Advisory Panel on Rare Disease Fall 2014 Meeting

Washington, DC
October 7, 2014 – 9:00 a.m. to 4:30 p.m. EST

Patient-Centered Outcomes Research Institute

Welcome and Plans for the Day

Bryan Luce, PhD, MBA, Chief Science Officer, PCORI

Marshall L. Summar, MD, Chair, Advisory Panel on Rare Disease, PCORI

Vincent Del Gaizo, Co-Chair, Advisory Panel on Rare Disease, PCORI

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Housekeeping

- Today's webinar is open to the public and is being recorded.
- Members of the public are invited to listen to this teleconference and view the webinar.
- Anyone may submit a comment through the webinar chat function or by emailing advisorypanels@pcori.org.
- Visit www.pcori.org/events for more information.

Today's Agenda

Start Time	Item	Speaker
9:00 a.m.	Welcome	B. Luce M. Summar V. Del Gaizo
9:05 a.m.	Update on CER Guide	E. Djabali
9:20 a.m.	PCORnet Rare Disease Task Force and RDAP	R. Richesson
10:00 a.m.	Discuss: Rare Disease Research Guide for Merit Reviewers	M. Summar
10:45 a.m.	Break	
11:00 a.m.	Discuss: Rare Disease Research Guide for Merit Reviewers (cont.)	M. Summar
12:00 p.m.	Lunch	
1:00 p.m.	PCORI's Focus on Rare Diseases	B. Luce

Today's Agenda

Start Time	Item	Speaker
2:30 p.m.	Cross-Cutting CER Topics	D. Hickman
4:00 p.m.	Break	
4:15 p.m.	Recap and Next Steps	B. Luce M. Summar V. Del Gaizo
4:30 p.m.	Adjourn	

Meeting Objectives

- Connect the RDAP with the RD PCORnet Task Force
- Get the panel's input on how to improve PCORI's Rare Disease portfolio
- Generate RD topics for potential prioritization

Update on CER Guide

Emma Djabali

Project Assistant, Office of the Chief Science Officer, PCORI

Patient-Centered Outcomes Research Institute

PCORnet Rare Diseases Task Force

Update to the Rare Diseases Advisory Panel

October 7, 2014

Presented by:
Rachel Richesson, PhD
Duke University School of Nursing

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The National Patient-Centered Clinical Research Network

Outline

- Task Force Membership
- Task Force Aims
- Major Activities
  - Inventory of Rare Disease Research issues
  - Rare Disease Phenotypes *(a stretch aim)*
- Future Directions
- Discussion

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Task Force Leadership

- Coordinating Center Leaders (Duke)
  - Rachel Richesson, PhD
  - Priya Kishnani, MD

- Co-Chairs
  - Holly Peay, MS (DuchenneConnect PPRN)
  - David Robertson, MD (Mid-South CDRN)




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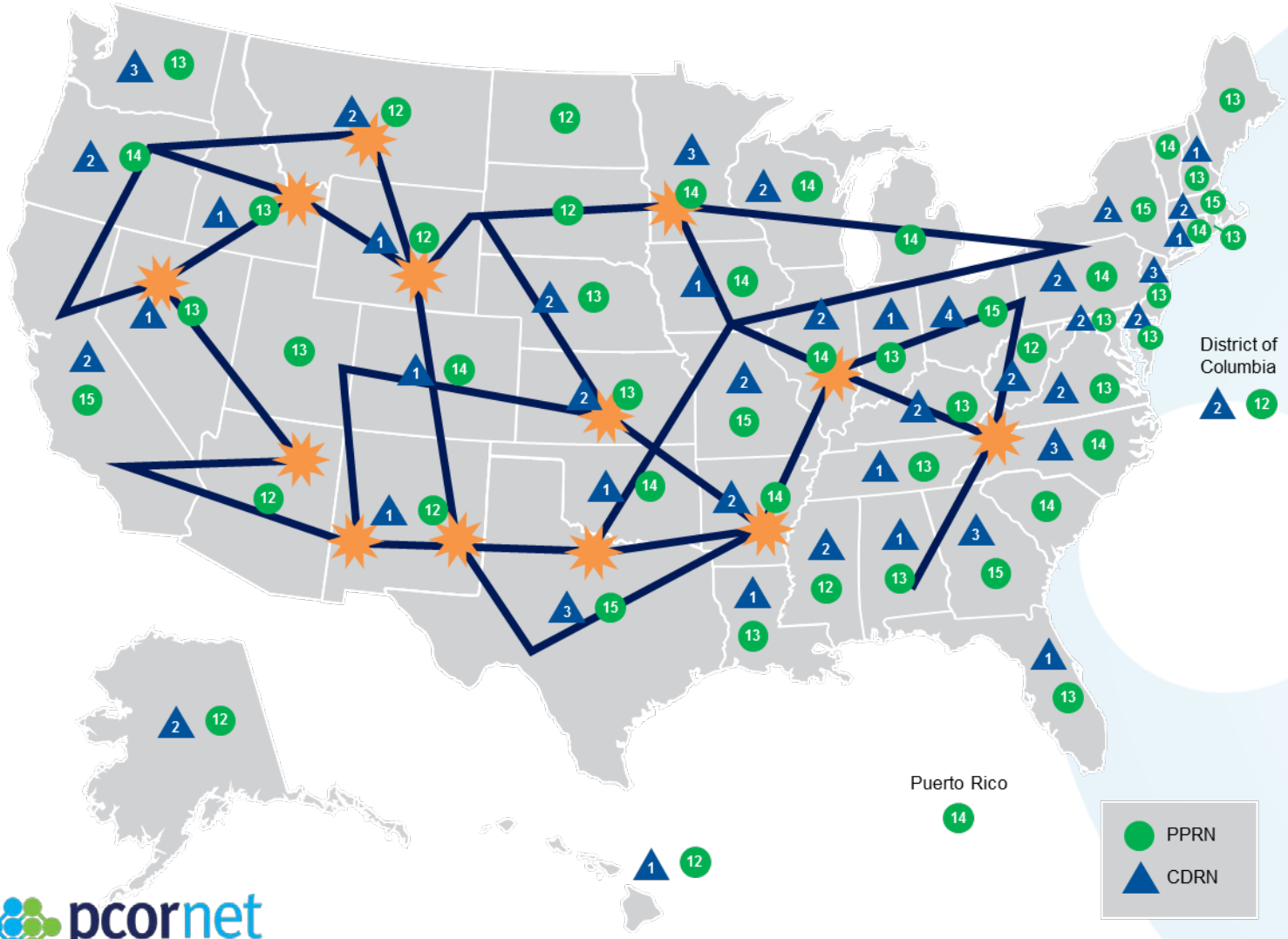
PCORnet: A Network of Networks



This map depicts the number of PCORI-funded Patient-Powered or Clinical Data Research Networks that have coverage in each state.

