National MPS Society

37TH ANNUAL FAMILY CONFERENCE

Bethesda, MD

SEPT. 28–30, 2023

CONNECTED TOGETHER

Trainee Lucille, R&D plateform, France©Vincent Fournier

We are an innovative global healthcare company, driven by one purpose: we chase the miracles of science to improve people's lives.

We are dedicated to transforming the practice of medicine by working to turn the impossible into the possible. We provide potentially life-changing treatment options and life-saving vaccine protection to millions of people globally, while putting sustainability and social responsibility at the center of our ambitions.

Honored to support the National MPS Society and the 37th Annual Family Conference.

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National MPS Society
Support for Families. Research for a Cure.

Mission Statement

The National MPS Society exists to cure, support and advocate for MPS and ML.

Contents

MPS and ML

Mucopolysaccharidoses (MPS) and Mucolipidosis (ML) are genetic lysosomal storage diseases (LSD) caused by the body's inability to produce specific enzymes.

MPS I

MPS I H Hurler

MPS I S Scheie

MPS I H-S Hurler-Scheie
Enzyme / α-L-Iduronidase

MPS II

MPS II Hunter
Enzyme / Iduronate sulfatase

MPS III

MPS IIIA Sanfilippo A
Enzyme / Heparan *N*-sulfatase

MPS IIIB Sanfilippo B
Enzyme / α-*N*-Acetylglucosaminidase

MPS IIIC Sanfilippo C
Enzyme / Acetyl CoA: α-glycosaminide acetyltransferase

MPS IIID Sanfilippo D
Enzyme / *N*-Acetylglucosamine 6-sulfatase

MPS IV

MPS IVA Morquio A
Enzyme / Galactose 6-sulfatase

MPS IVB Morquio B
Enzyme / β-Galactosidase

MPS VI

MPS VI Maroteaux-Lamy
Enzyme / (arylsulfatase B) *N*-Acetylgalac-tosamine 4-sulfatase

MPS VII

MPS VII Sly
Enzyme / β-Glucuronidase

MPS IX

Enzyme / Hyaluronidase

ML II/III

ML II I-Cell

ML III Psuedo-Hurler polydystrophy
Enzyme / *N*-acetylglucosamine-1-phosphotransferase

Thank You to Our Family Conference Committee

Tamara Cullere, *chair*
Carol Bryant
Patricia Espinal
Amy Holland
Lynn Hopkins
Kris Klenke
Terri Klein
Stacey Montgomery
Mercedes Ramirez Johnson
Leslie Urdaneta
Sheri Wise

Chair Letter

Welcome to Bethesda, MD, and the National MPS Society's 37th Annual Family and Science Conference!

Our conferences provide vital opportunities to engage with families, physicians, and researchers, with this conference offering unique experiences to participate with the FDA and NIH. I am confident you will go home with new resources and meaningful support to carry you on your journey.

The conference provides time to meet families in various breakout sessions, engage with physicians and researchers from around the world, and participate in educational conference sessions. You will have opportunities to meet with key advocacy leaders and changemakers, relax during meals, spend time with others, enjoy Camp Courage for children with MPS or ML and their siblings, and celebrate coming together at the family banquet. We also will have key educational opportunities regarding clinical studies, including updates on gene therapy, enzyme replacement therapies, and other science and research. We are in a new age of scientific discovery and clinical options for our families, and our conference is a great way to remain up to date on these breakthroughs.

I am excited to welcome our adult patient community to their SPIRIT conference that is concurrent on Saturday and includes wonderful speakers, clinicians, and researchers. We hope you enjoy reconnecting with one another and forge long friendships that will stay with you after the conference ends.

On Sunday we move to Capitol Hill to advocate for two days through our Speaker's Bureau Program. Thank you to everyone who has committed their time and efforts for these crucial meetings. This unique opportunity allows us to make our collective voice heard as we meet with lawmakers to discuss the needs of those with rare diseases.

In 2011, when our son Jack was diagnosed with this devastating disease, we found comfort and priceless education at our first conference. We discovered critical resources for Jack's continued care and therapy. We learned valuable information on how to navigate the MPS II environment, access treatment options, and how to assist Jack in receiving needed care. We made lifelong friends and gained a support system like nothing else. If this is your first time attending, I know your family will experience this too. Your "family" will grow exponentially overnight, and we welcome you.

Science is changing the future for those living with MPS and ML. As we explore new opportunities, let us walk together on our journey.

Lisa P. Todd
Chairman, board of directors

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Amy Downen

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General Information

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For emergencies during the conference, please contact the following team members:
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National MPS Society Child Care Policy

The National MPS Society offers complimentary child care, Camp Courage, at our conference. This enables parents and caregivers to attend and participate in conference presentations and concurrent sessions, confident that their children are being cared for in a safe environment. We offer Camp Courage for children ages 2 and up. All children, including children younger than 2, are welcome to join their parents in the educational sessions as desired.

We are excited to partner with KiddieCorp again this year. Parents or caregivers must check in and check out each child every time the child enters and leaves child care. Photo ID is required. One parent or caregiver must always remain in the meeting session rooms while their child is in child care. KiddieCorp must be able to reach the parent/caregiver, and parents/caregivers must remain on site and in conference settings. Children will receive wristbands indicating whether they can have snacks of crackers and juice. KiddieCorp staff and National MPS Society volunteers will **NOT** dispense prescription or over-the-counter medication to children, provide other food or feedings, or change diapers.

The child care supervisor or the National MPS Society's board of directors and staff are authorized to restrict a child from child care if it is determined that the child cannot be adequately cared for or if they are a danger to themselves or to others. Unfortunately, we are unable to provide one-on-one care, but parents or caregivers are welcome to stay with their child while in child care.

Children ages 5 and up are invited to a special outing this year. The KID Museum, located beside the Hyatt Regency Bethesda Hotel, will host a private event and activity time at their facility. Volunteers will be on hand to walk children to the KID Museum and stay for the duration of the field trip. Parents are welcome to attend.

CAMP COURAGE HOURS

Thursday, Sept. 28
6:30 pm – 9:45 pm

Friday, Sept. 29
7:15 am – 12:00 pm
12:45 pm – 5:30 pm

Saturday, Sept. 30
7:15 am – 12:00 pm
12:45 pm – 5:30 pm
8:15 pm – 10:30 pm

CHILD CARE ACTIVITIES

We are excited to feature entertainment throughout the conference, as well as a fun, kids-based activity about how to advocate, and a special activity for younger siblings of those with MPS or ML. Feel free to stop in to watch with your children.

Friday, Sept. 29
9:00 am—Mad Science Show
12:30 pm—KID Museum field trip (ages 5 and up)

Saturday, Sept. 30
9:30 am—Abracadabra Alex Magic Show

Schedule

Please note that the conference schedule is subject to change.

Social Media

Keep updated during the conference by following the National MPS Society on social media.

@NationalMPSSociety

@mpssociety

@mpssociety

National MPS Society

Use **#MPSFamily2023** to share your pictures and memories!

General Information

Medical Facilities/Services

Hospital/Emergency Room
Children's National Medical Center
1310 Southern Ave. SE
Washington, DC 200232
202.574.6000
childrensnational.org

Worship Services

Baptist
Capital Baptist Church
3504 Gallows Road
Annandale, VA 22003
capitalbaptist.org

Non-denominational
5033 Wilson Lane
Bethesda, MD 20814
churchinbethesda.org

Church of Jesus Christ of Latter-day Saints
The Church of Jesus Christ of Latter-day Saints
5460 Wester Ave.
Chevy Chase, MD 20815
local.churchofjesuschrist.org

Episcopal
St. Luke's Episcopal Church
6030 Grosvenor Lane
Bethesda, MD 20814
stlukesbethesda.org

Evangelical
Canaan Oromo Evangelical Church
5033 Wilson Lane
Bethesda, MD 20814
202.641.6865

Islamic
Maqaame Ibrahim-Jummah Only
6601 Bradley Blvd.
Bethesda, MD 20817
miichourseofibrahim.org

Jewish
Bethesda Jewish Congregation
6601 Bradley Blvd.
Bethesda, MD 20817
bethesdajewish.org

Lutheran
Emmanuel Lutheran Church
7730 Bradley Blvd.
Bethesda, MD 20817
elcbethesda.org

Methodist
Bethesda United Methodist Church
8300 Old Georgetown Road
Bethesda, MD 20814
washmorefeet.org

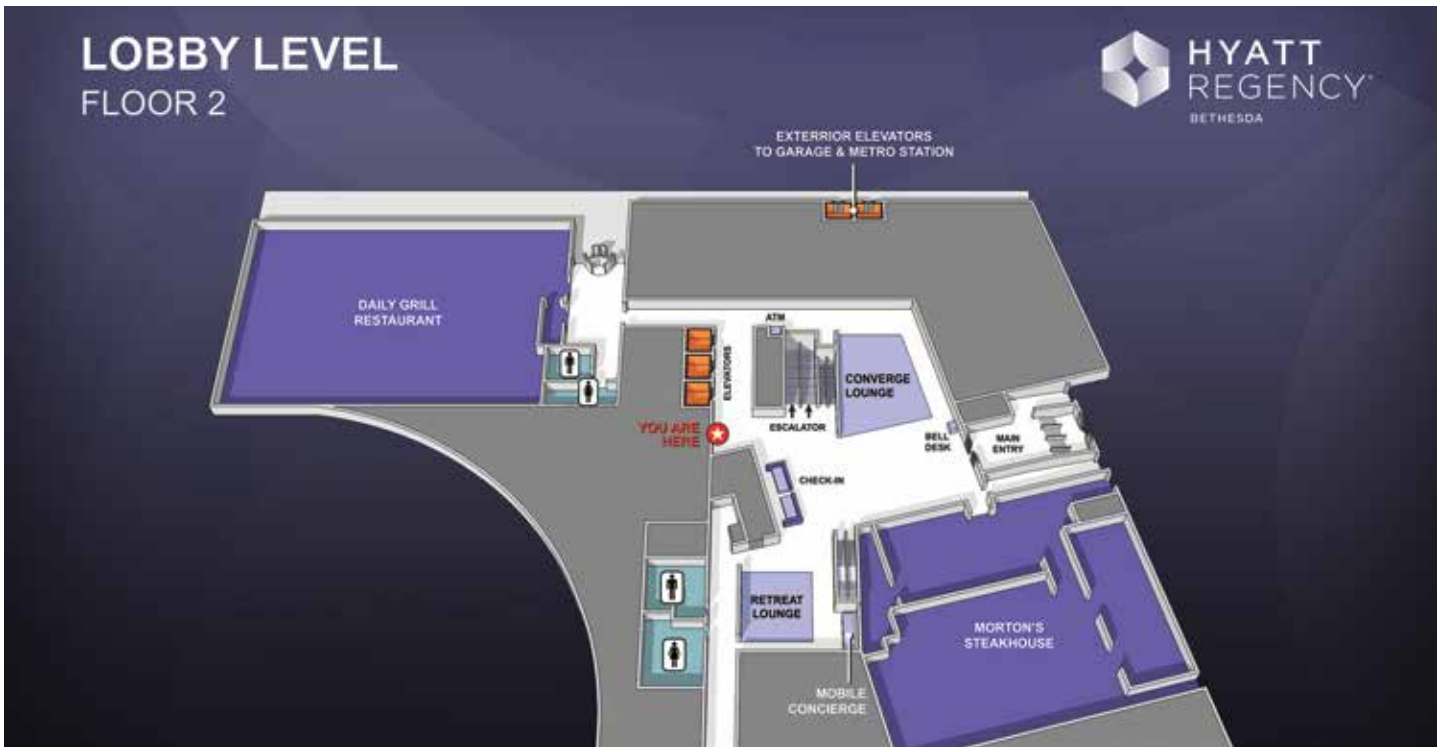
Presbyterian
Fourth Presbyterian Church
5500 River Road
Bethesda, MD 20816
4thpres.org

Support the Adult Resource Committee
by purchasing your Rare swag!



