The Rare Diseases Clinical Research Network (RDCRN): Registries and Longitudinal Studies

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RDCRN

- Established by the Office of Rare Diseases Research
- 2003--10 Consortia supported by ORDR, NCRR, NINDS, NIAMS, NICHD, NHLBI, NIDDK
- 2009--19 Consortia supported by ORDR, NINDS, NIAMS, NICHD, NHLBI, NIDDK, NIAID, NIDCR
- 167 institutions world-wide
- Collectively studying >200 diseases
- 5842 patients enrolled in 39 studies
The Data Management and Coordinating Center

Coalition of Patient Advocacy Groups (CPAG)

• Collaborative Clinical Research
• Public Resources and Education
• Centralized Data Coordination and Technology Development
• Training
Goals of the RDCRN

• Facilitate clinical research by:
  – Creation of disease focused Consortia
  – Cost-saving research infrastructures
  – Establishing uniform protocols for data collection
  – Making meaningful large-scale studies possible
    • Longitudinal cohorts, pilot projects, and randomized trials

• Directly engage patients and their advocates

• Train new investigators in rare diseases research
RDCRN Study Types

- Genotype – phenotype studies
- Longitudinal / natural history studies
- Biomarker identification
- Clinical trials
- Tissue banks/ bio-repositories
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<th>Name of study</th>
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<td>Longitudinal Study of Urea Cycle Disorders</td>
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<td>Rett Syndrome Natural History Clinical Protocol</td>
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<td>Longitudinal Study of Primary Ciliary Dyskinesia: Participants 5-18 Years of Age</td>
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<td>Longitudinal Study of Immune Mediated Disorders after Allogeneic HCT Protocol (Immune Mediated Disorders after Allo-HCT)</td>
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<td>A Natural History Study of CMT1B, CMT2A, CMT4A and CMT4C</td>
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<td>Natural History and Structural Functional Relationships in Fabry Renal Disease</td>
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<td>The Natural History of Mucolipidosis Type IV</td>
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<td>Characterizing the Neurobehavioral Phenotype(s) in MPS III (Pilot Study)</td>
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<td>Genotype-Phenotype Correlations of Late Infantile Neuronal Ceroid Lipofuscinosis</td>
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<td>A Prospective Natural History Study of Diagnosis, Treatment and Outcomes of Children with SCID Disorders</td>
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<td>Smith-Lemli-Opitz Syndrome: A Longitudinal Clinical Study of Patients Receiving Cholesterol Supplementation</td>
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<td>Longitudinal Study of the Porphyrias</td>
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<td>Natural History of and Genetic Modifiers in Spinocerebellar Ataxias</td>
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• Physician Enrolled with informed consent.
• Hypothesis Driven.
• Usual care augmented by directed tests and sample collection.
• Common goals:
  – Describe phenotypic variation.
  – Uniform outcome assessments.
  – Identify population for interventional studies.
Contact Registry’s Global Reach

10-15% of the Contact Registry Have non US, non Canada addresses
RDCRN

Patient Contact Registry

- Enrollment open to patients with diseases under study by Consortia
- Provides international on-line system for communication, recruitment, research
- 128 diseases represented
- 75544 Registrations
Contact Registry Characteristics

• Patient/family Enrolled with online informed consent.
• Limited data collection; includes PHI.
• Connect patients with providers.
• Common goals:
  – Provide information about RDCRN.
  – Provide information about studies.
  – Identify populations for interventional studies.
  – Vehicle for epidemiological studies.
Automated Communications

• Typical automations:
  – Welcome
  – New Study
  – New Clinical Site
  – Periodic (e.g., every 6 months)

• Automations can be customized by study, disease, or consortium
An automated communication system in a contact registry for persons with rare diseases: Scalable tools for identifying and recruiting clinical research participants

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ABSTRACT

Objectives: Strategies for study recruitment are useful in clinical research network settings. We describe a registry of individuals who have self-identified with one of a multiplicity of rare diseases, and who express a willingness to be contacted regarding possible enrollment in clinical research studies. We evaluate this registry and supporting tools in terms of registry enrollment and impact on participation rates in advertised clinical research studies.

Methods: A web-based automated system generates periodic and customized communications to notify registrants of relevant studies in the NIH Rare Diseases Clinical Research Network (RDCRN). Examination of contacts may be scheduled and can be tracked centrally.

Conclusions: The registry and supporting tools are useful to engage and retain patients.
• VCRC Reproductive Health Study: Using the Contact Registry, in less than 10 days of distributing study information to Contact Registry participants, the group has met over 10% of its 2000 participant accrual goal.

• BVMC Study of “Modifier Genes in Cerebral Cavernous Malformations”: Contact Registry helped assist site in recruiting participants from outside of the typical geographical region of families accrued into the study.
Thank you!
Technology

- Application service provider
- .Net / Oracle