



NASHVILLE TN

MTM-CNM FAMILY CONFERENCE



2017

★ RESEARCH ★
★ RESOURCES ★
★ RELATIONSHIPS ★
GET YOUR BOOTS ON
www.mtm-cnm.org

MTM-CNM FAMILY CONFERENCE
July 21-23, 2017 Nashville TN
MTM-CNM Family Connection

2017 MTM-CNM Family Conference

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As a community and as individuals, the journey with MTM-CNM can be a long and challenging one. We have been “on the road” for some time, and there are still many miles to go. We have held fast to the “anchors” of our strength, and pulled hard on the “oars” with great hope to get through rough waters. Now more than ever, it is important to come together as one and prepare for the next steps. And so we invite you all to

“Get Your Boots On”, whether yours are dusty cowboy boots, work boots, AFOs, or even rugged wheels on a chair. It’s time. **Get your boots on.**



Welcome!

On behalf of MTM-CNM Family Connection, welcome to the 2017 MTM-CNM Family Conference! We are so glad you have travelled to be with us in “Music City,” Nashville Tennessee!

The theme of our 5th national conference is “Get Your Boots On”! We wanted a theme that spoke to the active roles we all take on in our MTM-CNM community. Every day, our loved ones living with MTM or CNM “get their boots on” and overcome obstacles and challenges in extraordinary ways. Parents and family members “get their boots on” and provide support, protection, encouragement, and strength. Families who have lost a loved one to MTM or CNM “get their boots on” and stand strong in loving memory, and continue to carry their loved ones’ legacy. Researchers, medical professionals, and pharma industry partners “get their boots on” and are busy at work trying to bring about new treatments and medical advances for managing the challenges of MTM and CNM. We are coming together to celebrate all of these exceptional efforts to support our MTM-CNM Family Community.

This theme also illustrates our passion to help equip families with the connections and resources that will help us all stay grounded and prepared while coping with the day to day challenges – big and small – brought by MTM and CNM. At the same time, we are very excited about what the future may bring with regard to possible treatments. We aim to help families to “Get Your Boots On” to learn together about the treatments on the horizon, what we can be doing to prepare the way for treatment approvals, and how to advocate for our community.

It is with deep gratitude that we would like to thank our corporate sponsors and the many individual families and friends who have helped to support this Conference. In addition to financial contributions, we have many community members who are sharing their gifts and talents to help make this Conference a truly special event. It has been a coming together of resources and gifts that has brought us here today and it is our hope that we all leave with a renewed commitment and resolve to “Get Your Boots On” and move forward together with great hope. May you all have the most wonderful weekend and leave with memories to last a lifetime!

So glad you’re here, y’all!

MTM-CNM Family Connection, Inc.
Board of Directors & Conference Planning Team:

Erin Ward, President
Marie Wood, Vice-President
Mark Ward, Treasurer
Shannon Mashinchi, Clerk



Agenda

2017 MTM-CNM FAMILY CONFERENCE: "GET YOUR BOOTS ON" Nashville Airport Marriott, Nashville TN

THURSDAY, JULY 20TH

- Ongoing Participate in Research studies and Focus Groups
University of Florida, Nationwide Children's Hospital, Philips Respironics
- 3:00PM - 4:00PM..... Orientation & Welcome Reception for 1st Time Families
- 6:00PM - 8:00PM..... Community Dinner
Note: Participation in the Orientation and Welcome Reception and Community Dinner is limited to those who signed up during registration and have confirmed attendance with the Conference team.

FRIDAY, JULY 21ST

- Ongoing Participate in Research studies and Focus Groups
University of Florida, Nationwide Children's Hospital, Philips Respironics
- 2:00PM - 4:00PM..... Conference Sign-In and Community Connections Reception
Pickup welcome packets, enjoy light refreshments, and get to know the various resources and organizations represented at this year's Conference
- 5:00PM - 9:00PM..... Opening Dinner and Evening Program
Community Social (cash bar), Welcome Address, Buffet Dinner, Professional and Family Introductions

SATURDAY, JULY 22ND

- 8:00AM - 8:45AM Continental Breakfast
For those desiring a larger breakfast, a full menu is available at Champions Restaurant on-site (at your own expense).
- 8:45AM - 9:00AM Announcements and Welcome
- 9:00AM - 12:00PM..... Scientific Presentations
Light refreshments will be available mid-morning
- Introduction and Orientation
Alan Beggs, PhD, Boston Children's Hospital
- Gene Therapy Drives Whole-Body Correction of XLMTM in Dogs
Casey Childers, DO, PhD, David Mack, PhD, Mike Lawlor, MD, PhD
University of Washington and Medical College of Wisconsin

Agenda

SATURDAY, JULY 22ND (continued)

9:00AM - 12:00PM..... Scientific Presentations, continued

Applying RNA-Seq to Study Gene Expression in Dogs With XLMTM
Jean-Baptiste Dupont, University of Washington

Clinical Development of Gene Therapy for XLMTM
Sal Rico, Audentes Therapeutics

Company Overview - Audentes Therapeutics
Suyash Prasad, Audentes Therapeutics

Natural History Study, PIK3C2B Inhibition for XLMTM
Jim Dowling, MD, PhD, Hospital for Sick Kids



12:00PM - 1:00PM Lunch Break (box lunches)

1:00PM - 2:00PM..... Scientific Presentations

Development of ASO-Mediated DNMT2 Knockdown for Centronuclear Myopathies
Belinda Cowling, PhD, IGBMC, Dynacure

Company Overview - Dynacure
Stephane van Rooijen, MD, MBA, Dynacure

Company Overview - Valerion Therapeutics
Deborah Ramsdell, Valerion Therapeutics

2:00PM - 4:00PM..... Scientific & Medical Forum

Engage in extended Q&A and dialog between families and professionals regarding topics of interest in a moderated open-forum. Bring your questions!

2:30PM - 4:00PM..... Workshop: Creating Switch-Adapted Toys for Kids with MTM & CNM

Sponsored by a Quality Of Life Grant from the Dana and Christopher Reeve Foundation
Empowering through play! Learn how to adapt electronic toys for switch activation. Families will be given an age-appropriate toy, an accessibility switch, and instruction and assistance from two experienced parents, Daniel McDermott and Martin Wilhelm.



