Pompe Disease Patient-Focused Drug Development Virtual Meeting

July 13, 2020

In Partnership with:
Welcome!

Dear Pompe disease PFDD Participants,

On behalf of the Muscular Dystrophy Association (MDA), welcome to this virtual Pompe disease Patient-Focused Drug Development (PFDD) meeting. Thank you for joining us today.

PFDD meetings provide key stakeholders, including FDA, patient advocates, researchers, drug developers, healthcare providers, and others, with an opportunity to hear your voice and in order to better incorporate the patient voice into the drug development and evaluation process. We will be focusing on your perspectives on two topic areas: (1) the most significant symptoms of Pompe disease and its impact on your daily life; and, (2) current and future approaches to treatment.

We would like to thank the panelists, who are presenting at this event to share their stories, struggles, hopes and concerns. We are also grateful to everyone from the Pompe disease community who is joining us online. As people living with Pompe disease or caring for someone with Pompe disease, you are uniquely positioned to inform the understanding of the therapeutic context for drug development and evaluation.

We also thank the U.S. Food and Drug Administration, industry professionals, clinicians, and researchers for their collaboration in paving the path for scientific breakthroughs and potential therapies.

MDA is delighted to facilitate this PFDD meeting for Pompe disease. We look forward to an informative and insightful event.

Sincerely,

Lynn O'Connor Vos
President and CEO
Muscular Dystrophy Association
# Agenda

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| 12:00 - 12:05 p.m. | Welcome and Opening Remarks  
Paul Melmeyer, Director of Regulatory Affairs, MDA |
| 12:05 – 12:15 p.m. | Scientific and Clinical Overview of Pompe Disease  
Dr. Priya Kishnani, M.D., Chief, Division of Medical Genetics, Duke University |
| 12:15 - 12:20 p.m. | Regulatory Approach to Pompe Disease Therapies  
Dr. Kathleen Donohue, M.D., Medical Officer, Division of Gastroenterology and Inborn Errors Products (DGIEP), CDER, FDA |
| 12:20 - 12:25 p.m. | Introduction & Meeting Overview  
James Valentine, J.D., M.H.S., Meeting Moderator |

**First Session: Pompe Disease Patient and Caregivers Perspectives on Symptoms and Daily Impacts**

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<td>12:25 - 12:30 p.m.</td>
<td>Audience &amp; Remote Demographic Polling</td>
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<td>12:30 - 1:30 p.m.</td>
<td>Session 1: Symptoms and Daily Impact of Infantile/Early Onset Pompe Disease</td>
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<td>1:30 - 2:30 p.m.</td>
<td>Session 2: Symptoms and Daily Impact of Late Onset Pompe Disease</td>
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<td>2:30 - 2:45 p.m.</td>
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**Afternoon Session: Pompe Disease Patient Perspectives on Current and Future Treatment Options**

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| 2:45 - 2:50 p.m. | Afternoon Welcome and Overview  
Tiffany House, President and CEO, Acid Maltase Deficiency Association |
| 2:50 - 3:00 p.m. | Current and Future Treatment Options for Pompe Disease  
Dr. Barry Byrne, M.D., Director of the Powell Gene Therapy Center, University of Florida |
| 3:00 - 4:50 p.m. | Session 3: Current and Future Approaches to Treatments |
| 4:50 - 4:55 p.m. | Afternoon Session Closing Remarks  
Wilson Bryan, MD, Director, Office of Tissues and Advanced Therapies, CBER |
| 4:55 - 5:00 p.m. | Next Steps and Closing Remarks  
Paul Melmeyer, Director of Regulatory Affairs, MDA |
Meet Your Speakers

Paul Melmeyer, Director of Regulatory Affairs, MDA: Opening and Closing Remarks
Paul serves as the Director of Regulatory Affairs at the Muscular Dystrophy Association (MDA). In this role, Paul leads MDA’s policy and advocacy initiatives pertaining to therapeutic development with a particular focus on regulatory efforts. Prior to joining MDA, Paul spent over six years with the National Organization for Rare Disorders (NORD). At NORD, Paul led the Federal policy operations in developing and advocating for the enactment and implementation of pro-rare disease patient policy. Paul also holds a Master of Public Policy (MPP) from the George Washington University. Paul is constantly seeking opportunities to make a difference in the lives of the less fortunate through compassionate and effective policy change.

James Valentine, J.D., M.H.S., Hyman, Phelps, & McNamara, Meeting Moderator
James Valentine received a Master of Health Science in health policy from Johns Hopkins Bloomberg School of Public Health and a JD in health law from the University of Maryland Francis King Carey School of Law. For six years, he worked at what is now the FDA Office of Health and Constituent Affairs, where he launched the Patient-Focused Drug Development program and developed the FDA Patient Network. For the past four years, Mr. Valentine has been an associate at the Washington, DC, law firm of Hyman, Phelps & McNamara, where he 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. Mr. Valentine also works with clients on clinical trials operations and compliance matters.

