

Perspective of Rare Diseases Activities - A Global Approach

Stephen Groft, Pharm.D.

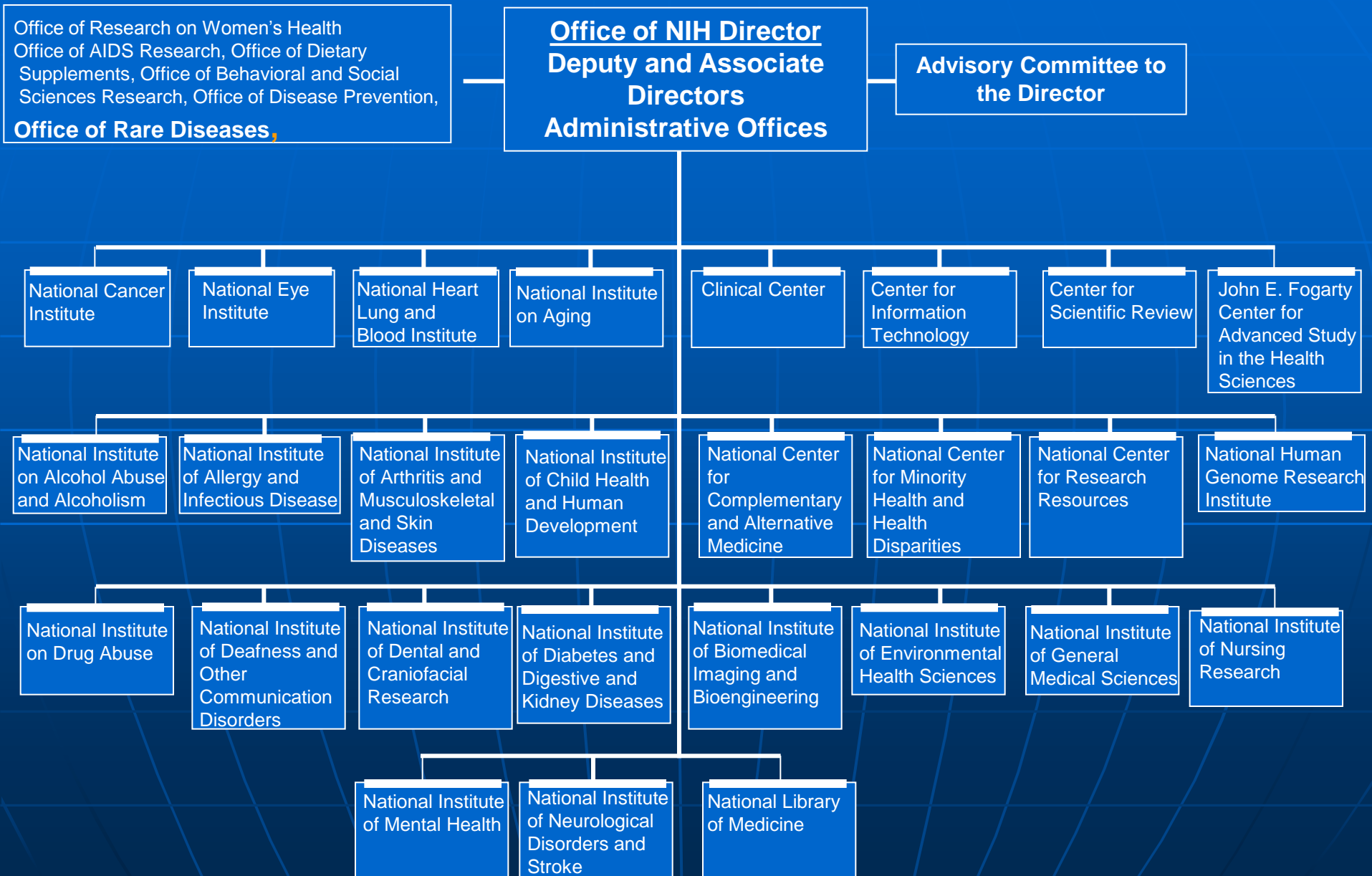
Office of Rare Diseases Research, NIH, DHHS

