

Rare Diseases and Orphan Products Experiences in the USA and A Global Perspective of Collaborative Research and Development Activities

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Rare Diseases Act of 2002

- **Public Law 107-280 (November 6, 2002)**
- Recommend Research and Public Education Agendas at NIH
- Promote Coordination and Collaboration of Rare Diseases Activities
- Develop Information Center (NHGRI)

Office of Rare Diseases

- Collaborative Clinical Research Programs
- Intramural Research Program
- Extramural Research Program
- Rare Diseases Clinical Research Network
- Information Development and Dissemination Activities
- Trans-NIH Working Group on Rare Diseases Research
- Genetic Testing
- Biospecimen Collection, Storage, and Distribution

Coordinated Efforts for Successful Orphan Product Development/Rare Diseases Research

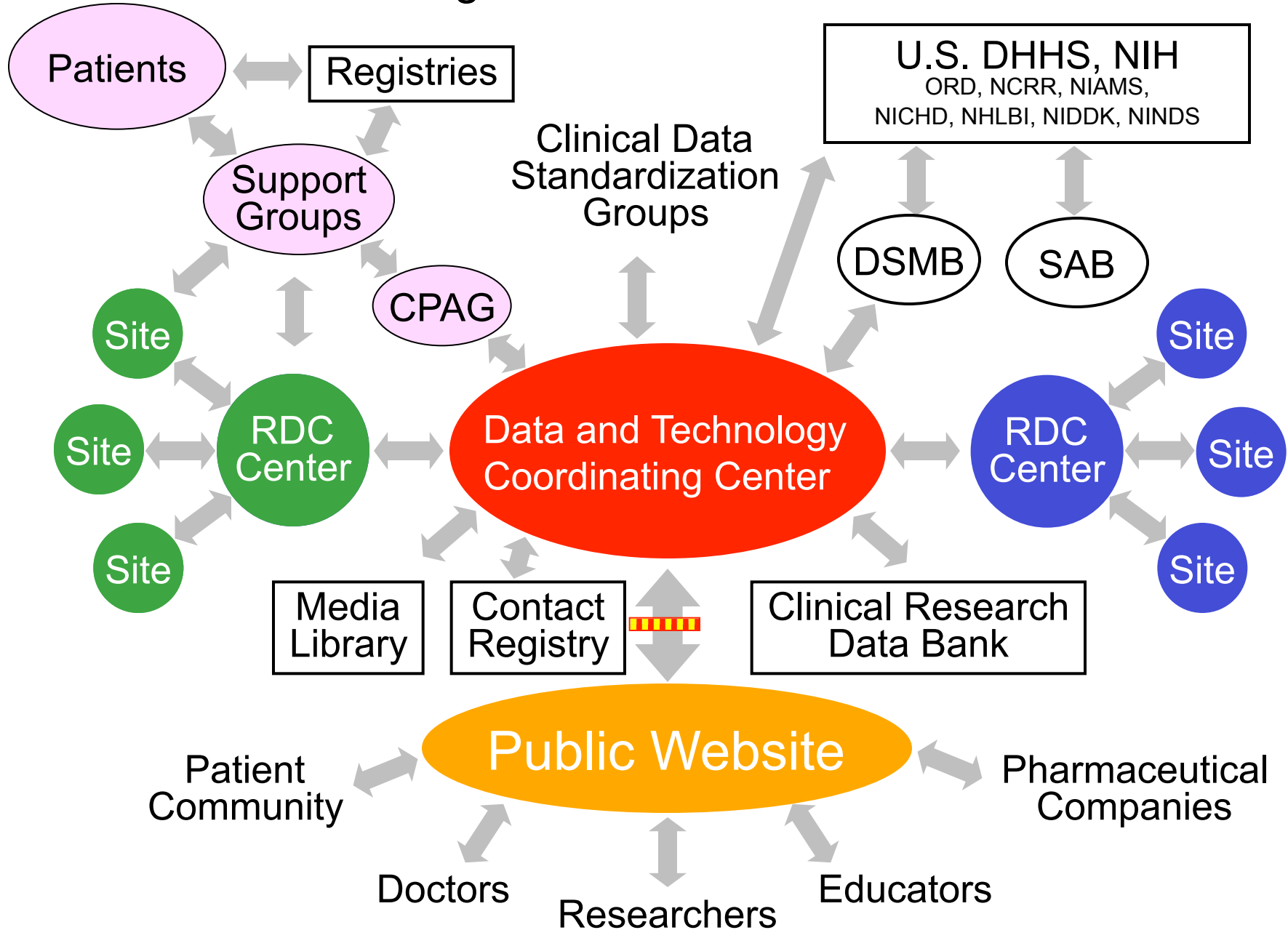
- **Industry (Domestic and International, Large and Small)**
- **Academic and Research Community-
Multidisciplinary Research Efforts**
- **Medical Specialty Societies**
- **Patient Advocacy Groups – The Hub or Glue**
- **Federal Government**
 - **Regulatory**
 - **Reimbursement**
 - **Research**
 - **Intramural Research Program**
 - **Extramural Research Program**

