

8th International Conference on Rare and Undiagnosed Diseases

The Aula, Comeniuslaan 2 6525 HP Nijmegen

Radboud Universitair Medisch Centrum

February 7-8, 2020, Nijmegen, Netherlands

Day 1, February 7, 2020

0800-0900 **Registration (coffee and tea)**

0900-0910 **Welcome**

**Wendy van Zelst-Stams
Helene Cederroth
William Gahl**

0910-1040 **Committee & Working Group Reports (Chair: William Gahl)**

0910-0920 Membership, how to enforce 5 cases

Eric Klee

0920-0930 Genetic Counseling Working Group

Janine Lewis

0930-0940 Communications/Website/White Paper #2

**Domenica Taruscio,
Marco Salvatore**

0940-1000 Education Working Group

**Béla Melegh, Domenica
Taruscio**

1000-1020 Functional Research Working Group

Eric Klee

Website, UDN Genes, ModelMatchmaker

Shinya Yamamoto

1020-1030 Patient Engagement

**Helene Cederroth, Vanessa
Boulanger**

1030-1040 Rare Barometer Voices (Survey)

Virginie Bros-Facer

1040-1110 **Break**

1110-1215 **Business meeting UDNI: Going Forward**

William Gahl

Ratify Charter, Appendices, Database, BOD proposals

Confirm Data Sharing Policies

Confirm New Committee Members & Criteria

Voting Eligibility and Rules

Discuss Other Working Groups

Patient Recruitment

Next Meeting

Board Members rotate off (1/3 switch every year)

Reaching out

Sustainability/Future of UDNI

Other Topics, Announcements

1215-1330 **Lunch**

Global Undiagnosed Disease Initiatives (Chair: Wendy van Zelst-Stams)

1330-1415 Undiagnosed & Rare Diseases initiatives in the Netherlands

1330-1345	WGS first/RADICON-NL	Lisenka Vissers
1345-1400	United for metabolic diseases	Clara van Karnebeek
1400-1415	X-omics	Alain van Gool

1415-1500 Undiagnosed & Rare Diseases initiatives in Europe

1415-1430	1+MG	Ruben Kok
1430-1445	Solve-RD	Alex Hoischen
1445-1500	ERNs	Irene Matthijssen

1500-1530 **Break**

1530-1630 International initiatives

1530-1540	IRDIRC, RD treatment access WG	William Gahl
1540-1550	Expanding the UDNI into Georgia and the Southern Caucasus Region	Oleg Kvlividze
1550-1600	NGO committee for RD	Anders Olauson
1600-1610	World Economic Forum	Lynsey Chediak
1610-1620	UDN in USA	Alexa McCray
1620-1630	Congenica	Andrea Haworth
1630-1640	Global Commission to End the Diagnostic Odyssey	Durhane Wong-Rieger

1640-1730 Presentation of unsolved cases (Chair: William Gahl)

1640-1650	Primary immune deficiency	Yuk Yin Ng
1650-1700	Hypertrophic cardiomyopathy in Thailand	Vorasuk Shotelersuk
1700-1710	Duodenal atresia, renal failure, cataract	Josephine Chong
1710-1720	Autoimmune lymphoproliferation	Ratna Puri
1720-1730	ASD, VSD, developmental delay	Carlo Marcelis

1730 Social gathering with drinks

1830 **Dinner for UDNI members & invitees (sponsored by Wilhelm Foundation)**

Day 2, February 8, 2020

0800-0900 **Registration (coffee and tea)**

0900-1030 **Learning from success (Chair: Han Brunner)**

0900-0915	Going Dutch	Han Brunner
0915-0930	The Australian way	Gareth Baynam
0930-0940	The Rare Disease Initiative in Germany	Heiko Krude
0940-0950	Care for Rare Canada	Michael Brudno
0950-1000	Hospital-supported UDP	Anna Hurst
1000-1010	Telethon Italy UDP	Raffaele Castello, Vincenzo Nigro
1010-1020	Japan's IRUD	Hisato Suzuki

1020-1050 **Break**

1050-1230 **Datasharing (Chair: David Adams)**

1050-1105	# PC cases; data federation	Michael Brudno
1105-1115	email listserv, share with UDN)	Alexa McCray
1115-1130	RD-Connect GPAP; ELIXIR	Sergi Beltran Agullo
1130-1140	Successful sharing	Christian Gilissen
1140-1150	From data sharing to data visiting	Peter-Bram 't Hoen
1150-1200	DART for sequence analysis	Daniele Barreca
Solved cases (10 min each)		
1200-1210	TAF6 and Alazami-Yuan syndrome	Ecenur Tuc
1210-1220	AUTS2 syndrome	Estrella Lopez Martin
1220-1230	CDH1 and gastric/breast cancer	Jose Garcia-Pelaez

1230-1330 **Lunch (sponsored by PhenoTips/Gene42)**

1330-1500 **Therapeutic models (Chair: Rob Collin)**

1330-1355	Cockayne syndrome	Jan Hoeijmakers
1355-1410	The future of new treatments in RD	Rob Collin
1410-1425	Usher syndrome	Erwin van Wijk
1425-1440	Brain in a dish	Nael Nadif Kasri
1440-1450	RNAseq and ASO therapeutic targets	Eric Klee

1450-1500	Taurine for SLC6A6 retinal/cardiac	Stylios Antonarakis
1500-1530	Break	
1530-1700	Functional models (Chair: Paul Lasko)	
1530-1545	RNA sequencing	Patrick Deelen
1545-1600	Zebrafish	Jo Vanoevelen
1600-1615	DNA methylation profiles	Marielle Alders
1615-1630	RDMM network	Paul Lasko
1630-1640	Tubulin detyrosination and HSP	Aurora Pujol
1640-1650	Hypotonia, cerebellar findings-ATG4D	May Malicdan
1650-1700	Closing session: Other topics (Chair: William Gahl)	
1730	Dinner for UDNI members and invitees (sponsored by Wilhelm Foundation)	