Dr. Priya Kishnani, M.D., Chief, Division of Medical Genetics, Duke University
Priya S. Kishnani, MD, is chief of the Division of Medical Genetics, Department of Pediatrics, at Duke University Medical Center in Durham, North Carolina, and director of its YT and Alice Chen Center for Genomic Research. She holds certification from the American Board of Medical Genetics and the American Board of Biochemical Genetics. She began her medical education in Bombay, India, followed by a residency and 2 fellowship programs at Duke University. Dr Kishnani’s primary focus has been the translation of laboratory science into the clinical arena.

She has a long-standing research and clinical interest in Pompe disease, and was the lead investigator for the pivotal trials of Pompe disease and responsible for the clinical translation from the bench, which led to FDA approval of Myozyme™ (2006)/Lumizyme™ (2010) as the first treatments for Pompe disease. Dr Kishnani continues to investigate the natural history of Pompe disease, uncovering several long-term complications, among them speech and swallowing dysfunction, cardiac arrhythmias, vascular involvement, sleep and gait disturbances, CNS involvement and small fiber neuropathy. She and the Duke team played an integral role in the nomination and approval for the addition of Pompe disease to the RUSP (Recommended Uniform Screening Panel) for newborn screening in the United States.
Meet Your Speakers

Tiffany House, President and CEO, Acid Maltase Deficiency Association
Tiffany House is the President of the Acid Maltase Deficiency Association (AMDA). In this role, Tiffany sets the organizational agenda for the year, communicates with industry, stays up to date on the latest advancements in treatment and patient care, and attends various scientific/patient conferences. Tiffany is a patient representative at the Food and Drug Administration (FDA) and chairs the International Pompe Association (IPA). Tiffany holds a J.D. from St. Mary’s University School of Law and resides in San Antonio, Texas.

Dr. Barry Byrne, M.D., Director of the Powell Gene Therapy Center, University of Florida, Chief Medical Advisor, MDA
Dr. Barry Byrne is the Associate Chair of Pediatrics and Director of the Powell Gene Therapy Center. He obtained his B.S. degree from Denison University, his M.D. and Ph.D. from the University of Illinois. He completed his Pediatrics residency, cardiology fellowship training and post-doctoral training in Biological Chemistry at the Johns Hopkins Hospital. He joined the University of Florida in 1997 and is now the Earl and Christy Powell University Chair in Genetics. His group has made significant contributions to the understanding and treatment of Pompe disease, which is a type of muscular dystrophy due to abnormal glycogen in the muscle. The research team has been developing new therapies using the missing cellular protein or the corrective gene to restore muscle function in Pompe and other inherited myopathies.

Dr. Kathleen Donohue, M.D., Medical Officer, Division of Gastroenterology and Inborn Errors Products (DGIEP), CDER, FDA
Dr. Katie Donohue graduated from medical school at Virginia Commonwealth University. She completed her residency in Internal Medicine and fellowship in Allergy & Immunology at Columbia University - New York Presbyterian Hospital. After completing her clinical training, she joined the faculty at the Columbia University College of Physicians and Surgeons. Dr. Donohue completed a Master’s Degree in Epidemiology at the Mailman School of Public Health at Columbia University, with a focus on statistics and clinical trial design. Her research focused on molecular epidemiology, leading to publications of her original research in peer-reviewed journals. Dr. Donohue joined the FDA in 2014, as a Medical Officer in the Division of Pulmonary, Allergy, and Rheumatology Products. In 2017, she was recruited as a Team Lead in the Division of Rare Diseases and Medical Genetics (DRDMG).

Dr. Wilson Bryan, MD, Director, Office of Tissues and Advanced Therapies, CBER, FDA
Wilson Bryan is a board-certified neurologist who graduated from the University of Chicago Pritzker School of Medicine. He served on the Neurology faculty of the University of Texas Southwestern Medical School for 13 years. He has been an investigator on clinical trials in cerebrovascular disease and neuromuscular disorders, particularly amyotrophic lateral sclerosis. Dr. Bryan joined the United States Food and Drug Administration (FDA) in 2000, and now serves as Director of the Office of Tissues and Advanced Therapies (OTAT) in the FDA’s Center for Biologics Evaluation and Research.
Meet Your Patient and Caregiver Panelists
Session 1: Symptoms and Daily Impact of Infantile/Early-Onset Pompe Disease

Paloma Juarez
Paloma Jaurez lives in Prairie Village, Kansas. Her and her husband Brian own and run a girls’ volleyball club called FENIX VB. Their 3-year-old son, Vaun, has infantile-onset Pompe disease.

