN consortium
- 3 drugs FDA approved during 8 year life-span, so far
 - Ravicti (Hyperion)
 - Carbaglu (Orphan Europe)
 - Ammonul (Ucyclyd)





Urea Cycle Disorders

- Medium rare (about 1/35,000)
- About 1000 patients in US in treatment
- Basic problem, Can't break down ammonia.
- Strong and single family organization (NUCDF which is a NORD member)
- Physicians in field for long time.
- Early players in Orphan Drug




This Is Why We Really Started





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- In the 1990s when Pharma first approached key opinion leaders the basic need was for consensus on clinical treatment with existing therapies. Common problem with rare diseases.
- In 2000 Ucyclyd took a chance and funded a consensus meeting where they just sat and watched. Clinician run.
- Researchers and clinicians were the same group
- Both groups could stand being in the room together for long periods of time.
- We realized after our first "Consensus" meeting that we didn't really know that much about what we were doing
- We wanted to do a better job and have better information.





WHAT WE LEARNED IS THAT WE DIDN'T KNOW ENOUGH ABOUT THE PATIENTS OR IF WHAT WE WERE DOING WORKED





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The Rare Disease Clinical Research Network: Begin With The End In Mind

- Develop better understanding of outcomes of UCD
- Conduct clinical trials of promising new drugs
- Develop resources and center's of excellence with information on UCD for clinicians, researchers, and patients
- Train next generation of investigators in UCD
- Figure out what we are missing.



Like they say in Real Estate: Location, Location, Location





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UCDC sites









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- 5101 Longitudinal Study
- 5102 Buphenyl_ASA
- 5103 Carbon 13 Acetate (Ureagensis)
- 5104 Assessing Neural Mechanisms of Injury
- 5105 NCLG Effect on ureagenesis in NAGS deficiency
- 5106 Pilot
- 5107 Brain Nitrogen Metabolism in Partial OTCD Using Imaging
- 5109 UCD Cytokines Study
- 5110 Nitric Oxide Flux in ASSD
- 5111 Carbaglu Surveillance Protocol





SURVIVAL DATA After Hyperammonemic Episode BY STUDY

Study	Neonatal Survival 1 year	Neonatal 5 year Survival	Late-onset Presentation	Late-onset 5 year Survival	Incidence
Japan (pre 1999)	43%	22%	75%	41%	1/46,000
France (pre 2000)	26%		72%		
U.S. FDA (1982-2003)	68%	55%	95%	88%	1/35,000
NIH-UCDC (180 pts, 6 deaths)	97%	96%	98%	98%	1/35,500





Outcome Data in Early Series: Batshaw and Brusilow





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Spectrum of Developmental Disabilities in Complete Urea Cycle Enzyme Deficiencies in 1984

Mental retardation	78%
Cerebral palsy	46%
Seizure disorder	17%
Multiple disabilities	46%





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Possible Age or Year of Diagnosis Effects-Neonatal group



First Consensus Meeting Formation of Consortium







Pharma

- If you build it they will come
- They came
 - 3 drugs all the way through FDA
- 1 Device under development
- 2 new drug trials underway
- Spin-off to two other rare disease groups and two common conditions
- Post-marketing study
- Funding for consortium





Why Should Pharma Come?

- Patients pre-concentrated
- Key leaders already organized
- Shortens time to approval
- Easier to pick key outcome variables
- Pre-existing data on natural history
- Family/patient organizations already involved
- Participants are already incentivized





Bromides: Lessons We (Re)Learned

- A Good Data Coordinator is more precious than Gold Platinum
- There are never as many patients who will enroll as you think (1200 down to 550). The law of additive optimism.
- Thank adaptive trial statistics, patient as own control etc. models. The classic RTC is very difficult to do here.
- The paperwork is the rate-limiting step for most things.
 - More IRBs
 - More centers
 - Academic medical center contracting agents are......challenging.
- Treatment will standardize by having the consortium this is good for the patients and good for pharma.
- Having a long-standing standardized database makes comparisons with other groups possible. E-IMD



What Pharma Must Deal With

- Exclusive ownership of data isn't feasible
- In small groups, single disruptive individuals have a disproportionate effect
- The clinical trial isn't the sole focus
- People will wheedle you for more money.
 Researchers are like teenagers, they always need money.