**DSD Research and Quality Improvement
Symposium**

April 1-3, 2009

Ann Arbor, Michigan

A View of the NIH Institutes and Centers



Rare Diseases Research and Orphan Products Development: A Collaborative Approach

- **Historical Alliances of Public-Private Collaborating Partners**
 - **DHEW Interagency Committee on Drugs of Limited Commercial Value (1974-1975)**
 - **Report on Significant Drugs of Limited Commercial Value (1979)**
 - **Orphan Drug Act (1983)**
 - **National Commission on Orphan Diseases(1986-1989)**
 - **Special Emphasis Panel on Coordination of Rare Disease Research(1997-1999)**
 - **Office of Rare Diseases Act and Rare Diseases Orphan Product Development Act (2002)**
 - **Determine Future Global Needs of Stakeholders**

A Collaborative Approach to Rare Diseases as a Public Health Problem

- **Identify Needs of Patients, Advocacy Groups, Health Care Providers, Researchers, Industry, Payers, Government Agencies.**
- **Determine Family and Societal Needs Across the Lifespan**
- **Decisions to Emphasize Needs and Develop Incentives to Meet Needs of All Patients Regardless of ...**
 - **Prevalence of Diseases**
 - **Severity of Diseases in All Age Groups**
 - **Expected Lifespan of Patients with Rare Diseases**
 - **Availability of Less than Optimal Treatments or Supportive Care**
 - **Cost of Treatment**

Rare Diseases – Perceptions

- Very Few People Have the Condition
- Little or No Information Available
- Little or No Research Interest
- No Treatments are Available
- Limited Access to Rare Diseases-Focused Clinicians
- Little or No Hope for the Future

Patients and Families Coping with a Rare Disease on Intellectual, Emotional, and Psychological Levels

- Gaining Access to Knowledgeable Physicians and other Health Care Providers
- Knowing there are other families living with the disease – Avoiding Stigmatization
- Finding a safe person to raise questions.
- Finding useful information on a gradual basis – the building blocks for decision making
- Understanding the process. Going from life as we knew it to life as it's going to be
- Meeting patients and families where they are and not try to rush them to where we (or they) want to be.

