Rare Diseases Collaborative Research and Development Activities

Stephen C. Groft, Pharm.D.
Office of Rare Diseases
National Institutes of Health
Department of Health and Human Services

American Academy of Dermatology
65th Annual Meeting
February 5, 2007
Washington, DC
Stephen C. Groft, Pharm.D.
Forum 556 Development of Molecularly-Targeted Therapies for Rare Diseases

I do not have any relevant relationships with industry.
Office of Rare Diseases (ORD) -

Collaborative Clinical Research Programs

- Intramural Research and Training Programs
  - Bench to Bedside Research Program with Extramural Research Program
  - Clinical and Biochemical Genetics Training Program (NHGRI)

- Extramural Research Program
  - Scientific Conferences
  - Rare Diseases Clinical Research Network (RDCRN)
  - CETT Genetic Testing Program

- Information Development and Dissemination Activities
- Trans-NIH Working Group on Rare Diseases Research
- Inventory of Bio-specimen Collection, Storage, and Distribution Systems (RAND Corporation)
- OTT/ORD Technology Transfer Activities
Purposes of Cooperative Rare Diseases Clinical Research Network (RDCRN)

- Facilitate Clinical Research in Rare Diseases
- Training of Clinical Investigators in Rare Diseases Research
- Test Site for Distributed Clinical Data Management
- Support Collaborative Clinical Research
  - Longitudinal Studies of Patients with Rare Diseases
  - Clinical Pilot or Demonstration Projects
  - Access to Information Related to Rare Diseases for Clinicians, Researchers, and the Lay Public
- Open Re-Competition in 2008/2009
- Web Site: http://rarediseasesnetwork.epi.usf.edu/
- Collaborative Clinical Research
- Centralized Data Coordination and Technology Development
- Public Resources and Education
- Training
Scientific Conferences Program

- Establish Research Priorities and Agenda
- Develop Program Announcements
- Establish Diagnostic and Monitoring Criteria
- Develop Animal Models
- Support Patient and Tissue Registries
- Develop Research Protocols and Collaborative Research Arrangements
- Initiate Clinical Trials
- Disseminate Outcomes of the Workshops to Targeted Professional and Voluntary Health Organizations
National Institute of Arthritis and Musculoskeletal and Skin Diseases-Scientific Conferences

- Ankyloblepharon-Ectodermal Dysplasia - Cleft Lip/Palate AEC Syndrome
- Translating Basic Knowledge of Genetic Skin Diseases Into Therapies
- Pachyonychia Congenita
- Infantile Hemangiomas
- Pemphigus
- Pseudoxanthoma Elasticum (PXE)
- Immunomodulatory Drugs in the Treatment of Skin Diseases
- Neonatal Onset Multi-system Inflammatory Disease (NOMID)
- Cutaneous Gene Therapy: Problems and Prospects (Previously “Skin as a Tool for Gene Therapy”

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
- Federal Government
  - Regulatory
  - Reimbursement
  - Health Care Services
  - Research
    - Intramural Research Program
    - Extramural Research Program
Promoting Quality Molecular and Biochemical Genetic Testing

- Formed - National Laboratory Network for Rare Disease Genetic Testing (NLN)
  http://www.rarediseasetesting.org
- Gaining acceptance of global testing services
- CLIA Certification Standards (USA)
- Interpretation of results with appropriate patient counseling
- Collaboration, Education, and Genetic Test Translation Program (CETT) Prototype
- Collaborative partnership and networks to improve research translation and data sharing
  - Clinical (CLIA-certified) laboratory
  - Researcher (laboratory and/or clinician)
  - Patient advocacy group
Technology Transfer Activities - OTT/ORD

- Office of Technology Transfer (Neglected Diseases, Rare Diseases) Available Technology from Government (>500 technologies) and Not for Profit Organizations - Academic Research Centers and Others


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
- Expand Global Linkages of Patient Advocacy Group Networks
- Develop Inclusive Web-Based Inventory of Global Rare Diseases Research/Intervention Activities and Information Resources
The Genetic and Rare Diseases Information Center (NHGRI/ORD)

- >17,000 Inquiries (2002 – 2007)
- > 4,700 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
Office of Rare Diseases - Staff

- Ms. Mary Demory
- Ms. Marita Eddy (Angel Flight)
- Dr. John Ferguson (Consultant)
- Dr. Stephen C. Groft
- Dr. Rashmi Gopal-Srivastava
- Mr. Christopher Griffin
- Ms. Henrietta Hyatt-Knorr
- Ms. Sharon Macauley
- Ms. Geraldine Pollen (Consultant)
- Dr. Giovanna Spinella (Consultant)
- Dr. William Gahl (Clinical Director, NHGRI)