4:30PM..... Group Photo

Wear your conference t-shirt (and cowboy boots, if you got 'em!)

5:00PM - 9:00PM..... Evening Program

Community Social (cash bar), Buffet Dinner, the world-famous MTM-CNM Family Variety Show (starring YOU!), and an evening with special Nashville guests.

(continued on next page)



Agenda

SUNDAY, JULY 23RD

8:15AM - 9:00AM Continental Breakfast

8:30AM - 8:50AM Nondenominational Inspiration & Reflection Service (optional)

9:00AM - 10:00AM Family Voices Panel

Members of our MTM-CNM family will share insight on a variety of topics related to life with MTM-CNM, advocacy, and community engagement.

Kimberly Brown, Rob Garland, Amanda Hollingsworth, Hillary Sirmon,
Joe Slaby, Erin Ward, Marie Wood

10:00AM - 12:00PM..... Scientific Presentations

Light refreshments will be available mid-morning

Study of Mothers of Children with Congenital Myopathies

Casie Genetti, MS, CGC, Boston Children's Hospital

Measurement of Respiratory Function in Neuromuscular Disease,
Muscle Function in Female Carriers of XLMTM

Barbara Smith, PT, PhD, University of Florida

Motion Assessments in Individuals with XLMTM

Lindsay Alfano, Nationwide Children's Hospital

Patient Advocacy

Barbara Wuebbels, Audentes Therapeutics

12:00PM - 1:00PM Lunch Buffet

From Silence to Resilience: Telling Your MTM-CNM Story

Levi Gershkowitz, Living in the Light

1:00PM - 2:00PM..... Closing Program

Final thoughts from the MTM-CNM Family Connection team, and an "Open Mic" forum for all to share reflections on the Conference and next steps ahead.

5:30PM - 8:00PM..... Community Dinner

Note: Participation in the Community Dinner is limited to those who signed up during registration and have confirmed attendance with the Conference team.

Agenda

THROUGHOUT THE CONFERENCE...

Participate in Research Studies (please contact a team member to arrange a time to participate):

University of Florida:

Observational Study of Respiratory Strength and Function in Centronuclear Myopathies
Muscle Function in Female Carriers of X-Linked Myotubular Myopathy

Nationwide Children's Hospital:

New assessments to test the ability to move in individuals with XLMTM

Boston Children's Hospital:

Molecular and Genetic Studies of Congenital Myopathies
MTM1 Carrier Study

Medical College of Wisconsin:

Learn about the Congenital Muscle Disease Tissue Repository

Visit the Reflection Room, a special tribute and memorial to the loved ones who have been lost to MTM-CNM

KIDS ACTIVITIES

Kids of all ages are always welcome in the main conference sessions, but will also enjoy these fun activities in the space directly adjacent to the main ballroom:

Balloon Animals & Facepainting
by Fantasy Face Artistry



Music with Ali

Arts and Crafts

Video Games

Movies

Board Games

Variety Show preparation
and more!



Note: while activities will be provided, the Conference will not provide nursing, healthcare supervision, babysitting, or childcare



Don't Miss!

Meet Myo, the MTM-CNM Traveling Turtle!

Myo's mission is to travel the world, visiting members of the Myotubular & Centronuclear Myopathy community, to help us all stay connected. Be sure to introduce yourself to him, and maybe even take a "shellfie" with him at the photo booth!



Perhaps Myo can come visit you after the Conference?

Learn more at www.mtm-cnm.org/traveling-turtle



#nashville2017

Thank you to RARE Science and the RARE Bear Army!



RARE Science has initiated the RARE Bear program, a grassroots community-driven outreach for kids with rare disease. Community volunteers create one-of-a-kind teddy bears for one-of-a-kind "rare" kids. We are happy to have partnered with RARE Science to provide RARE Bears to our MTM-CNM Families here at the conference.



Learn more at www.rarescience.org/rare-bear-program

#mtmcmfamily

With Switch-Adapted Toys, Everyone Can Play!

Thanks to a Quality of Life Grant from the Dana and Christopher Reeve Foundation, we are able to offer a workshop at this year's Conference. Parents and caregivers will learn the basics of adapting an electronic toy for switch-activation. All families attending will get to take home their new adapted toy and switch. It is our hope that by empowering families and children through the ability to adapt toys and other devices, the quality of life of all of our MTM-CNM community will be increased.



Special Guests

Alison Grace Rapetti, MT-BC - Starling Creative Arts



Alison Grace Rapetti is the founder and director of Starling Creative Arts. She oversees the team of incredible therapists, performers, and creators of various artistic disciplines. Ali actively practices music therapy in hospice care, teaches music lessons, and produces music, both as an artist/songwriter/performer and for other artists. She most loves to help craft personalized songs and legacy work.



Ali is a Graduate of the Berklee College of Music and is a seasoned singer, songwriter, and performer who has written and recorded with legendary artists. John Mayer, a personal mentor and co-writer, considers Ali's writing "rare stuff." She has sung and shared stages with Willie Nelson, Carol King, Bobby McFerrin, The Eagles, Annie Lennox, and West African sensation, Angeliqwe Kidjo. She is co-founder of the nationally recognized non-profit, Arts Education International, which creates arts programming for orphaned and abandoned children in Ghana, Sierra Leone, and Trinidad.

Alison Grace believes in the creative potential of each individual and encourages others to develop their inherent strengths and transform their vulnerabilities into creative confidence.

www.starlingcreativearts.com

Levi Gershkowitz - Living in the Light



Living in the Light, founded in 2012 by Levi Gershkowitz, began as an advocacy initiative utilizing the potency of photography and compelling personal narratives to educate about the realities of life faced by those with rare diseases. Since then, it has evolved to meet the complex needs of the rare disease community to ensure that its members are truly seen and heard as people, not patients.



With keen attention given to the diverse needs of patients, caregivers, family members, the broader rare disease community and industry, Living in the Light specializes in creating content that is clear, factual, ethical and establishes powerful personal connections that grow lasting partnerships.

