CDKL5 DEFICIENCY DISORDER

PATIENT-FOCUSED DRUG DEVELOPMENT MEETING

November 1, 2019

IFCR
International Foundation for CDKL5 Research

loulou FOUNDATION
Tackling CDKL5 Deficiency

1:30–5:00 pm
College Park Marriott Hotel &
Conference Center
Hyattsville, Maryland
www.cdkl5.com/pfdd

WHY IS THE CDD VOICE SO IMPORTANT

Dear CDKL5 Deficiency Disorder community member,

We would like to welcome you to the *Externally-Led Patient Focused Drug Development* (PFDD) Meeting for CDD. Disease-focused PFDD meetings give the Food and Drug Administration (FDA) and other important stakeholders such as industry professionals the opportunity to hear directly from you about what matters the most to families living with CDD.

We are honored to host this meeting where we will hear about the symptoms of CDD that matter the most to you, the impacts that the disease has on your entire family, and your experience with currently available medications. We also want to hear about your hopes for future treatments, since this input can inform the FDA and drug developers about the therapies that are needed.

Even if you could not join us today, we would love to hear about your experience so that the professionals at the FDA, and those developing therapies for CDD, can still hear your voice. You can do this by completing the online survey that will remain open until the end of November (www.cdkl5.com/pfdd). The results from the survey will be combined with the testimonies and discussions during the PFDD meeting into the *Voice of the Patient* report that we will release after the meeting.

We would like to thank everyone who has helped put together this meeting, in particular all the parents and grandparents who applied to participate as panelists. Whether you are here to speak and join the discussions, or simply completing the survey on line, what you have to say is very important for the entire community.

On behalf of the the thousands of families around the world living with CDD, thank you for sharing the CDD voice and making a difference.

Sincerely,

The IFCR & Loulou Foundation teams

ORGANIZING TEAM

Ana Mingorance, PhD
Chief Development Officer
Loulou Foundation

Daniel Lavery, PhD
Chief Scientific Officer
Loulou Foundation

Karen Utley
President & co-Founder
IFCR

Amanda Jaksha
Treasurer
IFCR

James Valentine, JD, MHS
Associate
Hyman, Phelps & McNamara

Larry Bauer, MA, RN
Senior Regulatory Drug Expert
Hyman, Phelps & McNamara

CDD PFDD MEETING | AGENDA | NOVEMBER 1, 2019

1:30 – 1:40pm **Welcome and overview**
Karen Utley, President IFCR

1:40 – 1:50pm **Opening remarks**
Dr Michelle Campbell, Division of Neurology Products, Office of New Drugs, CDER, FDA

1:50 – 2:05pm **Background on CDKL5 deficiency Disorder:**
Dr Tim Benke, Children's Hospital Colorado
Dr Orrin Devinsky, NYU Langone Comprehensive Epilepsy Center

2:05 – 2:15pm **Goals and objectives for the meeting, overview of discussion format**
James Valentine, Moderator

2:15- 3:25pm **Session 1: CDD patient voice: Symptoms and daily impacts**
- Panel: Diana Dinescu, Jenny Feinman, Marissa Bishop, Kathy DeSimone, Rick Upp
- Live Polling
- Moderated discussion among all meeting participants

3:25 – 3:35pm **Break**

3:35 – 4:45pm **Session 2: CDD patient voice: Current and future approaches to treatments**
- Panel: Kristin Kozera, Martha Rodgers Boyles, Rita Fredericks, Amanda Jaksha, Ed Fennell
- Live Polling
- Moderated discussion among all meeting participants

4:45 – 4:55 pm **Closing remarks**
Larry Bauer, Hyman, Phelps & McNamara

4:55 – 5:00 pm **Next steps**
Majid Jafar, Founder Loulou Foundation

The meeting will be live-streamed and remain available after November 1 at www.cdkl5.com/pfdd

SPEAKERS

Karen Utley, International Foundation for CDKL5 Research (IFCR)

Karen is mom to Samantha (13), with CDD, and after co-founding and serving as treasurer of IFCR for 8 years, she became the President in 2017. She also serves in leadership roles for the ELC (Epilepsy Leadership Council) and REN (Rare Epilepsy Network), and is active with rare disease organizations such as NORD, Global Genes, and EveryLife Foundation. As a strong believer that a cure is possible, Karen has begun to pursue a bachelor degree in nursing with the goal of being involved in clinical research for developmental epileptic encephalopathies.