Suggestions

- Check your coat and your egos at the door
- Collect only what you can consistently get. Revisit this frequently.
- Spend some time on defining what the criteria are for having a disease. Harder than you think.
- Plan for criteria for opening new sites and closing unproductive ones
- Like teaching toddlers. Try to avoid use of the word, MINE!!!! when thinking of data.





NORD Project

- Joint project with FDA and NIH
- Data housed at NORD (Switzerland)
- Common data elements for natural history and specific disease elements
- Entry by Patient/Families and Med Profs. Via web
- Data ownership by disease organization but can partner and also legacy option if organization fails etc.
- Common IRB nationally
- Current platform is RedCap
- Pilot rollout this year





How to Expand This

- NORD project is based on lessons from RDCRN
- Biggest difference will be patient/family entry rather than study site entry
 - Advantage: Funding, larger group, community participation
 - Questions: Data quality
- NORD will provide this as a service to its member organizations
- Partnering with FDA and using NIH tools avoids reinvention.
- Designed to do partnering with Pharma, researchers, regulatory bodies







Urea Cycle Disorders Consortium

What is the UCDC? Learn More	Take Action Take an active ro	le in le research
Take Action Information for Physicians News & Events	Receive the most on:	current information Your Participation Matters!
Participating Clinical Centers UCDC Contact Information	+ open recruitment for disease + opening of new clini rare diseases + activities from affilia	r clinical studies of your Find a Study: Help Move Research Forward by Participating in a Clinical Study ted awareness and advocacy Advocacy Group Information
RDCRN RESEARCH MEMBERS	groups + information future o research!	pportunities to participate in
UCDC en Español!	Find more Information about diseases and urea cycle resources Learn More	
	N-Acetylglutamate Synthase (NAGS) Deficiency Carbamyl Phosphate Synthetase (CPS) Deficiency Ornithine Transcarbamylase (OTC) Deficiency Argininosuccinate Synthetase Deficiency (Citrullinemia I) Citrin Deficiency (Citrullinemia II) Argininosuccinate Lyase Deficiency (Argininosuccinic Aciduria) Arginase Deficiency (Hyperargininemia)	Resources For Physicians: Learn More About Urea Cycle Disorders
	Ornithine Translocase Deficiency (HHH) Syndrome Glossary of Terms	Other Useful Resources Treatment Guidelines
	Frequently Asked Questions	Download Contact Registry Paper Form New > UCDC Publications

Welcome to the Urea Cycle Disorders Consortium!

Why am I here? The UCDC exists to bring together physicians and patients for the sake of Urea Cycle Disorders research.

What can I do?

UCDC Events | UCDC Publications

2011 National Urea Cycle Disorders Foundation Annual Conference





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Search

Ongoing Benefits to Patients

- Registry should offer tangible and immediate benefits to the patient community
 - Networking with other patients and families
 - Feedback on experiences of others with the disease
 - Sharing information about healthcare options and experience
 - Benchmarking individual status with grouped data
 - Data visualization for clinical characteristics and patient outcomes
 - Education





Megan O'Boyle



 Board of Directors, Phelan-McDermid Syndrome Foundation (PMSF)



 Registry Coordinator, Phelan-McDermid Syndrome International Registry (PMSIR)









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To submit questions, click the Q&A box and send to Shira Kramer.

What does Phelan-McDermid Syndrome (22q13 deletion/mutation) look like?















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Deletion 22q13





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What does Phelan-McDermid Syndrome (22q13 Deletion) look like?

- Global delays
- Absent or delayed speech
- Sleep Issues
- Gastro Intestinal issues
- Seizures
- Sensory Issues
- Behaviors

....and the list goes on....









Who Are We?

- We are a 12 year-old non-profit foundation with a 3 person office staff
- We do not have a Scientific Director
- We DO have a Scientific Advisory Committee
- We have members in over 30 countries
- We are the sole international foundation (no competing or "split-off" groups)











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For the purposes of this presentation, for the next 15 minutes please remember that ...

"Family" = "Patient"









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What do we offer as a patient organization?

