

DISCLOSURE OF RELEVANT
RELATIONSHIPS WITH
INDUSTRY

Stephen C. Groft, Pharm.D.

Fortran 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

National Cancer
Institute

National Eye
Institute

National Heart
Lung and
Blood Institute

National Institute
on Aging

Clinical Center

Center for
Information
Technology

Center for
Scientific Review

John E. Fogarty
Center for
Advanced Study
in the Health
Sciences

National Institute
on Alcohol Abuse
and Alcoholism

National Institute
of Allergy and
Infectious Disease

National Institute
of Arthritis and
Musculoskeletal
and Skin
Diseases

National Institute
of Child Health
and Human
Development

National Center
for
Complementary
and Alternative
Medicine

National Center
for Minority
Health and
Health
Disparities

National Center
for Research
Resources

National Human
Genome Research
Institute

National Institute
on Drug Abuse

National Institute
of Deafness and
Other
Communication
Disorders

National Institute
of Dental and
Craniofacial
Research

National Institute
of Diabetes and
Digestive and
Kidney Diseases

National Institute
of Biomedical
Imaging and
Bioengineering

National Institute
of Environmental
Health Sciences

National Institute
of General
Medical Sciences

National Institute
of Nursing
Research

National Institute
of Mental Health

National Institute
of Neurological
Disorders and
Stroke

National Library
of Medicine

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)
 - GEF - 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

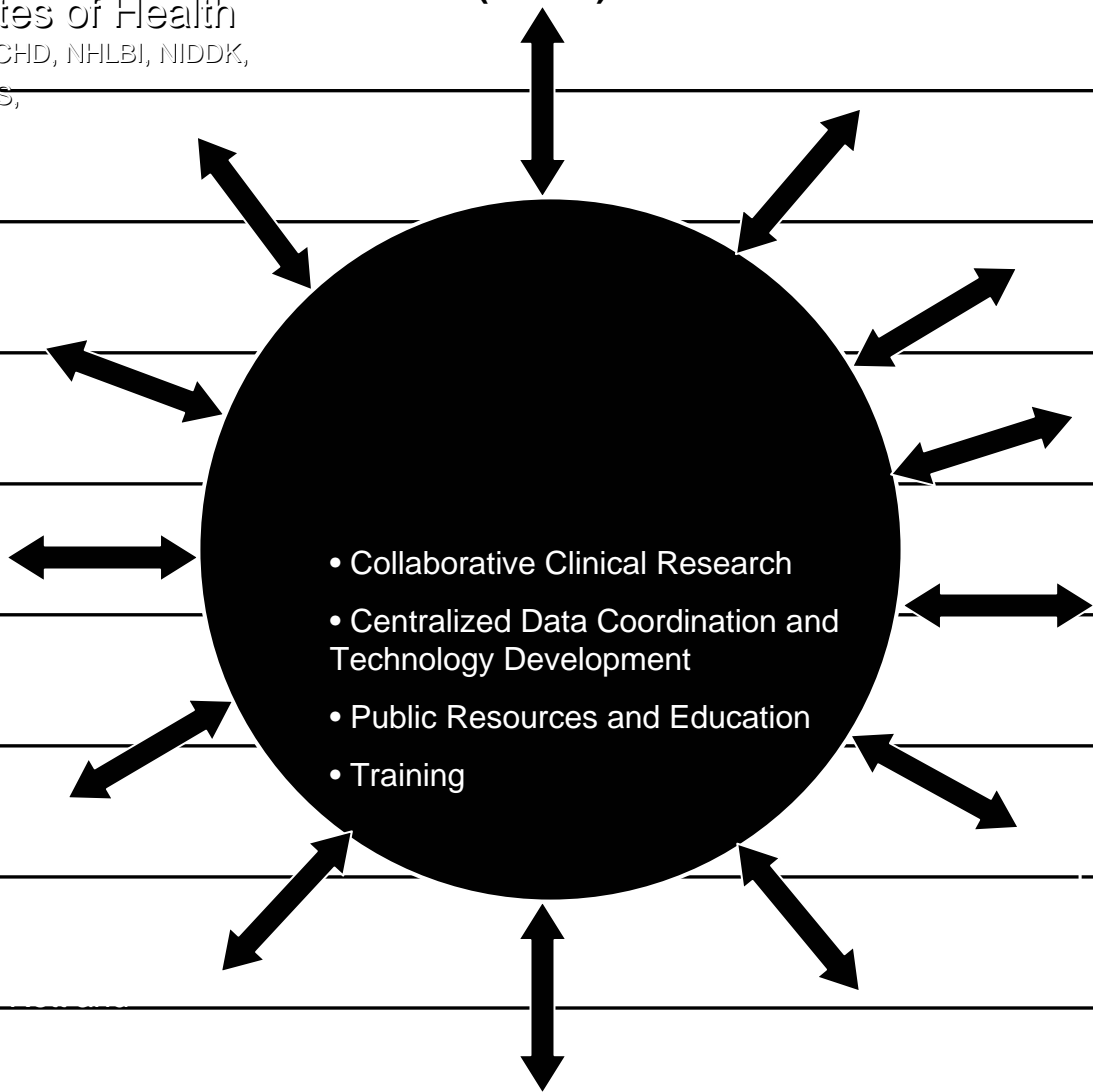
Purposes of Cooperative Rare Diseases Clinical Research Network (RDCRN)