Visit www.cafepress.com/rareawareshop
or scan the QR code

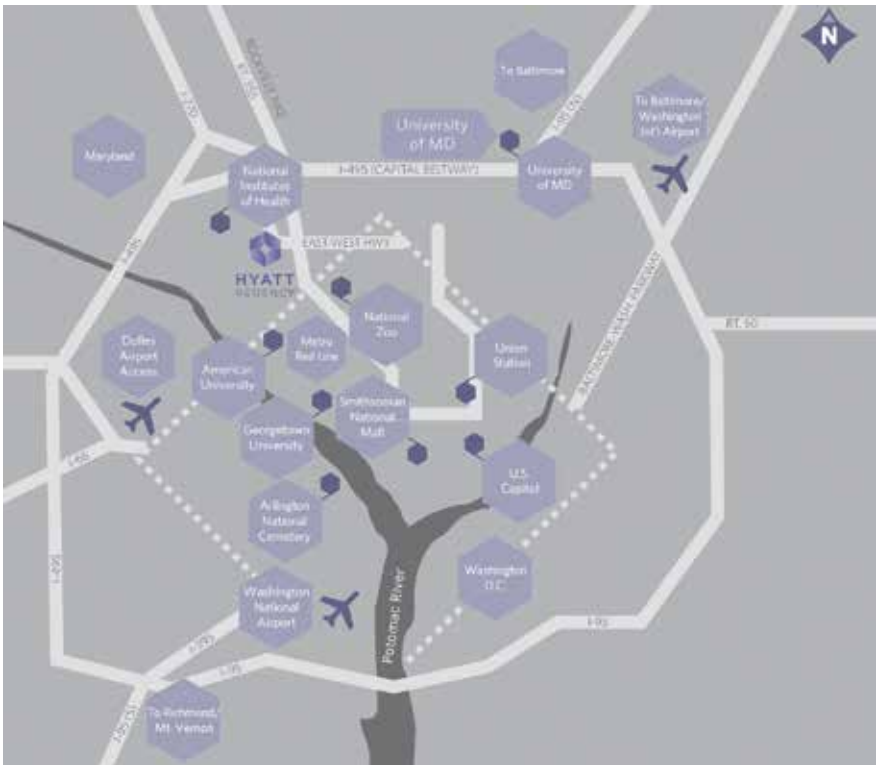
Hotel Layout



Hotel Layout

Highlights of Bethesda, MD

- Centered in the Bethesda Arts and Entertainment district with more than 200 restaurants, theaters, galleries, and boutiques adjacent to the hotel.
- Direct Metro subway access below the hotel to Washington, DC's monuments, museums, attractions, and Red Line Bethesda Station.
- Close to National Institutes of Health, Suburban Hospital, Walter Reed Military Medical Center, American University, Food & Drug Administration, Nuclear Regulatory Commission, and the National Zoo.
- Minutes to local shopping and boutiques.



Awards

Visionary Award

STEPHANIE BOZARTH AND STEVE HOLLAND

Legacy Award, Clinician

JOSEPH MUENZER, MD, PHD

Legacy Award, Research

MARK SANDS, PHD

President's Award

EKATERINA WRIGHT, MD

Chairman's Award

LARRY KIRCH

Directors' Award

STEPHANIE COZINE

"Friend"raising Award

MARIELLE MARINOFF

Schedule

THURSDAY, SEPT. 28

TIME	EVENT	LOCATION
4:00 pm – 9:00 pm	**Registration**	Regency Ball Foyer
6:30 pm – 9:45 pm	**Camp Courage**	Cabinet/Judiciary
5:30 pm – 7:30 pm	**Welcome Buffet Dinner**	Regency Ballroom
6:00 pm – 7:30 pm	**Newly Diagnosed Family Dinner *(diagnosed since August 2022)*** Joseph Muenzer, PhD, MD, and Nathalie Marie, MD **Overview and Coping with MPS and ML**	Old Georgetown/ Congressional
7:30 pm – 8:30 pm	**SYNDROME BREAKOUT SESSIONS**	
	MPS I	Regency Ballroom
	MPS II	Susquannah/Severn
	MPS III	Diplomat/Ambassador
	MPS IV	Embassy Suite
	MPS VI	Patuxent
	MPS VII	Potomac
	ML II/III	Chairmans Boardroom
8:30 pm – 9:30 pm	**FAMILY BREAKOUT SESSIONS**	
	Older Siblings (age 13+)	Diplomat/Ambassador
	Mothers	Regency Ballroom
	Fathers	Susquannah/Severn
	Grandparents	Patuxent
	Adults with MPS/ML	Old Georgetown/Congressional
	Bereaved Families	Embassy Suite
9:45 pm	**Camp Courage closes**	Cabinet/Judiciary

Schedule

FRIDAY, SEPT. 29

SCIENTIFIC CONFERENCE: SCIENCE, POLICY, AND ADVOCACY / BREAKFAST AND LUNCH SYMPOSIUMS

TIME	EVENT	LOCATION
6:30 am – 8:00 am	**Breakfast Buffet**	Regency Ballroom
7:45 am	**Camp Courage opens**	Cabinet/Judiciary
7:00 am – 7:05 am	**Welcome and Introduction**, Lisa P. Todd*, board chair, National MPS Society*	
7:00 am – 8:00 am	**BREAKFAST SYMPOSIUM—PART ONE / INDUSTRY UPDATES**	**Regency Ballroom**
7:00 am – 7:15 am	**Denali Therapeutics Update**, Bill Bakker*,* PharmD*, senior medical science liaison*	
7:15 am – 7:30 am	**REGENXBIO's Investigational Gene Therapies for the Treatment of MPS I and II** Laura Pisani, MD, *senior medical director of Global Clinical Development, REGENXBIO*	
7:30 am – 7:45 am	**Sanofi Update***,* Heather Mazzotta, RN*, senior patient education liaison, Rare Disease, Sanofi*	
7:45 am – 8:00 am	**Takeda Update***,* Erik Cline*, head of U.S. Marketing, Lysosomal Storage Disease Franchise*	
8:10 am – 10:10 am	**HSCT (TRANSPLANT) CONCURRENT SESSION**	**Patuxent/Embassy**
8:10 am – 9:00 am	**Considerations for Transplant**, Paul Orchard, MD, *University of Minnesota*	
9:00 am – 9:45 am	**Orthopedic Considerations for Transplant MPS Patients** Klane White, MD, MSc*, Orthopedic Institute, Children's Colorado Hospital*	
9:45 am – 10:00 am	**Q & A**	
10:00 am – 10:10 am	**Closing**	
8:00 am – 10:10 am	**SESSION I—REGULATORY AND POLICY HURDLES ON THE PATH TO OPTIMAL DIAGNOSIS AND TREATMENT**	**Regency Ballroom**
8:00 am – 8:10 am	**Visionary Award: Stephanie Bozarth and Steve Holland** **Legacy Award, Research: Mark Sands, PhD** **President Award, Volunteer: Katya Wright, MD**	
8:10 am – 8:30 am	**MPS Therapy, Newborn Screening, and the RUSP: An Overview** N. Matthew Ellinwood, PhD, DVM*, chief scientific officer, National MPS Society*	
8:30 am – 8:50 am	**Drug Approval in Rare Disease and the RUSP Bottleneck** Jamie Sullivan, MPH, *EveryLife Foundation*	
8:50 am – 9:10 am	**The Journey to Newborn Screening: Pilots, Legislation, RUSP, and Implementation** Amy Gaviglio, MS, CGC, *Connetics Consulting, LLC*	
9:10 am – 9:30 am	**From Public Health Screening to Uniform Equity in Care** Natasha Bonhomme, *Expecting Health*	
9:30 am – 10:00 am	**Round Table Discussion** *(speakers and moderator)*	
10:00 am – 10:10 am	**Q & A** *(speakers, moderator, audience)*	
10:10 am – 10:25 am	**Health Break**	
10:25 am – 12:00 pm	**SESSION II—KEYNOTE SESSION: THE FDA AND RARE FROM THE STAKEHOLDER PERSPECTIVE** *Moderators:* N. Matthew Ellinwood, DVM, PhD, and Amy Gaviglio, MS, CGC	**Regency Ballroom**
10:30 am – 10:55 am	**Challenges in FDA Structure and Practice Confronting the Rare Disease Community**, Annie Kennedy*, EveryLife Foundation*	

continued

Schedule

TIME	EVENT	LOCATION
10:55 am – 11:20 am	**Drug Development for Rare Diseases Must Catch Up with Scientific Advances: The Role of Primary Disease Biomarkers and Accelerated Approval to Advance Drugs for MPS and Other Rare Disorders**, Camille Bedrosian, MD, *Ultragenyx*	
11:20 am – 11:45 am	**Addressing the Unmet Needs of the Rare Disease Community**, Peter Marks, MD, PhD, *FDA*	
11:45 am – 12:00 pm	**Q & A** *(panel and audience)*	
12:00 pm	**Camp Courage closes**	Cabinet/Judiciary
12:10 pm – 1:00 pm	**LUNCH SYMPOSIUM—ORTHOPEDIC UPDATES IN MPS** Klane White, MD, MSc, *Orthopedic Institute, Children's Colorado Hospital*	**Regency Ballroom**
12:45 pm	**Camp Courage opens**	Cabinet/Judiciary
1:00 pm – 1:50 pm	**SESSION III—PROGRESS IN UNMET ORTHOPEDIC AND CONNECTIVE TISSUE DISEASE** *Moderator:* N. Matthew Ellinwood, DVM, PhD	**Regency Ballroom**
1:05 pm – 1:22 pm	**Leveraging Naturally Occurring Canine Models to Advance Novel Treatments for Skeletal Disease in the Mucopolysaccharidoses** Lachlan Smith, PhD, *University of Pennsylvania*	
1:22 pm – 1:50 pm	**Growth Failure in MPS Disease** Paul Harmatz, MD, *pediatric gastroenterologist, UCSF Benioff Children's Hospital*	
1:50 pm – 2:55 pm	**SESSION IV—PROGRESS IN MODELING DISEASE** *Moderator:* N. Matthew Ellinwood, DVM, PhD	**Regency Ballroom**
1:55 pm – 2:15 pm	**Progress in MPS VI Research with the Canine Model** Magret Casal, DVM, PhD, *University of Pennsylvania*	
2:15 pm – 2:40 pm	**The Feline Model of ML II** Allison Bradbury, PhD, *Nationwide Children's Hospital*	
2:40 pm – 2:55 pm	**Q & A Session III and IV** *(panel and audience)*	
2:55 pm – 3:15 pm	**Health Break**	
3:15 pm – 4:00 pm	**Camp Courage Youth Advocacy** Stephanie Cozine, *National MPS Society board of directors*	Old Georgetown/Congressional
3:15 pm – 4:05 pm	**SESSION V—THERAPEUTIC INNOVATION: ERT AND GENE THERAPY ADVANCEMENT** *Moderators:* N. Matthew Ellinwood, DVM, PhD, and Mark Sands, PhD	**Regency Ballroom**
3:20 pm – 3:35 pm	**Prenatal Enzyme Replacement Therapy for MPS and Other Lysosomal Storage Disorders** Emma Canepa, *UCSF*	
3:35 pm – 3:50 pm	**Progress in GlcNAc-1-Phosphotransferase Research and Its Implications on Therapy** Patricia Dickson, MD, *Washington University School of Medicine, St. Louis*	
3:50 pm – 4:05 pm	**An AAV-Based Clinical Trial for MPS IVA** Shunji Tomatsu, MD, PhD, *Nemours Children's Hospital*	
4:05 pm – 5:10 pm	**SESSION VI—THERAPEUTIC INNOVATION: HYBRID BMT**	**Regency Ballroom**
4:05 pm – 4:25 pm	**Human Genome-Edited Hematopoietic Stem Cell-Based Therapy for MPS I** Natalia Gomez-Ospina, MD, PhD, *Stanford Medicine*	
4:25 pm – 4:40 pm	***Ex vivo* Autologous Stem Cell Gene Therapy Clinical Trial for MPS IIIA: Update on Phase I/II Clinical Trial** Simon Jones, MBChB, BSc, MRCPCH, *University of Manchester*	

continued

Schedule

TIME	EVENT	LOCATION
4:40 pm – 4:55 pm	**Gene Modified Hematopoietic Stem Cell Transplantation for MPS IIIC** Rafael Badell Grau, PhD, *UCSD*	
4:55 pm – 5:10 pm	**Q & A Session V and VI** *(panel and audience)*	
5:10 pm – 5:30 pm	**Advocacy, Policy, and Research Recap** N. Matthew Ellinwood, DVM, PhD, *chief scientific officer, National MPS Society*	
5:30 pm	**Camp Courage closes**	Cabinet/Judiciary
6:00 pm – 7:30 pm	**Planned Giving Cocktail Event** *Speaker:* Terri Klein, *president and CEO, National MPS Society*	Rooftop, Level P

