UNLOCK CTX

CEREBROTENDINOUS XANTHOMATOSIS

An initiative of the United Leukodystrophy Foundation

Patient-Focused Drug Development Meeting on Cerebrotendinous Xanthomatosis (CTX)

September 14, 2021

11:00 a.m. – 3:00 p.m.

Eastern U.S. Daylight Time

Streamed live at:
https://ctxresource.org/unlockctx/

Social Media Hashtags:
#unlockCTX #leukodystrophy
#RareDisease #PFDD

Organized & Hosted By:

united LEUKODYSTROPHY FOUNDATION

TODAY'S AGENDA *(all times are Eastern U.S. Daylight Time)*

11:00 – 11:05 am **Welcome Remarks**
Robert Rauner, *President, United Leukodystrophy Foundation*

11:05 – 11:15 am **About this Meeting: Review of Meeting Agenda and Desired Outcomes**
Kim McCleary, *Moderator* and Neel Odedara, *Co-Moderator*

11:15 – 11:30 am **Overview of the Diagnosis, Clinical Features, and Treatment of CTX**
P. Barton Duell, M.D., *Professor of Medicine, Oregon Health and Science University*

Session 1: Diagnostic Odyssey, Disease Symptoms, and Daily Impacts of CTX

11:30 – 11:55 am **Panelist Remarks: Living with CTX**
John Wolf, Sangna Odedara, Debra and Tom Johnson, and Sue Stewart

11:55 – 1:00 pm **Facilitated Discussion**
Each part of the discussion will start with a community perspective
Narinder Ghara, Catherine Zonsky, Shannon DeLaMar, Kay Fischer, and Austin Larson
Other community members are invited to call in live with comments: 1.703.844.3231

Key Questions:

- What were some of the challenges of getting appropriate attention in the search for a diagnosis?
- Of all the symptoms experienced, which 1 to 3 symptoms are the greatest burdens?
- What are the biggest concerns or worries related to CTX – both today and for the future?
- What are the greatest impacts on daily living for both the person with CTX and the family?

1:00 – 1:20 pm **Break**

Session 2: Current and Future Treatments for CTX

1:20 – 1:40 pm **Panelist Remarks: CTX Treatment Experiences**
Bobbi Blanchard, Robin Huard, and Shawn Laurie

1:40 – 2:35 pm **Facilitated Discussion**
Each part of the discussion will start with a community perspective
George Bryce, Avril Dimond, Holly Rosengrant, Estefania Amyerich, and Bob Steiner
Other community members are invited to call in live with comments: 1.703.844.3231

Key Questions:

- What therapies are being used to help treat CTX, relieve symptoms, or improve quality of life?
- Which 1-2 aspects of the condition are most important to relieve/control/reverse?
- What issues have affected access to therapy and what effects are seen when primary therapy with CDCA is interrupted?
- What has your experience been with regard to clinical trials or experimental therapies?
- What are the unmet treatment needs? What would you like to see in future treatments?

2:35 – 2:50 pm **Remarks from FDA: Value of PFDD in CTX and Themes Heard from the CTX Community**
Vijay Kumar, M.D., *Office of Tissues and Advanced Therapies, CBER, FDA*

2:50 – 3:00 pm **Final Closing Remarks**
Robert Rauner and Kim McCleary

Please submit written comments using the comment form at **https://ctxresource.org/unlockctx/**.
This comment form will remain open until October 14, 2021.

Cerebrotendinous Xanthomatosis (CTX)

Cerebrotendinous Xanthomatosis (CTX) is an ultra-rare, genetic disease that affects the body's ability to metabolize fats known as cholesterols. Individuals with CTX experience a vast range of symptoms that progress across the lifespan, with increasing severity and impact on daily functioning, health, and well-being. Symptoms may begin in infancy, yet the disease is often not identified until neurological symptoms become more pronounced. Despite numerous means of detecting and diagnosing CTX, the average age of diagnosis is 35. This long delay in diagnosis and lost opportunity for earlier treatment can have tragic repercussions. While there is no cure for CTX, if it is identified and treated early, progression can be slowed or stopped.