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CDRN's Rare Disease Cohorts

CDRN	Rare Disease
Mid-South CDRN	Sickle Cell Disease
LACDRN	Sickle Cell Disease; rare cancers
CAPriCORN	Sickle Cell Disease; recurrent C. Difficile colitis
SCIHLS	Pulmonary arterial hypertension
PORTAL	Severe congenital heart disease
PEDSNet	Hypoplastic left heart syndrome
GPC	ALS
NYC-CDRN	Cystic fibrosis
ADVANCE	Alpha 1 Antitrypsin deficiency
PaTH	Idiopathic pulmonary fibrosis
pSCANNER	Kawasaki disease

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PPRN's Cohorts

Representatives from PPRNs in red are on our TF.

PPRN	Disease	Rare?
Health eHeart Alliance	CV health	No
ImproveCareNow	Pediatric Crohn's disease & ulcerative colitis	Yes
CCFA PPRN	Crohn's disease & ulcerative colitis	No
AR-PoWER	Arthritis, musculoskeletal disorders & inflammatory conditions	No
Sleep Apnea PPRN	Sleep apnea	No
The COPD PPRN	COPD	No (subtypes)
MS PPRN	Multiple Sclerosis	No
ABOUT Network	Hereditary breast & ovarian cancer	No
Mood	Major depressive disorder & bipolar disorder	No
PARTNERS Consortium	Juvenile Rheumatic Disease	Yes
ALD Connect	Adrenoleukodystrophy	Yes
PMS_PPRN	Phelan-McDermid syndrome	Yes
PI-CONNECT	Primary immunodeficiency diseases	Yes
The Vasculitis PPRN	Vasculitis	No (subtypes are)
DuchenneConnect	Duchenne & Becker muscular dystrophy	Yes
NephCure	Primary Nephrotic syndrome	Yes
REN	Aicardi, Lennox-Gastaut, Phelan-McDermid, Dravet Syndromes; Hypothalamic Hamartoma; Tuberors Sclerosis	Yes
CENA	Alström, Joubert, Klienfelter Syndromes; Gaucher Disease, PXE, etc.	Yes

Task Force Membership Overview

25 Task Force Members!

Representing **11 CDRNs & 13 PPRNs**

Experience with **> 50 Rare Diseases**

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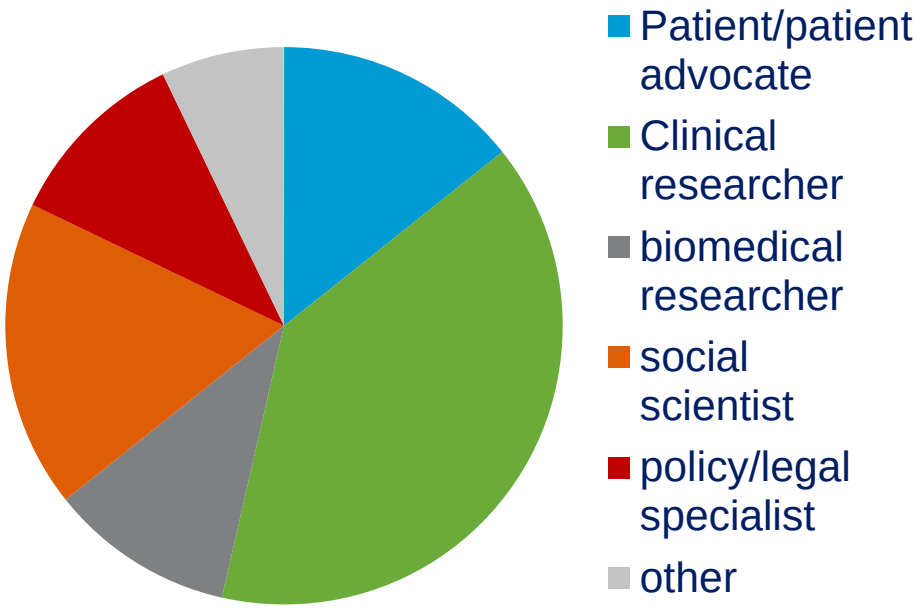
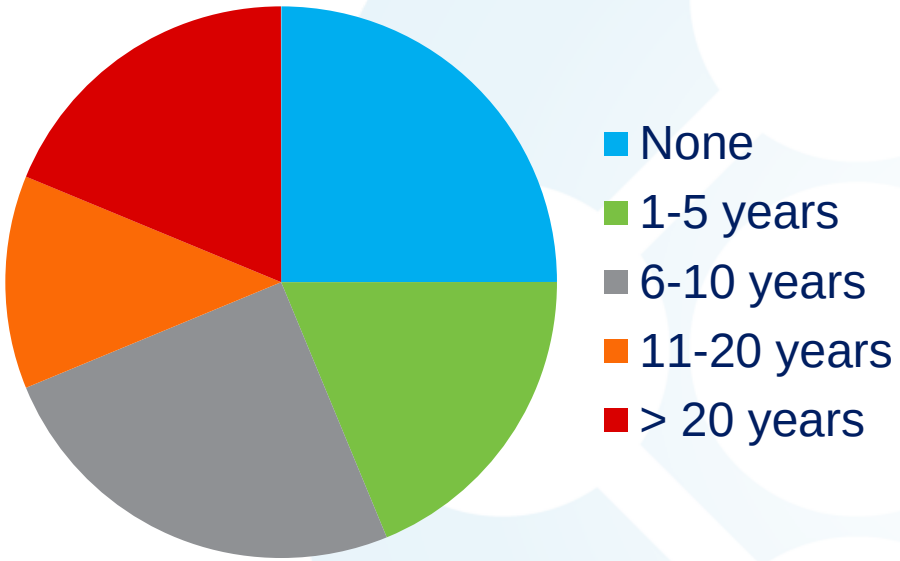
Members

Role within your organization



Years experience in rare diseases advocacy research



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Role of Rare Disease Task Force Members

- Serve as liaisons to their networks
  - Bring rare disease research-related needs, challenges, and successes of each network to the RDTF for inventory and discussion
  - Facilitate networks' success – we need to know how to help!
  - Share RDTF information on activities and updates with their networks.