SATURDAY, SEPT. 30

FAMILY CONFERENCE: CARE MANAGEMENT FOR MPS AND ML / BREAKFAST AND LUNCH SYMPOSIUMS

TIME	EVENT	LOCATION
6:30 am – 8:00 am	**Breakfast Buffet**	Regency Ballroom
7:15 am	**Camp Courage opens**	Cabinet/Judiciary
7:15 am – 7:45 am	**BREAKFAST SYMPOSIUM—ANNUAL GENERAL MEMBERSHIP MEETING**, Lisa P. Todd, *board chair, National MPS Society*	**Regency Ballroom**
7:45 am – 8:00 am	**Legacy Award, Clinician: Joseph Muenzer, PhD, MD** **Chairman Award: Larry Kirch** **Directors' Award: Stephanie Cozine**	
8:00 am – 10:45 am	**SESSION I—CARE MANAGEMENT FOR MPS AND ML**	**Regency Ballroom**
8:00 am – 8:45 am	**ML II/III and MPS Clinical Overview and Clinical Trial Updates** Joseph Muenzer, MD, PhD, *Bryson distinguished professor in pediatric genetics and professor in the Department of Pediatrics and Department of Genetics at the University of North Carolina at Chapel Hill*	
8:45 am – 9:30 am	**Expanding the Understanding of Function in MPS: Neurocognitive Endpoints and Beyond**, Julie Eisengart, PhD, *University of Minnesota*	
9:30 am – 10:15 am	**ENT and Airway Issues Associated with MPS** David Molter, MD, *Washington University School of Medicine, St. Louis*	
10:15 am – 10:45 am	**Cardiac Considerations and Updates** Elizabeth Braunlin, MD, *University of Minnesota*	
10:50 am – 12:00 pm	**SESSION II—NEWBORN SCREENING AND UNDERSTANDING DIFFERENT THERAPEUTIC APPROACHES**	**Regency Ballroom**
10:50 am – 11:25 am	**Newborn Screening—Updates on Diagnostics in Newborns** Barbara Burton, MD, *Lurie Children's Hospital*	
11:25 am – 12:00 pm	**Understanding Gene and Cell Therapies** Paul Harmatz, MD, *UCSF*	
12:00 pm	**Camp Courage closes**	Cabinet/Judiciary

continued

Schedule

TIME	EVENT	LOCATION
12:00 pm – 1:00 pm	**LUNCH SYMPOSIUM—PART II INDUSTRY UPDATES**	**Regency Ballroom**
12:00 pm – 12:15 pm	**JCR Pharmaceuticals Update**, Takayo Egawa, *corporate director, Patient Group Communications, director, International Affairs*	
12:15 pm – 12:30 pm	**Orchard Therapeutics: Hematopoietic Stem Cell Gene Therapy** Annamarie Dillon, *executive director, Patient Advocacy*	
12:30 pm – 12:45 pm	**Ultragenyx Update**, Deborah Marsden, MD, *global medical expert for genetic metabolic disorders*	
12:45 pm – 1:00 pm	**BioMarin Pharmaceuticals: BioMarin's Partnership with the MPS Community** Kate Delaney, *senior director, Global Patient Affairs*	
12:45 pm	**Camp Courage opens**	Cabinet/Judiciary
1:00 pm – 2:25 pm	**SESSION III—UNMET NEEDS IN RESEARCH**	**Regency Ballroom**
1:00 pm – 1:35 pm	**Mucolipidosis—Understanding the Research** Patricia Dickson, MD, *Washington University School of Medicine, St. Louis*	
1:35 pm – 2:15 pm	**Sanfilippo—Advancing Clinical Studies to Therapies, What Can We Do?** N. Matthew Ellinwood, DVM, PhD, *chief scientific officer, National MPS Society*	
2:15 pm – 2:25 pm	**Q & A**	
2:30 pm – 3:00 pm	**SPECIAL SESSION—FAMILY PLANNING AND GENETIC COUNSELING** Barbara Burton, MD, *Lurie Children's Hospital*	**Regency Ballroom**
3:00 pm – 3:15 pm	**Health Break**	Regency Foyer
3:15 pm – 5:10 pm	**SESSION IV—WARRIORS NAVIGATING LIFE; CHANGE AGENTS OF ECOSYSTEMS**	**Regency Ballroom**
	"Friend"raising Award: Marielle Marinoff	
3:20 pm – 3:40 pm	**Taking Control: Fundraising for Critical Research**, Marielle Marinoff, *MPS II mother*	
3:40 pm – 4:05 pm	**Strengthening Self-Reported Health Outcomes and Quality of Life Through Movement** Jennifer Klein, MS, *ML III patient*	
4:05 pm – 4:25 pm	**How to Share Your Story: Making an Impact on Capitol Hill** Mercedes Ramirez Johnson, *National MPS Society board of directors*	
4:25 pm – 5:10 pm	**State Advocacy Workshop** Stephanie Cozine and Carl Kapes, *National MPS Society board of directors*	
5:30 pm	**Camp Courage closes**	Cabinet/Judiciary
5:30 pm – 6:00 pm	**Remembrance Ceremony**	Outdoor Lawn
7:00 pm – 9:00 pm	**Banquet**	Regency Ballroom
8:15 pm – 10:30 pm	**Camp Courage**	Cabinet/Judiciary

SUNDAY, OCT. 1

TIME	EVENT	LOCATION
8:00 am – 12:00 pm	**Board of Directors Meeting**	Rooftop, Level P

Schedule

FINDING OUR SPIRIT:
STRENGTH, PURPOSE, INDEPENDENCE, RESILIENCE, AND INITIATIVE TOGETHER

All sessions held in Old Georgetown/Congressional Room

FRIDAY, SEPT. 29

TIME	EVENT
5:30 pm – 8:00 pm	**SPIRIT dinner**, Matchbox, 7278 Woodmont Ave., Bethesda, MD *(meet in hotel lobby at 5:15 pm and walk over)*
8:30 pm – 11:00 pm	**Game night**, Hyatt Regency Bethesda—Rooftop Room, Level P

SATURDAY, SEPT. 30

9:00 am – 9:05 am	**Welcome**
9:10 am – 10:00 am	**Navigating Social Security, Medicaid, and Medicare**, Tom Wier, *Social Security Administration*
10:00 am – 11:00 am	**Cannabidiol, Medical Marijuana, and Pain Management: Considerations for MPS** Lisa Garrity, PhD, *Cincinnati Children's Hospital Medical Center*
11:00 am – 12:00 pm	**Managing MPS and ML in Daily Living: Using Tools for Wellness** Carmen Sanchez, PhD, *licensed clinical psychologist*
12:00 pm – 1:00 pm	**Lunch** *(Served in Congressional/Old Georgetown Room)*
1:00 pm – 2:50 pm	**Gaining Clinician and Research Insights—Ask the Doctor**
1:00 pm – 1:30 pm	Barbara Burton, MD, *Lurie Children's Hospital*
1:30 pm – 2:00 pm	Paul Harmatz, MD, *UCSF Benioff Children's Hospital*
2:00 pm – 2:20 pm	Elizabeth Braunlin, MD, *University of Minnesota*
2:20 pm – 2:50 pm	N. Matthew Ellinwood, DVM, PhD, *National MPS Society*
3:00 pm – 3:45 pm	**Resume Building** Amy Downing, MBA, *National MPS Society*
3:45 pm – 4:15 pm	**Adult Transitions** Questionnaire, Sheri Wise
4:15 pm	**Closing remarks**

Speakers

Rafael Badell-Grau

Rafael Badell-Grau, PhD, is a postdoctoral fellow in the Department of Pediatrics at the University of California, San Diego in the Cherqui lab, which focuses on developing stem cell and gene therapy strategies for degenerative multi-systemic disorders. Dr. Badell-Grau obtained his master's degree and PhD at Cardiff University where he worked on investigating the cellular mechanisms of neurodegenerative diseases, such as Huntington and lysosomal storage diseases, with a specific focus on different forms of Batten and Niemann Pick diseases for the purposes of understanding these diseases and drug screening. During his Master's of Research he was awarded a letter of commendation and the Best Student Award for best overall performance. His PhD focused on improving the understanding of Batten disease, a devastating lysosomal storage disorder, to find and develop novel, small-molecule therapeutic approaches. Currently, Dr. Badell-Grau is continuing to develop novel therapeutic approaches for lysosomal storage diseases, specifically MPS IIIC; hematopoietic stem cell and gene therapy strategies; and *in vivo* mouse models of these diseases under the supervision of Dr. Stephanie Cherqui.

Gene-Modified Hematopoietic Stem Cell Transplantation for MPS IIIC

4:40 PM, SEPT. 29, REGENCY BALLROOM

MPS IIIC is a lysosomal storage disease characterized by the accumulation of long sugars called glycosaminoglycans (GAGs). This disease affects children, causing progressive neurological or brain defects due to errors in the instruction to make the protein heparan-α-glucosamine *N*-acetyltransferase (HGSNAT) which leads to this protein (located in the lysosomes in cells) not working properly. There is no available treatment for this disease. Our goal is to develop a therapy for MPS IIIC using bone marrow stem cells that have been genetically corrected. Our group has previously shown that this treatment method could rescue cystinosis, another lysosomal storage disease involving a similar protein, and has conducted a phase I/II clinical trial using this treatment in cystinosis with positive clinical outcomes. The stem cells in this treatment will turn into different types of cells within tissues that can deliver healthy lysosomes to diseased cells through long tunnels or tubes. We believe this will be the same for MPS IIIC. We created a new MPS IIIC mouse model which shows similar issues seen in patients, such as the accumulation of GAGs in several organs as well as an increase in the size of the spleen, and neurological defects. We have treated MPS IIIC mice with these stem cells from healthy donors and have seen positive outcomes in most MPS IIIC issues including the decrease in the organ size and improvement of neurological defects, but not a decrease in GAG accumulation. We also have created a delivery system to insert a healthy copy of the HGSNAT gene into MPS IIIC stem cells for transplant. We will test this treatment with stem cells that have been genetically corrected in the MPS IIIC mouse model to see if it can correct the disease. So far, results show that we have successfully made a mouse that represents MPS IIIC, and shown some beneficial impact on healthy stem cells; we are ready now to test the impact of the genetically modified stem cells. These studies are the first step toward a future clinical application of this therapy.

Speakers

Bill Bakker

Bill Bakker serves as senior medical science liaison at Denali Therapeutics. He received his PharmD from Ohio State University and lives in Reading, VT. He has been working in industry for more than 20 years and with the lysosomal storage disorder community for the last 10 years. He is grateful to support and engage with the lysosomal storage disorder clinical and patient communities daily in this role at Denali.

Denali Therapeutics Update

7:00 AM, SEPT. 29, REGENCY BALLROOM

Denali Therapeutics is a biopharmaceutical company focused on developing treatments that make a meaningful difference for families living with MPS and other lysosomal storage disorders. Denali's Enzyme Transport Vehicle technology is designed to enable treatments to cross the blood-brain barrier and reach the brain in addition to the rest of the body. This presentation will include an update on Denali's DNL310 clinical program in Hunter syndrome and its lysosomal storage disorders pipeline. DNL310 is an investigational IV enzyme replacement therapy that aims to treat the behavioral, cognitive, and physical aspects of Hunter syndrome.

Speakers

Camille Bedrosian

Camille Bedrosian, MD, is strategic development advisor at Ultragenyx Pharmaceutical, where she recently served for more than five years as chief medical officer and executive vice president. Since joining the biotech industry nearly 30 years ago from academia, she has focused on the development of medicines for individuals with rare diseases that spans the areas of hematology, rare cancers, rare bone diseases, nephrology, inborn errors of metabolism, and neurology.

Prior to joining Ultragenyx, Dr. Bedrosian served for nearly a decade as senior vice president and chief medical officer at Alexion Pharmaceuticals, Inc. Prior to Alexion, she spent more than five years as the chief medical officer for ARIAD Pharmaceuticals, Inc., and earlier served in the Clinical Research and Development Department of Genetics Institute, Inc. (now part of Pfizer). Previously, Dr. Bedrosian was an assistant professor of medicine at Duke University Medical Center and a member of the Duke Comprehensive Cancer Center.

Dr. Bedrosian earned an AB, magna cum laude, in honors chemistry from Harvard University, an MD from Harvard Medical School, and an MS in biophysics from the Massachusetts Institute of Technology (MIT). She currently serves as a member of the MIT Corporation Visiting Committee for the Department of Biology, and on the board of directors of Rhythm Pharmaceuticals and Crinetics Pharmaceuticals.

Drug Development for Rare Diseases Must Catch Up with Scientific Advances: *The Role of Primary Disease Biomarkers and Accelerated Approval to Advance Drugs for MPS and Other Rare Disorders*

10:55 AM, SEPT. 29, REGENCY BALLROOM

Despite several drugs and modalities in development, progress in various lysosomal storage diseases (LSDs) has been stymied, with many drugs being shelved or caught up in lengthy trials reliant upon clinical outcomes in order to meet rigid regulatory requirements. The genetic loss of specific lysosomal enzymes that leads to various LSDs is known and over recent years the assays to measure the resulting toxic molecules, such as glycosaminoglycans, have improved substantially. These molecules are primary disease activity biomarkers that are hallmarks of the diseases. Furthermore, scientific advancements also have led to cutting-edge treatment methodologies to address the more challenging aspects of LSDs, such as the neurological manifestations of certain mucopolysaccharidoses (MPS). Yet no drugs have been approved to date to address the neurological consequences of MPS. In the meantime, individuals with these diseases experience the irreversible consequences of the disease process.

