Invitation to the Joint Outreach Day of NeurOmics, EURenOmics und RD-Connect

3. May 2017, Ramada Hotel, Berlin Alexanderplatz, Germany

Rare Diseases affect millions of people in Europe, an estimated 8% of the general population, and consume 20% of health care budgets. They are chronic, disabling, often life-limiting, usually without a cure or an effective treatment, and burden affected people and their families. The European Commission, patient organizations and others have recognized the specific need for coordinated research into Rare Diseases and joined forces through the International Rare Disease Research Consortium (IRDiRC). Flagship funded projects RD-Connect, NeurOmics and EURenOmics started out in 2012 to develop better diagnosis and treatment for Rare Diseases through cutting-edge research, in particular the revolutionary -omics technologies. On behalf RD-Connect, NeurOmics and EURenOmics you are invited to attend a rare disease outreach day on 3rd May in Berlin, Germany. The venue will be Ramada Hotel Berlin Alexanderplatz.

The aim of this day is to bring together stakeholders from the rare disease community to share the value of the tools created and the knowledge generated through these projects. In particular to discuss how their utility can grow beyond the original scope and interact with other organisations and infrastructures in order to make an even larger impact on the field of rare diseases. The day will provide lots of time for discussion and interaction with researchers, policy makers, pharmaceutical representatives and patient organisations.

The day will focus on three main themes:

Data sharing

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.

Diagnostics

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.

Therapy

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.

There is no charge for attendance but we ask that you register here: Registration Page

A draft agenda is available here: Draft Programme

Please don't hesitate to get in touch if we can provide any more information,

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About RD-Connect:

RD-Connect is a global research and infrastructure resource for rare diseases. Set up to overcome the siloing, fragmentation and inaccessibility of datasets from different projects, it links omics data with phenotypic data and information in registries and biobanks at both an individual-patient and whole-cohort level to enable researchers to analyse their own data and gain a complete view of their disease and patient population of interest.

About NeurOmics:

NeurOmics is a research project investigating ten rare neurodegenerative and neuromuscular diseases entities. It aims to improve the lives of affected patients by identifying new disease causing genes, by establishing new and standardized diagnosis tools, by finding new modifiers of disease onset and course, and finally by developing novel therapy approaches.

About EURenOmics:

EURenOmics is a health project devoted to improving the lives of patients affected by rare kidney diseases. It aims to develop novel tools that will allow to make more accurate diagnoses, predict the disease course and the efficacy of available treatments, and help developing new and better therapies for rare kidney diseases.