George Fox
George is the father of Phoenix, a 17 year old boy with infantile-onset Pompe disease, and the husband of Gina Fox. The family currently resides in Gainesville, Florida. Due to Gina’s successful business, George can be Phoenix’s primary caregiver. George also sits on the Florida newborn screening advisory council as a consumer and voices the concerns of families with children born with rare diseases.

Deana Bridges
Deana is a stay at home mother and former nurse who resides in Durham, North Carolina with son Dakota. She is passionate about helping others who have family members with Pompe disease and looks to advocate in any way that she can to ensure Dakota is well taken care of.

Sarah Musgrove
Sarah has been navigating the rare disease world since 2011 as mom to her two infantile-onset Pompe warriors, Bruce and Myra. As a registered nurse, Sarah has incorporated mom life with nurse life and is “Chaos Coordinator” for their weekly home infusions on Lumizyme. Sarah lives with her partner, John, and their kids in northern Missouri where they raise beef cattle and spend their best days together in the sun on the ranch.
Meet Your Patient and Caregiver Panelists
Session 2: Symptoms and Daily Impact of Late-Onset Pompe Disease

Sean Doerr
Sean was diagnosed with late-onset Pompe disease in 2018. Presently he works as a fire dispatcher for the city of Detroit and holds a Bachelor of Fine Arts in photography from the College for Creative Studies. Sean is very active in Pompe advocacy, sits on multiple patient advocacy boards, and served as the Fill the Boot campaign advocate for his fire department. Wanting to make the most of life, Sean bought a former ambulance and turned it into an RV to see the world (the PompeTrek).

Vanessa O’Connell
Vanessa O’Connell has lived in the Orlando area for over 25 years. She is a retired property manager and has late-onset Pompe disease. She adores her 16 and 25 year old sons. She enjoys exercising, working out and tries to stay active. She’s always trying healthy, new cooking recipes on her unsuspecting family. Vanessa is crazy about her very charismatic Maine Coon cat Russell-Leo (aka Boo).

Monique Griffin
Monique Griffin was diagnosed with Pompe disease in 2010 after living with the misdiagnosis of inflammatory myopathy for over a decade. There were no early signs of Pompe disease - in fact she was a competitive gymnast, dancer, and swimmer growing up. As a young adult she remained active and worked in theater, film, and television until headaches, difficulty climbing stairs, and her inflammatory myopathy diagnosis forced her into a desk job. While her condition literally slowed her down in many ways, it also put her on another path. A path that led to new opportunities like living in Europe and Asia for school and work, and getting involved in the patient advocacy community shortly after her diagnosis. She was the first US patient to receive ERT for Pompe disease once it received its FDA approval. With that first treatment came a lot of media attention, which set her on the path to where she is today.

Lucas Garrett
Lucas Garrett is from Glens Falls, a small city in Upstate New York. He works in his community primarily as a musician - playing out and around Upstate New York around 20 to 30 times a year - as well as serving as the Entertainment Director for a local cafe and bakery. Lucas also works as a tutor in the area, teaching high school and college students various subjects in mathematics and science. When he’s not working, Lucas enjoys watching television, doing logic puzzles, or hanging out with friends; most of whom are also musicians! Although he doesn’t have many friends in the Pompe community, Lucas maintains a great friendship with Tiffany House, President and CEO of the Acid Maltase Deficiency Association, who encouraged him to speak on this panel.
Meet Your Patient and Caregiver Panelists
Session 3: Perspectives on Current and Future Treatment Options

Dave Manger
Dave Manger is from Chicago, Illinois, where he lives with his two daughters, Caroline and Evelyn, and his wife, Kate. He works in investment management and is passionate about fitness, participating in the occasional triathlon. Dave advocates for Pompe disease on behalf of his daughter, Caroline.

Haley Hayes
Haley is a 14 year old from Virginia. She enjoys socializing with her friends and swimming. Haley was diagnosed with infantile-onset Pompe disease at the age of 6 months.

Krystal Hayes
Krystal is a registered nurse and lives in Virginia with her husband, youngest daughter, cat, and dog. Her oldest daughter attends University of Virginia. She is very passionate about advocating for others and has been ever since her youngest daughter, Haley, was diagnosed with infantile-onset Pompe disease in 2006.
Meet Your Patient and Caregiver Panelists
Session 3: Perspectives on Current and Future Treatment Options (continued)

**Lisa Schlein**
Lisa is a veterinarian and pathologist working on her Ph.D. in cancer biology. She studies rare and deadly cancers that occur in both dogs and people, and works to improve treatments for both species. Two years ago, she learned that she is homozygous for the common “leaky splice” mutation that is common in late-onset Pompe disease patients. She shares her home with her husband, young daughter, two dogs, and a cat. In her spare time, she enjoys playing clarinet, crafting with polymer clay, and savoring sunny days in the beautiful Colorado mountains.