Rare Disease Consortia

<http://www.rarediseasesnetwork.org>

- Angelman, Rett, & Prader-Willi Syndromes C. (ARPWSC)
- Bone Marrow Failure Disease C. (BMFDC)
- Cholestatic Liver Disease C. (CLiC)
- C. Clinical Investigations of Neurological Channelopathies (CINCH)
- Genetic Disorders of Mucociliary Clearance C. (GDMCC)
- Rare Genetic Steroid Diseases C. (RGSDC)
- Rare Lung Diseases C. (RLDC)
- Rare Thrombotic Diseases C. (RTDC)
- Urea Cycle Disorders C. (UCDC)
- Vasculitis Clinical Research C. (VCRC)

Organization of the RDCRN



Selected Tasks of Patient Advocacy Groups

- Establish Medical and Scientific Advisory Boards
- Support Research and Training Programs
- Provide Ready Access to Media
- Serve as Direct Links to Patients and Physicians
- Translate Research Results to Communities
- Organize Research Based Conferences and Meetings for Patients/Families/Caregivers

Promoting Quality Genetic Testing

- Gaining acceptance of global testing services
- CLIA Certification Standards (USA)
- Interpretation of results with appropriate patient counseling
- Partnership and networks to improve research translation and data sharing
 - Between and among research and clinical laboratories
 - Among research investigators, clinical laboratories, patient groups, clinicians, payers

Gaining Access to Clinical Studies

- **National Library of Medicine - ClinicalTrials.gov**
- **Search By Name of Condition or Disease Group, Treatment, Location, or Sponsor**
- **Browse by Condition, Sponsor, or Recruitment Status**
- **12,550 Total Studies (Phase 1, 2, 3, 4)**
- **4,311 Studies Actively Recruiting**
- **236 Not Yet Recruiting**
- **4,059 No Longer Recruiting**
- **3,944 Completed Studies**

ClinicalTrials.gov - Sponsors - Actively Recruiting (Total)

- Industry - 969 Recruiting (2848)
- University/Organization – 1838 Recruiting (4938)
- Other Federal Agencies – 150 Recruiting (402)
- NIH – 2999 Recruiting (8377)
- Co-Sponsorship

ClinicalTrials.gov – Rare Diseases

- Rare Diseases
- Total Studies (Including Those No Longer Recruiting – Significant to Include)
 - 5, 851 Studies
 - 841 Rare Diseases
- Active Recruiting Studies
 - 2,317 Studies
 - 650 Rare Diseases

Challenges and Strategies

- **Provide Global Access to Clinical Studies and Clinical Trials**
- **Develop Globalization of Research Efforts and Common Protocols with Multidisciplinary Research Teams**
- **Continue Efforts for Harmonization of Research Data for Regulatory Purposes**
- **Establish Better Definitions of Patient Responders with Development of Appropriate Biomarkers and Surrogate Endpoints for Safety and Efficacy**

Challenges and Strategies

- Utilize Screening Processes of Industry and Government Chemical Libraries
- Product Development and Manufacturing Capabilities
- Identify Research and Development Needs, Challenges, and Solutions for ...
 - Academic Research Investigators,
 - Pharmaceutical, Biotechnology, and Medical Device Industries
 - Government Agencies
 - Patient Advocacy Groups

Challenges and Strategies

- **Expand Newborn Screening Programs and the Development of Genetic and Diagnostic Tests with Appropriate Counseling**
- **Increase Educational Efforts for the Public and Health Care Providers' Communities**
 - **Diagnostic Criteria Rare Diseases and Conditions**
 - **Disease Specific and Available Treatments**
 - **Standards of Care for Emergency and Critical Care Treatments**
 - **Basis of Genetics and Inherited Disorders**
- **Expand Global Linkages of Patient Advocacy Group Networks**
- **Develop Inclusive Web-Based Inventory of Global Rare Diseases Research/Intervention Activities and Information Resources**

Challenges and Strategies

- **Identify and Expand Worldwide Partnerships and Collaborations**
- **Identify Economic Impact of Rare Diseases**
- **Expand Training Programs on Living and Coping with Rare and Genetic Diseases - Gaining Acceptance for Disabilities**
- **Explore Across-life Implications**
- **Maximize Access to Rehabilitation Therapies – Physical, Hearing, Speech, Vocational, Occupational, etc.**
- **Assure Worldwide Access to Safe and Effective Products for the Prevention, Diagnosis, and Treatment of Rare Diseases**

The Genetic and Rare Diseases Information Center (NHGRI/ORD)

- **>10,000 Inquiries (2002 – 2004)**
- **> 3,500 Rare Diseases or Conditions**
- **Toll-free 1-888-205-3223 (USA)**
- **International Access Number:
301-519-3194**
- **Fax: 240-632-9164**
- **E-mail: GARDinfo@nih.gov**

ORD Website

<http://rarediseases.info.nih.gov/>

- **Rare Diseases Information – Pub Med**
- **Research and Clinical Trials - CRISP, ClinicalTrials.gov**
- **Patient Support Groups - CHID Database > 1200 Patient Advocacy Groups, NORD, Genetic Alliance**
- **Patient Travel & Lodging**
- **Genetics Information – Gene Tests, OMIM, NCHPEG**
- **Research Resources**
- **Scientific Workshops, Archived Reports**
- **Website Trends**
 - ~ 75,000 Users per Month
 - Average Visit – 17.24 Minutes

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