Utilization of these primary disease biomarkers as endpoints in conjunction with the accelerated approval pathway is an important approach to address this dilemma. The accelerated approval pathway was conceived to shorten time to drug approval in cases where it is impractical to collect sufficient clinical outcome data within a reasonable timeframe. The use of specific primary disease biomarkers that directly and precisely measure the underlying disease state is a more accurate way to measure disease than clinical endpoints and can transform the development process—rapidly accelerating the cycle time for drug discovery, clinical study, and approval—resulting in improved drugs that optimally treat the underlying disease.

Natasha Bonhomme

Natasha Bonhomme is the founder of Expecting Health and has more than a decade of nonprofit and maternal and child health experience. She launched Expecting Health to bring a range of consumer and professional stakeholders together to address the need for clear, science-based information for families and providers through tangible, actionable messages. Natasha also serves as chief strategy officer at Genetic Alliance where she oversees the organization's maternal and child health initiatives, with a particular focus on bringing the families' perspectives into policy setting around screening, diagnosis, and maternal and child health overall. Natasha led an extensive study of women (with more than 2,000 expectant and new mothers) to understand their attitudes toward newborn screening and their preferences on how and when to be educated. As director of the nation's center on newborn screening education, Baby's First Test, Natasha has testified before the U.S. Senate Health, Education, Labor, and Pension Committee's Subcommittee on Children and Families on the importance of public education for newborn screening. She serves on a range of committees, including as co-chair of the Genetics and Bioethics Committee and American Public Health Association. She recently co-chaired the National Academies of Sciences, Engineering, and Medicine's workshop *Next-Generation Screening—The Promise and Perils of DNA Sequencing of Newborns at Birth*. Outside of the office, Natasha has been involved with the Planned Parenthood of Metropolitan Washington for several years and was a founding co-chair of its Developing Leaders Program. She is a board member of Whitman Walker Health, a DC-based federally qualified health center focused on the healthcare needs of the LGBTQ+ community.

From Public Health Screening to Uniform Equity in Care

9:10 AM, SEPT. 29, REGENCY BALLROOM

The listing of a condition on the federal Recommended Uniform Screening Panel and the adoption of screening by individual states and territories is only the beginning of the process of establishing optimal therapy for screened conditions. This presentation will provide an overview of a host of barriers that exist to equitable access and equity in delivery of therapy for babies and their families. These barriers exist at every step of the system of newborn screening. Being mindful and aware of these barriers provides organizations the tools necessary to ensure the highest level of equity to access for these babies and their families.

Speakers

Allison Bradbury

Allison Bradbury, MS, PhD, is an assistant professor in the Department of Pediatrics at Ohio State University and a principal investigator in the Center for Gene Therapy at the Abigail Wexner Research Institute at Nationwide Children's Hospital. Dr. Bradbury earned her PhD in biomedical sciences from Auburn University and subsequently completed a NIH National Research Service Award postdoctoral fellowship at the University of Pennsylvania. The acmes of her training career include the completion of preclinical studies that resulted in adeno-associated virus-mediated gene therapy clinical trials for GM2 gangliosidosis and Krabbe disease. She received the NIH Pathway to Independence award (K99/R00) and joined the faculty at OSU and NCH. The Bradbury Laboratory is dedicated to understanding disease mechanisms and the development of safe and efficient therapeutic approaches for rare, pediatric neurologic disorders.

The Feline Model of ML II

2:15 PM, SEPT. 29, REGENCY BALLROOM

Mucolipidosis II (ML II) is a complex disorder that affects multiple organ systems, most notably the skeletal system and the cardiovascular system. Heart disease is a leading cause of death in ML II patients, which typically occurs in the first decade of life. There currently are no treatment options for ML II. Recent studies in the ML II zebrafish and a human patient have revealed promising therapeutic targets. These findings are important and compelling, but anatomical differences in the zebrafish heart and a limited patient population necessitate evaluation in the only mammalian model of ML II heart disease, the cat. We began evaluating cardiac disease in the naturally occurring feline model of ML II which biochemically, physiologically, and anatomically models the human disease. This will provide the natural history data necessary to undertake a larger preclinical study evaluating therapeutic strategies to treat cardiac disease in ML II. One such therapeutic strategy that has been ongoing in the feline model of ML II is systemic delivery of adeno-associated virus (AAV) mediated gene replacement therapy. AAV is a small, nonpathogenic virus that can serve as a delivery vehicle to provide a healthy copy of the defective GNPTAB gene. ML II cats have been treated at one month of age by intravenous delivery of AAV at three different doses. We have monitored the effect of the therapy on the heart, skeletal system, visual system, and central nervous system. Our aim is to use this valuable large animal model of ML II to elucidate disease mechanisms and evaluate therapeutic interventions, with the ultimate goal of translating successful therapies to the clinic.

Speakers

Elizabeth Braunlin

Elizabeth Braunlin, MD, PhD, is a professor of pediatrics at the University of Minnesota Medical School in Minneapolis, MN. She is a board-certified pediatric cardiologist with a long-standing interest in cardiac features found in MPS disorders. She has followed more than 200 patients who have undergone bone marrow transplantation for MPS at the university since 1983 and has authored several papers on this topic. She is also engaged in basic research—defining the cardiac features of MPS mouse models by cardiac echo and working with other scientists to develop treatments to prevent the residual disease that remains after current treatments for MPS.

Cardiac Considerations and Updates

10:15 AM, SEPT. 30, REGENCY BALLROOM

The mucopolysaccharidoses (MPS) affect all organs of the body, including the heart. This session will review the typical cardiac features found in MPS, and discuss how they are evaluated and how they respond to current therapies. We will review the need for cardiac surgery in MPS and what is important to know before undergoing cardiac surgery. The effects of newborn screening on cardiac findings and how newborn screening may affect treatment outcomes will be discussed, as well as new treatments that may help prevent residual disease in MPS.

Gaining Clinician and Research Insights—Ask the Doctor

2:00–2:20 PM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Speakers

Barbara Burton

Barbara Burton, MD, is a professor of pediatrics at the Northwestern University Feinberg School of Medicine, and an attending physician in the Division of Genetics, Birth Defects and Metabolism at the Ann & Robert H. Lurie Children's Hospital of Chicago where she directs the MPS/ML Treatment Center. She is board certified in pediatrics, clinical genetics, and clinical biochemical genetics. Dr. Burton is an investigator for numerous natural history studies and clinical trials of new therapies for various genetic disorders, including many trials in the MPS disorders. She has published more than 300 peer-reviewed articles, 50 chapters in books, and is an editor of two textbooks.

Dr. Burton is active in professional organizations and is former president of the Society for Inherited Metabolic Disorders and the Chicago Pediatric Society. She served for four years as a member of the Secretary's Advisory Committee on Heritable Disorders in Infants and Children, the federal advisory committee that makes recommendations regarding newborn screening in the United States. She is an emeritus member of the board of directors of the Greater Chicago Area March of Dimes and received a Lifetime Achievement Award from the March of Dimes in 2018. She is a member of the Scientific Advisory Board of the National MPS Society and of Project Alive, and serves on the medical advisory board of a number of other patient advocacy organizations.

Newborn Screening—Updates on Diagnostics in Newborns

10:50 AM, SEPT. 30, REGENCY BALLROOM

Newborn screening refers to testing performed on all newborns shortly after birth to detect a group of serious disorders for which treatment is most effective when initiated prior to clinical recognition of the disease. Each state health department determines which disorders are included, although there is a federal advisory committee that approves disorders for inclusion on the Recommended Uniform Screening Panel (RUSP). MPS I was added to the RUSP in 2016 and currently more than 80% of newborns in the United States are screened for this condition. MPS II was added to the RUSP in 2022 based on data from Illinois and Missouri, the only two states currently screening all newborns for this disorder. It is anticipated that many more states will add MPS II to their screening panel in the next few years. Screening tests are available for all of the other MPS disorders. This presentation will review the newborn screening process and the current status of newborn screening for MPS disorders, emphasizing both the benefits and the lessons learned. We will discuss the role of families and advocacy organizations in promoting newborn screening.

Family Planning and Genetic Counseling

2:30 PM, SEPT. 30, REGENCY BALLROOM

MPS disorders are genetically determined. This presentation will discuss basic genetics and review the pattern of inheritance of each of the MPS disorders. We will address the risk of recurrence faced by parents and healthy siblings of affected individuals as well as adult patients with MPS disorders. The genetic counseling process will be explained. Reproductive options that may be considered by couples at increased risk of having a child with a genetic disorder will be reviewed.

Gaining Clinician and Research Insights—Ask the Doctor

1:00–1:30 PM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Speakers

Emma Canepa

Emma Canepa, MS, CCRP, is a program manager in the Center for Maternal Fetal Precision Medicine at University of California, San Francisco. Emma's work in clinical research has focused on rare genetic disorders and spanned fetal, pediatric, and adult studies from both industry- and investigator-initiated protocols. She received her BA in molecular and cell biology with a focus on immunology from the University of California, Berkeley, and her MS in health policy and law from UCSF/UC Hastings.

Prenatal Enzyme Replacement Therapy for MPS and Other Lysosomal Storage Disorders

3:20 PM, SEPT. 29, REGENCY BALLROOM

Optimal therapeutic response to lysosomal storage disease may reasonably require in utero therapy. Utilizing FDA-approved enzyme replacement therapy (ERT) treatments, we have embarked on a phase I in utero clinical trial of lysosomal storage diseases including for multiple MPS syndromes. We will review the emerging phase I clinical trial in utero ERT for fetuses diagnosed with MPS disorders, and provide an overview of the study rationale, as well as potential risks and benefits of enrollment. Presented data will include a review of the interim data from the first three treated participants, including pregnancies affected by both MPS I and MPS II.

Speakers

Margret Casal

Margret Casal, DVM, MS, PhD, is specialized in veterinary genetics, pediatrics, and reproduction. She is professor of medical genetics, reproduction, and pediatrics at the University of Pennsylvania School of Veterinary Medicine. She earned her DVM from the University of Zürich, Switzerland, her MS from University of Bern, Switzerland, and her PhD (pathology) from the University of Pennsylvania. Dr. Casal is board certified in clinical animal reproduction with the European College of Animal Reproduction. Following her PhD research on MPS clinical presentations in animal models and the influence of background genetics, she has pursued an active career in comparative medical genetics with a focus on treatment interventions. A current focus is the clinical and cognitive responses to gene therapy and other treatment modalities in the dog models of MPS types I, VI, and VII, lysosomal storage diseases that are quite devastating in humans. These studies are performed in collaboration with other researchers at the University of Pennsylvania as well as colleagues outside of the university.

Progress in MPS VI Research with the Canine Model

1:55 PM, SEPT. 29, REGENCY BALLROOM

The canine model of MPS VI is an extremely attractive model system to both assess general pathology and therapy for bone disease in the MPS diseases generally, as well as for specific MPS VI issues of disease and treatment. This session will outline two ongoing programs utilizing the canine MPS VI model to 1) assess autologous hematopoietic stem cell transplant with *ex vivo* lentiviral treated cells and 2) assess the compound odiparcil to treat canine MPS VI disease.

Speakers

Erik Cline

With 23 years of leadership in the bio/pharma industry with a focus on helping patients and their caregivers, Mr. Cline's work includes both FDA-approved treatments and early stage therapies across central nervous system therapeutic areas and rare disease. In his most recent role, leading U.S. marketing for Takeda's lysosomal storage disorders franchise, Mr. Cline's work focuses on supporting patients and their families affected by MPS II and Gaucher disease.

Takeda Update

7:45 AM, SEPT. 29, REGENCY BALLROOM

In addition to an overview of Takeda's research and development efforts, this session will provide information about services available for certain patients who are receiving a Takeda prescription medication. Exciting ways that the company is collaborating with the MPS II community to help reduce the time to diagnosis and improve access to care will also be discussed.

Speakers

Stephanie Cozine

Stephanie Cozine is an MPS I mother from Delaware. Her son Ethan was diagnosed in 2016 and received a bone marrow transplant a few months later. She attended her first family conference in 2017 and fell in love with the National MPS Society's members and mission. In 2018, she attended her first advocacy trip to Capitol Hill during Rare Disease Week. On this trip she met many incredible people and felt if they could share their stories, they could inspire so many more. Her passion for advocacy grew, primarily in the area of newborn screening at both the state and federal levels. This was the inspiration for the National MPS Society's *Our Voices* podcast. Stephanie has been serving on the board of directors since 2020, and continues to serve as Advocacy Committee chair.

Youth Advocacy Workshop

2:00–3:00 PM, SEPT. 29

Stephanie Cozine and Zach Thomas, youth advocate, will discuss how to develop and share your story to pursue changes in both state and federal laws. Activities will include the infamous "School House Rock" and relating where your story can make an impact in government processes. We will take time to help each youth develop and practice how to share their story with lawmakers. Materials will be available for elementary, middle, and high school levels. Visual and adaptive supports will be available. Recommended ages 5+; parents must attend for youth under age 15.