**Ryan Colburn**
Ryan Colburn is a rare disease patient with a professional background in engineering and operations management, spending portions of his career working on racecars, airplanes, and rockets. Since being diagnosed with Pompe disease in 2015, he has spent time learning about rare disease topics, including research, advocacy, and drug development in order to better understand how to participate in the rare disease ecosystem. He is passionate about patient empowerment and engagement, and actively developing relationships with other patients, advocacy groups, researchers, and pharmaceutical companies. He is driven to find the most effective ways to tackle the challenges of rare disease and break down barriers that inhibit progress.

**Morgan Burroughs**
Morgan Burroughs is 24 years old and lives with Pompe disease. She is originally from south Louisiana, but moved to Asheville, North Carolina after Hurricane Katrina. She enjoys spending time with her friends and family, reading a good book, trying new restaurants, and relaxing at home in her pj’s. She is extremely passionate about advocating for disability rights and for those whose voices are not always heard!
OUR IMPACT
Is Measured Not in Just Numbers...

70
YEARS
OF SUPPORTING PEOPLE WITH NEUROMUSCULAR DISEASE

150+
TOP MEDICAL INSTITUTIONS WITH CARE CENTERS

1B
RESEARCH DOLLARS INVESTED

2,000+
CLINICAL PROVIDERS

43+
NEUROMUSCULAR DISEASES SUPPORTED BY OUR RESEARCH

8
MEDICINES BROUGHT TO MARKET IN PAST 4 YEARS WITH MORE TO COME

OUR IMPACT
Is Measured By Lives Transformed

KIDS

TEENS

ADULTS

VOLUNTEERS

CAREGIVERS

HCPS

Transforming Lives Through Innovations in Science and Care

Muscular Dystrophy Association
Partnering Organizations

The Acid Maltase Deficiency Association
The Acid Maltase Deficiency Association was established in 1995 to assist in funding research and to promote public awareness of Pompe disease. Pompe disease is one of a family of 49 rare genetic disorders known as Lysosomal Storage Diseases or LSDs. Pompe disease is also known as Acid Maltase Deficiency or Glycogen Storage Disease type II. It affects an estimated 5,000 to 10,000 people in the developed world.

The United Pompe Foundation
The United Pompe Foundation was formed to assist patients and/or their families with medical costs and other expenses that these patients and families face and may not be able to cover, or fully cover, through their insurance.
MDA would like to thank our generous sponsors for their support of the Pompe disease Patient-Focused Drug Development Meeting

Thank you to our Sponsors:

Platinum Sponsor:
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Amicus Therapeutics

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AUDENTES Therapeutics
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**Make Your Voice Heard. Make A Difference.**

MDA fearlessly advocates for policies and programs that transform the lives of people living with Pompe disease. But we cannot do it alone.

Whether you are living with Pompe, or love someone with Pompe. Whether you are a researcher, a health care provider, a teacher, or a community leader. We need your help.

MDA grassroots advocates are crucial in helping improve the lives of the neuromuscular disease community, but we need all our voices.

Join our grassroots network today. Visit mda.org/advocacy to learn more.

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**Don’t navigate Pompe disease alone.**

Call MDA’s Resource Center for one-on-one support.

MDA’s National Resource Center can provide you with information about navigating Pompe disease and connecting with the neuromuscular community. Get in touch with questions about:

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833-ASK-MDA1  
1-833-275-6321

ResourceCenter@mdausa.org

mda.org
At MDA, we aim to bring knowledge and resources to you in accessible and understandable ways. Through MDA's Engage community education program, you will have access to a variety of important, up-to-date, and relevant educational content. While the pandemic has caused us to shift from being in-person to virtual events, the scope and availability of resources has not changed. We invite you to explore our educational offerings.

**MDA Engage Seminars**
These virtual seminars will feature information on neuromuscular disease around such topics as best practices in clinical care, genetics, and research. Each seminar will feature regional specialists as presenters and will have a focus on a specialty area. While these events will highlight regional medical professionals, families are encouraged to attend any of the seminars and their sessions.

**MDA Engage Disease Specific Symposia**
These virtual symposia will focus specifically on a single diagnosis and will include presenters from around the country who focus on care, genetics, and research. Each symposium will also highlight a variety of other topics related to that diagnosis.

**MDA Engage Webinars**
These online education sessions are up to two hours and focus on a single topic that is presented by experts in the field. Webinars are offered live and on-demand.

For more information on the MDA Engage program, please visit https://www.mda.org/care/mda-engage or email mdaengage@mdausa.org.

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**Notes**