

## NeurOmics Final Meeting 3-5 May 2017 | Berlin, Germany

Venue: H4 Hotel Berlin-Alexanderplatz, Karl-Liebknecht-Straße 32, 10178 Berlin

### Wednesday, May 3<sup>rd</sup>: Joint Outreach Day with RD-Connect and EURenOmics

	<b>Registration</b> ( <i>Foyer Conference Centre</i> ) The registration will be open from 9:00-11:00 and from 15:00-18:00.		
<b>10:00</b>	<b>Joint Plenary session</b> ( <i>Room: Alexander II+III</i> ) <ul style="list-style-type: none"> <li>▪ Welcome and overview of the day (<i>Holm Graessner</i> – 10 min)</li> <li>▪ Welcome from the European Commission (<i>Iiro Eerola</i> – 10 min)</li> <li>▪ Impact of NeurOmics (<i>Olaf Riess</i> – 10 min)</li> <li>▪ Impact of EURenOmics (<i>Franz Schaefer</i> – 10 min)</li> <li>▪ Impact of RD-Connect (<i>Hanns Lochmüller</i> – 10 min)</li> </ul>		
<b>11:00</b>	<b>Data sharing</b> ( <i>Room: Alexander II+III</i> ) <ul style="list-style-type: none"> <li>▪ Introduction (<i>Ivo Gut</i>, RD-Connect - 15 min)</li> <li>▪ Panel discussion (45 min) Panel: <i>Peter-Bram 't Hoen</i> (RD-Connect), <i>Matthias Kretzler</i> (EURenOmics), <i>Cathy Turner</i> (NeurOmics), <i>Joseph Irwin</i>, <i>Veronica Popa</i></li> </ul>		
<b>12:00</b>	<b>Lunch</b> ( <i>Room: Alexander I</i> )		
<b>13:00</b>	<b>Diagnostics</b> ( <i>Room: Alexander II+III</i> ) <ul style="list-style-type: none"> <li>▪ Introduction (<i>Nine Knoers</i>, EURenOmics – 15 min)</li> <li>▪ Panel discussion (45 min) Panel: <i>Milan Macek</i> (RD-Connect), <i>Nine Knoers</i> (EURenOmics), <i>Peter Bauer</i> (NeurOmics), <i>Daniel Renault</i></li> </ul>	<b>Training workshop: RD-Connect platform</b> ( <i>Room: Grenander II – Chair: Sergi Beltran, Steve Laurie</i> )	
<b>14:00</b>	<b>Therapy</b> ( <i>Room: Alexander II+III</i> ) <ul style="list-style-type: none"> <li>▪ Introduction (<i>Annemieke Aartsma-Rus</i>, NeurOmics – 15 min)</li> <li>▪ Panel discussion (45 min) Panel: <i>Lucia Monaco</i> (RD-Connect), <i>Jack Wetzels</i> (EURenOmics), <i>Annemieke Aartsma-Rus</i> (NeurOmics), <i>Jenny Versnel</i></li> </ul>		
<b>15:00</b>	<b>Coffee break</b> ( <i>Room: Alexander I</i> )		
<b>15:30</b>	<b>Training workshop: RD-Connect platform</b> (repeat) ( <i>Room: Grenander II – Chair: Sergi Beltran, Steve Laurie</i> )	<b>Training workshop: RD-Connect Sample Catalogue for Biobanks</b> ( <i>Room: alexander II+III – Chair: Mary Wang</i> )	<b>PAC meeting</b> ( <i>Room: Grenander IV</i> )
<b>16:30</b>			<b>RD-PEC meeting</b> ( <i>Room: Grenander IV</i> )
<b>17:30</b>	<b>Poster set up</b> ( <i>Room: Alexander I</i> )		
<b>18:00</b>	<b>Poster session with finger food and wine</b> ( <i>Room: Alexander I</i> ) 18:00 - 19:00 Posters with <u>even numbers</u> will be presented 19:00 - 20:00 Poster with <u>odd numbers</u> will be presented		