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Assumptions

- Membership represents a range of experience in rare diseases research and advocacy
- Work products and expertise from other PCORnet Task Forces will be leveraged, and applied, refined, or extended as needed to support the objectives of CDRN and PPRD awardees.
- Initial focus will be on achieving the PCORnet Program milestones, TF goals, and the goals outlined in the Network proposals

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PCORI Milestones related to Rare Disease Research

HIGH LEVEL OBJECTIVE: PATIENTS, HEALTH SYSTEMS, AND CLINICIANS ARE ENGAGED IN GOVERNANCE GOALS AND USE OF THE NETWORK
Approaches to engage with the rare disease community that is relevant to the CDRN's application are developed and submitted to PCORI (RC 6)
Approved approaches to engage with a rare disease community are implemented (RC 6)
HIGH LEVEL OBJECTIVE: THREE PATIENT COHORTS HAVE BEEN SUCCESSFULLY IDENTIFIED, CHARACTERIZED, AND SURVEYED
Approaches to defining membership and characterizing members for rare disease cohort are developed and submitted to PCORI (RC6)
Approved approaches to defining membership and characterizing members for rare disease cohort are implemented (RC6)
Patients characterized as having the rare disease are contacted and recruited to participate in the cohort and in a brief baseline survey (RC6)

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Task Force Aims

- **Support CDRNs & PPRNs in identifying populations, developing research priorities, designing, and implementing studies for rare disorders**
  - Act as source of information for rare diseases research, including stakeholder engagement, regulatory, study design, recruitment, data collection and standards, analytic methods.
  - Act as discussion and advocacy forum to identify and advocate for needs specific to rare diseases research.

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What are challenges that are *unique* to rare disease research?

Framed within the PCORnet Task Force areas:

- Governance & Collaboration
- Health Systems Involvement & Sustainability
- Ethics & Regulatory
- Data Standards, Security & Network Infrastructure
- Data Privacy
- Patient-reported Outcomes
- Patient & Consumer Engagement
- Biorepositories
- Obesity
- Clinical Trials

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Challenges Identified to Date

Issue / Challenge Reported	Primary TF Overlap
Patient friendly ICF	Ethics & Regulatory
Human research compliance	
Electronic-based consent process	
Mobile apps for collecting PRO	Patient Reported Outcomes
Engaging patients who do not see specialists	Patient & Consumer Engagement
Patient-friendly definition of a learning health system	
Define PCORI / PCORnet and relevance to patients	
Dealing with small sample sizes in rare disease data sets	Data Privacy
Utilizing patient and family data in distributed research network	
Coordination/cooperation of CDRNs/PPRNS	Health Systems Involvement & Sustainability
Models for collaboration amongst rare disease organizations that reduce competition and share resources	
Pushing/pulling data from EHR	Data Standards, Security & Network Infrastructure
Coordinate EHR-phenotyping projects and definitions	

How should we address these issues?

- Coordinate/enhance reports and activities of other PCORnet Task Forces
- Identify other resources that address these challenges
- Provide written guidance documents in certain areas
- …
- ***Bring to the PCORI Rare Diseases Advisory Panel!***

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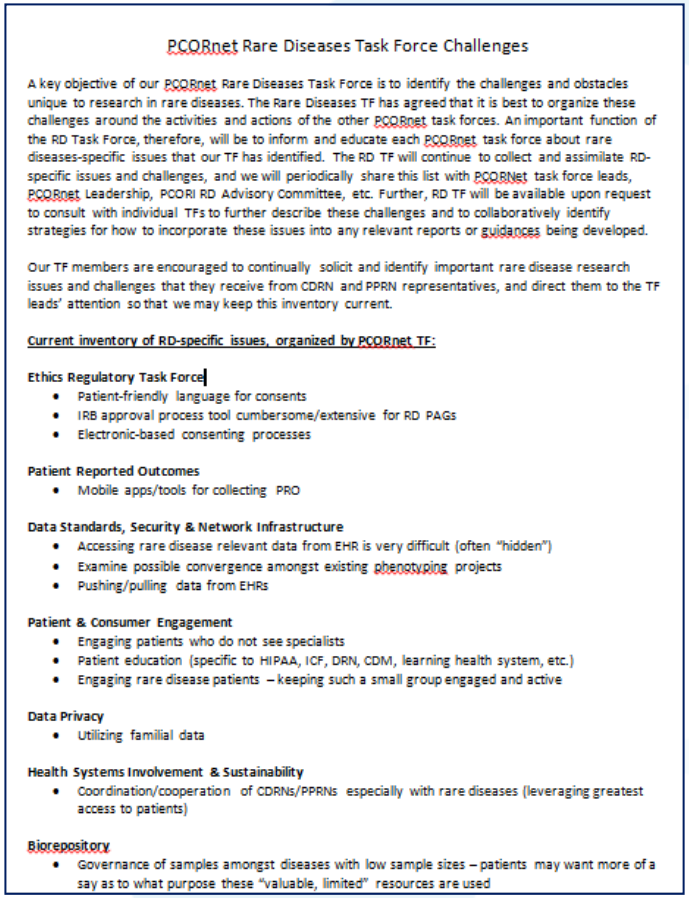
Strategy

- A key objective of our PCORnet Rare Diseases Task Force is to identify the challenges and obstacles unique to research in rare diseases. The Rare Diseases TF has agreed that it is best to organize these challenges around the activities and actions of the other PCORnet task forces. An important function of the RD Task Force, therefore, will be to inform and educate each PCORnet task force about rare diseases-specific issues that our TF has identified.
- The RD TF will continue to collect and assimilate RD-specific issues and challenges, and we will periodically share this list with PCORNet task force leads, PCORnet Leadership, PCORI RD Advisory Committee, etc.
- Further, RD TF will be available upon request to consult with individual TFs to further describe these challenges and to collaboratively identify strategies for how to incorporate these issues into any relevant reports or guidances being developed.
- Our TF members are encouraged to continually solicit and identify important rare disease research issues and challenges that they receive from CDRN and PPRN representatives, and direct them to the TF leads' attention so that we may keep this inventory current.



