7TH ANNUAL

LAFORA DISEASE SCIENCE SYMPOSIUM REPORT

September 26-27, 2022

Sanford Consortium, San Diego, California

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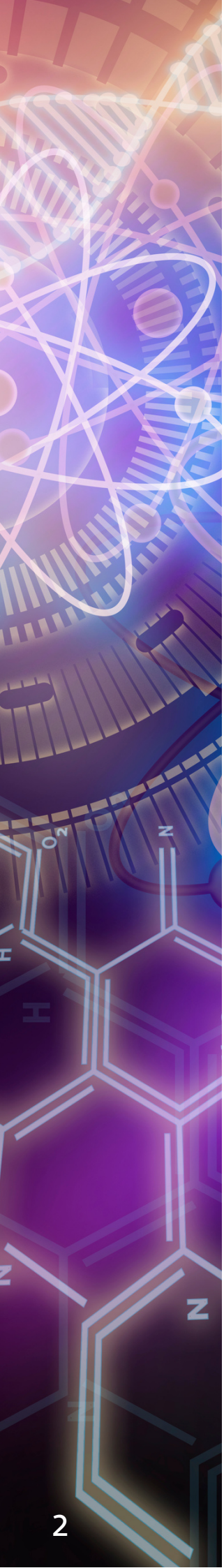
Chelsea's Hope
Lafora Children Research Fund



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Introduction

On Monday, September 26th, 2022, in San Diego, USA, the 7th Annual meeting gathered including researchers, clinicians, pharmaceutical representatives, and caregivers. There was excitement to be in person again to connect and share.

Day One

The afternoon began with **Frank Harris**, the president of **Chelsea's Hope Lafora Children Research Fund**, welcoming all the attendees and thanking them for their participation.

The group then watched a **patient spotlight video** from the Merriam family, located in Arizona, USA. It shows sister Mariah Merriam discussing having two siblings affected by Lafora disease and its impact on her. Mother Jenifer Merriam also talks about her family, the diagnosis, breakthroughs in research, and the challenges in finding treatments.

Next up was the **keynote address by Dr. Matthew Gentry**, University of Florida, who talked about **defining metabolic signatures of Lafora disease.**

The **Pharmaceutical Session** followed the Keynote address. Frank Harris introduced the speakers for this session:

- **Dr. Alok Tayi from Vibe Bio;**
- **Dr. Suyash Prasad from Taysha GTx;**
- **Dr. Karyn O'Neil from Aro Biotherapeutics;**
- **Dr. Michelle Boyd from Cerebral Therapeutics;**
- **Dr. Dustin Armstrong from Parasail.**

Honoring Dr. Peter Roach

The last presentation for the day was to **honor Dr. Peter Roach**, presented by Dr. Matthew Gentry. He discussed the surprises of genetic engineering: a possible model of polyglucosan disease continuing onto Dr. Roach's life's work (along with his wife Anna) throughout his career and the discoveries in biochemistry that contributed to understanding the glycogen metabolism for Lafora disease.

Dr. Peter Roach
(1948-2022)

Day Two

The next day, Tuesday, September 27th, 2022, started with the **Translational Science Research Session** chaired by Dr. Matthew Gentry. Here we heard from Dr. Craig Vander Kooi, University of Florida, on the **molecular basis for disfunction in Lafora Disease**, followed by Dr. Jose Serratosa, Universidad Autónoma de Madrid, who presented his **evaluation of new knock-in mouse models of Lafora Disease.**

Dr. Jordi Duran, Institute for Research in Biomedicine, spoke about using **Malin restoration as proof of concept for gene therapy for Lafora disease.** The last presenter of this session was Dr. Berge Minassian, University of Texas Southwestern, who shared his work on **AAV Gene Therapy for Lafora Disease and Adult Polyglucosan Body Disorder.**

The session chair for the **Basic Science Research Session** was Dr. Jordi Duran. The first to present was Dr. Marta Riba, Universitat de Barcelona, who discussed the transition of carbohydrate aggregates **from corpora amylacea to wasteosomes**, followed by Dr. Felix Nitschke, University of Texas Southwestern Medical Center, who discussed **cross-correction-enabled gene therapy for Polyglucosan Body Diseases.**

Dr. Thomas Hurley, Indiana University School of Medicine, presented tools for **understanding glycogen storage using chemical probes, proteomics and metabolomics** and Dr. Sharmistha Mitra, University of Texas Southwestern Medical Center, highlighted a function of the laforin-malin complex in her presentation about how **laforin targets malin to glycogen in a new FLAG-malin mouse model.** Finally, Dr. Pascual Sanz, Instituto de Biomedicina de Valencia, proposed a pathway for **understanding the role of malin in cell physiology.**

Dr. Marta Riba (left) and Dr. Feliz Nitschke (right)

The session chair for the **Patient Care & Advocacy Session** was Glenna Steele from Glut1DS. Cristol Barrett O'Loughlin from Angel Aid started the session with **Caring for the Caregivers: From brokenhearted to belonging using the Raregivers™ Emotional Journey Map**, an emotional presentation for many caregivers attending in person.

Next, Dr. Alison Dolce, University of Texas Southwestern Medical Center, shared her findings from a small survey on **anti-seizure medications for Lafora disease patients** followed by a session on **how to talk with your neurologist** with Dr. Jennifer Griffith, Washington University.

The last session of the day was the **Clinical Science Research Session** chaired by Dr. Berge Minassian. We heard from Dr. Lorenzo Muccioli, University of Bologna, about using **FDG-PET assessment as a possible biomarker of disease progression**. Dr. Nirbhay Yadav, Johns Hopkins University, then shared a novel method that allows for **non-invasive MRI imaging of glycogen in vivo**. Next, Dr. Antonio Delgato-Escueta, David Geffen School of Medicine at UCLA, presented information on **early clinical and EEG biomarkers**.

Dr. Roberto Michelucci, Istituto delle Scienze Neurologiche di Bologna, shared the **clinical, EEG and neuropsychological findings in a cohort of patients enrolled in the natural history study**, then Maria Macchio, Universidad Autónoma de Madrid, presented **on a registry-based prospective follow-up** to the natural history study. Dr. Viet Nguyen, Chapman University, gave the final presentation of the session detailing **clinical milestones in Lafora disease**.

Conclusion

We heard from a motivated community about new findings and advances toward treatments for Lafora disease. Researchers, clinicians, and pharmaceutical representatives from around the world shared their important insights. Family member involvement in the research, especially with the natural history studies, is valuable to the whole process.

Thank you to all our participants! We are grateful for your work to help reach a cure for Lafora disease.

THANK YOU TO OUR SPONSORS!

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