Patient-Focused Drug Development & the Unlock CTX Meeting

In 2013, the U.S. Food and Drug Administration (FDA) began holding a series of meetings to hear directly from patients, caregivers, advocates, and other key community stakeholders about what it's like to live with various medical conditions and how well available treatments are meeting patients' needs. This meeting series, called the "Patient-Focused Drug Development" initiative or PFDD, has now been expanded to enable patient organizations to host PFDD meetings in collaboration with FDA. (See http://bit.ly/FDA-PFDDinfo)

In February 2021, the United Leukodystrophy Foundation (ULF) applied to host an externally led PFDD meeting on CTX. The FDA approved the proposal in March 2021, encouraging participation beyond the United States and the inclusion of individuals and families living in other countries.

The purpose of PFDD meetings in general and the Unlock CTX meeting is for FDA staff, researchers, and developers of drugs, gene therapies, and medical devices to understand more fully the burdens of living with a particular medical condition, as well as what patients most value in terms of treatment benefits and the risks and side effects they are willing to tolerate. An example of how PFDD can benefit people living with CTX is to deepen understanding about all the ways in which CTX impacts people's lives and the unmet medical needs that persist in spite of treatment with available medications.

Your active participation during this meeting will help depict the full range of experiences with CTX and will allow these key audiences to help unlock CTX.

PANELISTS AND DISCUSSION STARTERS

The purpose of today's meeting is to better understand the experience of individuals living with CTX. Panelists and "discussion starters" have dedicated additional time to prepare to speak to the challenges, expectations, and hopes of the community as a way of inviting an even broader range of experiences through the moderated discussion. We deeply appreciate their courage and dedication to the success of this meeting.

Panelists *(in program order)*

John Wolf, speaking on behalf of Ashley (OR)

Sangna Odedara (VA)

Debra and Thomas Johnson (NC)

Sue Stewart, speaking on behalf of Eric (OR)

Bobbi Blanchard, speaking on behalf of Jordan and Joelle (WI)

Robin Huard, speaking on behalf of Duane (WA)

Shawn Laurie, speaking on behalf of James (TX)

Discussion Starters *(in program order)*

Session 1:

Narinder Ghara (NY)

Catherine Zonsky (NY)

Shannon DeLaMar (IL)

Kay Fischer (Norway)

Austin Larson, M.D. (CO)

Session 2:

George Bryce (NC)

Avril Dimond (England)

Holly Rosengrant (IL)

Estefania Amyerich (Spain)

Bob Steiner, M.D. (WI)

SPEAKERS *(in program order)*

Robert Rauner
United Leukodystrophy Foundation

Robert Rauner is the parent of a son, Kevin, who passed away from adrenoleukodystrophy (ALD). His son Paul has adrenomyeloneuropathy (AMN).

Mr. Rauner has been involved with the ULF since 1994 and has been a Board Member since 2000. He has served as Treasurer of the ULF and now serves as President. He is a retired UPS truck driver and is now devoting his retirement to the future of the ULF. His hope for the future is to help researchers find treatments and cures for the leukodystrophies, as well as to support affected families. His hobbies include golf and fishing.

K. Kimberly McCleary
Kith Collective

Kim McCleary has been at the forefront of patient engagement for 30 years. She is a nationally regarded subject matter expert on patient-focused medical product development, patient-centered benefit-risk assessment, and organizational change. Ms. McCleary has authored scores of publications, served on dozens of steering committees and advisory boards, is a sought-after speaker, and has an extensive network of leadership contacts. Her passion for this work is rooted in personal experience, lived and as a family caregiver.

Ms. McCleary founded the Kith Collective in 2018 to speed adoption of patient-centricity by life science companies, not-for-profit organizations, and academic research teams. She has been involved in numerous patient-focused drug development meetings, including sessions led by the FDA and by patient advocacy organizations.

Ms. McCleary lives in Arlington, Virginia and is a graduate of the University of North Carolina.

Neel Odedara
CTX Alliance

Neel Odedara currently serves as a Board Member and Secretary for the CTX Alliance. Neel is an Associate Principal with Trinity Life Sciences and has over 10 years of experience advising small to large global biopharmaceutical clients across a wide range of therapeutic areas including rare/orphan diseases, neurology, and cardiometabolic conditions. Neel's experience spans multiple functions within the life sciences including commercial assessment, new product planning, brand/commercial strategy, launch, access, and patient engagement. Neel graduated from Brown University in 2009 with a B.A. in International Relations.