PCORnet Rare Diseases Task Force Challenges

A key objective of our PCORnet Rare Diseases Task Force is to identify the challenges and obstacles unique to research in rare diseases. The Rare Diseases TF has agreed that it is best to organize these challenges around the activities and actions of the other PCORnet task forces. An important function of the RD Task Force, therefore, will be to inform and educate each PCORnet task force about rare diseases-specific issues that our TF has identified. The RD TF will continue to collect and assimilate RD-specific issues and challenges, and we will periodically share this list with PCORNet task force leads, PCORnet Leadership, PCORI RD Advisory Committee, etc. Further, RD TF will be available upon request to consult with individual TFs to further describe these challenges and to collaboratively identify strategies for how to incorporate these issues into any relevant reports or guidances being developed.

Our TF members are encouraged to continually solicit and identify important rare disease research issues and challenges that they receive from CDRN and PPRN representatives, and direct them to the TF leads' attention so that we may keep this inventory current.

Current inventory of RD-specific issues, organized by PCORnet TF:

Ethics Regulatory Task Force
- Patient-friendly language for consents
- IRB approval process tool cumbersome/extensive for RD PAGs
- Electronic-based consenting processes

Patient Reported Outcomes
- Mobile apps/tools for collecting PRO

Data Standards, Security & Network Infrastructure
- Accessing rare disease relevant data from EHR is very difficult (often "hidden")
- Examine possible convergence amongst existing phenotyping projects
- Pushing/pulling data from EHRs

Patient & Consumer Engagement
- Engaging patients who do not see specialists
- Patient education (specific to HIPAA, ICF, DRN, CDM, learning health system, etc.)
- Engaging rare disease patients – keeping such a small group engaged and active

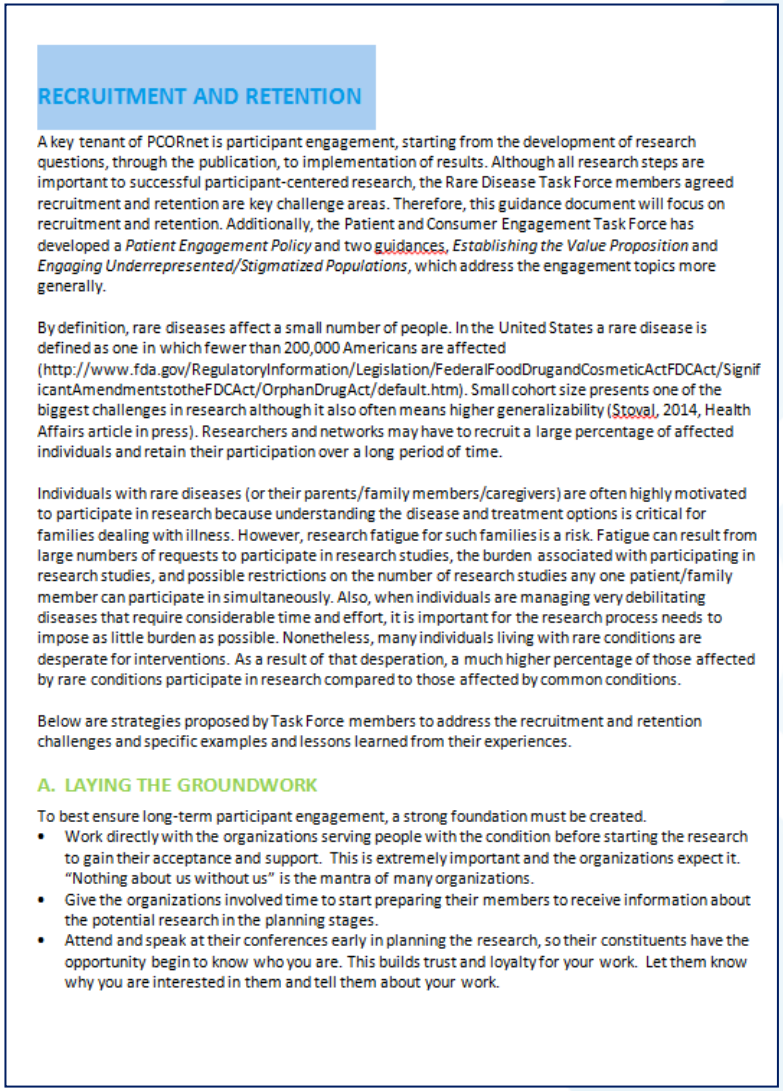
Data Privacy
- Utilizing familial data

Health Systems Involvement & Sustainability
- Coordination/cooperation of CDRNs/PPRNs especially with rare diseases (leveraging greatest access to patients)

Biorepository
- Governance of samples amongst diseases with low sample sizes – patients may want more of a say as to what purpose these "valuable, limited" resources are used

Challenge: Recruitment and Retention

- High Need / Low Effort
- Relevant to all conditions and many task forces
  - Compiled best practice document
  - Task force discussed and enhanced on Oct. 6



RECRUITMENT AND RETENTION

A key tenant of PCORnet is participant engagement, starting from the development of research questions, through the publication, to implementation of results. Although all research steps are important to successful participant-centered research, the Rare Disease Task Force members agreed recruitment and retention are key challenge areas. Therefore, this guidance document will focus on recruitment and retention. Additionally, the Patient and Consumer Engagement Task Force has developed a *Patient Engagement Policy* and two guidances, *Establishing the Value Proposition* and *Engaging Underrepresented/Stigmatized Populations*, which address the engagement topics more generally.

By definition, rare diseases affect a small number of people. In the United States a rare disease is defined as one in which fewer than 200,000 Americans are affected (http://www.fda.gov/RegulatoryInformation/Legislation/FederalFoodDrugandCosmeticActFDCAct/SignificantAmendmentstotheFDCAct/OrphanDrugAct/default.htm). Small cohort size presents one of the biggest challenges in research although it also often means higher generalizability (Stoval, 2014, Health Affairs article in press). Researchers and networks may have to recruit a large percentage of affected individuals and retain their participation over a long period of time.