Michelle Campbell, PhD, Division of Neurology Products, Office of New Drugs, CDER, FDA

Michelle Campbell is the Sr. Clinical Analyst for Stakeholder Engagement and Clinical Outcomes for the Division of Neurology Products, Office of New Drugs (OND), Center for Drug Evaluation and Research (CDER), U.S. Food and Drug Administration (FDA). Previously, Dr. Campbell was a reviewer on the Clinical Outcome Assessments (COA) Staff and Scientific Coordinator of the COA Qualification Program in OND. Dr. Campbell's focus is in patient-focused drug development and the use of patient experience data in the regulatory setting.

Tim Benke, MD, PhD, University of Colorado/Children's Hospital Colorado

Tim Benke, MD, PhD is the Ponzio Family Endowed Chair in Neurology Research at University of Colorado/Children's Hospital Colorado. Dr. Benke received his MS in electrical engineering at Rice University and his MD, PhD at Baylor college of Medicine. He completed his clinical training in pediatrics and pediatric neurology at Texas Children's Hospital, and did a post-doctoral fellowship at the MRC Centre for Synaptic Plasticity/University of Bristol in the UK. After he joined the faculty at the University of Colorado School of Medicine, his lab has focused on the molecular mechanisms that are impacted by early-life seizures that go on to cause intellectual disabilities and autism. Dr Benke leads the Colorado CDKL5 Center of Excellence.

SPEAKERS

Orrin Devinsky, MD, NYU School of Medicine

Dr Orrin Devinsky is Professor of Neurology, Neurosurgery, and Psychiatry at the NYU School of Medicine. He received his B.S. and M.S. from Yale University, M.D. from Harvard Medical School and interned at Boston's Beth Israel Hospital. He completed neurology training at the New York Hospital-Cornell Medical Center and his epilepsy fellowship at the NIH. He is the Principal Investigator for the North American SUDEP Registry and for the SUDC Registry and Research Collaborative. He founded Finding A Cure for Epilepsy and Seizures (FACES) and co-founded the Epilepsy Therapy Project and epilepsy.com. Dr Devinsky leads the NYU CDKL5 Center of Excellence.

James Valentine, JD, MHS

James Valentine assists medical product industry and patient advocacy organization clients in a wide range of new drug and biologic development and approval matters. James has been central to the transition of the FDA Patient-Focused Drug Development (PFDD) program to externally-led meetings, having helped plan and moderated two-thirds of those. Before joining his current firm in 2014, James worked at FDA as a patient liaison where he facilitated patient input in benefit-risk decision-making and regulatory policy issues. There, James administered the FDA Patient Representative Program, helped launch the PFDD program, and developed the FDA Patient Network. He holds a law degree from University of Maryland and a masters from Johns Hopkins School of Public Health.

Larry Bauer, MA, RN

Larry Bauer is a Sr. Regulatory Drug Expert with Hyman, Phelps, & McNamara, P.C. and assists medical product industry and patient advocacy organization clients in a wide range of regulatory matters, including new drug and biologic development and approval issues. Prior to this position he worked at the FDA in CDER's Rare Diseases Program working on policy, education, and science related to rare disease drug development. He has expertise in Rare Pediatric Disease priority review vouchers and designations, expedited programs, and patient engagement including extensive experience guiding patient advocacy groups. He also serves on the National Organization for Rare Disorders (NORD) Advocacy Committee.

SPEAKERS

Majid Jafar, Loulou Foundation
Majid Jafar is the father of Loulou (5), with CDD, and the Founder of the Loulou Foundation. Majid works in the oil and gas business as the CEO of Crescent Petroleum which operates in the Middle East, having previously worked for Shell International in Europe. Majid is also a supporter of initiatives in the field of education and tackling youth unemployment and sits on various non-profit boards including the Board of Fellows of Harvard Medical School, and is a member of the Academy of the University of Pennsylvania, the Advisory Board of Cambridge University Children's Research Hospital Project, and the Global Precision Medicine Council of the World Economic Forum. He holds engineering degrees from Cambridge University in England and an MBA from Harvard Business School.