Contact: Levi@FromPatientToPerson.com, (339) 225-2812
www.frompatienttoperson.com

Research & Medical Professionals

AUDENTES

Suyash Prasad, MD - Audentes Therapeutics



Dr. Prasad has served as Senior Vice President and Chief Medical Officer since February 2014. He has a wide range of experience and achievement in international drug development across Phase I to IV, with a specific focus in the clinical development of therapies to treat rare pediatric disorders. Dr. Prasad has worked in drug development for the past 14 years in positions of increasing responsibility at BioMarin

Pharmaceutical, Inc., Genzyme Corporation, and Eli Lilly and Company. He has broad therapeutic expertise in neuromuscular disease, metabolic medicine, bone, neuroscience, endocrinology, immunology, and genetics. Dr. Prasad has significant experience with the development and commercialization of enzyme replacement therapies to treat lysosomal storage disorders, including Cerezyme®, Aldurazyme®, Fabrazyme®, and Myozyme®. For Pompe Disease, he led the global medical planning activities for Lumizyme® for the treatment of adult Pompe Disease and for Myozyme® to treat infantile Pompe. Most recently, he was responsible for the clinical development of novel treatments for phenylketonuria (PKU) and achondroplasia.

Dr. Prasad graduated in Medicine at the University of Newcastle-upon-Tyne, UK, where he received commendations for Pediatrics, Obstetrics and Gynecology, and Medical Ethics. He is a United Kingdom board certified physician with a sub-specialty interest in Pediatric Critical Care, and is a member of the Royal College of Physicians (MRCP), the Royal College of Pediatrics and Child Health (MRCPC), and the Faculty of Pharmaceutical Medicine (MFPM). Dr. Prasad is a past recipient of the Outstanding Contribution Award from the Faculty of Pharmaceutical Medicine of the UK Royal College of Physicians.

Sal Rico - Audentes Therapeutics



Sal Rico is Vice President of Clinical Development at Audentes Therapeutics. He is a clinical pharmacologist with 12 years of clinical research experience in the pharmaceutical industry, contract research organizations and academia. He has participated in the design, conduct, and analysis of phase 1-4 clinical trials in multiple therapeutic areas including rare diseases, hema-

tology/oncology, transfusion medicine, psychiatry, sleep medicine, cardiovascular medicine, infectious diseases, pain, allergy, and dermatology. Prior to joining Audentes he led the clinical development teams at Cerus Corporation and Transcept Pharmaceuticals, and was an investigator at the Centre for Drug Research, Hospital de la Santa Creu i Sant Pau in Barcelona, Spain. His clinical research and development experience includes drugs, biological products and medical devices, such as the INTERCEPT Blood System®, INTERMEZZO®, HIBOR®, CHAMPIX®, PROLIA® and KERYDIN®, among others.

Dr. Rico was awarded a Doctor of Medicine and Surgery degree from the National Autonomous University of Mexico and holds a Masters in Science and a Ph.D. (Summa Cum Laude) in Pharmacology from the Universitat Autònoma de Barcelona. Additionally, he has received post-doctoral training in clinical pharmacology, transfusion medicine (National Autonomous University of Mexico), and pharmaceutical medicine (Universitat Autònoma de Barcelona).

Research & Medical Professionals

Barbara Wuebbels, RN - Audentes Therapeutics

Ms. Wuebbels has served as Vice President Patient Advocacy since October 2013. She has more than 15 years of experience working with patients, patient organizations and clinicians in the rare disease community. Ms. Wuebbels joined Audentes following 6 years at BioMarin Pharmaceutical, Inc. where she held positions of increasing responsibility in patient advocacy and medical affairs. This work included the creation of a global patient advocacy and investigator relations department. Through this new department, Ms. Wuebbels linked the critical perspectives of patient advocates with the internal planning of clinical and commercial activities. Earlier at BioMarin, she had oversight of BioMarin's global registry for Maroteaux-Lamy Syndrome (MPS VI) patients. Prior to BioMarin, Ms. Wuebbels directed clinical education within the Orphan Drug Division at Medicis Pharmaceutical Corporation, and served as Director of Clinical Affairs at Vivra Health Advantage. Ms. Wuebbels started her career as a registered nurse.

Ms. Wuebbels earned a Master of Science degree in Adult Health Nursing from Arizona State University, and a Bachelor of Science in Nursing from St. Louis University.



Kimberly Trant, RN - Audentes Therapeutics

Kimberly Trant joined Audentes Therapeutics as the Director of Patient Advocacy in May 2017. Kimberly began her career in healthcare as a pediatric and neonatal Registered Nurse (RN) where she had the privilege to provide direct care to patients. She transitioned from patient care to industry when she earned her Master of Business Administration (MBA) degree.

Kimberly has spent the past 16 years in the pharmaceutical/biopharmaceutical industry in various roles with increasing leadership where she has had the opportunity to partner with and learn from patients, families, and patient advocacy organizations. She has extensive experience developing patient education and resources, creating patient communication strategies, and integrating the patient and family perspective into clinical development and commercialization activities.

Kimberly is honored to serve the patient community and strives to make a meaningful impact helping patients and their families.



Shawn Jones - Audentes Therapeutics

Shawn Jones has worked in Biopharmaceutical industry for the past 25 years and currently leads the clinical operations group at Audentes.

Research & Medical Professionals

Deborah Ramsdell - CEO, Valerion Therapeutics

Deborah Ramsdell has over 30 years of experience in the design and implementation of world-wide development strategies for therapeutic drugs and biotechnology products. Over the course of her career, she has been responsible for the strategic design and operational oversight of more than 70 drug candidates in the fields of oncology, rare metabolic diseases, dermatology, pain, infectious diseases, transplantation, cardiology, renal diseases, and endocrinology. She most recently has been involved in clinical and regulatory development for Valerion Therapeutics, Enobia Pharma, Lotus Tissue Repair (now Phoenix), Pellepharm Pharma, Orphan Technologies, aTyr Pharma, and PreciThera, all rare genetic disease companies.



She has been a strategic drug development consultant for over 30 years, and is currently a co-founder of Alopexx Enterprises, a family-funded investment and management company and consulting CEO for Valerion Therapeutics, one of their key assets. She also is on the Board of Directors for Orphan Technologies, a biotech company developing therapies for rare metabolic diseases.

She has been a senior level executive in the biotechnology industry as well as a strategic consultant to biotechnology and pharmaceutical companies for most of her career.

