Perspective of Rare Diseases Activities - A Global Approach

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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)
  - 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


- 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

INDUSTRY

ACADEMIC RESEARCH

MEDICAL SPECIALISTS

HEALTHCARE PROVIDERS

REIMBURSEMENT

HEALTHCARE SERVICES

PHILANTHROPIC FOUNDATIONS

RESEARCH AGENCIES

REGULATORY AGENCIES

Federal/NATIONAL GOVERNMENTS REGIONAL/LOCAL

PATIENT ADVOCACY GROUPS
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 - Staff

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- John Ferguson, M.D.
- Rashmi Gopal-Srivastava, Ph.D.
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