- We have our families ...
 - Contact information
 - Trust
- We have...
 - Means of communications with families
 - Consent from families to be re-contacted
 - Ability to inform and recruit patients



Phelan-McDermid Syndrome Foundation Newsletter - No. 52 / July 2013



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To submit questions, click the Q&A box and send to Shira Kramer.



Why a <u>patient-group</u> sponsored registry?

To submit questions, click the Q&A box and send to Shira Kramer.





- Decrease patient survey fatigue
- Data can be shared with more researchers faster and for less \$
- Increase in researcher access to data
- Ability to return results to families immediately (no waiting for data to be published in a scientific publication that charges for articles!)
- Researcher can post Q&A on the PMSIR registry faster and for less





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What is the PMSIR?

• PMS International Registry...



- Collects contact info (for foundation use only)
- Collects Genetic Reports (curated/de-identified by a trained genetic counselor)
- Asks 100 clinical (medical) questions
- Asks 100 developmental questions
- Asks 100 additional questions submitted by a researcher (data will be "protected" for one year it will then become part of the data available to the entire research community)



What else?

• PMS International Registry...



- Has an Informed Consent
 - Participants agreed to the terms of the IC including having de-identified data shared in public data bases
- Is approved by an Internal Review Board (IRB)
 - Obtained IRB approval from a commercial IRB









What platform is used?







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How do we recruit patients?

 We marketed to PMS Foundation families via Constant Contact, Facebook and foundation websites



• We explained the purpose of the registry and hyped it for weeks prior to the launch









How do we recruit patients?





 We have sponsored contests to encourage registration and survey completion (iPad giveaway)



We post statistics from the registry in the foundation's monthly newsletter





How do you follow-up with patients once they register?

- Targeted e-mails generated by the registry (Asking that surveys be completed or updated annually. Reminding families to send in genetic reports.)
- How often do you follow-up with registered patients to collect new/more data, etc.
- Varies on the amount of other communications that families have received from the foundation. Need to pace the contacts or the families will ignore the requests.










Why did we put our limited time & resources into building a patient registry?

- Better characterize syndrome (genotype-phenotype studies)
- Develop cohorts
- Validate animal models
- Identify meaningful clinical endpoints
- Understand the natural history of the syndrome (this requires annual updates from families)
- Collecting patient contact information (for communications and recruitment)
- Educate families and empower families







Contributors?

- PMS Foundation
- PMS Families





- Researchers that discovered the syndrome (shared the questions they have been asking families for 8 years)
- Autism Speaks (shared AGRE Q&A)
- Researchers in areas of specific organ/symptoms (Suggested Q&A and reviewed draft Q&A)



Other disease groups that had already established registries

Parent Project Muscular Dystrophy

LEADING THE FIGHT TO END DUCHENNE





Who did the planning and implementation of the registry?

- Parent Volunteers
- Patient Crossroads









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How did we accomplish what others said we couldn't?

We...

- ✓ Found a vendor that met our needs
- ✓ Compiled potential Q&A



- \checkmark Consulted with researchers in the field about the Q&A
- Created necessary documents: Informed Consent, IRB protocol, marketing materials, etc. (with the help of outside advisers)
- $\checkmark\,$ Beta tested the registry with a variety of families
- ✓ Launched the registry (May 2011)
- ✓ Marketed the registry with regular communications
- ✓ Returned data to patients/families whenever possible
- \checkmark Will re-access the registry after 1-2 years. Change as needed.





Beta Testing was done twice with a variety of stakeholders:

- Drs. Katy Phelan & Curtis Rogers (the genetic team with the most years of experience with the syndrome)
- Families recently diagnosed (raw and emotional)
- Families diagnosed several years ago (more experience and less emotional)
- Older families with little or no computer experience
- Younger more tech savvy families







What did we accomplish?

- In less than 2 years the PMSIR has...
 - Has registered 620+ patients of known ~900 foundation (diagnosed) members worldwide
 - Translated the registry Q&A to Spanish, French and Italian
 - Posted 100 questions from a researcher
 - The attention of the research community
 - Registrants from 39 countries









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What we've learned...