PANELISTS Session 1: CDD patient voice: Symptoms and daily impacts

Diana Dinescu Strother, Front Royal, VA
Diana lives in Front Royal with her husband, Bryan, and their 17 month-old daughter, Lina. Lina started having seizures before she was 5 weeks old, and was diagnosed with CDKL5 at 3.5 months of age. Lina is a kind, warm, and gentle soul, and inspires love in everyone who meets her. She loves lights, music, and cuddles. Diana is a clinical psychologist working 2 days per week, and dedicates the rest of her time to making sure that Lina receives every opportunity to thrive.

Jenny Feinman, Greenwood, IN
Jenny lives in a suburb of Indianapolis with her husband Steve, and children Nick (14), Hannah (12) and Allie (7). Allie was diagnosed with CDD at 6 months of age and loves swimming, lights and swinging. Jenny is the Human Resources - Office Manager for Decamp Enterprises as well as the Director of the Noah Kriese Foundation.

PANELISTS Session 1:
CDD patient voice: Symptoms and daily impacts

Marissa Bishop, CT
Marissa lives in Connecticut with her husband, Jeff, and son, Gregory (3). Gregory was diagnosed at six months old. He is a sweet boy who loves listening to stories and music. Marissa is a former school social worker, but is now full-time caregiver to Gregory. She and Gregory fundraise for CDKL5 research through their art project “Art For Hope Love Cure.” Marissa also designs products for the IFCR Zazzle store.

Kathy DeSimone, Nesconset, NY
Kathy lives in Nesconset on Long Island, NY with her husband Gary and children Ryan (12) and Katelynn (10). Katelynn was diagnosed with CDD at 26 months of age. Katelynn loves the breeze in her face, cuddling, and when her brother plays the piano. Kathy works as a Payroll Tax Accountant.

Rick Upp, Spokane, WA
Rick and his wife Cynthia have raised 5 children, including Olivia (17), who was diagnosed with CDD at the age of 11, and Emily (29), who is Olivia's state-licensed caregiver. Olivia's passion is babies and she has her own subscription to People magazine, just for the baby pictures. Rick is vice president of the IFCR and past chair of the CDKL5 Alliance.

PANELISTS Session 2: CDD patient voice:
Current and future approaches to treatments

Kristin Kozera, Philadelphia, PA
Kristin lives outside of Philadelphia with her husband, Brian, and their 3 daughters Paige (10), Josie (7) and Avery (5). Avery was diagnosed with CDD at 4 months of age and loves swimming, biking with her daddy, running in her jogger, and playing with her sisters. Kristin works as a QA specialist in the pharmaceutical field and has been a member of the board of IFCR for 1 year.

Martha Rodgers Boyles Lexington KY
Martha lives near Lexington KY with her husband Paul and daughter Elsah (13). Elsah was diagnosed with CDD at 5 years old and she loves music, swimming in warm Gulf waters, and giving cuddles. Martha is an equine veterinarian in private practice with a special interest in equine locomotor pathology and rehabilitation.

PANELISTS Session 2: CDD patient voice: Current and future approaches to treatments

Rita Fredericks, Grimes, IA
Rita lives outside of Des Moines the Capitol of Iowa with her husband Nathan, a public school teacher and their two sons Liam (9) and Miles (4). Miles was diagnosed with a CDKL5 mutation at 4 months of age. He loves to swim & swing and look at lights and feel vibrations. Rita works with families across Iowa by providing support and resources to ensure they have their basic, medical and educational needs met. The Fredericks Family is grateful to be a part of the CDD family and to be here to provide valuable information and a real life perspective.

Amanda Jaksha, Denver, CO
Amanda lives in the greater Denver metro area with her husband Joe and children Ava (14) and Aliza (10). In 2012 at the age of 6.5, Ava was diagnosed with CDKL5 Deficiency Disorder. She enjoys riding horses, music therapy, family time, her school mates, and fabulous food. Amanda is a managerial accountant and has served on the Board of Directors for the International Foundation for CDKL5 Research for the past 5-years.