## Thursday, May 4<sup>th</sup>

	<b>Registration</b> ( <i>Foyer Conference Centre</i> ) The registration will be open from 8:30-9:00.
<b>9:00</b>	<b>Joint plenary session</b> ( <i>Room: Alexander II+III – Chair: O. Riess / F. Schaefer</i> ) <ul style="list-style-type: none"> <li>▪ Welcome (<i>O. Riess, F. Schaefer</i> – 10 min)</li> <li>▪ RD-Connect (<i>Hanns Lochmüller</i> – 15 min)</li> <li>▪ NeurOmics 2012-2017 (<i>Olaf Riess</i> – 15 min)</li> <li>▪ EURenOmics 2012-2017 (<i>Franz Schaefer</i> – 15 min)</li> <li>▪ Best of NeurOmics (UK Cologne, <i>Brunhilde Wirth</i> – 10+5 min)</li> <li>▪ Best of EURenOmics (INSERM, <i>Pierre Ronco</i> – 10+5 min)</li> <li>▪ Best of NeurOmics (INSERM, <i>Fanny Mochel</i> – 10+5 min)</li> <li>▪ Best of EURenOmics (INSERM, <i>Corinne Antignac</i> – 10+5 min)</li> </ul>
<b>11:00</b>	<b>Coffee break</b> ( <i>Foyer Conference Centre</i> )
<b>11:30</b>	<b>Aim 2: Panel</b> ( <i>Room: Alexander III – Chair: O. Riess</i> ) <ul style="list-style-type: none"> <li>▪ Ataxia and HSP panel (EKUT, <i>Florian Harmuth</i> – 8+2 min)</li> <li>▪ Genetic characteristics of sporadic adult onset degenerative ataxia (DZNE, <i>Brigitte Paap</i> -8+2 min)</li> <li>▪ SMA/LMND panel (UK Cologne, <i>Mert Karakaya</i> – 8+2 min)</li> <li>▪ NMD panel (AMUMS, <i>Martin Krahn</i> – 8+2 min)</li> <li>▪ Experience of the INSERM team in diagnosis of hereditary spastic paraplegia (INSERM, <i>Giovanni Stevanin</i> – 8+2 min)</li> <li>▪ The Western Australian Neurogenetic Diseases Diagnostic Panel (UWA, <i>Nigel Laing</i> – 8+2 min)</li> <li>▪ Data integration and variant effect predictions (Bio-Product, <i>Bas Vroling</i> – 8+2 min)</li> <li>▪ Joint discussion on panel sequencing (30 min)</li> </ul>
<b>13:00</b>	<b>Group photo</b> <b>Lunch</b> ( <i>Restaurant Ramada Hotel</i> )
<b>14:30</b>	<b>Aim 5: Modifier</b> ( <i>Room: Alexander III – Chair: S. Tabrizi</i> ) <ul style="list-style-type: none"> <li>▪ CMS modifier (UNEW, <i>Yoshiteru Azuma</i> – 8+2 min)</li> <li>▪ Identification of genetic modifiers of Huntington's disease and other polyglutamine repeat disorders (UCL, <i>Davina Hensman Moss</i> – 8+2 min)</li> <li>▪ HSP/SPG4 modifier EKUT and INSERM (<i>Rebecca Schüle-Freyer</i> 4 min, <i>Livia Parodi</i> 4+2 min)</li> <li>▪ Disease modifiers WP6 (AMUMS, <i>NN</i> – 8+2 min)</li> <li>▪ Joint discussion on modifier identification (20 min)</li> </ul>
<b>15:30</b>	<b>Aim 1: Gene identification</b> ( <i>Room: Alexander III – Chair: B. Wirth</i> ) <ul style="list-style-type: none"> <li>▪ Technical overview on the sequencing platform (deCODE, <i>Olafur Magnusson</i> – 8+2 min)</li> <li>▪ New genes demonstrate broad overlap between HSP and ataxia (EKUT, <i>Rebecca Schüle-Freyer</i> – 8+2 min)</li> <li>▪ Gene identification (LUMC, <i>Silvere van der Maarel</i> – 8+2 min)</li> </ul>
<b>16:00</b>	<b>Coffee break</b> ( <i>Foyer Conference Centre</i> )
<b>16:30</b>	<b>Aim 1: Gene identification – continued</b> ( <i>Room: Alexander III – Chair: B. Wirth</i> ) <ul style="list-style-type: none"> <li>▪ Gene identification (UK Cologne, <i>Mert Karakaya</i> – 8+2 min)</li> <li>▪ Gene identification (UNEW, <i>Ana Topf</i> – 8+2 min)</li> <li>▪ Identification of new channelopathy genes (UCL, <i>Henry Houlden</i> – 8+2 min)</li> <li>▪ Gene identification (UCL, <i>Irina Zaharieva</i> – 8+2 min)</li> <li>▪ Increasing genetic heterogeneity in spinocerebellar degenerations: new genes, unexpected inheritance modes and novel mechanisms (INSERM, <i>Marie Coutelier</i> – 8+2 min)</li> <li>▪ Gene identification (VIB, <i>Danique Beijer</i> – 8+2 min)</li> <li>▪ New genes bridging muscle and heart (UNIFE, <i>Alessandra Ferlini</i> – 8+2 min)</li> <li>▪ NeurOmics Gene Identification Western Australian Style (UWA, <i>Nigel Laing</i> – 8+2 min)</li> <li>▪ Joint discussion on gene identification</li> </ul>
<b>18:30</b>	<b>On own</b>
<b>19:30</b>	<b>NeurOmics meeting dinner</b> ( <i>Frannz</i> )

