

## Program LSD subnetwork meeting 3 November 2018

### Networking diner (Friday, 2 November 2018)

19:00 – 20:00 *Welcome & drinks*

20:00 – 23:00 *Diner*

### Program (Saturday, 3 November 2018):

08:30 – 09:00 *Registration & coffee*

09:00 – 09:05 Opening (Ans van der Ploeg)

09:05 – 10:45 Update on LSD subnetwork activities and next steps

09:05 – 09:20 WP1 (Ans van der Ploeg / Hidde Huidekoper)

09:20 – 09:35 WP2 (Mireia del Toro)

09:35 – 09:45 WP3

09:35 – 09:45 WP4 (Giancarlo Parenti)

09:45 – 10:00 WP5 (Andrea Dardis)

10:00 – 10:15 WP6 (Dominique Germain)

10:15 – 10:30 WP7 (Mireia del Toro)

10:30 – 10:45 *Coffee break*

10:45 – 11:00 WP8

11:00 – 11:15 WP9 (Christina Lampe)

11:15 – 12:30 Interaction with other professional networks and patient organizations

- ERNDIM (George Ruijter, chair ERNDIM)

- ESGLD (Pim Pijnappel, secretary ESGLD)

- EPOC (Ans van der Ploeg, co-chair EPOC)

- Brains for Brain (Maurizio Scarpa, president Brains for Brain)

- Patient organizations

12:30 – 13:30 *Lunch*

13:30 – 15:30 Proposals for collaborative projects to work on within the LSD subnetwork

13:30 – 13:50 Variants of unknown significance in Fabry disease (D. Germain)

13:50 – 14:10 Start and stop criteria for use of orphan drugs in LSDs (Maja di Rocco)

14:10 – 14:30 Patients experiences as a basis for mapping centres of expertise in LSDs (Hanka Dekker)

14:30 – 14:50 Identification of new biomarkers for Pompe's disease (G. Parenti)

14:50 – 15:10 Natural history of Mucopolidosis II/III (Esmee Oussoren)

15:10 – 15:30 Newborn screening for LSDs (Flavia Tubili)

15:30 – 16:00 *Coffee break*

16:00 – 17:00 Discussion on projects

17:00 – 17:15 Closing remarks (Ans van der Ploeg)



**European  
Reference  
Network**

for rare or low prevalence  
complex diseases

 **Network**  
Hereditary Metabolic  
Disorders (MetabERN)