Edward Fennell, Schodack, NY
Ed lives near Albany NY with his wife Lynne. He is grandfather and caregiver to his CDD grandchild, Haley. Haley is 12 yrs old and lives with her mother, father and younger sister Emily (9). Haley was diagnosed at 2 years of age. She loves school, music, shopping and travel. Ed is retired from his government finance consulting firm.

CORPORATE SPONSORS

The Loulou Foundation and IFCR thank the following corporate sponsors for their generous support towards covering CDD families travel and accommodation to participate in the meeting:

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MEETING PARTNERS
International Foundation for CDKL5 Research

IFCR
International Foundation for CDKL5 Research

The The International Foundation for CDKL5 Research began as a group of parents whose children had CDKL5 Deficiency Disorder. We came together and dared to dream of **a new future for our children**. With education and research, we believe a life-changing cure will be found.

In 2009, we incorporated the International Foundation for CDKL5 Research as a non-profit organization. In 10 years, the IFCR has grown from being a group of parents of children with CDKL5 Deficiency Disorder to leading the world in developing **CDKL5 Centers of Excellence** and advocating for the CDD community.

In 2010 IFCR started building a **tool kit for researchers** by funding animal and cell model projects. A collaborative effort in 2012 launched an international database that has been instrumental to our current understanding of CDD symptoms. The IFCR has eight CDKL5 Centers of Excellence in the United States. We are the driving force that establishes and maintains these Centers of Excellence. Our Centers also provide guidance on CDD to physicians and health systems internationally.

We are the leader in CDD patient education in the English-speaking world. Our **resource library** provides families living with CDD with the information and resources they need to help their affected loved ones. We also sponsor the **biennial CDKL5 Family Conference**, where affected families can come to learn about CDD and support one another.

OUR MISSION

To treat and cure CDKL5 Deficiency Disorder by funding scientific research, while helping affected individuals and their families to thrive.

CONTACT US

info@cdkl5.com
www.cdkl5.com
Twitter: @CDKL5USA
facebook.com/CDKL5foundation/

Our community partners include the Epilepsy Leadership Council (ELC), Rare and Catastrophic Seizure Collaborative (RCSC), Rare Epilepsy Network (REN), The Haystack Project, Alliance for Genetic Etiologies of Neurodevelopmental Disorders and Autism collaboration (AGENDA), the Consortium for Outcome Measures & Biomarkers for Neurodevelopmental Disorders (COMBINEDbrain), NORD, and Global Genes.

MEETING PARTNERS
Loulou Foundation

The **Loulou Foundation** is a private non-profit UK foundation founded in 2015 by the parents of an affected child.

Dedicated to advancing research into the understanding and development of therapeutics for CDKL5 Deficiency Disorder.

OUR MISSION

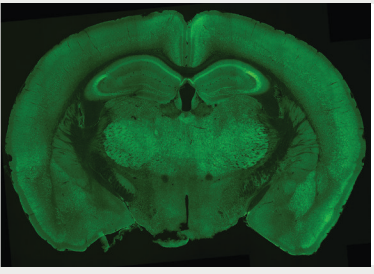
To de-risk the disorder to enable industry interest and partner with all stakeholders in order to realize treatments for CDKL5 Deficiency Disorder by 2020 and cures by 2025.

BREAKING DOWN BARRIERS

- Annual **CDKL5 Forum** research congress (London and Boston).
- **www.cdkl5forum.org** on-line portal for research collaborations.
- Working closely with the CDKL5 Deficiency **International Patient Alliance.**
- Working with biotech and pharmaceutical companies as well as regulators to **de-risk clinical trials** and attract interest.

BUILDING THE KNOWLEDGE AND RESEARCH TOOLBOX

Open-access mouse models. Patient-derived iPSCs. Brain atlas. Substrate identification. Biomarkers. Clinical endpoints. Trial site identification. Protocol design support.



FUNDING RESEARCH AND NOVEL THERAPIES

CDKL5 Program of Excellence through strategic partnership with Penn Medicine Orphan Disease Center since 2016 with annual Pilot Grant program and direct funding.

Over 40 labs and 120 scientists funded.

loulou FOUNDATION

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