Hal Landy, MD - CMO, Valerion Therapeutics

Dr. Landy is a board certified Pediatric Endocrinologist with 30 years of clinical research experience in academics and industry, primarily in rare orphan diseases. Educated at Harvard College (AB '72) and Columbia University (MD '81), he trained at Massachusetts General Hospital and at Children's Hospital, Boston and was on staff at Children's Hospital and Harvard Medical School until 1996.



In 1992, he joined Serono Laboratories (Norwell, MA and Geneva, Switzerland) where he worked on numerous successful regulatory submissions in growth-related disorders, AIDS wasting and short bowel syndrome. He subsequently moved to Genzyme (Cambridge, MA), where he was VP, Clinical Research and assisted in the registration of Aldurazyme for MPS-1 and Myozyme for Pompe Disease. In 2007, Dr. Landy joined Enobia Pharma (Cambridge, MA) as Chief Medical Officer, where he led its clinical program in hypophosphatasia, a rare and devastating disorder of skeletal mineralization, until its acquisition by Alexion in 2012 for over \$1 billion. Subsequently, Dr. Landy served as a consultant to Lotus Tissue Repair where he helped design clinical and pre-clinical development strategies for human recombinant collagen 7 for the treatment of dystrophic epidermolysis bullosa until its acquisition by Shire in 2013 for over \$300 million.

Dr. Landy currently consults for several biotech companies and venture funds in Boston, New York, Washington DC, California and Europe and is consulting Chief Medical Officer for Valerion Therapeutics. He has authored or co-authored over 20 peer-reviewed articles and chapters.



Research & Medical Professionals

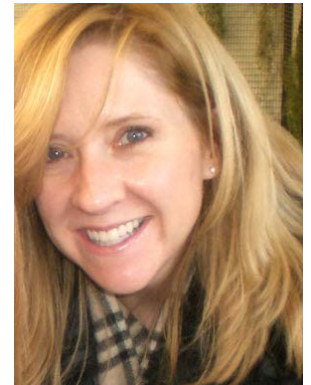


valerion

Kristen Cunningham

Regulatory Affairs, Valerion Therapeutics

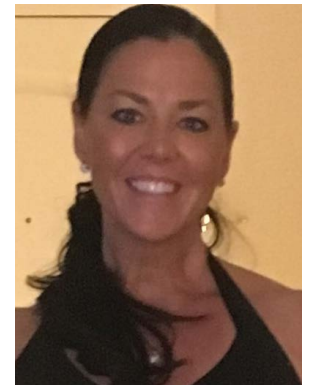
Kristen Cunningham has over 20 years of experience working in Drug Development, specializing in Regulatory and Clinical Operations. She is currently an independent consultant and has been providing regulatory, clinical and program management support for the past 9 years to numerous biopharmaceutical client companies. Previous affiliations include Alexion Pharmaceuticals, Enobia Pharma, Cubist Pharmaceuticals and Idera Pharmaceuticals (formerly Hybridon, Inc.).



Chrissy Burton, BSN, RN

Clinical Operations, Valerion Therapeutics

Chrissy Burton has been an RN for over 16 years currently specializing in critical care with extensive background in surgical, medical, cardiac and hospice. She joined Alopexx Enterprises (parent company of Valerion) 4 years ago to assist with the oversight of all aspects of clinical Operations and performs as CRA for current clinical trials.



Michelle Nelken

Clinical Operations, Valerion Therapeutics

Michelle Nelken has over 15 years experience in the pharmaceutical and biotechnology industry focusing on management of early and late phase clinical studies in multiple therapeutic areas, with a strong focus on rare diseases. During her career, Ms. Nelken's contribution has help lead to the approval of Genzyme's Myozyme® for treatment of Pompe disease, Genzyme's OSOM® Trichomonas Rapid Test, and most recently the Japanese approval of Strensiq® for Alexion Pharmaceuticals.

Research & Medical Professionals

Dynacure

Stephane van Rooijen, MD, MBA - CEO, Dynacure



Stephane is a general management biotech executive with in-depth experience in life sciences working across European and North American organizations. He studied medicine at the KU Leuven, and obtained an MBA at the Erasmus University Rotterdam and Georgetown University, Washington DC.

He joined Dynacure in 2016 as Chief Executive officer, a drug development company focusing on innovative therapies for rare myopathies. Founded as spin-off of Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC); with lead investor Kurma Partners.

Stephane started his career at Arthur D. Little's strategy consultants' team. After joining Genzyme Corporation (2004, Amsterdam/Brussels) working in business development and finance, he became European Cardiovascular Business Unit Leader. In 2012 he joined Viropharma (London) as member of Viropharma's European Business Council and global pipeline project team, responsible for leading the company's main commercial program Cinryze (orphan drug indicated for hereditary angioedema). In 2014, as founding CEO of Confo Therapeutics (VIB-VUB spin-off), he set-up the company, raised funding and recruited an outstanding team.

Belinda Cowling, PhD - IGBMC



Belinda's research has focused on the investigation of the normal role of proteins in skeletal muscle, how these roles are disturbed in muscle disease, and identifying novel therapies for congenital myopathies.

She completed a PhD at Monash University, Melbourne, Australia with Prof. Christina Mitchell, during which time she identified a novel regulator of skeletal muscle mass. She then moved to the IGBMC, France, to work with Dr. Jocelyn Laporte. The IGBMC is one of the leading European centers for biomedical research. The institute is located near Strasbourg, the capital of the European Union, and hosts 49 research teams working on rare and common human disorders. During this time she investigated the normal function of dynamin 2 in muscle, and how defects can cause centronuclear myopathy. She identified down regulation of dynamin 2 as a novel therapeutic target for myotubular myopathy.

As of 2014 she is a project leader at the IGBMC, and working with a motivated team devoted to understanding muscle diseases and developing novel therapies. She is also co-founder and scientific advisor of the company Dynacure, and since 2016 has been working closely with the research and development team at Dynacure.

