

Qui di seguito gli interventi e gli orari a cui per il nostro fuso orario dobbiamo aggiungere 5 ore.



**Lafora Disease Zoom Meeting**  
**October 29, 2020, 11AM-4:30PM EST**  
**Zoom link:**

**Session I: Introduction, NIH Priorities, & Anti-sense oligonucleotides**

11:00 am, **Matthew Gentry**, PhD, U. of Kentucky College of Medicine: *Opening comments*

11:10 am, **Miriam Leenders**, PhD, NINDS/NIH Program Officer  
*The NINDS commitment to basic research and rare diseases*

11:30 am, **Y. Paul Goldberg**, MD/PhD, Ionis Pharmaceuticals, VP, Clinical Development  
*Progress to the Clinic: Antisense Oligonucleotide Suppression of Glycogen Synthase 1 to Treat Lafora Disease*

12:00 pm, **Berge A. Minassian**, MD, UT-Southwestern, Pediatrics, Neurology, and Neuroscience  
**Silvie Nitschke**, PhD, Minassian Group, UT-Southwestern  
*The winding pre-clinical road to an ASO therapy for Lafora disease*

**Session II: Clinical Insights, PME, Targeting Glycoqen Synthesis**

12:30 pm, **Alison Dolce**, MD, – Minassian Group, UT-Southwestern  
*Short talk: Lafora disease EEG in a pair of siblings, one affected, one yet not*

12:45 pm, **Subramaniam Ganesh**, PhD, Indian Institute of Technology  
*PME genes, neuroinflammation, and epilepsy*

1:15 pm, **Olga Varea**, PhD – Guinovart Group, IRB-Barcelona  
*Short talk: Suppression of glycogen synthesis as a treatment for LD*

1:30 pm, **Tom Hurley**, PhD, Indiana University School of Medicine  
*Small molecule suppression of glycogen storage: structural and mechanistic studies*

**Session III: Repurposing. Novel Mechanisms. Novel Models. Clinical Insights**

2:00 pm, **Pascual Sanz**, PhD, Institute of Biomedicine in Valencia-CSIC  
*Repurposing drugs: a disease-modifying strategy to ameliorate Lafora disease pathophysiology*

2:30 pm, **Joan Guinovart**, PhD, IRB-Barcelona: *Role of p62 in Lafora disease*

3:00 pm, **Jose M. Serratosa**, MD/PhD, Fundación Jimenez Diaz Hospital  
*Generation and characterization of the first LD patient mutation mouse model*

3:30 pm, **Maria Machio**, MD, – Fundación Jimenez Diaz Hospital, *Short talk: Defining early stage LD EEG*

3:45 pm, **Antonio Delgado-Escueta**, MD, UCLA Neurology: *The five stages of Lafora disease*

4:15 pm, **Matthew S. Gentry**, PhD, U. of Kentucky College of Medicine  
*Glycosylation defects in Lafora disease due to aberrant glucosamine metabolism*