Camp Courage Youth Advocacy

3:15–4:00 PM, SEPT. 29, OLD GEORGETOWN/CONGRESSIONAL

Stephanie Cozine will present the basic concepts of sharing your story and needs with legislators and how that relationship can foster changes in laws. This session will include talks about how a bill becomes law, exploring the basics of government, and a fun activity to engage our young advocates and learners. Concepts developmentally appropriate for youth age 5+; and activity developmentally appropriate for all ages and abilities. Recommended ages 3+.

State Advocacy Workshop

4:25 PM, SEPT. 30, REGENCY BALLROOM

Stephanie Cozine and Carl Kapes, board of directors Advocacy Committee co-chairs, will discuss the importance of state advocacy, and the need for stronger networks and better relationships with state governments. This session will give a brief overview and update of the National MPS Society's newborn screening efforts across the nation, including basic concepts of newborn screening and obstacles faced. Presenters will encourage and guide advocates to develop relationships with their state legislators, as well as explain how their story can make an impact. This "How to" guide to making positive changes in your state will empower advocates. Those interested are encouraged to also attend Barbara Burton's presentation, "Newborn Screening—Changing the landscape for MPS," and "How to Share Your Story—Making an Impact on Capitol Hill." Registration is recommended. State-specific information will be provided to each advocacy unit to make getting started easier.

Speakers

Kate Delaney

BioMarin Pharmaceuticals: BioMarin's Partnership with the MPS Community

12:45 PM, SEPT. 30, REGENCY BALLROOM

Patricia Dickson

Patricia Dickson, MD, is a pediatrician and medical biochemical geneticist with active board certifications in both specialties. She received her bachelor's degree in classics from the University of Chicago, and her medical degree from Columbia University College of Physicians and Surgeons in New York. Her main research focus is intra-cerebrospinal fluid (intraventricular and intrathecal) enzyme replacement therapy (ERT) for the mucopolysaccharidoses, with the goal of finding new and better treatments for patients. Her laboratory conducts bench-to-bedside research into treatments for MPS disorders. She currently is the centennial professor of pediatrics and genetics and the division director of Genetics and Genomic Medicine at the Washington University School of Medicine Department of Pediatrics.

Progress in GlcNAc-1-Phosphotransferase Research and Its Implications for Therapy

3:35 PM, SEPT. 29, REGENCY BALLROOM

The molecular machinery of the cell that tags lysosomal enzymes for delivery is a critical step in the lifetime of enzymes. Recently, significant advances have been made in understanding the structure and function of the GlcNAc-1-phosphotransferase, which is a critical enzyme-modifying enzyme that tags lysosomal enzymes with a mannose-6-phosphate, which targets enzyme to the lysosome, as well as being the key molecular tag that allows for cross correction. Exploiting this newfound understanding opens up important therapeutic opportunities for much more efficient enzyme replacement therapy, as well as offering a potential path to therapy for ML II/III.

Mucolipidosis—Understanding the Research

1:00 PM, SEPT. 30, REGENCY BALLROOM

Speakers

Annamarie Dillon

Annamarie Dillon is passionate about partnering with and learning from rare disease patient communities. During her 17+ years in rare disease patient advocacy roles within industry, she continually seeks to incorporate the patient perspective by soliciting input to improve understanding of the patient journey to support decision making, including clinical trial design and data generation for regulatory and health technology assessment submissions. She embraces opportunities to foster mutually beneficial partnerships with patient organizations in areas of common interest, such as disease awareness, diagnostic initiatives, capacity building, and supporting networking opportunities. Annamarie currently serves as executive director of patient advocacy at Orchard Therapeutics and prior to this held similar roles at ProQR Therapeutics and Genzyme.

Orchard Therapeutics: Hematopoietic Stem Cell Gene Therapy

12:15 PM, SEPT. 30, REGENCY BALLROOM

Orchard Therapeutics' vision is to end the devastation caused by genetic and other severe diseases. They aim to do this by discovering, developing, and commercializing new treatments that tap into the transformative potential of hematopoietic stem cell (HSC) gene therapy. In this approach, a patient's own blood stem cells are genetically modified outside of the body and then reinserted, with the goal of correcting the underlying cause of disease in a single treatment. Today, Orchard is advancing a pipeline spanning pre-clinical, clinical, and commercial stage HSC gene therapies designed to address serious diseases, including MPS I and MPS IIIA, where the burden is immense for patients, families, and society, and current treatment options are limited or do not exist.

Speakers

Takayo Egawa

Takayo Egawa joined JCR Pharmaceuticals in 1981 with a role in business communications and transactions with non-Japanese companies. Her current role as director of International Affairs encompasses multiple activities, including alliance management of non-Japanese business partners and, notably in the last decade, relationship building with patient advocacy groups in the lysosomal storage disorders. She values communication with patient families to whom her deepest respect goes, along with caregivers, healthcare professionals, and researchers devoted to the rare disease fields.

JCR Pharmaceuticals Update

12:00 PM, SEPT. 30, REGENCY BALLROOM

What is JCR Pharmaceuticals? What do we do? What have we achieved so far? How can we deliver on our promises? Since its inception in 1975, JCR Pharmaceuticals has focused on the research, development, and manufacture of biotherapeutics. Our engagement in lysosomal storage disorders (LSDs) led to the discovery and establishment of a blood-brain barrier penetrating technology, which is applied to multiple programs in our LSD pipeline for Japan and the rest of the world. In this session you will learn the story behind what led us to where we are today.

Speakers

Julie Eisengart

Julie Eisengart is an associate professor of pediatrics at the University of Minnesota Medical School and director of the Neurodevelopmental Program in Rare Disease. As a pediatric neuropsychologist, she specializes in rare neurodegenerative disorders of childhood, evaluating functional course of disease and response to therapy. Her research has focused on the MPS disorders, particularly examining outcomes of early diagnosis, newborn screening, and novel therapies. Her research extends to defining and measuring aspects of disease that are under-represented in the clinical and research communities but are important and meaningful to patients and families, such as neurobehavioral symptoms and the family experience.

Dr. Eisengart has been a principal or co-investigator and research mentor on a number of observational studies or clinical trials for MPS and other rare diseases. She has served on the Technical Expert Panel for the U.S. Secretary of Health and Human Services to consider the addition of MPS II to the Recommended Uniform Screening Panel, and on the Working Group for the Minnesota Department of Health to implement newborn screening for MPS I and other rare disorders. Presently she sits on the National Organization for Rare Disorders Training Work Group and the Critical Path Institute Rare Disease Clinical Outcome Assessment Consortium.

Expanding the Understanding of Function in MPS: Neurocognitive Endpoints and Beyond

8:45 AM, SEPT. 30, REGENCY BALLROOM

MPS disorders are associated with a wide spectrum of neurocognitive effects, from progressive neurocognitive and neurobehavioral decline to “invisible” challenges with attention and mental health. Studies of neurocognition are necessary to determine the rates of change, which are crucial to select endpoints for clinical trials of brain treatments. Increased representation of the community’s functional needs is also vital. This presentation will address more than just “IQ” or cognitive/intellectual level, and will expand focus on the many ways a person functions, including cognition, communication, attention, independence skills, behaviors, social skills, and more. Challenges must be understood not exclusively as MPS disease effects on the brain, but also as the complex multi-system experience of living with MPS. Supporting the functioning of all people with MPS and their families requires increased scientific and clinical understanding of this complexity.

Speakers

N. Matthew Ellinwood

N. Matthew Ellinwood, DVM, PhD, has longstanding research experience with MPS and ML. He has worked with the National MPS Society for more than 20 years, and currently serves as its chief scientific officer guiding research to benefit patients. He is a professor emeritus at Iowa State University, where he conducted basic and applied research on the neuropathic mucopolysaccharidoses.

MPS Therapy, Newborn Screening, and the RUSP: An Overview

8:10 AM, SEPT. 29, REGENCY BALLROOM

Optimal therapy for MPS and ML II/III disorders requires, among other things, initiation of therapy at the earliest possible age. This makes newborn screening a vital tool in addressing unmet clinical need in these disorders. However, there is complex interdependence of therapy development and approval and newborn screening adoption. This overview will present in a historical fashion the accomplishments of the Society, and the continued unmet needs and institutional and regulatory barriers to overcome.

Advocacy, Policy, and Research Recap

5:10 PM, SEPT. 29, REGENCY BALLROOM

Sanfilippo—Advancing Clinical Studies to Therapies, What Can We Do?

1:35 PM, SEPT. 30, REGENCY BALLROOM

Gaining Clinician and Research Insights—Ask the Doctor

2:20–2:50 PM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Speakers

Lisa Garrity

Lisa Garrity is a pharmacy clinical specialist in neurology at Cincinnati Children's Hospital Medical Center, working primarily with outpatients in the Comprehensive Epilepsy Clinic. She graduated from Miami University in 2001 with a bachelor of science in biochemistry, earned a master of science in chemistry in 2004 at the Massachusetts Institute of Technology, and a doctorate in pharmacy from the University at Buffalo in 2008. She completed a pharmacotherapy residency in 2010 at the Cleveland Clinic. Her areas of interest include status epilepticus, care of women with epilepsy, and improving medication access for patients with epilepsy. She also has lead work on developing policies for medical marijuana and cannabidiol and has presented at national meetings on use of cannabinoids in epilepsy.

Cannabidiol, Medical Marijuana, and Pain Management: Considerations for MPS

10:00 AM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Cannabidiol (CBD) and medical marijuana have been touted as "all natural" cures for epilepsy, pain, anxiety, sleep issues, and many other problems. Despite the promise of cannabidiol benefit for seizures, there are many issues to consider around use of CBD and medical marijuana, including variability in products, legal status, drug interactions, and side effects. This session will review the differences between prescription and artisanal CBD products, as well as potential concerns associated with use of medical marijuana. We will also discuss various over-the-counter and prescription medications that may be utilized for pain associated with MPS.

Amy Gaviglio

Amy Gaviglio, MS, CGC, is a certified genetic counselor and founder of Connetics Consulting, which provides public health genetics, genomics, and rare disease services across the country. She has been working in the areas of newborn screening and rare disease for the past 15 years. Amy currently works with the Centers for Disease Control and Prevention's Newborn Screening and Molecular Biology Branch, the Association of Public Health Laboratories (APHL), Expecting Health, Gillette Children's Hospital, and several other rare disease organizations. She is co-chair of APHL's New Disorders in Newborn Screening workgroup and is a member of additional national groups, including the Rare Disease Diversity Coalition and EveryLife Foundation's Community Congress. She also serves as an advisor for the Midwest Genetics Network and is on the MPS Society's Scientific Advisory Board. Additionally, Amy serves as chair of the Newborn Screening Expert Panel for the Clinical and Laboratory Standards Institute and is chair of Minnesota's Rare Disease Advisory Council.

The Journey to Newborn Screening: Pilots, Legislation, RUSP, and Implementation

8:50 AM, SEPT. 29, REGENCY BALLROOM

The path to adding a disease to newborn screening is a multi-step journey requiring the coordination of numerous partners. This presentation will walk through the process of newborn screening advocacy—from pilots to the federal Recommended Uniform Screening Panel (RUSP) to state-based legislation and universal implementation.

In order for a disease to be considered for newborn screening, it must meet certain criteria—including the development of a suitable assay, an understanding of the natural history of the disease, and an effective presymptomatic treatment. Pilot studies can help generate the evidence needed for a disease to meet these criteria and are usually performed through a consent-based model on a limited number of families. From there, advocates can choose to nominate the condition to be added to the RUSP or work on state-based legislation requiring screening. Often, both paths are utilized, with the RUSP process being preferred with supplemental legislation to assist programs in funding and other resources needed to implement universal screening.

Implementation of universal screening within a program requires a phased approach that spans pre-analytical, analytical, and post-analytical components. Advocates are poised to assist in many aspects, and successful implementation ensures that families are supported throughout the entirety of the newborn screening, diagnostic, and therapeutic processes.

Speakers

Natalia Gomez-Ospina

Natalia Gomez-Ospina, MD, PhD, is a board-certified medical geneticist and assistant professor in the Department of Pediatrics at Stanford University. As a physician-scientist, she brings passion and expertise to the clinic and the laboratory to diagnose and treat genetic diseases, particularly lysosomal storage disorders (LSDs). She manages the enzyme replacement service at Lucile Packard Children's Hospital and leads the Program for Inherited Metabolic Disorders at Stanford's Center for Definitive and Curative Medicine which strives to advance gene- and cell-based therapies for LSDs. One of Dr. Gomez-Ospina's main scientific contributions is commandeering the hematopoietic system to express proteins needed in other organs, including the brain. Her groundbreaking work includes an adaptable platform for treating lysosomal enzyme deficiencies and pioneering a first-of-its-kind preclinical study for autologous transplantation of genome-edited cells to treat patients with MPS I (Hurler syndrome). Beyond this, her platform holds tremendous promise for delivering lysosomal enzymes to the brain and other organs to treat other LSDs.

