



Agenda

FIFTH INTERNATIONAL CONFERENCE ON RARE AND UNDIAGNOSED DISEASES

Karolinska Institutet
Stockholm, Sweden
August 30-31, 2017

Tuesday, August 29

6:30 Bus departs from Elite Palace to Welcome Dinner at the Vasa museum for those available

Wednesday, August 30

8:30 Welcome, Introductions, Listing of Official UDNI Members
Ann Nordgren, Anna Wedell (Karolinska)
William Gahl (NIH)
Helene and Mikk Cederroth (Wilhelm Foundation)

9:00-9:15 Goals of this meeting: Updates of Sites
Data Sharing (Thursday AM)
Planning (After video)

William Gahl

2-Minute video from Australia
Gareth Baynam

9:15-10:00 UDNI Organization (**Gahl, Domenica Taruscio**)
Board of Directors
Voting Today and in Future
Charter & Appendices
Tokyo changes
BOD's proposals
Membership

Non-Clinical Members (**Eric Klee**)
Advocacy Groups (**Paul Melmeyer**)
Committees or Working Groups? Prepare to nominate.
Membership
Program
Functional Research
Development
Website, Journal (if time)

10:00-10:30

Break (Wilhelm Foundation)

10:30-12:15

Updates from Various Centers (10 min TOTAL on #cases seen/describe a case/invite collaboration)

UDN (Anastasia Wise)

Australia (Gareth Baynam)

Sweden (Anna Lindstrand)

Japan (Kenjiro Kosaki)

Canada (Paul Lasko)

Netherlands (Wendy van Zelst-Stams)

Catalan (Aurora Pujol)

Alabama-Birmingham (Bruce Korf)

Utah (Lorenzo Botto)

12:15-1:15

Lunch (Wilhelm Foundation)

1:15-3:00

More updates from Centers (Same as above)

Vienna (Vanja Nagy)

Italy (Domenica Taruscio, Francesco Brancati, Vincenzo Nigro, G. Casari, Marco Tartaglia)

Sri Lanka (Vajira Dissanayake)

India (Ratna Puri)

Spain (Manuel Posada)

Mayo (Steve Ekker)

Emory (Clyde Partin)

South Africa (Judith Kumuthini)

3:00-3:30

Break (Wilhelm Foundation)

3:30-3:50

Proposal for Advocacy Group Involvement in UDNI

Eurordis (Patient Data Sharing Opinions)

NORD

3:50-4:05

Whole genome sequencing for diagnosis and discovery of monogenic disorders

Anna Wedell

4:05-4:20

Discussion

4:20-4:30

Rest room break

4:30

**Bus leaves from Nobel Forum to boat trip and dinner
(UDN/UDNI Members Only)**

Thursday, August 31

8:30-

Industry brochures available

9:00-10:00

Update on Data Sharing – 10 min Summary of Progress
Alexa McCray, UDN Coordinating Center

Panel Discussion

Chair: David Adams, NIH UDP

**Participants: Alexa McCray, Harvard
Tudor Groza, Patient Archive
Orion Buske, Phenome Central**

10:00-10:30

Break (Wilhelm Foundation)

10:30-12:00

Business Discussion (**Gahl**)
Ratify Charter
Choose Database
Nominate Committee Members
For Working Groups
Discuss Funding
Website
Newsletter (**Domenica Taruscio**)
Journal
Next Meeting

12:00-1:00

Lunch (Wilhelm Foundation)

1:00-2:00

**Volunteer short cases or functional studies
Matt Wheeler
Giedre Grigelioniene
Metabolomics compound sharing (D. Koeller – 5 min)**

2:00-2:15

Break (Wilhelm Foundation)

- 2:15-3:30 Sustainability of the UDNI
- Use and Outcomes Data
 - Administrative Requirements and Staff
 - Patient Recruitment
 - Measures of Success: Applications, Acceptances, Publications
- Gahl, Groft, Wise**
- 3:30-3:45 **Anders Olauson**, NGO Committee for Rare Diseases
- 3:45-5:00 Drinks and snacks at Nobel Forum
- 5:30-8:00 **Early dinner at Restaurant Stallmästaregården**