Individuals with rare diseases (or their parents/family members/caregivers) are often highly motivated to participate in research because understanding the disease and treatment options is critical for families dealing with illness. However, research fatigue for such families is a risk. Fatigue can result from large numbers of requests to participate in research studies, the burden associated with participating in research studies, and possible restrictions on the number of research studies any one patient/family member can participate in simultaneously. Also, when individuals are managing very debilitating diseases that require considerable time and effort, it is important for the research process needs to impose as little burden as possible. Nonetheless, many individuals living with rare conditions are desperate for interventions. As a result of that desperation, a much higher percentage of those affected by rare conditions participate in research compared to those affected by common conditions.

Below are strategies proposed by Task Force members to address the recruitment and retention challenges and specific examples and lessons learned from their experiences.

A. LAYING THE GROUNDWORK

To best ensure long-term participant engagement, a strong foundation must be created.

- Work directly with the organizations serving people with the condition before starting the research to gain their acceptance and support. This is extremely important and the organizations expect it. "Nothing about us without us" is the mantra of many organizations.
- Give the organizations involved time to start preparing their members to receive information about the potential research in the planning stages.
- Attend and speak at their conferences early in planning the research, so their constituents have the opportunity begin to know who you are. This builds trust and loyalty for your work. Let them know why you are interested in them and tell them about your work.

Challenge: Identifying Rare Diseases Cohorts (from Electronic Health Records)

- High Need / High Effort
- Relevant to all conditions and many task forces
  - Compiling background information and instruction
  - Helping networks as we can
    - CDRNs ok
    - PPRN might need help
    - Possible collaborative activity
  - Facilitating cooperation and discussion
    - standardization

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What is a Phenotype?

- Expression of genetic factors, influenced by environment
- Measurable biological (physiological, biochemical, and anatomical features), behavioral, or cognitive markers that are found more often in individuals with a disease than in the general population (MeSH definition)

- **“Computable Phenotypes” / Phenotyping / EHR-based condition definitions / ~ cohort identification**
  - using data from EHRs to identify persons or populations with a condition or clinical profile.
  - data sources: ICD, CPT, labs, meds, vital signs, narrative notes

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Phenotypes

Group - Any -

Include Methods ICD 10 Codes

Exclude Methods

Mine Only - Any -

Apply

Title	Groups	Institutions	Data and Methods	Status
Atrial Fibrillation - Demonstration Project	Vanderbilt - SD/RD Group	Vanderbilt University	CPT Codes, ICD 9 Codes, Natural Language Processing	Final
Cardiac Conduction (QRS)	eMERGE Phenotype WG	Vanderbilt University	CPT Codes, ICD 9 Codes, Laboratories, Medications, Natural Language Processing	Final
Cataracts	eMERGE Phenotype WG	Marshfield Clinic Research Foundation	CPT Codes, ICD 9 Codes, Medications, Natural Language Processing	Final
Clopidogrel Poor Metabolizers	Denny's Group at Vandy, VESPA - Vanderbilt Electronic Systems for Pharmacogenomic Assessment		CPT Codes, ICD 9 Codes, Laboratories, Medications, Natural Language Processing	Final
Crohn's Disease - Demonstration Project	Vanderbilt - SD/RD Group	Vanderbilt University	ICD 9 Codes, Medications, Natural Language Processing	Final
Dementia	eMERGE Phenotype WG	Group Health Cooperative	ICD 9 Codes, Medications	Final
Diabetic Retinopathy	eMERGE Phenotype WG	Marshfield Clinic Research Foundation	CPT Codes, ICD 9 Codes, Medications, Natural Language Processing	Final
Drug Induced Liver Injury	eMERGE Phenotype WG	Columbia University	ICD 9 Codes, Laboratories, Medications, Natural	Final

Most Recent Phenotypes

- Severe Early Childhood Obesity
- Warfarin dose/response
- Drug Induced Liver Injury
- Clopidogrel Poor Metabolizers
- Rheumatoid Arthritis - Demonstration Project