Despite her relatively young career, Dr. Gomez-Ospina has already made significant contributions, with lead authorship in prestigious research publications like the *New England Journal of Medicine*, *Cell*, *Nature Communications*, and the *American Journal of Medical Genetics*. Her achievements have not gone unnoticed, as she has received several prestigious awards, including the Outstanding Young Investigator Award from the American Society for Cell and Gene Therapy, the Young Scientist Award from the American Society for Clinical Investigation, the Young Investigator Award from the Western Society for Pediatric Research, and the prestigious William K. Bowes Jr. Award in Medical Genetics from Partners HealthCare Personalized Medicine.

Human Genome-Edited Hematopoietic Stem Cell-Based Therapy for MPS I

4:05 PM, SEPT. 29, REGENCY BALLROOM

Current treatments for MPS I are not very effective and can at best slow down disease progression. Two common treatments are enzyme replacement therapy (ERT) and blood stem cell transplantation. However, ERT does not help with neurological symptoms, and blood stem cell transplantation has challenges, including the need for donors, delays in treatment, and limited effects on certain complications. We are exploring a novel strategy: genetically modifying the patient's own blood stem cells using CRISPR/Cas9 to make them produce higher levels of the enzyme iduronidase. This approach has potential advantages, including improved safety and effectiveness compared to transplants from a genetically distinct donor.

Our previous research demonstrated success in mice, where genome edited human blood stem cells produced increased enzyme levels and ameliorated MPS I symptoms. The goal now is to conduct a phase I/II clinical trial of these edited human cells. We currently are preparing for the Investigational New Drug submission, which involved establishing a manufacturing process that meets quality standards and ensuring the edited cells are safe and stable through rigorous testing.

This innovative approach, known as gtCCR5-MPS1, holds promise for a safer, more effective MPS I therapy. Additionally, the research may contribute valuable insights applicable to other similar genetic disorders. We aim to leverage our findings to advance the treatment landscape for MPS I and related MPS disorders.

Speakers

Paul Harmatz

Paul Harmatz, MD, is professor in residence, Department of Pediatrics, University of California, San Francisco and UCSF Benioff Children's Hospital Oakland. He is medical director of the Pediatric Clinical Research Program in MPS and Related Disorders. Dr. Harmatz completed his pediatric internship and residency training at Harbor-UCLA Medical Center. Following a clinical and research fellowship in pediatric gastroenterology and nutrition at Massachusetts General Hospital, he remained in Boston until 1992 as faculty in pediatrics at Harvard Medical School. During the last 20 years, Dr. Harmatz has participated in clinical trials with MPS I, MPS II, IIIA, IIIB, IVA, VI, and VII, and has managed clinical care for patients with MPS living in northern California.

Growth Failure in MPS Disease

1:22 PM, SEPT. 29, REGENCY BALLROOM

Many of the MPS syndromes with bone involvement also have severe growth restrictions which impacts many physiological systems. Understanding the mechanism of this impact may offer an improved diagnosis and treatment of this aspect of disease.

Understanding Gene and Cell Therapies

11:25 AM, SEPT. 30, REGENCY BALLROOM

Gaining Clinician and Research Insights—Ask the Doctor

1:30–200 PM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Mercedes Ramirez Johnson

In 1995, Mercedes Ramirez Johnson narrowly survived a commercial airplane crash that killed 160 people, including her parents. She presents her proprietary Second Chance Living concept, an innovative mindset and approach with a proven track record, to organizations such as General Electric, Exxon Mobil, Microsoft, NASA, Chevron, and various branches of the U.S. Armed Forces.

Before becoming a speaker, Mercedes spent nearly a decade in high-level sales in the pharmaceutical and medical software industries. A record-breaking, multi-million dollar producer, she became the youngest female and sole Latino account executive at Cerner Corporation. She is an active leader in philanthropic organizations advocating for special needs children and children's health initiatives, notably for Make-A-Wish and the National MPS Society.

Mercedes and her story have been the subject of considerable national and international media coverage.

Her extensive media experience includes appearances on the BBC, National Geographic Channel, Discovery Health Channel, and as a recurring guest on numerous nationally syndicated daytime talk shows on NBC and ABC. In addition, numerous national magazines and major-market newspapers have run cover stories and special-interest pieces about her, and she was recognized as one of the country's top young Hispanic up-and-comers by *People Magazine*'s Spanish edition, *People en Español*.

Mercedes and her children reside in Dallas, TX.

How to Share Your Story: Making an Impact on Capitol Hill

4:05 PM, SEPT. 30, REGENCY BALLROOM

Speakers

Simon Jones

Simon Jones, MBChB, BSc, MRCPCH, was born in Northern Ireland and attended Edinburgh University Medical School, qualifying with a BSc in neurosciences. He moved to London and trained in pediatrics at Guy's and St. Thomas' Hospital. Dr. Jones has been working at the Willink Biochemical Genetics Unit, now part of Genetic Medicine at St. Mary's Hospital, Manchester, since September 2005, where he is a consultant in pediatric inherited metabolic disease as well as honorary professor of pediatrics and translational medicine. He has been principal investigator or actively involved in many phase I–IV international multi-center trials of both gene therapy and enzyme replacement therapy for lysosomal storage disorders. Since 2008 he has been a consultant in pediatric inherited metabolic diseases at the Willink Unit. Dr. Jones also serves as medical director for the National Institute for Health Research Clinical Research Facility at NIHR.

Ex vivo Autologous Stem Cell Gene Therapy Clinical Trial for MPS IIIA: Update on Phase I/II Clinical Trial

4:25 PM, SEPT. 29, REGENCY BALLROOM

MPS IIIA is among the most challenging MPS disorders, both in terms of clinical management as well as a target for an effective therapy. Utilizing a platform involving *ex vivo* autologous stem cell gene therapy, we will report on the most current available update of a clinical trial involving OTL-201 as a treatment for MPS IIIA.

Speakers

Carl Kapes

Carl Kapes works as an electrical engineering manager for Burns Engineering in Philadelphia, PA. He attended Villanova University where he majored in electrical engineering and played on the Wildcats baseball team. He has raised more than $1 million for Sanfilippo syndrome through various fundraisers. Carl summitted Mt. Kilimanjaro in 2012, Mt. Rainier in 2016, and Pico de Orizaba in Mexico in 2018, all as fundraisers. In addition to the National MPS Society, Carl is also a board member of the Team Sanfilippo Foundation and Naamans Little League in Wilmington, DE. He is dad of Ryan (forever 16, MPS IIIA), Brayden (15, MPS IIIA), Bryce (7), and Brooklyn (5).

State Advocacy Workshop

4:25 PM, SEPT. 30, REGENCY BALLROOM

Stephanie Cozine and Carl Kapes, board of directors Advocacy Committee co-chairs, will discuss the importance of state advocacy, and the need for stronger networks and better relationships with state governments. This session will give a brief overview and update of the National MPS Society's newborn screening efforts across the nation, including basic concepts of newborn screening and obstacles faced. Presenters will encourage and guide advocates to develop relationships with their state legislators, as well as explain how their story can make an impact. This "How to" guide to making positive changes in your state will empower advocates. Those interested are encouraged to also attend Barbara Burton's presentation, "Newborn Screening—Changing the landscape for MPS," and "How to Share Your Story—Making an Impact on Capitol Hill." Registration is recommended. State-specific information will be provided to each advocacy unit to make getting started easier.

Annie Kennedy

Annie Kennedy is chief of policy, advocacy, and patient engagement at EveryLife Foundation for Rare Diseases. Focused on improving health outcomes for people living with rare diseases by advancing the development of treatment and diagnostic opportunities for rare disease patients through science-driven public policy, Annie's work includes building strong partnerships with policy makers, federal agencies, industry, and alliances. She has served within the community for nearly three decades through her roles with Parent Project Muscular Dystrophy and the Muscular Dystrophy Association. During that time, she helped lead legislative efforts around passage and implementation of the MD-CARE Act (2001, 2008, 2014) and the Patient-Focused Impact Assessment Act, which became the Patient Experience Data provision within the 21st Century Cures Act (section 3001). She has engaged with the Food and Drug Administration and industry around regulatory policy and therapeutic pipelines, led access efforts as the first therapies were approved for Duchenne muscular dystrophy, and engaged with the Institute for Clinical and Economic Review around the development of the modified framework for the valuation of ultra-rare diseases.

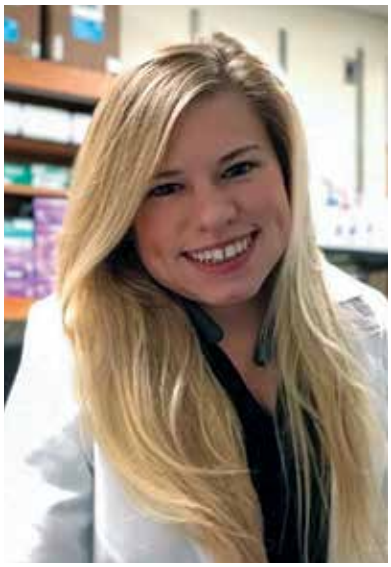
Annie's community roles include service on the board of directors of Cure SMA, the PFDD Works coalition, the Patient-Driven Values in Healthcare Evaluation Steering Committee, FasterCures Cures for Life initiative, the National Health Council's PCORI Valuation Group, the Innovation and Value Initiative Patient Advisory Committee, the National Duchenne Newborn Screening Pilot Program Steering Committee, the Institute for Gene Therapies Patient Advocacy Advisory Council, the State Rare Disease Education Initiative National Steering Committee, and as a member of the National Institutes of Health's National Center for Advancing Translational Sciences Advisory Council and the Cures Accelerator Network Advisory Board.

Challenges in FDA Structure and Practice Confronting the Rare Disease Community

10:30 AM, SEPT. 29, REGENCY BALLROOM

The rare and ultra-rare disease communities are confronted with multiple challenges when one considers the process of FDA drug approvals. These challenges involve extremely small patient populations, patient clinical trial recruitment challenges, and efficacy endpoints. Coupled with these challenges is the extreme nature of the clinical need in this community. To move forward in an efficient and ethical way, new ways of considering trial structure and endpoints needs to more effectively incorporate both expert opinion and patient experience. This presentation will outline some of these challenges and consider past and potential future successes.

Speakers



Jenny Klein

Jenny Klein is a rare disease patient battling ML III and a research scientist. She received undergraduate degrees in human biology and psychology and a master's degree in physiology from North Carolina State University. Upon graduation, she entered the biotech industry where she expanded the drug discovery pipeline for rare diseases at Collaborations Pharmaceuticals, Inc. Jenny currently serves as head of operations and program management at Odylia Therapeutics, Inc. where she is developing gene therapy products from early pre-clinical through phase I clinical trial for rare diseases. In 2021, Jenny earned her 200-hour yoga teacher training in Vinyasa Flow from YOGA SOUL·LEC·TIVE in Raleigh, NC.

Strengthening Self-Reported Health Outcomes and Quality of Life Through Movement

3:40 PM, SEPT. 30, REGENCY BALLROOM

Vinyasa Flow is a yoga practice focused on breath to movement. Jenny cites this, in combination with restorative yoga and weight lifting, as playing a critical role in maintaining her health and well-being. As a graduate student once focused on anatomy and physiology, Jenny has taken what she's learned in the classroom and incorporated it into her active daily lifestyle. Learn more on how low impact movements have shaped Jenny's health outcomes for the better.

Speakers

Nathalie Marie

Nathalie Marie, MD, was born and raised in Miami, FL, and currently resides outside of Boston, MA. She completed her BA degree at Boston College in Massachusetts where she double majored in philosophy and pre-medical sciences. There she graduated magna cum laude and was the recipient of the Dean's Scholar Award. She attended medical school at the Miller School of Medicine in Miami, FL, and was the recipient of the Distinction in Psychiatry Award, selected by faculty as having both academic and clinical distinction within a graduating class. Dr. Marie completed her psychiatric residency in 2008 in Houston, TX, at Baylor College of Medicine Menninger Department of Psychiatry. She received multiple distinctions during her residency training including the Association of Women Psychiatrist Alexandra-Simmons Fellowship Award for distinction in women's mental health and the Dr. Henry Page Durkee Laughlin Foundation Merit Award. After residency, Dr. Marie served the veteran population for six years at the Michael E. DeBakey VA Medical Center where she also held dual appointment with Baylor College of Medicine. There she worked as an outpatient psychiatrist within the Trauma and Recovery Program specializing in post-traumatic stress and anxiety disorders, as well as other mood disorders. In 2011, she was a Federal Executive Horizon Award Nominee in the category of leadership. Dr. Marie has since worked in community psychiatry and in private practice and currently works as a telehealth outpatient psychiatrist. She holds special interests in trauma, women's mental health, and in working with adults with differing abilities and their families. She is fluent in three languages, enjoys ballroom dancing, travel, and spending time with her husband and 11-year-old son who was diagnosed with MPS II at age 3.