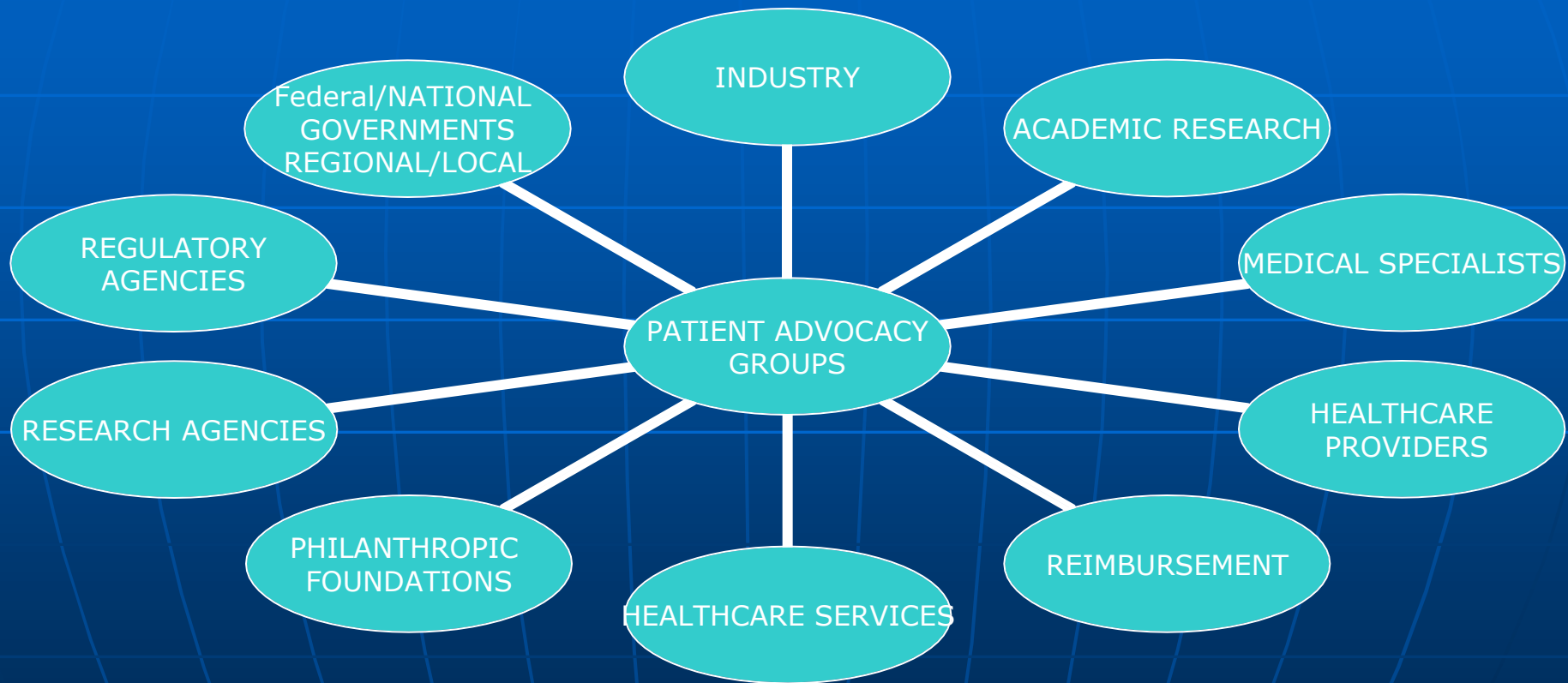
Meeting Patient and Family Needs

- **Expand Training Programs on Living and Coping with Rare and Genetic Diseases**
- **Gain Acceptance for Disabilities and Improving Educational Opportunities for Patients**
- **Maximize Access to Rehabilitation Therapies**
 - **Physical, Hearing, Speech, Vocational, Occupational**
- **Assure Worldwide Access to Safe and Effective Products for the Prevention, Diagnosis, and Treatment of Rare Diseases**

Coordinated Efforts for Successful Orphan Product Development/Rare Diseases Research

- **Pharmaceutical, Biotechnology, and Medical Devices Industries**
- **Academic and Private Foundation Research Communities-Multidisciplinary Research Efforts**
- **Medical Specialty Societies and Healthcare Providers**
- **Patient Advocacy Groups**
- **Federal Government**
 - **Regulatory**
 - **Reimbursement**
 - **Research**
 - **Health Care Services**
 - **Prevention**

Coordinated Efforts for Successful Orphan Product Development/Rare Diseases Research – A Different View



Office of Rare Diseases (ORD) -

Collaborative Clinical Research Programs

- **Intramural Research and Training Programs**
 - **Bench to Bedside Research Program with Extramural Research Programs/Grantees**
 - **Clinical and Biochemical Genetics Training Program**
 - **Unknown Diagnosis (Diseases of Unknown Origin)**
- **Extramural Research Program**
 - **Scientific Conferences >900**
 - **CETT Genetic Test Development Program**
 - **Genetic Test Materials Standards: OECD and CDC**

Office of Rare Diseases (ORD) - Collaborative Clinical Research Programs

- **National Coalition for Health Professional Education in Genetics (NCHPEG)**
- **Genetic and Rare Diseases Information Center**
- **Trans-NIH Working Group on Rare Diseases Research**
- **Angel Flight/Mercy Medical Airlift – Patient Travel**
- **WHO International Classification of Diseases - Rare Diseases Emphasis for ICDX+ and ICD XI**
- **Educational Program on Rare Diseases for Middle School Children with Office of Science Education/NIH**

Translating Rare Diseases Research Advances to Orphan Products

- **Identify and Expand Worldwide Partnerships and Collaborations of Patient Advocacy Groups – NORD, EURORDIS, Genetic Alliance. KORD, NZORD, CORD**
- **Provide Global Access to Clinical Trials at Multi-National Research Sites**
- **Develop Globalization of Research Efforts and Common Protocols with Multidisciplinary Research Teams (Treat NMD, Prader-Willi Syndrome, Progeria)**
- **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**
- **Develop Inclusive Web-Based Inventory of Global Rare Diseases Research/Intervention Activities and Information Resources**

