PROGRAMME

CDKL5 FORUM
BOSTON | 29-30 NOV 2017

WEDNESDAY 29TH – THURSDAY 30TH NOVEMBER 2017
Royal Sonesta Boston Hotel, Cambridge MA

Tuesday 28th November:

06:00 - 08:00pm	**Informal reception and early registration** Royal Sonesta Boston Hotel: Forum registration, reception

Wednesday 29th November:
Morning session

08:30 - 09:00am	**Registration, coffee**
09:00 - 09:15am	**Welcome** Majid and Lynn Jafar, Co-Founders, Loulou Foundation
09:15 - 10:30am	**Session 1: CDKL5 Function** Chair: Dario Alessi, University of Dundee **Speakers:** • *Identification and validation of the first physiological substrates of the CDKL5 kinase mutated in CDKL5 disorder* Ivan Munoz, University of Dundee • *CDKL5, its substrates and neuronal microtubules* Sila Ultanir, Francis Crick Institute • *Presynaptic dysfunction in CDKL5 deficiency* Michael Cousin, University of Edinburgh • *Identification of novel CDKL5 complex partners and kinase substrate candidates* Vera Kalscheuer, Max Planck Institute, Berlin
10:30 - 11:00am	**Break**
11:00 - 12:00pm	**Keynote session: Genome Editing** Chair: James Wilson, University of Pennsylvania 11:00 - 11:20am *Expanding the Genome Editing Toolbox* Feng Zhang, Broad Institute/ MIT 11:20 - 11:40am *Base Editing: Genome Editing Without Double-Stranded DNA Cleavage* David Liu, Broad Institute/Harvard University 11:40 - 12:00pm *Discussion moderated by James Wilson on the therapeutic use of genome editing*
12:00 - 01:15pm	**Session 2: Model systems and phenotyping** Chair: Peter Kind, University of Edinburgh

Speakers:

- *Improving the identification of reproducible and reliable phenotypic endpoints: early findings from a natural history study of a mouse model of CDKL5 disorder*
  Rodney Samaco, Baylor College of Medicine
- *Callosal refinement in CDKL5 disorders*
  Michela Fagiolini, Boston Children's Hospital
- *Modeling CDKL5 syndrome with brain organoids*
  Alysson Muotri, UCSD

Afternoon session

01:15 - 02:30pm — **Lunch and posters**

02:30 – 03:45pm — **Session 3: Clinical trials and clinical endpoints**
Chair: Orrin Devinsky, NYU
Speakers:

- *Read-through and antiepileptic drug trials in CDKL5*
  Orrin Devinsky, NYU
- *Clinical experience of CDKL5 Deficiency Syndrome in the IFCR COEs: Towards development of a Severity Scale*
  Tim Benke, UC-Denver
- *Ganaxolone, a novel investigational treatment for children with CDKL5 epilepsy: Results from an open label phase 2 study*
  Lorianne Masuoka, Marinus Pharmaceuticals
- *Optimising therapeutic development in rare epilepsies: lessons for CDKL5*
  Helen Cross, UCL/Great Ormond St Hospital

03:45 - 04:15pm — **Break**

04:15 - 05:30pm — **Breakout discussions**
Breakout topics:

A. *CDKL5 Models: what is the relevance of animal and cell models and phenotypes to human disease?*
Moderator: Michael Green, UMass Medical School

B. *Bioinformatics and drug screening/repurposing*
Moderator: Isaac Kohane, Harvard Medical School

C. *Imaging and EEG as tools for probing CDKL5 function and disease natural history*
Moderator: Helen Cross, Great Ormond St Hospital

D. *Biomarker discovery and validation in the periphery and CNS*
Moderator: Laura Mamounas, NINDS/NIH

E. *Improving clinical outcome measures for CDKL5 deficiency disorder*
Moderator: Tim Benke, University of Colorado-Denver

Evening session

07:30pm

Forum dinner at Museum of Science, Boston
Dinner remarks: Eric Lander, President and Founding Director, Broad Institute
Awards Ceremony and Announcements

Thursday 30th November: Morning session

09:00 - 09:30am

The Patient Perspective:
- *Believing in the Promise of Today and Hope for Tomorrow*
  Katheryn Elibri Frame, DO

09:30 - 10:45am

Session 4: Novel therapies
Chair: Yael Weiss, Ultragenyx
Speakers:
- *X chromosome reactivation strategies*
  Jeannie Lee, Massachusetts General Hospital
- *Developing New Therapeutics for CDKL5: Lessons Learned from Lysosomal Storage Disorders*
  Hung Do, Amicus Therapeutics
- *Evidence of Excitatory: Inhibitory Imbalance in CDKL5 Disorder*
  Frances Jensen, Perlman School of Medicine, University of Pennsylvania
- *Strategies for delivering gene therapy to the brain*
  Jan Nolta, UC-Davis

10:45 - 11:15am

Break

11:15 - 12:00pm

Keynote presentation: Gene Therapy
11:15 - 11:45am *Viral-mediated gene therapy in the central nervous system*
James Wilson, University of Pennsylvania
11:45 - 12:00pm *Q & A for Prof Wilson*

12:00 - 12:45pm

Moderated Discussion on Therapeutic Strategies

12:45 - 02:00pm

Lunch & posters

Afternoon session

02:00 - 03:15pm

Session 5: Industry Panel
Moderator: Majid Jafar, Co-Founder, Loulou Foundation
Panelists:
- Omar Khwaja, Global Head Rare Diseases, Roche
- Jeremy Levin, Chairman and CEO, Ovid Therapeutics
- Andrew Plump, Chief Medical and Scientific Officer, Takeda
- Philip Reilly, Partner, Third Rock Ventures

03:15 - 04:00pm	**Review of breakout sessions, Part I, from breakout discussion leaders**
04:00 – 04:15pm	**Break**
04:15 – 05.00pm	**Review of breakout sessions, Part II, from breakout discussion leaders**
05:00 - 05:30pm	**Conclusions and closing comments**