Research & Medical Professionals

Martin “Casey” Childers DO, PhD - University of Washington



Dr. Martin (Casey) K. Childers DO, PhD is a professor in the Department of Rehabilitation Medicine and an Investigator at the Institute for Stem Cell and Regenerative Medicine at the University of Washington. Dr. Childers rehabilitation medicine practice at the University of Washington Medical Center (UWMC) and is dedicated to cure patients with neuromuscular diseases. He serves as a Member of Scientific and Clinical Advisory Board at Audentes Therapeutics, Inc. Professor Childers' laboratory is currently pursuing two areas of investigation, systemic gene replacement delivery for patients with X-Linked Myotubular Myopathy (XLMTM) and induced pluripotent stem (iPS) cell technology to study heart disease in patients with Duchenne muscular dystrophy (DMD). Dr. Childers rehabilitation medicine practice at the University of Washington Medical Center (UWMC) is dedicated to serve patients with neuromuscular diseases. He received his B.A. from Seattle Pacific University, his D.O. from Western University, and his Ph.D. in Physiology and Pharmacology from the University of Missouri.

David Mack, PhD - University of Washington



Dr. David Mack is an Assistant Professor in the Department of Rehabilitation Medicine and Bioengineering, as well as a faculty member of the Institute for Stem Cell and Regenerative Medicine at the University of Washington. The goal of the Mack laboratory is to apply their understanding of how stem cells make cell fate decisions during embryonic development to the development of stem cell and gene therapy treatments for neuromuscular diseases.

David's foundation is a Ph.D. in molecular genetics from the Indiana University School of Medicine, where he studied transcriptional regulation of T-cell development and how this process goes awry to cause leukemia. As a postdoctoral fellow with Dr. Gilbert Smith at the National Cancer Institute in Bethesda, he studied how tissue-specific stem cells interact with their microenvironment and how this impacts cell fate choices during mammary gland development and pregnancy. David then switched from cancer research to the relatively new field of regenerative medicine by accepting a senior postdoc position at the Wake Forest Institute for Regenerative Medicine. Under the leadership of Dr. Anthony Atala, David's work focused on how to control embryonic and fetal stem cell differentiation by using natural and artificial scaffolds in concert with direct manipulation of the cells' genetic program. All of these efforts have the overriding purpose of developing therapies to enhance tissue repair and regeneration following injury or disease.



Jean-Baptiste Dupont, PhD University of Washington



Dr. Jean-Baptiste Dupont is a postdoctoral fellow at the Institute for Stem Cell and Regenerative Medicine, University of Washington. The focus of his research gravitates toward the understanding of genetic disorders affecting muscles, and the discovery of new treatments for these devastating diseases. He has obtained a Ph.D. from the University of Nantes, France,

working on the biology of gene therapy vectors in the context of Duchenne muscular dystrophy (DMD). In July 2016, he moved to Seattle and started his postdoctoral fellowship, guided by Dr. Childers and Dr. Mack. Part of his job is to investigate the impact of XLMTM on the transcriptome – the overall result of genome expression – and to see how gene therapy can correct it. The results of this study should not only shed light on XLMTM biology and progression, but also provide new indicators of gene therapy efficiency in preclinical studies.

Research & Medical Professionals



Boston Children's Hospital



Alan Beggs, PhD - Boston Children's Hospital, Harvard

Alan Beggs is director of The Manton Center for Orphan Disease Research at Children's Hospital Boston and the Sir Edward and Lady Manton Professor of Pediatrics at Harvard Medical School. He received his AB in biology at Cornell University and his PhD in human genetics at Johns Hopkins University. He then completed postdoctoral fellowships in medical genetics at Johns Hopkins University and in clinical molecular genetics at Harvard Medical School, and has directed an independent research laboratory in the Genetics Division at Children's Hospital since 1992.



Casie Genetti, MS, CGC - Boston Children's Hospital

Casie Genetti is a genetic counselor and program manager specializing in the genetics of congenital myopathies as well as rare and undiagnosed disorders. She received her Master's degree in Genetic Counseling from Boston University in May 2015 and began working with the Beggs Congenital Myopathy Research Program at Boston Children's Hospital in June 2015. She is thrilled to attend her second family conference and to have the opportunity to meet more members of the MTM-CNM community! Casie coordinates the recruitment and enrollment of families for the Beggs Lab research study, and is involved with the clinical studies that focus on identifying the genes and characterizing the symptoms associated with congenital myopathies.



Neeha Nori, BA - Boston Children's Hospital

Neeha Nori is a research assistant specializing in the genetics of congenital myopathies and rare and undiagnosed disorders. She graduated as a Psychology Major from Boston University in May of 2016. Shortly after she graduated, she started working as a research assistant for the Beggs Congenital Myopathy Research Program and has been a part of the team for almost a year. Neeha primarily recruits and enrolls families into the Beggs lab research study by obtaining informed consent, gathering a medical and family history, as well as coordinating sample collection. She is very excited to attend her first family conference and meet the members of the MTM-CNM community!

Research & Medical Professionals

Jim Dowling, MD, PhD - Hospital for Sick Kids, Toronto

Dr. Dowling is a clinician-scientist who is focused on gene discovery and therapy development for childhood muscle diseases.

Dowling received his BSc and MSc from Yale University and his MD/PhD from the University of Chicago. His PhD work was performed in the laboratory of Elaine Fuchs. He did his residency in child neurology at Children's Hospital of Philadelphia and completed postdoctoral research with Jeff Golden (UPenn) and Eva Feldman (University of Michigan). Until recently, he was an assistant professor at the University of Michigan. His clinical expertise is in childhood neuromuscular disorders and he is considered one of the leading authorities on the diagnosis and management of congenital myopathies. Dowling's research has examined questions of disease pathogenesis and therapy development for congenital myopathies and childhood muscular dystrophies. His laboratory has helped pioneer the use of the zebrafish model for these disorders.

Dowling participates in multiple clinics related to paediatric neuromuscular disease. With Dr. Ronni Cohn, he has developed and is co-directing the neuromuscular undiagnosed diseases program (or UDP). This clinic is the first of its kind in Canada and aims to provide expert diagnostic assessment for children with unsolved muscle disease. Dowling and Dr. Grace Yoon run a clinic for paediatric neurogenetic/neuromuscular conditions. Dowling also participates in the ward consultation service through the Division of Neurology.