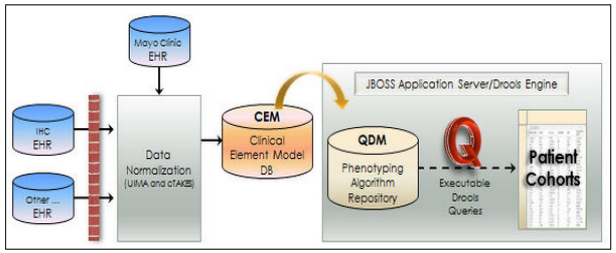
SHARPn.org Phenotype Portal

Log In | Register

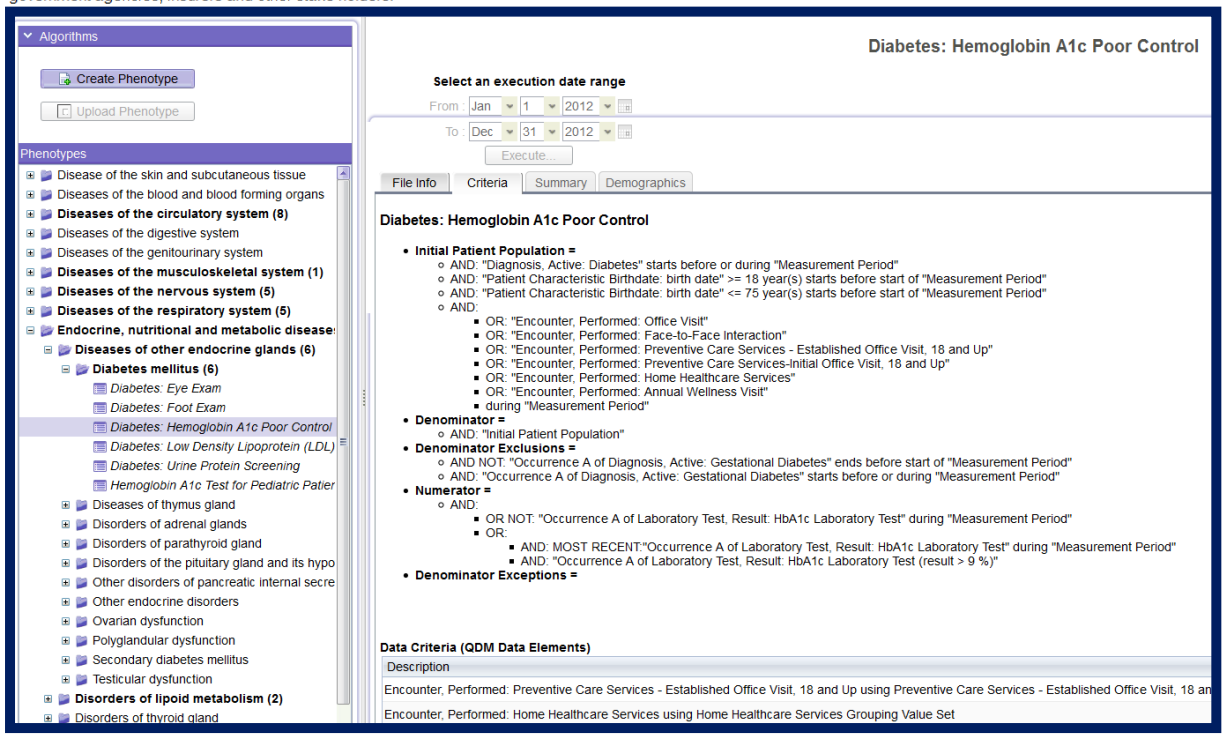
Home Phenotypes Help

What is the Phenotype Portal?

Phenotyping is the process of identifying a cohort of patients based on certain diseases, symptoms or clinical findings. The Phenotype Portal is a tool funded by the SHARPn Project from the Office of the National Coordinator (ONC). It will enable clinicians and investigators to identify patient cohorts using electronic health record (EHR) data by leveraging informatics-based phenotyping processes. In turn, these cohorts will facilitate clinical trial enrollment, outcomes research, and inform clinical decision support. Currently, the field has various barriers in technological research and tool development, and Phenotype Portal is the first such platform for generating and executing Meaningful Use standards-based phenotyping algorithms that can be shared across multiple institutions and investigators.



Traditionally, a patient's medical information is stored inconsistently and in multiple locations, both electronically and non-electronically. The Phenotype Portal will work towards creating a unified framework for normalizing and standardizing clinical data, which will allow for the exchange of patient information among care providers, government agencies, insurers and other stake holders.



News and Updates

Date	News
July 10, 2013	NQF 2014 eMeasures have been uploaded.
September 24, 2013	QDM Phenotyping Translator in now integrated with the portal.
September 22, 2013	All of the Eligible Provider Clinical Quality Measures (CQMs)
December 10, 2012	Phenotype Portal now uses CTS2 value set service.
August 12, 2013	NQF 2014 beta translator now integrated with the portal.
June 17, 2012	Updates with new algorithms.
June 07, 2012	Release version 1.0 of Phenotype Portal.
June 01, 2012	Part of the Office of the National Coordinator for Health
June 01, 2012	We propose research that will generate a framework of

Recently Uploaded

Date	Algorithm
2013-09-20	Appropriate Testing for Children with Pharyngitis
2013-09-20	Initiation and Engagement of Alcohol and Other Drug Dependence
2013-09-20	Controlling High Blood Pressure
2013-09-20	Preventive Care and Screening: Tobacco Use: Screening and
2013-09-20	Chlamydia Screening for Women

Current Database Statistics

gender | age | race | ethnicity

DB Stats Chart for gender

“Standardized” Phenotype Definitions

- Explicit, consistent, and computable definitions can support:
  - development and conduct of new multi-site studies (interventional and observational)
  - comparability of EHR-derived data sets
  - comparison of study results and aggregation of evidence
  - reporting of data sets or results (e.g., ClinicalTrials.gov)
  - better practices for describing research populations in publication submissions to medical journals

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Phenotype for Rare Disease

- Potentially more complex algorithms
- Rare diseases less likely to have diagnosis codes
  - Require other types of data
- Two types of phenotype definitions needed
  - Screening definition
  - Confirmed diagnosis
- Different approach for validation in rare diseases
- Increased importance for identifying individuals with rare disorders across PCORnet