<b>Friday, May 5<sup>th</sup></b>	
<b>9:00</b>	<b>Aim 3: biomarker</b> ( <i>Room: Alexander III – Chair: A. Durr</i> ) <ul style="list-style-type: none"> <li>▪ Technical overview on the metabolomics/lipidomics platform (medDay, <i>Alexandre Seyer</i> – 8+2 min)</li> <li>▪ Ataxia, FTL, HD, HSP (INSERM/ medDay, <i>Alexandre Seyer</i> – 15+5 min)</li> <li>▪ RNAseq and multi-omics analysis platform (LUMC, <i>Peter-Bram 't Hoen</i> – 8+2 min)</li> <li>▪ Biomarker study in SCA3 (EKUT, <i>Kathrin Gonsior</i>– 8+2 min)</li> <li>▪ Transcriptional dysregulation in Huntington's disease blood (UCL, <i>Michael Flower</i> – 8+2 min)</li> <li>▪ NMD biomarker (LUMC, <i>Pietro Spitali</i> – 8+2 min)</li> <li>▪ Joint discussion on biomarker identification (20 min)</li> </ul>
<b>11:00</b>	<b>Coffee break</b> ( <i>Foyer Conference Centre</i> )
<b>11:30</b>	<b>Aim 6: therapies</b> ( <i>Room: Alexander III – Chair: A. Aartsma-Rus</i> ) <ul style="list-style-type: none"> <li>▪ STOP-SPG5: A randomized controlled trial in SPG5 targeting neurotoxic oxysterols (EKUT, <i>Ludger Schöls</i> – 8+2 min)</li> <li>▪ SPG5 clinical trial (INSERM, <i>Fanny Mochele</i> – 8+2 min)</li> <li>▪ Antisense oligonucleotide therapy in spinocerebellar ataxia type 3 mice (LUMC, <i>Lodewijk Toonen</i> – 8+2 min)</li> <li>▪ MD mouse models (LUMC, <i>Annemieke Aartsma-Rus</i> – 8+2 min)</li> <li>▪ LGMD2B (AMUMS, <i>Marc Bartoli</i> – 8+2 min)</li> <li>▪ Hyperglycosylation for CMD (UCL, <i>Francesco Muntoni</i> – 8+2 min)</li> <li>▪ Joint discussion on therapy development (20 min)</li> </ul>
<b>12:50</b>	<b>Lunch</b> ( <i>Restaurant Ramada Hotel</i> )
<b>14:10</b>	<b>Aim 4: Pathophysiology</b> ( <i>Room: Alexander III – Chair: D. Rubinsztein</i> ) <ul style="list-style-type: none"> <li>▪ Integration of Lipidome and Gene Data to Monitor Disease Effects (Ariadne Diagnostics, <i>Ilya Mazo</i> – 8+2 min)</li> <li>▪ SPG5 hiPSC (EKUT, <i>Stefan Hauser</i> – 8+2 min)</li> <li>▪ Lipid metabolism dysregulation in SPG11 (INSERM, <i>Frederic Darios</i> – 8+2 min)</li> <li>▪ SMA hiPSC (UK Cologne, <i>Brunhilde Wirth</i> – 8+2 min)</li> <li>▪ Exploring axonal CMT through iPSC derived neurons (VIB, <i>Manisha Juneja</i> – 8+2 min)</li> <li>▪ Impairment of neural differentiation in HD-iPS cells (UMIL, <i>Marta Valenza</i>– 8+2 min)</li> <li>▪ QPCT, a druggable modulator of polyglutamine toxicity (UNICAM, <i>David Rubinsztein</i> – 8+2 min)</li> <li>▪ Analyzing the influence of parkin in the pathogenesis of SCA3 (EKUT, <i>Jeannette Hübener-Schmid</i> – 8+2 min)</li> <li>▪ Joint discussion (20 min)</li> </ul>
<b>15:50</b>	<b>Coffee break</b> ( <i>Foyer Conference Centre</i> )
<b>16:20</b>	<b>Final plenary session</b> ( <i>Room: Alexander III – Chair: V. Straub</i> ) <ul style="list-style-type: none"> <li>▪ NeurOmics – lessons learned (<i>Volker Straub</i> – 5 min)</li> <li>▪ Poster prize and joint discussion (35 min)</li> </ul>
<b>17:00</b>	<b>End of NeurOmics Final Meeting 2017</b>
<b>17:15</b>	<b>NeurOmics Steering Committee &amp; Scientific Advisory Board meeting</b> <i>(Room: Alexander III – Chair: O. Riess)</i>

The NeurOmics project has received funding from the European Union's Seventh Framework Programme for research, technological development and demonstration under grant agreement no 2012-305121.