The overarching goal of the Dowling laboratory is to develop therapies for childhood muscle diseases. Dowling and his team are particularly focused on congenital myopathies and muscular dystrophies. His research spans the continuum from new gene discovery to disease pathogenesis studies in model organisms to both targeted and non-biased drug discovery. His laboratory employs both the zebrafish and mouse model systems and complementary in vitro analyses. A common cell biologic theme to these studies is the examination of the role(s) of endo-lysosomes and their regulators in muscle development and in disease.

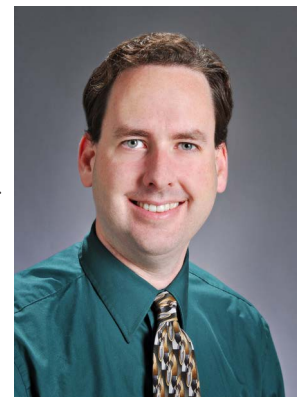
Mike Lawlor, M.D., PhD - Medical College of Wisconsin

Since completing his postdoctoral research training in the laboratory of Dr. Alan Beggs at Boston Children's Hospital, and moving to the Medical College of Wisconsin in 2011, Dr. Mike Lawlor has continued to work closely with the Beggs laboratory while establishing clinical and research neuromuscular pathology laboratories. The work performed in his research laboratory at MCW has included the pathological analyses for a number of preclinical trial studies for animal models of X-linked myotubular myopathy (XLMTM) that are currently being performed worldwide, including anti-myostatin therapy, gene therapy, and protein replacement therapy. Dr. Lawlor is currently working with Audentes Therapeutics to plan muscle biopsy and pathology procedures for a human gene therapy clinical trial for XLMTM. He is also working to design and coordinate transitions to clinical trials for gene therapies in several other disorders. Additional projects being pursued in the Lawlor laboratory are focused on Duchenne muscular dystrophy, nemaline myopathy, congenital contractures, and mitochondrial myopathy.

In the spring of 2013, Dr. Lawlor's laboratory became the site of the Congenital Muscle Disease Tissue Repository, which is intended to provide a central place for the donation and distribution of patient tissues. This resource is supported by patient families and non-profit organizations including Cure CMD, A Foundation Building Strength, the Joshua Frase Foundation, and Where There's A Will There's A Cure. It is our hope that such a central resource for tissue storage and distribution will improve the pace of research in our field.



SickKids
THE HOSPITAL FOR
SICK CHILDREN




**MEDICAL
COLLEGE
OF WISCONSIN**

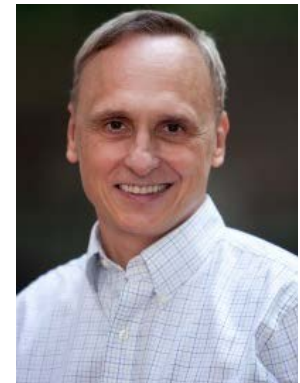
Research & Medical Professionals



The University of Florida is a leader in the clinical evaluation and treatment of neuromuscular disease, grounded in its MDA-funded interdisciplinary neuromuscular clinic and the Powell Gene Therapy Center, an institute dedicated to the development and implementation of gene therapy for rare inherited diseases. The UF team studies the potential interactive effects of rehabilitation and regenerative therapies to enhance neuromuscular function for individuals with inherited neuromuscular disorders.

Barry Byrne, MD, PhD - University of Florida

Dr. Byrne leads a team of more than 30 scientists and clinicians dedicated to understanding the pathophysiology of rare neuromuscular diseases and leading therapeutic strategies using gene therapy. Specifically, the lab has developed novel gene therapies using AAV vectors, to achieve sustained correction of gene deficiencies in animal models of inherited muscle diseases. Most exciting for our community, the methodology they have applied to test respiratory motor performance holds promise for quantifying respiratory muscle function in other severe pediatric neuromuscular disorders, including XLMTM.



Barbara Smith, PT, PhD - University of Florida

Those of you who attended a conference between 2011-2015 will remember Dr. Barbara Smith, PT, PhD, who collaborates with Dr. Childers on measurements for pulmonary testing in the centronuclear myopathies. Her research focuses on evaluation of respiratory motor control and the effects of exercise and regenerative therapies to facilitate ventilatory recovery for patients with neuromuscular disease. Dr. Smith has clinical and research experience with XLMTM, nemaline myopathy, Pompe disease, and Duchenne muscular dystrophy, and she also collaborates with Dr. Barry Byrne on gene therapy clinical trials. With over 20 years of clinical practice and 10 years of full-time research experience, her specialty in respiratory muscle assessment will be an asset for understanding the control of breathing in rare neuromuscular diseases, particularly in patients who use mechanical ventilation.

Research & Medical Professionals



Terry Sexton, ARNP - University of Florida and Shriners' Hospital, Greenville, SC
Terry Sexton joined the UF team's respiratory study at the MTM conference in 2013, and she brings with her more than 30 years of pediatric experience working with medically complex patients. Although Terry relocated to Greenville, SC, she remains affiliated with UF on a part-time basis as a co-Investigator in Dr. Smith's study of respiratory function in XLMTM. Terry would not miss the opportunity to reconnect with her friends again at the 2017 conference!

Lee Kugelmann, BA - Research Coordinator, University of Florida
Lee Kugelmann is excited to return to the family conference, after first attending in 2015. Lee graduated from Emory University in 2014 and has worked since with Drs. Byrne and Smith as a Clinical Research Coordinator. Lee works on a variety of the team's studies in neuromuscular diseases, including four projects focused on XLMTM.



Markus Renno, MD, MPH - Vanderbilt University/University of Florida
Dr. Renno is completing a year of advanced cardiovascular imaging during his final year of pediatric cardiology fellowship at Vanderbilt Children's Hospital right here in Nashville. He earned his medical doctorate at the University of Arizona and completed his pediatric residency training at the University of Florida, where his research first introduced him to the care of children with CNMs. Since moving to Nashville, Dr. Renno has continued to collaborate with the University of Florida team in studying the respiratory physiology in children with CNMs. He is excited to welcome everyone to his hometown and will provide medical oversight of the UF team's respiratory tests during the conference.