Neel's journey with CTX began in 2018 when his older sister Sangna was diagnosed at the age of 34 following genetic testing at National Institutes of Health. This followed decades of missed symptoms and misdiagnoses across various physicians and health institutions. For Neel and his family, this experience has ignited a commitment to supporting awareness, advocacy, and engagement on behalf of CTX patients.

P. Barton Duell, M.D.
Knight Cardiovascular Institute and Division of Endocrinology, Diabetes and Clinical Nutrition
Oregon Health & Science University

Dr. Barton Duell has cared for patients with CTX for more than 25 years. Dr. Duell is currently a Professor of Medicine in the Center for Preventative Cardiology at the Oregon Health and Science University. Additionally, he serves as the director of the Lipid-Atherosclerosis Laboratory and Sterols Analysis Laboratory. He is also the director of the LDL Apheresis Unit.

He has served as a clinician. attending physician, and researcher at the Oregon Health and Science University for over 29 years. His other roles include associate editor of the Journal of Clinical Lipidology, editor of the American Journal of Preventive Cardiology, chair of the Clinical Lipidology Committee for the American Heart Association, member of the Board of Directors for the National Lipid Association and American Board of clinical Lipidology, on the publications committee for the FH Foundation, among others.

Dr. Duell attended medical school at the Oregon Health and Science University and did his internship and residency in internal medicine at the University of Chicago and the Oregon Health and Science University. He did a fellowship in Metabolism, Endocrinology, and Nutrition at the University of Washington. He currently resides in Portland, Oregon.

Vijay Kumar, M.D.
Center for Biologics Evaluation and Research
US Food and Drug Administration

Dr. Vijay Kumar is a nephrologist and currently serves as a Medical Officer in the Office of Tissue and Advanced Therapeutics (OTAT) at Center for Biologics Evaluation and Research (CBER) within FDA. OTAT regulates new cell, tissue and gene therapies, many of which are targeted to the treatment of rare diseases.

Prior to joining the agency, Dr. Kumar was engaged in the practice in nephrology for over two decades. He completed his medical degree in India. Subsequently, he completed his internal medicine residency at Wayne State University in Michigan and his nephrology fellowship at University of Texas -Southwestern School of Medicine. He is board certified in nephrology and maintains an active physician license in the state of Maryland.

ACKNOWLEDGEMENTS

With gratitude for the following individuals who have enhanced the Unlock CTX PFDD Meeting:

Keely Mata and Christopher Rice of the United Leukodystrophy Foundation for support with outreach, planning, and communications for today's meeting.

William Lewallen of the FDA's Office of Strategic Initiatives, Center for Drug Evaluation and Research for providing counsel and encouragement throughout the entire planning process.

Samantha Mayberry of the Kith Collective for support with content development, planning, preparation, and communications for today's meeting.

Jean Pickford of Pickpoint LLC for assistance connecting with founding members of the CTX Alliance and additional meeting support.

Julie Rathjens of Hello Brand for design of the Unlock CTX logo and production of other creative materials, including this meeting program.

John Dudley, Eric Quigley, and Kyle Rhoderick of Dudley Digital Works for creative and technical services to broadcast today's meeting live and preserve it for later on-demand viewing.

Corporate Sponsors

Thank you to these generous sponsors for their unrestricted educational grants in support of the Unlock CTX meeting:

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THE UNITED LEUKODYSTROPHY FOUNDATION

Leukodystrophies comprise a group of genetic nervous system disorders affecting the myelin sheath, which insulates the axon through which nerve impulses are conducted.

OUR MISSION is to provide support to the leukodystrophy community and enable platforms to accelerate improving patient quality of life and finding cures.

OUR VISION is to meet the needs of patients and families, whether newly diagnosed or living with a leukodystrophy. Through unified collaboration with advocacy groups, medical and research professionals, and pharma/biotech companies, we provide support, networking, and education to help navigate the journey of the disease.

About

Located in DeKalb, Illinois, the United Leukodystrophy Foundation (ULF), is a nonprofit, voluntary health organization dedicated to providing patients and their families with information about their disease and assistance in identifying sources of medical care, social services, and genetic counseling. The ULF works to establish a communication network among families; as well as increases public awareness and acts as an information source for health care providers by promoting and supporting research into causes, treatments, and prevention of the leukodystrophies. Incorporated in 1982, the United Leukodystrophy Foundation is solely supported and funded by donations.

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