Promoting Quality Genetic Testing

<http://www.cettprogram.org/>

- Gaining acceptance of global testing standards and services (OECD-CDC)
- CLIA Certification Standards (USA)
- Interpretation of results with appropriate patient counseling
- Development of Model Collaboration, Education, and Genetic Test Translation Program (CETT) Program
- Partnership and networks to improve research translation and data sharing
 - Results Reporting Forms for..
 - Patients
 - Referring Physicians

Challenges and Strategic Needs

— The Diagnostic Odyssey

- Obtaining the Diagnosis
 - <1 year - 51% Diagnosed
 - >1 year but <5 Years – 31% Diagnosed
 - >5 years – 15% Diagnosed
 - ~6% of Requests to GARD for Undiagnosed Diseases
- Increased Development of Genetic and Diagnostic Tests with Appropriate Counseling
- Better Diagnostic Criteria for Rare Diseases
- Standards of Care for Treatment

Future - More Efficient Utilization of Research Resources

- Continue to expand International Regulatory Harmonization of Research Data
- Genetic Predictors of Patient Responders
- Study Design and Biostatistics Analyses Accepted for Small Sample Sizes
- Global Patient Recruitment Strategies for Multi-National Clinical Trials
- Develop Better Tools for Web-Based Patient Recruitment and Referral
- Provide Greater Emphasis on Longitudinal Studies of Patients with Rare Diseases to Identify Appropriate Biomarkers and Endpoints of Efficacy and Safety
- Increase Clinical Research Pilot Studies or Demonstration Projects

NIH Emphasis on Patient Advocacy Groups as Collaborators

- Advisory Councils/Special Committees
- Council of Public Representatives
- Institutional Review Boards
- Advice on NIH Research Priorities
 - NHLBI, NINDS, NCI, ORD Conferences
 - Identify Public Concerns
- Participate in Scientific Conferences
- Review of Disease-Specific Information
 - Fact Sheets
 - Policy Statements

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
- Recruit Patients for Clinical Studies
- Identify Cohorts of Patients with Range of Phenotypic Expression

Meeting Societal Needs

- Mobilize Health Professionals and Society to Take Action
- Obtain Cooperative and Collaborative Support From All Nations and Partners in Health Research and Product Development
- Improve Health Literacy of Populations to Enable Information-Based Decision Making
- Reduce Disparities in Global Health with Ready Access to Information and Interventions for Diagnosis, Prevention, and Treatment of Rare Diseases

Selected Outcomes of the Scientific Conferences Program

- Identify Research Opportunities
- Establish Research Priorities
- Develop Program Announcements and Solicit R01 Applications
- Establish Diagnostic and Monitoring Criteria
- Develop Animal Models
- Support Registries - Patient and Tissue
- Develop Research Protocols, Collaborative Research Arrangements, and Plan Clinical Trials
- Disseminate Results to Targeted Professional and Voluntary Health Organizations
- Co-Sponsor with Patient Advocacy Groups, Academic Investigators, Industry, Foundations, Intramural and Extramural Research Programs
- Observation: Greater attendance from Global Collaborators

Office of Rare Diseases National Institutes of Health

6100 Executive Boulevard

Room 3B-01, MSC - 7518

Bethesda, MD 20892-7518

Voice: 301-402-4336

Fax: 301-480-9655

E-mail: ORD@nih.gov

Website:

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

Office of Rare Diseases - Staff

- Ms. Mary Demory
- Ms. Marita Eddy (Angel Flight)
- David Eckstein, Ph.D.
- John Ferguson, M.D.
- Rashmi Gopal-Srivastava, Ph.D.
- Mr. Christopher Griffin
- Ms. Henrietta Hyatt-Knorr, M.A.
- Ms. Susan Lowe
- Ms. Geraldine Pollen, M.A.
- Yaffa Rubinstein, Ph.D.
- Giovanna Spinella , M.D.
- William Gahl, M.D., Ph.D. (Clinical Director, NHGRI)