Cristina Liberati, MD - Research Fellow, University of Florida
Dr. Liberati graduated from the Medical School of the Sapienza University of Rome in 2015. After a year of training in general medicine, she moved to the USA and joined the UF research team in September 2016. Currently, she is acting as study coordinator and medical supervisor for a variety of studies while improving her medical skills and expanding her knowledge by following the lead of Dr. Byrne, Dr. Smith and Dr. Corti at UF. She has recently had the chance to work on Dr. Smith's study of respiratory function in XLMTM. This will be her first conference, and she is excited for the opportunity to reconnect with old friends and make new ones!



Research & Medical Professionals



Samantha Mays, BS

Research Assistant, University of Florida

Samantha graduated from the University of Florida with a Bachelor's degree in Exercise Physiology. She has spent the past year assisting Dr. Smith on her research studies evaluating respiratory strength in people with neuromuscular diseases. In the fall, she will be starting graduate school at the University of Florida in Information Systems and Operations Management.



Melissa Smith, BS

Student Researcher, University of Florida

Melissa completed her undergraduate honors thesis and project under Dr. Smith on quality of life in patients with XLMTM. She graduated from the University of Florida this past spring with a Bachelors in Health Science. She is currently attending the University of Miami's Doctor of Physical Therapy program and hopes to become a pediatric physical therapist.



Gee Kim, BS

Research Coordinator, University of Florida

Gee joined Dr. Byrne's clinical research team in 2015 and works on an assortment of neuromuscular disorder studies. This is her first time attending the MTM-CNM Family Conference. She graduated from the University of Florida (Go Gators!) and has over 5 years of experience working with pediatric patients.

Research & Medical Professionals

Nationwide Children's Hospital - Columbus, Ohio

The team from Nationwide Children's Hospital, Drs. Lindsay Alfano, Natalie Miller, and Megan Iammarino work in the Center for Gene Therapy and perform clinical evaluations of children with neuromuscular disorders. Their experience working with traditional tests that require fatiguing position changes led them to develop the ACTIVE system (Abilities Captured Through Interactive Video Evaluation) to measure movement in children with muscle weakness. They designed ACTIVE-mini to measure the movement ability of children lying on their back. This test measures the spontaneous movement of a child over 2-minute trials without position changes. Additionally, they've created a video game that measures a child's ability to reach. This game quantifies a child's arm movement while sitting and interacting with a custom-designed video game. The objective of their research is to further develop these tools for use in both clinics and research trials to reduce the burden of testing of children.



Lindsay Alfano, DPT



Megan Iammarino, DPT



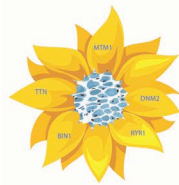
Natalie Miller, DPT



**NATIONWIDE
CHILDREN'S**

Community Organizations

We are grateful for the many family and community organizations working very hard to support the MTM-CNM Community. It is with great appreciation and in the spirit of collaboration, that we share the following resources. You will have an opportunity to meet in person with many of these community leaders throughout the weekend or connect with them following the conference via their respective websites.



CNM - Together Strong!
Association for Centronuclear Myopathies

German CNM Association “Together Strong! (www.znm-zusammenstark.org/)

Inspired by the 2014 Myotubular Trust Family Conference, three German families organized the first German Family Conference in June 2015. It was a great success! 21 families got together and founded the German CNM Association “CNM-Together Strong!” (ZNM - Zusammen Stark!). Their organization aims to raise awareness, help support families, and raise funds for research. Every year a family conference is organized. Today “ZNM” represents 43 member families in Germany and the Netherlands. In their general meetings these members decided to support with the association funds the research grants of the Myotubular Trust. Every two years the members elect a management board. At our conference, Arlene Wuestner is sent as envoy of the association. She is also a physician and speaker of the association's working group on medical questions. We are delighted Arlene has traveled to be with us at our conference and please take a moment to meet her throughout the weekend.



Information Point for Centronuclear and Myotubular Myopathy (www.centronuclear.org.uk)

The Information Point for Centronuclear and Myotubular Myopathy is an excellent source of information, resources, family stories, and community events for our global MTM-CNM community. The Information Point was created by, and is run through, the volunteer work of Toni Abram, an individual who lives with centronuclear myopathy. For 16 years, Toni has helped to connect our community through her informative newsletters and website. Please take some time to visit the website and read past issues of her wonderful newsletters.



Joshua Frase Foundation (www.joshuafrase.org)

The Joshua Frase Foundation, ceated by Alison and Paul Frase in honor of their son Joshua, has been raising funds to support research in hopes of finding a treatment and/or cure for Centronuclear Myopathies for over twenty years. Over the past decade, they have assembled a team of leading researchers from a variety of fields, combining their respective areas of expertise into a multi-faceted approach, aimed at understanding how and why the body responds to myotubular myopathy so that effective therapies can be formulated. They are also dedicated to supporting families who struggle with the realities of MTM. Please take a moment to introduce yourself to Alison, Paul, and Josh's sister Isabella during the conference.



Community Organizations



Myotubular Trust (www.myotubulartrust.org)

The Myotubular Trust, created in 2006 by two MTM moms, Anne Lennox and Wendy Hughes, has been instrumental in funding ground-breaking research for our community. The Myotubular Trust offers scientists within the community competitive scientific grants. These awards are made following an international peer review process. The Myotubular Trust also hosts European conferences for the MTM-CNM community, most recently in collaboration with ZNM-Zusammen Stark! Our team, along with the community, learned so much from their joint conference held last year! The Myotubular Trust also co-funds, along with MDUK (Muscular Dystrophy UK), the Myotubular and Centronuclear Myopathy Patient Registry – the first international database specific to our condition. The registry was developed by the Trust over a number of years, in partnership with TREAT-NMD, and with a number of leading neuromuscular researchers. Please visit their website for more information about their great work on behalf of our community.



RYR-1 Foundation (www.ryr1.org)

The Foundation was started by members of the Goldberg Family, who have been affected by RYR-1 muscle disease (myopathy). Michael and Morton Goldberg attended our 2015 Family Conference and became inspired to host their first Family Conference for RYR-1 in the summer of 2016. Currently, there is no other organization that exists solely to advocate for and serve the needs of patients with RYR-1 myopathy. The goal of their Foundation is to fill this much needed void. We are happy to have Rachel Bronstein, Program Coordinator for RYR-1 joining us at our conference to share RYR-1 information.



