8:30 a.m. **First Morning Session**

- **Opening Remarks**  
  *John I. Gallin, M.D.* — Director, Clinical Center, National Institutes of Health (NIH)

- **Welcoming Remarks**  
  *Francis S. Collins, M.D., Ph.D.* — Director, NIH

- **The Accomplishments and Goals of the Rare Disease Caucus**  
  *Congressman Leonard Lance* — Representative (R-NJ 7th District)

- **National Center for Advancing Translational Sciences (NCATS) Director Remarks**  
  *Christopher Austin, M.D.* — Director, NCATS, NIH

- **Common Devices for Rare Diseases**  
  *William Heetderks, M.D., Ph.D.* — Director, Extramural Science Program, National Institute of Biomedical Imaging and Bioengineering (NIBIB), NIH

- **Radiology Imaging of Rare Diseases: New Frontiers**  
  *Ronald M. Summers, M.D., Ph.D.* — Senior Investigator and Staff Radiologist Chief, Clinical Image Processing Service Chief, Imaging Biomarkers and Computer-Aided Diagnosis Laboratory Radiology and Imaging Sciences, Clinical Center, NIH

- **An Open Notebook: A Reporter’s Perspective on Covering Rare Diseases and the NIH Cyclodextrin Trial**  
  *Amy Dockser Marcus* — Reporter, Wall Street Journal

- **Introducing the Undiagnosed Diseases Network**  
  *Anastasia Wise, Ph.D.* — Program Director, National Human Genome Research Institute (NHGRI), NIH

10:15 a.m. **Morning Break**

10:30 a.m. **Second Morning Session**

- **Introduction and Short Talk on Intramural Research**  
  *Michael Gottesman, M.D.* — Deputy Director for Intramural Research, NIH

- **Brain-Directed Adeno-Associated Virus (AAV) Gene Therapy for Neurometabolic Rare Diseases**  
  *Stephen G. Kaler, M.D.* — Senior Investigator, Molecular Medicine Program, Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), NIH

- **A Recurrent Mutation Causing Ablepharon Macrostomia Syndrome**  
  *Taylor Davis* — Post-Baccalaureate Intramural Research Training Award (IRTA) Fellow, Undiagnosed Diseases Program (UDP), NHGRI, NIH

- **Development of Effective Therapies for Neurofibromatosis Type 1 Related Tumors: Preclinical and Clinical Trials**  
  *Brigitte Widemann, M.D.* — Senior Investigator, National Cancer Institute (NCI), NIH

- **Targeting the Metabolic Basis of Kidney Cancer**  
  *W. Marston Linehan, M.D.* — Chief, Urologic Surgery, NCI, NIH
12:00 p.m. Lunch and Poster Session, Clinical Center Tours

1:30 p.m. First Afternoon Session

- NICHD/NHGRI Newborn Sequencing Program
  Tiina Urv, Ph.D. — Program Director, Intellectual and Developmental Disabilities Branch (IDDB), NICHD, NIH

- Clinical Center Sequencing Program
  Dan Kastner, M.D., Ph.D. — Scientific Director, Division of Intramural Research, NHGRI, NIH

- The Diploid Alignment Pipeline: Using Individual, Parental, and Population Information to Improve Sequence Alignment
  Elise Flynn — Post-Baccalaureate IRTA Fellow, UDP, NHGRI, NIH

- UDPICS: Translational Research Information System Developed by UDP
  Murat Sincan, M.D. — Research Fellow, NHGRI, NIH

- Uplifting Athletes Update
  Scott Shirley — Executive Director, Uplifting Athletes

- Next Steps for Newly Diagnosed
  Tamandra Morgan, M.S. — Genetic and Rare Diseases Information Center (GARD), Office of Rare Diseases Research (ORDR), NCATS, NIH

3:00 p.m. Afternoon Break

3:15 p.m. Second Afternoon Session

- Biopharmaceutical Pipeline and Orphan Diseases
  Genia Long — Senior Advisor, Analysis Group, Inc.

- Rare Diseases and the Biopharmaceutical Industry
  William W. Chin, M.D. — Executive Vice President, Science and Regulatory Affairs, The Pharmaceutical Research Manufacturers of America (PhRMA)

- Food and Drug Administration (FDA) Updates
  Gayatri Rao, M.D., J.D. — Director, Office of Orphan Product Development (OOPD), FDA

- Genetic Alliance Updates
  James O’Leary, M.B.A. — Chief Innovation Officer, Genetic Alliance

- National Organization for Rare Disorders (NORD) Updates
  Diane Dorman — Vice President of Public Policy, NORD

4:45 p.m. Wrap Up

5:00 p.m. Adjourn