Agenda

SIXTHINTERNATIONAL CONFERENCE ON RARE AND UNDIAGNOSED DISEASES

Telethon Institute of Genetics and Medicine
Via Campi Flegrei 34, 80078
Pozzuoli (NA), Italy
June 19-21, 2018

Tuesday, June 19

6:30 pm **Welcome Dinner- Participants Abroad (Courtesy of TIGEM)**

Wednesday, June 20

SESSION 1: *Chairs: William Gahl, Giorgio Casari*

9:00	Welcome **Giorgio Casari** (Coordinator, Telethon UDP, TIGEM) **Helene and MikkCederroth**(Wilhelm Foundation) Introductions, List of Official UDNI Members, Board **William Gahl**(NIH)
9:30-9:45	UDNI Organization Meeting Goals: Committee/Working Group Reports Confirm Data Sharing Policies Discuss Science and Cases Address Future of UDNI What we will vote on (Charter, Appendices, BOD proposals) Who votes (Today and in Future) **William Gahl**
9:45-10:30	Committee Reports 9.15-9.30 Membership**(Eric Klee)** 9.30-9.45 Program**(Vincenzo Nigro, William Gahl)** 9.45-10.00 Communications**(Domenica Taruscio)**
10:30-11:00	Working Group Reports

10.00-10.15 Functional Research **(Eric Klee)**
10.15-10.30 Patient Engagement **(Paul Melmeyer)**

11:00-11:30 **Coffee/Vitamin Break (TIGEM)**

SESSION 2: *Chairs: Vincenzo Nigro and Nicola Brunetti-Pierri*

11:30-12:00 Science/Cases – Solved **(**6 min: 4 min present+ 2 min Q&A)

M. Posada: Data sharing: lights and shadows shown through two undiagnosed cases

D. Babovic-Vuksanovic:*RINT1*biallelic alterations: a novel cause of infantile onset recurrent liver failure with dysostosis multiplex

A. Pujol(DEGS1): Biallelic mutations of the dihydroceramidedesaturase DEGS1 gene cause a novel hypomyelinatingleukodystrophy with atherapeutic hope

12:00-1:00pm Cases – Mysteries (6 min: 4 min presentation + 2 min Q&A

C. Radio: The Undiagnosed Patients Program @OPBG

Y. Anikster: Affected siblings sharing homozygous nonsensemutations in new genes: Does n=1 or 2?

A. Nordgren: An unknown syndrome in a boy with intellectual disability, structural brain anomalies, ulnar polydactyly, tetralogy of Fallot, skeletal abnormalities, hypospadias and dysmorphic features

U. Ozbek: Missense EPG5 mutation segregation in 2 siblings withisolated neurological involvement. Isolated form of Vici syndrome?

N. Brunetti-Pierri:*RAB10:* a candidate gene for a severe Phenotypewith developmental delay, hypotonia, epilepsy and microcephaly

N. Brunetti-Pierri: Ectodermal dysplasia and syndactyly of Handsandfeet in a subject harboring compound heterozygous variants in the *ZFYVE16* gene

R. Puri: Undiagnosed case of dysmorphic features

G. Baynam: Undiagnoseddysmorphic case

1:00 pm-2.00pm Lunch (TIGEM)

SESSION 3: *Chairs Domenica Taruscio and Gareth Baynam*

2:00pm-4:00 Collaborative Networks, Services, Innovations

2:00-2:20 WuXi NEXTCODE
Christina Waters

2:20-2:40 Models and Mechanisms
Paul Lasko(Canada)

2:40-3:00 Studies with indigenous populations
Gareth Baynam (Australia)
Paul Lasko (Canada)

3:00-3:20 Genetic Counseling in the UDNI
Resource Packs +/- a Diagnosis
Janine Lewis, GARD
Cases
Stephanie Broley, Australia

3:20-3:40 COST; Geographical bottlenecks
Bela Melegh, ZeynepTumer, Manuel Posada, Domenica Taruscio

3:40-4:00 A Genetic Evaluation Pipeline
Kevin Strauss, USA

4:00-4:30 Coffee/Vitamin Break (TIGEM)

Session 4-*Chairs: Holm Graessner and KenjiroKosaki*

4.30-4:45 Solve-RD Consortium
Holm Graessner, Tubingen

4:45-5:00 Efforts in rare and undiagnosed Diseases
Claudia Gonzaga-Jauregui, Regeneron

5:00-5:15 Japan's IRUD, data sharing, model organism team
KenjiroKosaki, AMED

5:15-5:20 Korea's UDP-web for international data sharing
DongsungRyu

5.20-5:35 Innovative NGS Methods to Diagnose the Undiagnosed
Vincenzo Nigro(Telethon)

5:35-5:40 NIH Undiagnosed Diseases Network Affiliate Members
Anastasia Wise

5:40-6.00 Discussion on all Networks, Services, Innovations

6.00 Adjourn

6:30 pm **Dinner (Wilhelm Foundation)**

Thursday, June 21

8:30- 9:00 Industry brochures available

SESSION 5: *Chairs: Raffaele Castello and Michael Brudno*

9:00-10:00

9:00-9:20 Data Sharing
David Adams, NIH UDP
Tudor Groza, Patient Archive
Michael Brudno

9:20-9:40 Options for data sharing
David Adams

9:40-10:00 Progress Report on Phenome Central Entries
Michael Brudno
Panel 1: Within Nation Sharing Experiences
Panel II: International Sharing Experience

10:00-10:15 RD-Connect, Solve-RD, and the UDNI
Manuel Posada, Domenica Taruscio

10:15-10:30 Discussion of UDNI-wide federated systems

10:30-11:00 **Coffee/Vitamin Break (TIGEM)**

11:00-12:15 Business Discussion **(Gahl)**
- Ratify Charter, Appendices
- Choose Database (Noda Bene?)
- ConfirmNew Committee Members
- Discuss Other Working Groups
- Patient Recruitment
- Next Meeting
- Other topics, Announcements

12:15-1:30pm **Lunch (TIGEM)**

SESSION 6: *Chairs: William Gahl and David Adams*

1.30-1-50	Management of the (quite) unmanageable: Pros and Cons of a super-network based Undiagnosed Diseases Program **Raffaele Castello**
1:50-2:35	Considerations of Philanthropy **Gareth Baynam, Steve Groft, Paul Lasko**
2:35-3:45	Sustainability of the UDNI - Use and Outcomes Data - Administrative Requirements and Staff - Measures of Success: Applications, Acceptances, Publications **Gahl, Groft, Wise**
3.45-4.15	Coffee/Vitamin Break
4:15-4:45	Next meeting proposals and vote.
4:45-5:15	Tour of the Institute
5:15-6:30	TBD. Cases or discussion from floor or adjourn
6.30-pm	Goodbye; Network Strengthening Dinner (Wilhelm Foundation)

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