Newly Diagnosed Family Dinner

6:00 PM, SEPT. 28, OLD GEORGETOWN/CONGRESSIONAL

Speakers

Marielle Marinoff

MPS II mother

Taking Control: Fundraising for Critical Research

3:20 PM, SEPT. 30, REGENCY BALLROOM

This presentation will focus on how families can use a terrible, life-changing diagnosis of a loved one as the driving motivator to make a significant difference in disease research through awareness campaigns and fundraising. With awareness comes public interest. With interest comes fundraising for research. Research yields improved treatments and hope for a cure.

Speakers

Peter Marks

Peter Marks, MD, PhD, is the director of the Center for Biologics Evaluation and Research (CBER) at the U.S. Food and Drug Administration. The center is responsible for assuring the safety and effectiveness of biological products, including vaccines, allergenic products, blood and blood products, cellular, tissue, and gene therapies.

Dr. Marks received his graduate degree in cell and molecular biology and his medical degree at New York University. Following this, he completed an internal medicine residency and hematology/medical oncology fellowship at Brigham and Women's Hospital in Boston, where he subsequently joined the attending staff as a clinician-scientist and eventually served as clinical director of hematology.

He then worked for several years in the pharmaceutical industry on the clinical development of hematology and oncology products prior to returning to academic medicine at Yale University where he led the Adult Leukemia Service and served as chief clinical officer of Smilow Cancer Hospital. He joined the FDA in 2012 as deputy center director for CBER and became center director in 2016. Dr. Marks is board certified in internal medicine, hematology, and medical oncology, and is a fellow of the American College of Physicians. In 2022, he became a member of the National Academy of Medicine, one of the highest honors in the fields of health, science, and medicine.

Addressing the Unmet Needs of the Rare Disease Community

11:20 AM, SEPT. 29, REGENCY BALLROOM

Out of thousands of rare hereditary diseases there are hundreds of disorders affecting one to a few hundred individuals per year. Structuring approaches and platforms to address the critical unmet needs of those with rare diseases is a specific focus of multiple programs and approaches both at the FDA and in partnerships between the FDA and other entities. This presentation will outline these current approaches which reflect the thinking and actions of the FDA as it seeks to better serve rare disease communities.

Deborah Marsden

Deborah Marsden, MD, is a pediatrician with specialty training in clinical biochemical genetics (Fellow of the American College of Genetics and Genomics). She graduated from the University of New South Wales Medical School in Sydney, Australia, with postgraduate training in Australia and the UK. She also completed a fellowship in biochemical genetics at the University of California, San Diego, and joined the faculty there for several years.

After moving to New England, Dr. Marsden became the medical director of the New England Newborn Screening Program, introducing expanded newborn screening for multiple rare inborn errors of metabolism. She was subsequently the medical director of the Metabolic Clinic (Department of Genetics and Genomics) at Boston Children's Hospital. She moved to industry 12 years ago, working on rare genetic disorders in medical affairs and clinical development. She also serves as a part-time staff physician in the Metabolic Clinic and Lab Medicine at Boston Children's Hospital, and as a corresponding member of the faculty at Harvard Medical School. She is currently employed by Ultragenyx Pharmaceutical as global medical expert for genetic metabolic disorders, Medical Affairs.

Ultragenyx Update

12:30 PM, SEPT. 30, REGENCY BALLROOM

Speakers

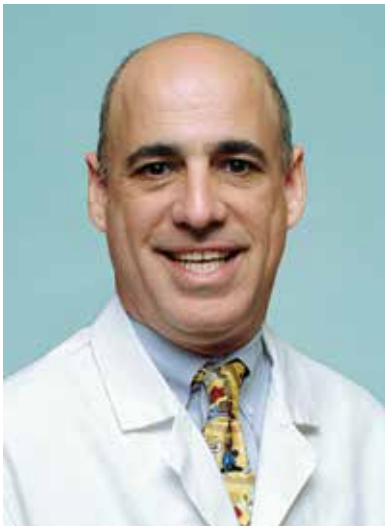
Heather Mazzota

Heather Mazzotta, RN, is a senior patient education liaison (PEL), Rare Disease, at Sanofi. She has been a nurse for 30 years and a PEL for almost seven years. Heather's territory covers South Jersey and half of the state of Pennsylvania, including Philadelphia.

Sanofi Update

7:30 AM, SEPT. 29, REGENCY BALLROOM

This session will provide an introduction to the CareConnectPSS Team and will discuss how they can support patients at various points in their diagnostic and treatment journey.



David Molter

David Molter, MD, is board certified in otolaryngology-head and neck surgery, and in medical informatics. His primary research interests include applications of biomedical engineering and computer science. His clinical interests include airway management and reconstruction, management of patients with complex medical needs, velopharyngeal imaging and reconstruction, aerodigestive care, cleft palate and craniofacial concerns, pediatric sleep apnea, management of ear, nose, and throat issues in MPS patients, hearing disorders, and juvenile laryngeal papilloma.

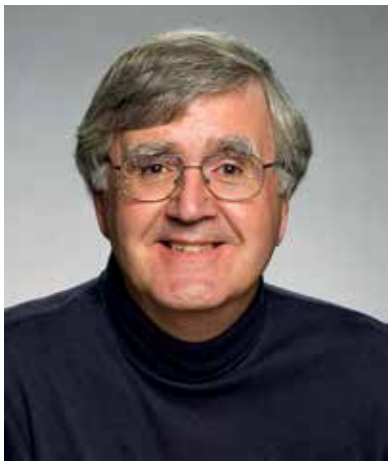
Dr. Molter graduated from medical school in 1988 and completed his residency in otolaryngology in 1994, both at Duke University School of Medicine in Durham, NC. He then completed a pediatric otolaryngology fellowship at Cincinnati Children's Hospital and joined the faculty at the University of Maryland, where he taught for five years.

In 2000, Dr. Molter moved to the Department of Otolaryngology at the Washington University School of Medicine. He currently is a professor in the Division of Pediatric Otolaryngology at St. Louis Children's Hospital. He has been listed in *Best Doctors* yearly since 2007. Dr. Molter's publications are primarily related to cleft and craniofacial concerns, clinical MPS management, and the pediatric airway.

ENT and Airway Issues Associated with MPS

9:30 AM, SEPT. 30, REGENCY BALLROOM

Speakers



Joseph Muenzer

Joseph Muenzer, MD, PhD, is the Bryson Distinguished Professor in Pediatric Genetics and a Professor in the Department of Pediatrics and Department of Genetics at the University of North Carolina at Chapel Hill, where he has practiced since 1993. He received an MD (1976) and PhD in biochemistry (1979) from Case Western Reserve University in Cleveland, OH. He completed a residency in pediatrics at the University of Wisconsin Hospitals, Madison, and a genetic/endocrine fellowship at the National Institute of Child Health and Human Development, NIH, in Bethesda, MD. Dr. Muenzer is director of the recently created Joseph Muenzer MPS Research and Treatment Center at the University of North Carolina at Chapel Hill. He has been the chairperson for the North Carolina Newborn Screening Advisory Committee for more than 28 years.

Dr. Muenzer is involved in the diagnosis, management, and treatment of patients with inborn errors of metabolism, especially MPS, and newborn screening for MPS I and MPS II. He is board certified in pediatrics and in clinical biochemical/molecular genetics. He has been actively involved in developing new treatments for MPS disorders his entire professional career. He has created a mouse model for Hunter syndrome that has been widely used to develop new treatment for MPS II. He has been a principal investigator for IV enzyme replacement clinical trials (ERT) for both MPS I and MPS II resulting in FDA approval. His recent clinical research has focused on the development of new treatments for brain disease in MPS. He has been the principal investigator for more than 20 MPS clinical trials/observational studies. Dr. Muenzer currently is the principal investigator for phase I/II and phase II/III intrathecal enzyme replacement clinical trials for MPS II, a phase I/II gene editing clinical trial for MPS II, and phase I/II and phase II/III IV ERT clinical trials to treat brain disease in MPS II.

Newly Diagnosed Family Dinner

6:00 PM, SEPT. 28, OLD GEORGETOWN/CONGRESSIONAL

ML II/III and MPS Clinical Overview and Clinical Trial Updates

8:00 AM, SEPT, 30, REGENCY BALLROOM

An overview of MPS and ML II/III will be presented, including clinical features of the common MPS disorders, diagnosis, and current treatment options. The status of current clinical trials for the MPS disorders will be presented, including *ex vivo* gene therapy, intrathecal gene therapy, intrathecal enzyme replacement therapy, enzyme replacement using gene engineered B cells, and intravenous enzyme replacement therapy using enzymes engineered to cross the blood-brain barrier.

Speakers

Paul Orchard

Dr. Paul Orchard is the medical director of the Inherited Metabolic and Storage Disease Program, and a professor in the Department of Pediatrics in the Division of Blood and Marrow Transplant & Cellular Therapy at the University of Minnesota. He is interested in the use of hematopoietic stem cell transplantation and other cell therapies, including gene therapy, to improve outcomes. In addition to his clinical work with patients who have inherited metabolic diseases, Dr. Orchard is engaged in research designed to identify strategies that enhance the delivery of enzymes to the brain and the peripheral nervous system for patients who lack specific enzymes. Other interests include the modification of stem cell transplant approaches and combination therapies to improve outcomes for patients with inherited diseases, and the potential to develop multi-institutional cooperative studies for these disorders.

Considerations for HSCT Transplant

8:10 AM, SEPT. 29, PATUXENT/EMBASSY

Speakers

Laura Pisani

Laura Pisani, MD, senior medical director of Global Clinical Development at REGENXBIO, is a passionate rare disease expert and advocate, bringing her clinical and academic background to clinical development. Laura received her BS in medical biotechnology, her MD from Federico II University, Naples, Italy, where she was first exposed to the world of rare diseases and clinical trials by working in Niemann-Pick C disease, and MBA from Fayetteville State University, NC. She completed residency training in pediatrics at Northwell Health, followed by fellowship training in medical genetics at the Icahn School of Medicine at Mount Sinai, both in New York. After working at Columbia University as a metabolic geneticist, she moved back to Northwell Health, where she successfully opened and directed an inherited metabolic disorder newborn screening center. She continued her commitment to rare diseases in her industry work, first at Ultragenyx, then finally working on gene therapy at REGENXBIO to further her commitment to help bring novel therapeutics to children and adults with rare disorders. She is an avid long-distance runner, travel and language enthusiast, and big sci-fi and fantasy fan who can be spotted on a weekend at the start line of a marathon, as well as at a Comic Con, cosplaying with her daughters Freya and Arwen.

REGENXBIO's Investigational Gene Therapies for the Treatment of MPS I and II

7:15 AM, SEPT. 29, REGENCY BALLROOM

Speakers

Carmen Sanchez

Carmen Sanchez, PhD, is a clinical psychologist with a 20-year background driving quality clinical research projects. With a soft spot for rare and genetic diseases, she is an endless, patient-focused learner who immerses fully in each project to grasp the unique challenges faced by rare disease patients.

Managing MPS and ML in Daily Living: Using Tools for Wellness

11:00 AM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Not all coping strategies work for all situations. It is important to have a “tool belt” of coping approaches to use during stressful life events. We will identify your main coping response and discuss successful strategies to add to your skillset.

Speakers

Lachlan Smith

Lachlan Smith, PhD, began his training at the University of Adelaide, Australia, obtaining his undergraduate degree in mechatronic engineering, followed by a PhD in pathology working under the direction of Drs. Nick Fazzalari, John Costi, and Sharon Byers. Subsequently, he completed postdoctoral training in orthopaedic bioengineering at the University of Pennsylvania working with Drs. Dawn Elliott and Rob Mauck before joining the faculty in 2013 as associate professor, Department of Orthopaedic Surgery, University of Pennsylvania Perelman School of Medicine. The focus of Dr. Smith's research program is the pathophysiology and treatment of degenerative and developmental disorders affecting the spine and synovial joints. The scope of his research includes basic mechanistic studies, translational studies in animal models, and clinical studies in human patients. In the translational space, his work bridges the fields of tissue engineering, biomaterials, drug delivery, and stem cells, and is focused on arresting disease progression, restoring spine and joint function, and potentiating long-term tissue regeneration. His lab applies novel, naturally occurring, and inducible large animal models to study disease etiology and evaluate therapeutics.