Patients/families have limited time:

- Preference for short, specific questionnaires over longer questionnaires
- Preference to answer questions in multiple sessions
- Too many questions can LOOK and be overwhelming, possibly scaring registrants off
- Older parents can't remember answers and that makes them feel like "bad" parents
- If the first experience took too long then they are less likely to update annually – diminishing the longitudinal data









What we've learned...

- Genetic data is VITAL
- Older genetic reports (FISH) are not <u>as</u> useful as newer ones (micro arrays) but still helpful
- Micro arrays alone are not always enough. ALL reports the family has (Karyotype, FISH and microarray are best if possible)





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What we've learned...

Genetic reports are harder to get than we thought

- Families of older DX don't have the newer tests
- Families of older DX can't find genetic reports
- Some families can't deal with the technology of uploading, faxing, scanning or e-mailing
- Families with sick kids are overwhelmed and don't have time to dig through medical records
- Some countries (socialized medicine) don't always send written reports





Contests motivate patients/families to register and/or update their registry profile

In order to obtain a free entry into the contest, the following criteria must be met*:

- •All clinical questions answered
- •All developmental questions answered
- •All special project questionnaires answered
- •All genetic lab reports uploaded/faxed/emailed

<image>

*Please note: "no response" or blank answers will not be accepted. If you do not know the answer, please complete by selecting "no", "unsure" or "not applicable".





What we learned...

Social Networking and Contests



Registrants by Month



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Cost to maintain the PMSI Registry to date...

 Year One: set up, fees, IRB -\$51,500

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- Year Two: fees & IRB -
- Year Three: fees & IRB -
- not included

\$37,175

\$37,250

Registry Coordinator:

Translations –

Volunteer - \$0.00

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To submit questions, click the Q&A box and send to Shira Kramer.



Funding?

- What are the source(s) of seed funding and ongoing funding?
 - The registry is currently funded 100% by the PMS Foundation!
- Is there a plan for sustaining funding donations/philanthropy)?



- We MUST find funding to sustain this registry!
- Efforts are being made to solicit stakeholders that will benefit from the data







Who gets what?

- Patient Support Group gets...
 - Info about the condition
- Researchers get ...
 - Data about the patients



- Pharmaceutical companies get ...
 - Data to improve selection of appropriate candidates for clinical trials





Value to families....

Retuning data to patients/families is very helpful in improving the care and safety of the patient





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Value to families...

- Participating in the registry empowers parents/families.
 - Families are using PMSIR to inform their physicians about the syndrome and specific conditions
 We Can Do It!





They print out or send charts from the PMSIR showing the prevalence of conditions to encourage proper testing and treatment





Value to researchers...

- Get data from more patients
- Get data faster
- Get data for less \$\$ (saved funds can be spent on more studies)
- Be able to re-contact patients through support group
- See data that may not have been of interest but becomes valuable









What are the expected yields from the registry in the future?

- Attracting the attention of the research community
- Genotype-phenotype studies
- Development of cohorts (epilepsy, renal issues, GI issues, etc.)
- Increased # of studies and publications
- Better understanding of the syndrome (families, researchers, clinicians)
- Attract the attention of pharma/biotech since we are well organized, have data and the ability to easily recruit
- Use of the registry data to design protocols for clinical studies
- Natural history data







What are the plans and next steps for the registry?

- Overhaul the current surveys to lessen the burden to families so they will return annually
- Add a survey about Adults
- Add a survey about Quality of Life
- Obtain outside funding
- Transition from volunteer registry coordinator to hired registry coordinator
- Find another IRB
- Finalize a Research Data Access Policy
- Release data to the research community
- Add more translations
- Celebrate our success!







Advice for a patient group just starting out with a registry.....





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When is the right time to start a patient registry?

- Now!
- If resources are limited, use a "no-cost to patient group" registry
- If you don't know where to start, ask for help













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Play well with others...

Collaboration is key!

Families, other groups representing the same disease and researchers and clinicians must remember that the patient's best interest is the most important end point.







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You can't improve on something if you never start it!





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Don't Let the Perfect Be the Enemy of the Good.







Never lose sight of the patient/family!





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Questions or Comments?

If you would like to submit questions or topics for Webinar #2 (October 23, 2013), email Carrie Ostrea: <u>carrieo@rareproject.org</u>





