

# Rare Diseases Collaborative Research and Development Activities

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*American Academy of Dermatology  
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# DISCLOSURE OF RELEVANT RELATIONSHIPS WITH INDUSTRY

65<sup>th</sup> ANNUAL MEETING

WASHINGTON D.C.



FEBRUARY 2-6, 2007

Stephen C. Groft, Pharm.D.  
Forum 556 Development of  
Molecularly-Targeted Therapies for  
Rare Diseases

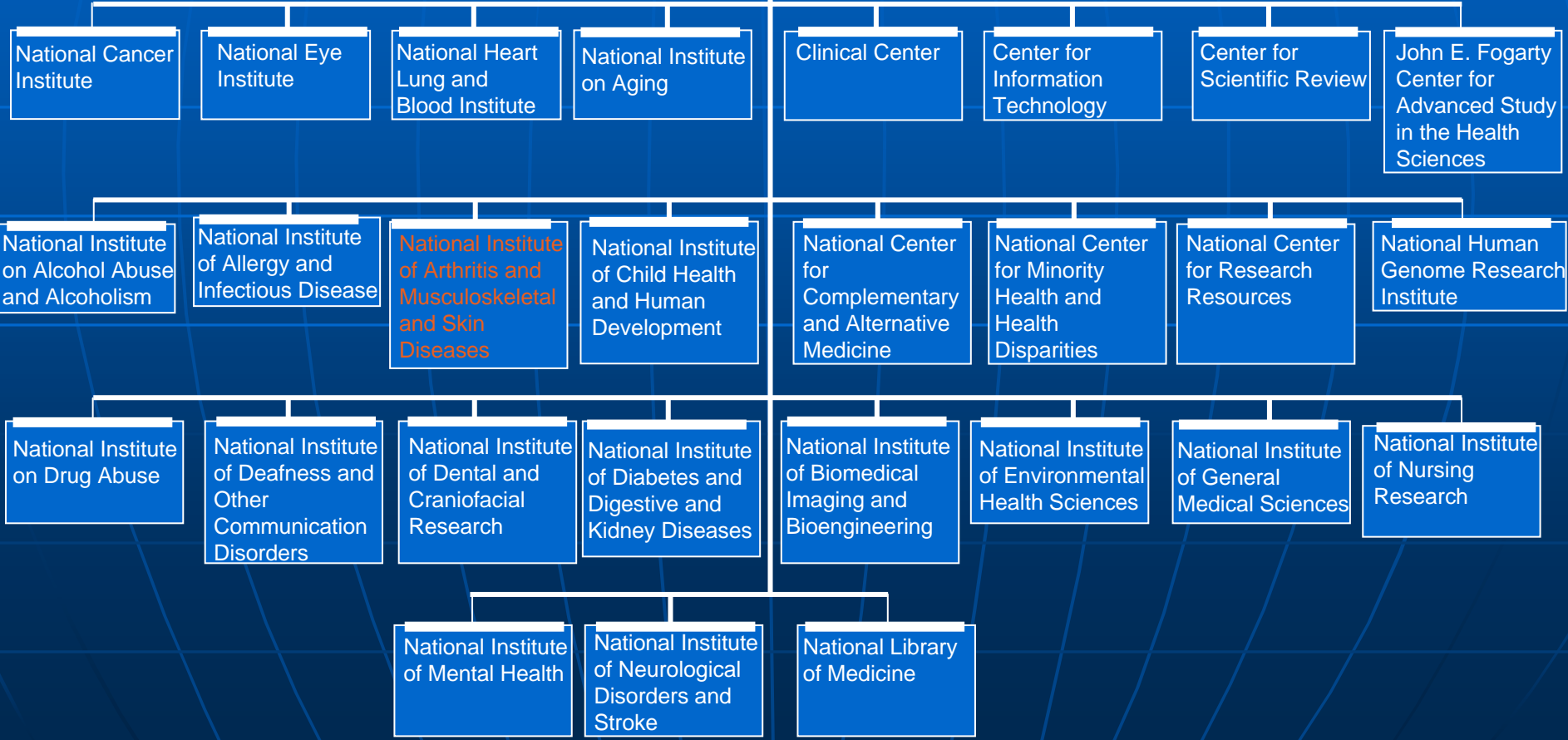
I do not have any relevant  
relationships with industry.

# The View of the NIH - A Complex Organization

Office of Research on Women's Health  
Office of AIDS Research, Office of Dietary Supplements, Office of Behavioral and Social Sciences Research, Office of Disease Prevention,  
**Office of Rare Diseases,**

**OD**  
**Deputy and Associate Directors**  
**Administrative Offices**

**Advisory Committee to the Director**



# **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/>

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES  
**National Institutes of Health**  
ORD, NCRR, NIAMS, NICHD, NHLBI, NIDDK,  
NINDS,

**Coalition of Patient  
Advocacy Groups  
(CPAG)**

**The Data Technology  
Coordinating Center**



Cholestatic Liver  
Disease Consortium  
CLIC



Rare Lung  
Disease Consortium



- Collaborative Clinical Research
- Centralized Data Coordination and Technology Development
- Public Resources and Education
- Training



Rare Thrombotic  
Diseases  
Consortium

Rare Genetic Steroid  
Disorders Consortium

Angelman, Rett and  
Prader-Willi Syndromes Consortium

Genetic Diseases of  
Mucociliary Clearance Consortium



# 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
- Web site:  
[http://www.ott.nih.gov/licensing\\_royalties/raredisease\\_ovrww.asp](http://www.ott.nih.gov/licensing_royalties/raredisease_ovrww.asp)
- Press release:  
<http://www.nih.gov/news/pr/dec2006/ord-11.htm>

# 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](mailto:GARDinfo@nih.gov)

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