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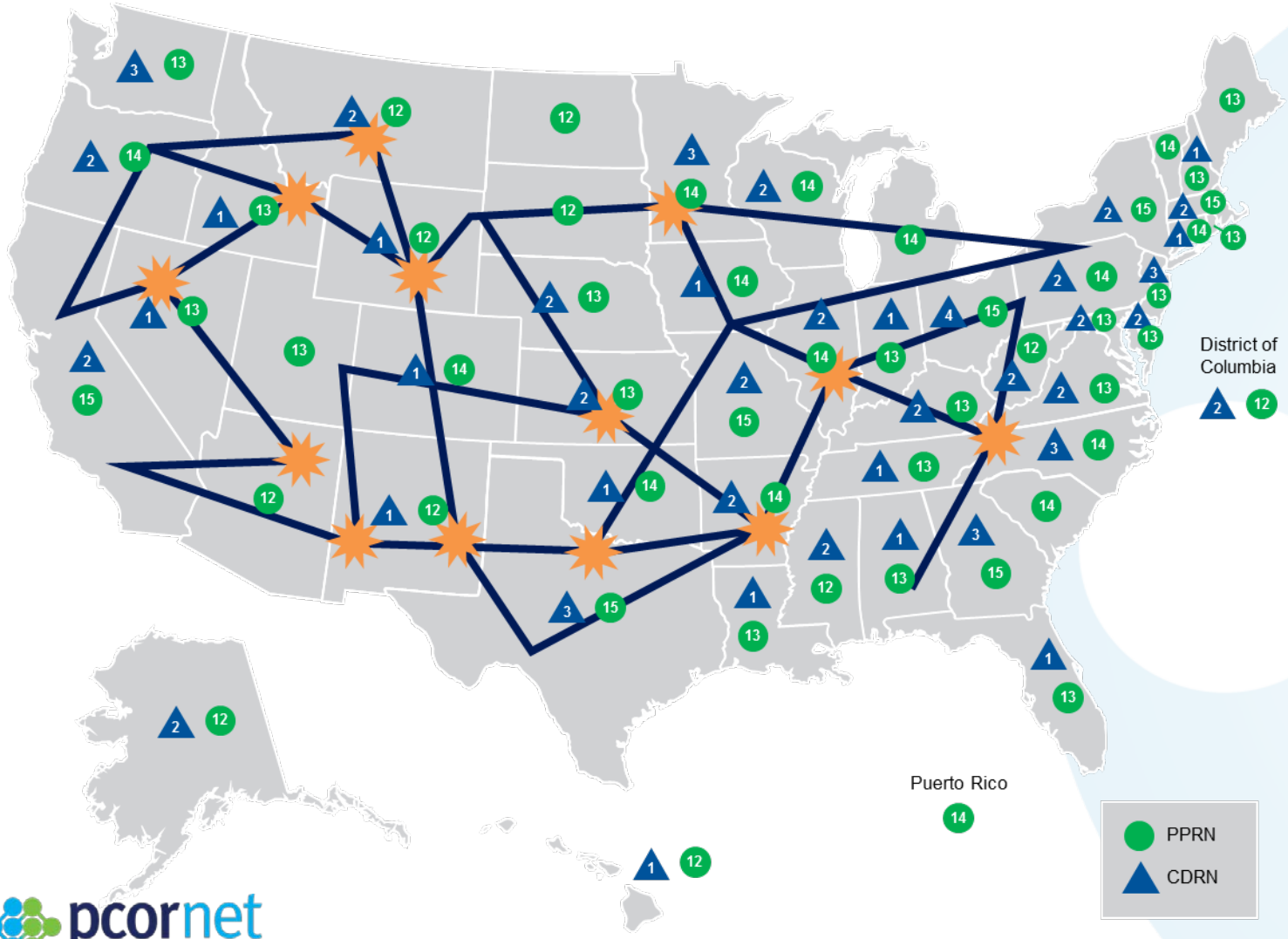
PCORnet: A Network of Networks



This map depicts the number of PCORI-funded Patient-Powered or Clinical Data Research Networks that have coverage in each state.

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Common Data Model (CDM) Specification, Version 1.0

Released by the Data Standards, Security and Network Infrastructure (DSSNI) Task Force on May 30, 2014

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   - 2.2. Overview .....
   - 2.3. Design and Modeling for the PCORne
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   - 2.5. Future State and Revision Process .....
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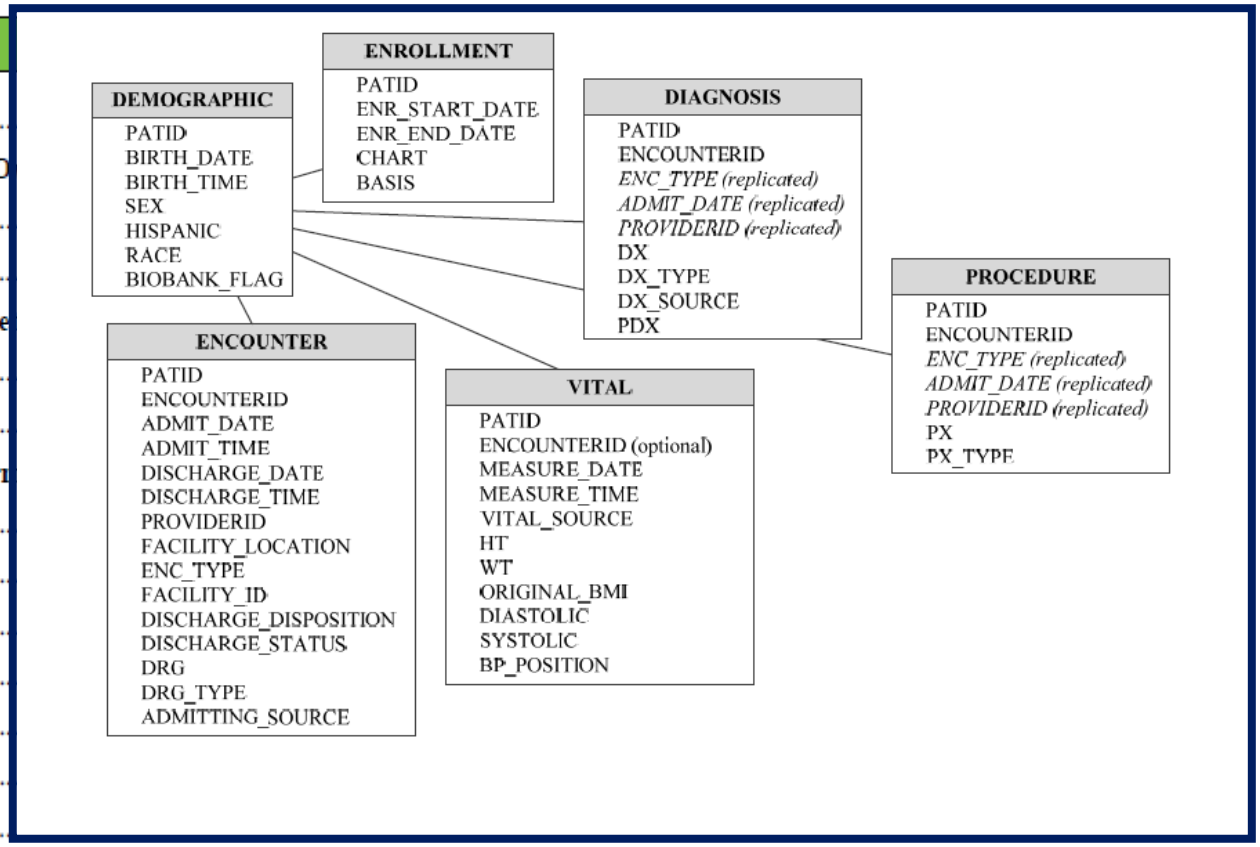

Some Next Steps

- Assess status of phenotyping for rare diseases in PCORnet
- Support “standardization”
  - Consistent documentation format (“template”)
  - Post for all future PCORnet studies to use
- Relate to Common Data Model (CDM)
  - Version 1 has diagnoses and procedures
- RDTF
  - Statement about coding systems (ICD-9-CM, ICD-10-CM, SNOMED CT)

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RDTF Products and PCORI RDAP

- **An inventory of reported needs, discussion priorities, and challenges elicited from PPRN and CDRN network members having affiliations with rare disease advocacy or research.**
- Guidance document for identifying rare disease research cohorts
- Can inform PCORI RDAP activities and discussion priorities
- Can inform future PCORI RFP and requirements for funded projects
- RDAP can review or help develop various guidance documents.

