

Joint Outreach Day
3rd May 2017
H4 Hotel, Alexanderplatz, Berlin

Wifi Login: VA-Rare / Password: Rare052017

09.00 Registration

10.00 Introduction and scene setting *Room Alexander II+III*

Welcome

Holm Graessner

Welcome from the European Commission

Iiro Eerola

Impact of NeurOmics

Olaf Riess

Impact of EURenOmics

Franz Schaefer

Impact of RD-Connect

Hanns Lochmüller

11.00 Data Sharing *Room Alexander II+III*

This session will explore the current trends in data sharing, as Rare Disease data is often fragmented, siloed and inaccessible for research. The session will focus on the integrated genomics analysis platform, and other data sharing mechanisms, tools and requirements developed by RD-Connect and how this has been utilised to improve diagnostics and therapy. Discussion will look towards future challenges, both the technical and the legal, ethical and social implications of increased data sharing.

This session will have a 15 minute introductory presentation followed by 45 minute panel discussion.

Chair

Ivo Gut (RD-Connect)

Peter-Bram 't Hoen

Cathy Turner

Panel Members

Matthias Kretzler

Joseph Irwin

Veronica Popa

12.00 Lunch (*H4 Hotel Restaurant*)

13.00 **Diagnostics** *Room Alexander II+III*

Genetic testing has an increasing role in diagnosing rare disorders and is important for genetic counselling, carrier testing, prenatal diagnosis, preimplantation genetic diagnosis (PGD), identifying risk factors and may be useful for determining appropriate treatment. With the availability of Next Generation Sequencing (NGS) techniques (such as disease-specific multi-gene panels, whole exome sequencing [WES] and whole genome sequencing [WGS]) the diagnostic yield of genetic testing has significantly improved. The discussion will focus on gene discovery, yet undiagnosed patients and the next frontiers in Rare Disease diagnostics.

This session will have a 15 minute introductory presentation followed by 45 minute panel discussion.

Chair	Nine Knoers (EURenOmics)
	Milan Macek
Panel Members	Peter Bauer
	Daniel Renault

14.00 **Therapy** *Room Alexander II+III*

This session will discuss the recent advances made in -omics research into the development of genetic and other targeted therapies. Recent years have seen new drugs and approaches making their way from the lab to the patient in rare neurological and renal diseases. This is an effort requiring patients, academics and industry. In this session we will reflect on past efforts to identify success factors but also our mistakes, for the benefit of future therapy development in Rare Diseases.

This session will have a 15 minute introductory presentation followed by 45 minute panel discussion.

Chair	Annemieke Artsma-Rus (NeurOmics)
	Lucia Monaco
Panel Members	Jack Wetzels
	Jenny Versnel

15.00 **Coffee Break (Foyer Conference Centre)**

15.30 **Optional training workshops for researchers**

RD-Connect Sample Catalogue for Biobanks
Mary Wang
Room: Alexander II+III

RD-Connect platform
Sergi Beltran
Room: Grenander II

18.00 **Joint RD-Connect, NeurOmics, EURenOmics and E-RARE poster session and drinks reception (Alexander I)**