- ┆ 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/>

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

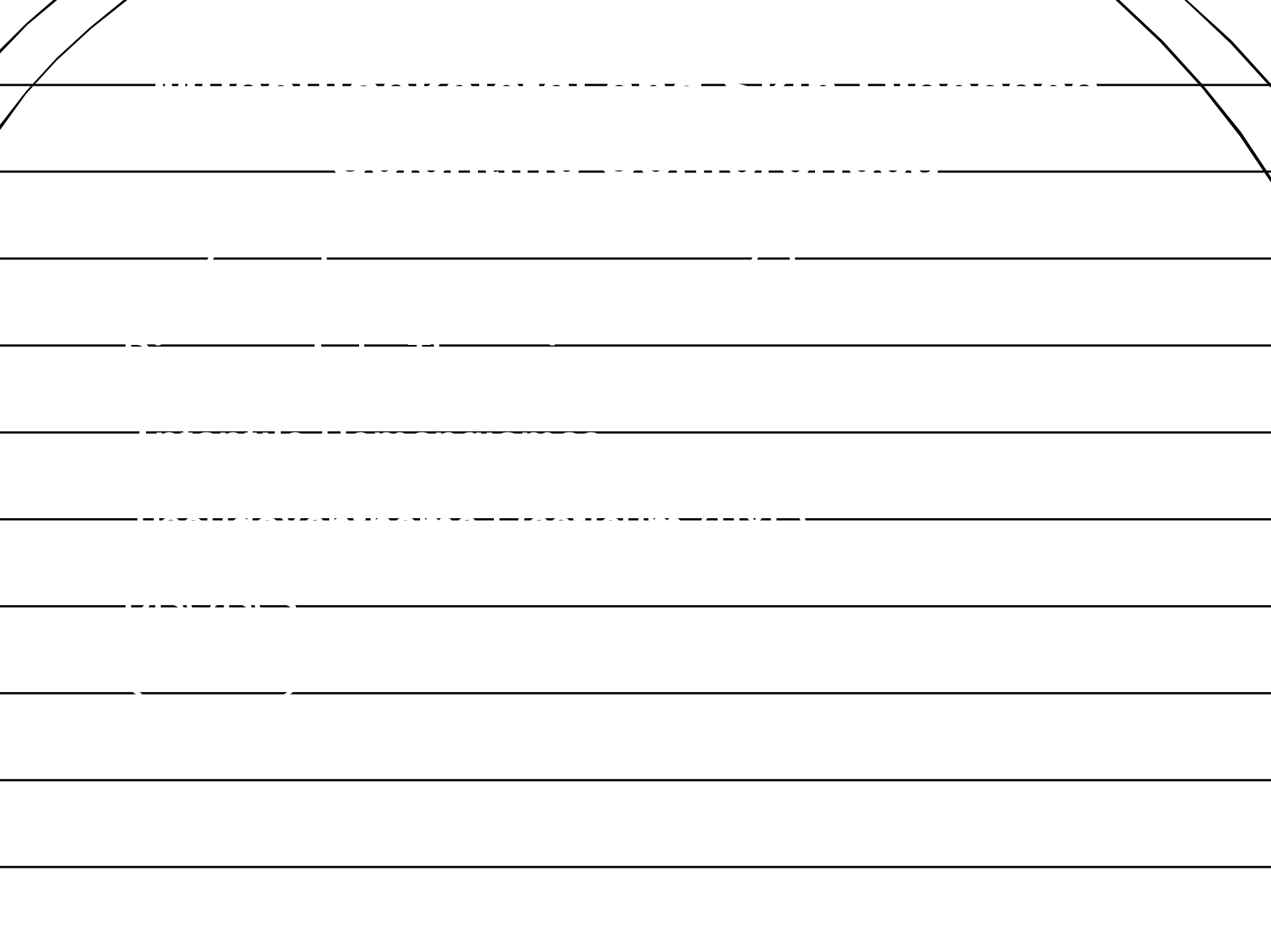
U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

National Institutes of Health
ORD, NCCR, NIAMS, NICHD, NHLBI, NIDDK,
NINDS,



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



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 OFFORD

- 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_ovrvw.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

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