### Day 1 – Friday, 11 November

Welcome lunch, 12:00–13:00

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| **Time** | **Session** | **Speaker** |
| 13:00–13:15 | Welcome to Steps Forward in Pompe Disease 2016 | Ans van der Ploeg (*Erasmus MC University Hospital, Rotterdam, The Netherlands*) and Benedikt Schoser (*Friedrich-Baur Institute, Ludwig-Maximilians University, Munich, Germany*) |
| Session 1 | Natural and modified course of Pompe disease | Chairs: Pascal Laforêt & Giancarlo Parenti |
| 13:15–13:30 | Effects of ERT in infantile Pompe disease and the new transition phenotype | Andreas Hahn (*University of Giessen, Giessen, Germany*) |
| 13:30–13:45 | The CNS in infantile Pompe disease | Hannerieke van den Hout (*Erasmus Medical Center, Rotterdam, The Netherlands*) |
| 13:45–13:55(10) | Israel and Gaza's long-term (13 years) experience of the treatment of Infantile Pompe disease | Hanna Mandel (*Rambam Health Care Center, Rappaport School of Medicine, Technion, Haifa, Israel*) |
| 13:55–14:10 | Effects of ERT in children with non-classic phenotypes | Chris van der Meijden (*Erasmus Medical Center, Rotterdam, The Netherlands*) |
| 14:10–14:25 | Natural course and effects of ERT in adults | Pascal Laforêt (*Institute of Myology, Pitié-Salpêtrière Hospital, Paris, France*) |
| 14:25–14:40 | Results of 10 years of the Genzyme Registry | Eugen Mengel (*Johannes Gutenberg University Mainz, Children's Hospital of the University of Mainz, Mainz, Germany*) |
| 14:40–14:55 | EPOC | Benedikt Schoser |
| *14:55–15:05* | *Panel discussion with audience Q&A* |  |
| 15:05–15:30 | *Break* |
| Session 2 | Pathophysiology | Chairs: Nina Raben and Pim Pijnappel |
| 15:30–15:50 | Role of autophagy – TFEB | Nina Raben (*National Institutes of Health, Bethesda, MD, USA*) |
| 15:50–16:10 | Role of muscle regeneration and repair | Pim Pijnappel (*Erasmus MC University Medical Center, Rotterdam, The Netherlands*) |
| 16:10–16:30 | Genetic and modifying factors | Giancarlo Parenti (*Telethon Institute of Genetics and Medicine, Napoli, Italy*) |
| *16:30–16:45* | *Audience Q&A* |  |
| 16:45–17:05 | Immunomodulation in infantile Pompe disease | Priya Kishnani (*Duke University School of Medicine, Durham, North Carolina, United States*) |
| 17:05–17:25 | Exploring the pros and cons of immunomodulation in adult Pompe disease | Pascal Laforêt |
| *17:25–17:40* | *Panel discussion: The role of immune tolerance and modulation in Pompe disease* |  |
| 17:40–17:50 | Close Day 1 | Ans van der Ploeg and Benedikt Schoser |
| 19:00 – 21.00 | *Dinner* |

### Day 2 – Saturday 12 November

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| **Time** | **Session** | **Speaker** |
| 09:00–09:05 | Welcome to Day 2 | Ans van der Ploeg and Benedikt Schoser |
| Session 3 | Novel diagnostic approaches | Chairs: Antonio Toscano & Pascal Laforêt |
| 09:05–09:20 | MRI as a diagnostic tool in neuromuscular disorders | Robert-Yves Carlier (*Hôpital Raymond Poincaré, Garches, France*) |
| 09:20–09:30 | MRI of the cerebral vessels and brain | Antonio Toscano (*University of Messina, Messina, Italy*) |
| 09:30–09:40 | MRI of the diaphragm | Nadine van der Beek (*Erasmus Medical Center, Rotterdam, The Netherlands*) |
| 09:40–09:55 | Assessment of high-risk patients in respiratory clinics | Michael Polkey (*Royal Brompton Hospital & National Heart & Lung Institute, London, UK*) |
| 09:55–10:10 | Blood-based testing and whole genome screening – what is the current state in neuromuscular disorders? | Volker Straub (*Institute of Human Genetics, University of Newcastle upon Tyne, Newcastle Upon Tyne, UK*) |
| *10:10–10:30* | *Audience Q&A* |  |
| *10:30–11:00* | *Break* |
| Session 4 | 10 years’ of Enzyme Replacement Therapy | Chairs: Ans van der Ploeg & Benedikt Schoser |
| 11:00–11:05 | Introduction |  |
| 11:05–11:25 | A company perspective | Henk Schuring (*Vice President – Head Rare Neurological diseases, Sanofi Genzyme*) |
| 11:25–11:55 | A doctor’s perspective | Ans van der Ploeg |
| 11:55–12:15 | A patient´s perspective | Ria Broekgaarden *(Dutch Pompe patient organization VSN [Vereniging Spierziekten Nederland])* |
| *12:15–12:40* | *Panel discussion and audience Q&A: How has ERT changed Pompe care?* | *Moderated by Ans van der Ploeg & Benedikt Schoser* |
| *12:40–13:25* | *Lunch* |
| **13:25–14:25** | **Poster session** |
| Session 5 | Next therapeutic approaches | Chair: Giancarlo Parenti & Nina Raben |
| 14:25–14:45 | Results of NeoGAA and other ERT therapies under development | Mark Roberts (*Salford Royal NHS Foundation Trust, Salford, UK*) |
| 14:45–15:05 | Exosomes – a new way to transport large molecules? | Mark Tarnopolsky (*McMaster University, Hamilton, Ontario*, *Canada*) |
| 15:05–15:20 | Gene therapy – AAV/lentiviral | Federico Mingozzi (*University Pierre and Marie Curie, Paris, France*) |
| 15:20–15:35 | Stem cells what is possible today? | Francesco Saverio Tedesco (*University College London, London, UK*) |
| *15:35–15:50* | *Audience Q&A: Future perspectives in Pompe disease* |  |
| 15:50–16:00 | Close of meeting | Ans van der Ploeg and Benedikt Schoser |