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Thank you.

- Vincent Del Gaizo, Co-Chair, PCORI Advisory Panel on Rare Disease

- Sarita Wahba, Program Officer, PCORI

- Questions or Comments:
  - Rachel Richesson (rachel.richesson@dm.duke.edu)
  - Darcy Louzao (darcy.louzao@duke.edu)

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Break

10:45 – 11:00 a.m. EST

Patient-Centered Outcomes Research Institute

Discuss: Rare Disease Research Guidance for Merit Reviewers

Marshall L. Summar, MD

Chair, Advisory Panel on Rare Disease, PCORI

Patient-Centered Outcomes Research Institute

Draft Guidance for PCORI Merit Reviewers

- Geographically distributed over wider ranges, require more time and/or more sites to enroll patients
- Power calculations are problematic, true numbers often unavailable
- Statistical methodologies are still very young
- Focus on patient pool documentation
- Monitor number of ongoing studies competing for recruitment of the same patients
- Benefit that the PI is affiliated or part of a preexisting registry or longitudinal program for that rare disease
- Risk benefit calculations and explanations are different, many have severe and often rapid outcomes for the patients

Lunch

12:00 – 1:00 p.m. EST

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PCORI's Focus on Rare Diseases

Bryan Luce, PhD, MBA
Chief Science Officer, PCORI

Patient-Centered Outcomes Research Institute

Current Status of Funded Studies in Rare Disease

- PCORI's legislation calls for the RDAP
- PCORI topic prioritization
  - 54 rare disease topics submitted
  - 5 submitted for prioritized by PCORI's priority setting advisory panels
  - Zero received high priority score
- Current portfolio:
  - 17 RD projects;
  - 9 RD PPRNs

Current RD Projects

Decision Support for Parents Receiving Genetic Information about Child's Rare Disease	CDR
Relapsed childhood neuroblastoma as a model for parental end-of-life decision-making	CDR
Enhancing Genomic Laboratory Reports to Enhance Communication and Empower Patients	CDR
Comparative Effectiveness of a Decision Aid for Therapeutic Options in Sickle Cell Disease	CER
Individualized Patient Decision Making for Treatment Choices among Minorities with Lupus	CER
Patient Centered Comprehensive Medication Adherence Management System to Improve Effectiveness of Disease Modifying Therapy with Hydroxyurea in Patients with Sickle Cell Disease	CER

Current RD Projects (cont.)

Collaborative Assessment of Pediatric Transverse Myelitis: Understand, Reveal, Educate (CAPTURE) Study	CER
Development of a Patient-Centered Decision Tool for Nephrotic Syndrome Management	Pilot Projects
Patient-Reported Outcomes for Vasculitits	Pilot Projects
A Community Partnership Approach for Advancing Burden Measurement in Rare Genetic Conditions	Pilot Projects
Comparing patient centered outcomes in the management of pain between emergency departments and dedicated acute care facilities for adults with sickle cell disease	IHS
Comparative Efficacy of Therapies for EosiNophilic Esophagitis	APDTO

Current RD Projects (cont.)

A randomized controlled trial of anterior versus posterior entry site for cerebrospinal fluid shunt insertion	APDTO
Taking Charge of Systemic Sclerosis: Improving Patient Outcomes Through Self-Management	APDTO
The Impact of Self-Management with Probiotics on Urinary Symptoms and the Urine Microbiome in Individuals with Spinal Cord Injury (SCI) and Spina Bifida (SB)	AD
Establishing a Patient-Centered Research Community for Cystic Fibrosis	Pipeline to Proposal
Addressing Obesity in Latino Adolescents with Spina Bifida	Pipeline to Proposal

Rare Disese PPRNs

Empowering Patients and Families for Community-Driven Research: The DuchenneConnect Patient-Report Registry Infrastructure Project
ALD Connect
The Vasculitis Patient Powered Research Network
Collaborative Patient-Centered Rare Epilepsy Network
Phelan-McDermid Syndrome Data Network
NephCure Kidney Network for Patients with Nephrotic Syndrome
The Patients, Advocates and Rheumatology Teams Network for Research and Service (PARTNERS) Consortium
Advancing the medical care of patients with PI by creating a PPRN that welds personal patient information with clinical outcomes.
PATIENT-POWERED RESEARCH NETWORKS (PPRN): A Networked Registry for All

Options to Increase Rare Disease CER

- Additional emphasis in broad funding announcements
  - Possible set-aside funding
- Dedicated funding announcement
- Dedicated Merit Review panel

Discussion

- Funding & merit review options
- Role of RDAP

Cross-Cutting CER Topics

David Hickam, MD

Program Director, Clinical Effectiveness Research, PCORI

Patient-Centered Outcomes Research Institute

Calling on the RDAP

- What is the CER question?
- What are potential study designs?
- What do cross-cutting issues look like as CER questions?

Rare Disease Methodologic Issues

- Methodologic issues and standards in research in rare diseases
- Strength of evidence framework for systematic review
- Standard definition/taxonomy

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Rare Disease or Treatment Symptoms

- Fatigue
- GI symptoms
- Neuropathies
- Depression/anxiety
- Adverse events
- Sexual activity

Rare Diseases and Navigating Care

- Coordinating complex care
- Diagnosis and referral
- Self-management
- Pediatric vs. adult
- Cost of care

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Rare Diseases and Social Environments

- Employment
- Family Relationships
- Social Relationships

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Break

4:00 – 4:15 p.m. EST

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Recap and Next Steps

Marshall L. Summar, MD
Chair, Advisory Panel on Rare Disease, PCORI
Vincent Del Gaizo
Co-Chair, Advisory Panel on Rare Disease, PCORI
Bryan Luce, PhD, MBA
Chief Science Officer, PCORI

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Adjourn

Thank you for your participation!