Leveraging Naturally Occurring Canine Models to Advance Novel Treatments for Skeletal Disease in the Mucopolysaccharidoses

1:05 PM, SEPT. 29, REGENCY BALLROOM

Patients with MPS commonly exhibit progressive skeletal disease that reduces mobility, independence, and overall quality of life. Current treatments exhibit limited efficacy for improving skeletal disease in MPS, particular for bone and cartilage. Naturally occurring canine models represent powerful, clinically relevant platforms to study MPS skeletal disease pathophysiology and to advance novel treatments toward clinical application. Like humans, MPS dogs exhibit progressive spine and joint disease, and a corresponding decline in mobility. Compared to rodent models, the size and lifespan of MPS dogs makes them ideal for long-term treatment studies that incorporate gold standard *in vivo* imaging and functional assessments. This presentation will outline recent studies by Dr. Smith's group that leverage the canine models of MPS I, VI, and VII to advance understanding of spine, bone, and joint natural history and pathophysiology, and to evaluate the safety and efficacy of a range of current and emerging treatment modalities targeting these tissues.

Speakers

Jamie Sullivan

Jamie Sullivan, MPH, joined the EveryLife Foundation for Rare Diseases in July 2020, serving as senior director of policy. She previously served in a variety of roles for the COPD Foundation for more than a decade. Jamie's work has focused on achieving patient-centered federal, state, and regulatory policy changes in the areas of health appropriations, public health, and regulatory infrastructure, and access to care. In her role at the COPD Foundation, Jamie also focused on building programs to engage and train patient advocates, and advocating for the robust inclusion of patient and caregiver perspectives in all aspects of treatment development and review. Prior to joining the COPD Foundation, Jamie supported the research and clinical programs of the Alpha-1 Foundation. She obtained her master's in public health policy and management at Florida International University.

Drug Approval in Rare Disease and the RUSP Bottleneck

8:30 AM, SEPT. 29, REGENCY BALLROOM

The federal Recommended Uniform Screening Panel (RUSP) for newborn screening is a highly complex process. One of the key requirements of submitting a disease condition nomination to the Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, which assesses RUSP nominations, is that there is an accepted therapy to treat the condition in question. For MPS and ML disorders, this very often means a drug approval. Hence the drug approval process of the FDA can, in effect, act as a gatekeeper to RUSP nomination eligibility. This session will explore FDA drug approvals and how they can impact the RUSP process.

Lisa Todd

Lisa Todd lives in Albuquerque, NM, with her husband, Jerry, and their three sons—Jack (MPS II), Jake, and Jaden. Lisa supports the Society by serving as chair for the board of directors and serving on the Governance Committee.

She is a CPA specializing in not-for-profit entities. Through her professional role, Lisa provides training to many types of non-profits on a variety of governance and financial issues. Lisa has been a member of the Society since Jack was first diagnosed with MPS in June 2011 and has been a board member since January 2012.

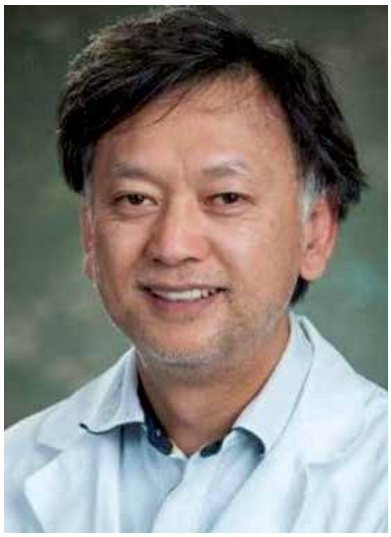
Welcome and Introduction

7:00 AM, SEPT. 29, REGENCY BALLROOM

Annual General Membership Meeting

7:15 AM, SEPT. 30, REGENCY BALLROOM

Speakers



Shunji Tomatsu

Shunji Tomatsu, MD, PhD, received his medical training from Gifu University School of Medicine, Gifu, Japan, where he also earned his PhD training. He currently serves as head of the Skeletal Dysplasia Research Lab, Nemours Children's Hospital, DE. His research career, which spans more than 35 years, focuses on bench-to-bed diagnosis and treatment for MPS, especially type IV (Morquio A syndrome). He and his team are the recent recipients of an award from the Foundation for the NIH (FNIH) as part of the Bespoke Gene Therapy Consortium (BGTC) to pursue a first-of-its-kind gene therapy clinical trial for MPS IVA.

An AAV-Based Clinical Trial for MPS IVA

3:50 PM, SEPT. 29, REGENCY BALLROOM

Current approved therapy for MPS IVA does not completely address disease in clinically diagnosed patients. To fill this need, and in collaboration with the FNIH Accelerating Medicines Partnership® (AMP®) BGTC, we are working toward a first in-human clinical trial treating MPS IVA using AAV gene therapy. The FNIH AMP® BGTC program is a public-private partnership between the National Institutes of Health, U.S. Food and Drug Administration, biopharmaceutical and life science companies, and non-profit and other organizations. Our laboratory and Nemours Children's Hospital were selected as part of this initiative to move gene therapy forward for skeletal diseases like MPS IVA.

Speakers

Klane White

Klane White, MD, MSc, is an internationally recognized expert and advocate in the care of mucopolysaccharidosis and skeletal dysplasia. He serves on the Medical Advisory Board of Little People of America, the Scientific Advisory Board of the National MPS Society, and is an executive founding member of the Skeletal Dysplasia Management Consortium.

In addition to skeletal dysplasia and MPS, Dr. White's clinical and research interests include the diagnosis and management of early onset scoliosis, metabolic bone disease, and complex spine deformity. Dr. White has authored more than 90 peer-reviewed publications, articles, and book chapters, serves as reviewer for multiple medical journals, and is principal investigator in several multicenter research studies for rare disease.

Orthopedic Considerations for HSCT Transplant MPS Patients

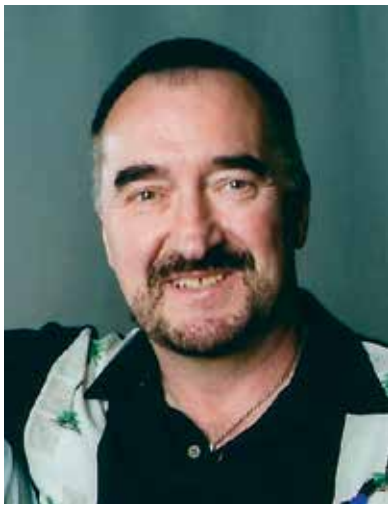
9:00 AM, SEPT. 29, PATUXENT/EMBASSY

Orthopedic Updates in MPS

12:10 PM, SEPT. 29, REGENCY BALLROOM

Skeletal abnormalities are early and prominent features of MPS. The orthopedic surgeon is often the first healthcare provider to raise suspicion for this diagnosis and an integral member of the MPS healthcare team. Medical therapies for the management of MPS (i.e., hematopoietic stem cell transplantation, intravenous enzyme replacement therapy) have led to increased lifespan but have had limited beneficial effect on the development of skeletal deformities. Patients must be monitored carefully and treated surgically as necessary. Conditions that may require surgical management include spinal cord compression in the cervical spine, gibbus deformity, hip dysplasia and osteonecrosis, genu valgum, and carpal and tarsal tunnel syndromes. Anesthetic and perioperative evaluation are critical for safe and successful orthopedic surgery.

Speakers



Thomas Wier

Thomas Wier is a retired dual service (Army/Navy) veteran of 22 years whose final tour included serving with a counterintelligence unit at Fort Sheridan, IL/Fort Meade, MD. After receiving an honorable discharge from the U.S. Army in 2009, he has been employed as a representative for the Social Security Administration in Woodstock, IL, serving the community, performing in-depth analysis of benefits, and answering specific questions and concerns from the public at large. Thomas has been married for 36 years to his wife, Blanca, and is the proud father of four sons, including Frankie (MPS II) who passed away in 2014.

Navigating Social Security, Medicaid, and Medicare

9:10 AM, SEPT. 30, OLD GEORGETOWN/CONGRESSIONAL

Navigating through Medicaid and Supplemental Security Income can be challenging for many MPS adults, parents, and legal caregivers. Not knowing where to start or what benefits one may qualify for at times can seem very daunting and stressful. Working at the Social Security Administration for the past 14 years, Mr. Wier provides answers and education to those navigating through the system on a daily basis. This session will address basic steps to start the process as well as possible benefits available. Mr. Wier also will discuss the different programs offered to empower those individuals as they embark on this journey. There will be a Q & A to answer specific questions from the audience following the presentation.

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Elizabeth and her daughter Zoe who is living with MPS IIIA

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Ultragenyx is proud to sponsor the National MPS Society's 37th Annual Family & Scientific Conference and to support its ongoing work on behalf of the MPS community. Together, we are charting a course toward a bright future, and we are honored to be on this extraordinary journey with you.

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MRCM-UX003-00329 September 2023

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For more information:
→ jcrpharm.co.jp ir-info@jp.jcrpharm.com
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- Pediatric Special Care
- Genetic Counseling
- Social Work
- Rehabilitation Medicine
- Physical Therapy
- Endocrinology

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Attendees

MPS I

NAME(S)	EMAIL ADDRESS	RELATIONSHIP	STATE
Charlotte, Thomas, Titus, and Eliza Barrett-Weber	char_208@hotmail.com	Family	KY
Alicia, Jacob, and Mia Bohley	abohley7615@yahoo.com	Family	NC
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MPS II

NAME(S)	EMAIL ADDRESS	RELATIONSHIP	STATE
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continued

Attendees

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Daiza and Philip Torbert	daizagordon@icloud.com	Family	PA
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Blanca and Thomas Wier amd Carmen Torres	fourwiers@yahoo.com	Family	IL

MPS III

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Austin, Stephanie, and Annabelle Bozarth	stephbozarth@yahoo.com	Family	VA
Ruofan (Camilla) Gu	jjaini1999@gmail.com	Self	MD
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Lina Ricci	lina.sayuri@gmail.com	Family	TX
Shahid Ullah and Salma, Maria, and Ahmed Bin Shahid	shahid11khan@yahoo.com	Family	NY
Danielle Spadafora	fixemallup@verizon.net	Grandparent	PA
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Fanny Zambrano	fezbsop26@msn.com	Self	TX

continued

Attendees

MPS VI

NAME(S)	EMAIL ADDRESS	RELATIONSHIP	STATE
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Rebekah Holland	rebekah.holland22@gmail.com	Family	OK

MPS VII

NAME(S)	EMAIL ADDRESS	RELATIONSHIP	STATE
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ML

NAME(S)	EMAIL ADDRESS	RELATIONSHIP	STATE
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PROFESSIONALS

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Rafael Baddell-Grau	UCSD	CA
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Autumn Benson	Takeda	MA
Natasha Bonhomme	Expecting Health	Washington, DC
Alison Bradbury	Nationwide Children's Hospital	OH
Elizabeth Braunlin	University of Minnesota	MN
Carol Bryant	National MPS Society	NC
Katie Burns	SmithSolve	NJ
Emma Canepa	UCSF	CA
Erik Cline	Takeda	MA
Kate Delaney	BioMarin	CA
Annamarie Dillon	Orchard Therapeutics	UK
Amy Downen		NC
Jill Dwyer	UCSF	CA
N. Matthew Ellinwood	National MPS Society	NC
Michael Fountain		TX
Jill Frauenheim	Takeda	IL
Jonathan Frei	BioMarin	PA
Jonathan Gall	Denali Therapeutics	CA
Sydney Gardner	Denali Therapeutics	AZ
Lisa Garrity	Cincinnati Children's Hospital	OH
Amy Gaviglio		MN
Natalia Gomez-Ospina	Stanford	CA
Belen Gonzalez Sutil	Ultragenyx	FL

continued

Attendees

NAME(S)	ORGANIZATION	STATE
Jacqline Gray	Takeda	MA
Cara Harrington	Takeda	MA
Amy Katz	BioMarin	GA
Andrew Krikorian	Takeda	MA
Joyianna Luc	Takeda	MA
Leslie Lynch	UCSF	CA
Haley Lynn	BioMarin	WI
Deborah Marsden	Ultragenyx	CA
Sarah Martin	Denali Therapeutics	CA
Kakneka Mason	BioMarin	MD
Jill Morris	NIH	MD
Jill Mursewick	BioMarin	WA
Keiko Nakai	Denali Therapeutics	CA
Jennifer Noonan	Accessia Health	VA
Ravi Pathak	Takeda	MA
Kim Ramsey	Denali Therapeutics	CA
Elizabeth Rice	University of North Carolina at Chapel Hill	NC
Laura Rieken	Ultragenyx	CA
Lachlan Smith	University of Pennsylvania	PA
Matt Thura	UCSF	CA
Kenneth Trapp Jr.	BioMarin	CA
Rose Treon	Takeda	MA
Nnenna Ukwu	Takeda	MA
Leslie Urdaneta	National MPS Society	NC
Jennifer VanHoutan	REGENXBIO	IL
Akriti Virdi	REGENXBIO	MD
Ryan Watts	Denali Therapeutics	UT
Klane White	Colorado Children's Hospital	CO
Chen Yu	BioMarin	CA

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- Toni-Ann,
mother of Aiden and AJ

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