Where There's A Will There's A Cure (www.will-cure.org)

Founded by the family and friends of William Richard Whiston, Where There's a Will There's a Cure is dedicated to raising funds for the purpose of supporting scientific research that is actively pursuing cures and/or treatments for those affected with Myotubular Myopathy. Please visit the Will-Cure website to read more about the active and ongoing research they are funding within the community.

MTM-CNM Family Connection Team

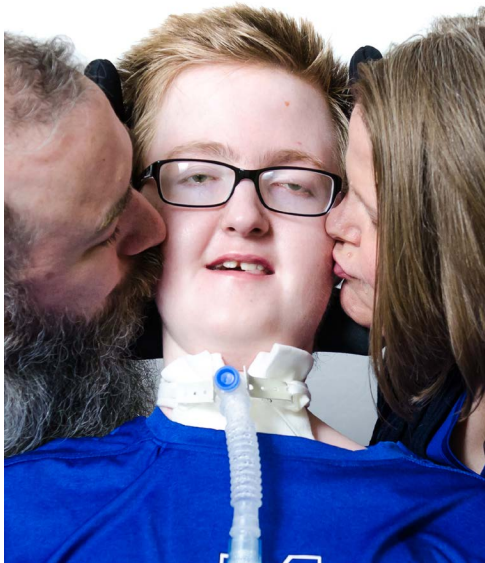
MTM-CNM Family Connection, Inc (www.mtm-cnm.org) is a non-profit charitable organization with a mission to connect families affected by Myotubular Myopathy (MTM) and/or Centronuclear Myopathy (CNM) to resources, research, and relationships within the MTM-CNM community. Our mission is two-fold:



Family Connection, Inc
Resources • Research • Relationships

- 1) With a deep awareness of the unique challenges in having or caring for someone with MTM-CNM, we seek to provide an enjoyable and enriching family-oriented conference experience that educates and unites the individuals and families affected by Myotubular & Centronuclear Myopathy in supportive friendships and life-enhancing networking opportunities.
- 2) With hearts full of hope for the future, we seek to connect families in person to the top researchers in potential MTM-CNM treatments in order to get them acquainted with the latest advances in medicine and assistive technology and to get families signed in and registered in the various studies that will push research forward.

The Ward Family



Mark, Will, Erin

Our son Will was born in 2001 and spent the first few months of his life in the Boston Children's Hospital NICU overlooking Fenway Park. As fate would have it, he is a Red Sox fan for life! He was diagnosed with MTM at four weeks old. At four months old, Will came home with a tracheostomy and G-tube on July 3rd, which we will forever celebrate as "Will's Independence Day"! Will has thrived at home and faces each day with incredible resilience and his "strong will".

An amazing sixteen years later, Will has just finished his sophomore year of High School with honors. He loves the Boston Red Sox and he plays on a Challenger baseball team. Will enjoys music, books, and going to the movies. Will also has two very special dogs, Gracie & Simba.

While we celebrate every day with Will just as he is, and have learned many ways to adapt and embrace life to the fullest, we also hold on to the hope that there will be treatments and potentially a cure for all individuals with MTM, as well as other myopathies. We have learned the importance of helping to strengthen our community, coming together to support each other and celebrating our loved ones affected by muscle disease. We hold onto the hope that anything is possible and together we can truly make a difference.

With this mission in mind, we helped to found MTM-CNM Family Connection, Inc., and Erin serves in the volunteer role of President and Mark volunteers as Treasurer. We feel extremely blessed that we've had this opportunity to plan another conference for our community and to serve our "family" through the work of MTM-CNM Family Connection. We can't wait to see y'all and hope that everyone has an amazing experience this weekend!

other conference for our community and to serve our "family" through the work of MTM-CNM Family Connection. We can't wait to see y'all and hope that everyone has an amazing experience this weekend!

MTM-CNM Family Connection Team

The Wood Family

We are the Wood family from NYC! David, Marie, Lucian (13), Blaise (11), Reid (9), and Paley (7)! Marie has been part of the MTM-CNM Family Conference planning team since 2009 and loves to serve our #mtm-mcnmfamily. Dr. David Wood, PhD. is an international speaker and writer on philosophy and religion. Lucian and Blaise are funny, bright kids who train regularly in Brazilian jiu jitsu. While their sibling rivalry with each other is strong, they completely melt over their little brothers, Reid and Paley, who both have XLMTM. Reid loves music, beginning every morning playing on the keyboard. Paley loves music, too, and is a huge bookworm! Lately, he's intrigued by geography, spending the whole day sometimes looking at maps. Their favorite place to go is the Bronx Zoo: Paley is shy around people but loves animals, whereas Reid seems to like the zoo more to socialize and befriend new people! Reid and Paley have endured more in their young lives than most people ever will—whether sicknesses, surgeries, or brushes with mortality—yet they're still here fighting and they can still do so much to enjoy life. They bring beauty and meaning to our lives, reawakening our wonder and gratitude in new and surprising ways, simply by being the unique, curious, sweet little people they are.

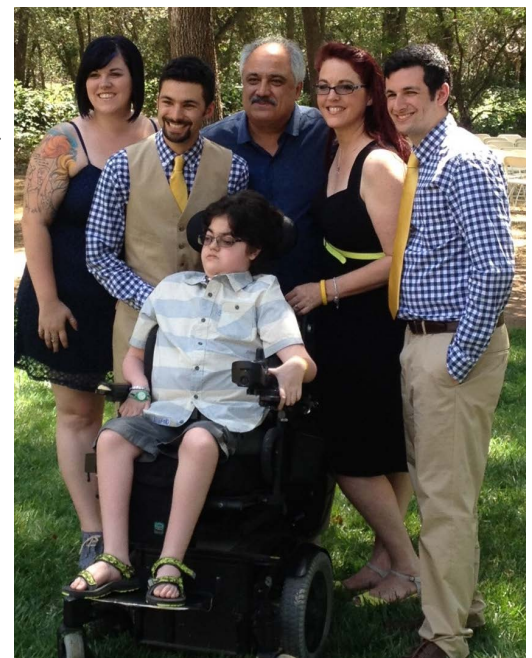


David, Reid, Marie, Blaise, Lucian

The Mashinchi Family

Javad Mashinchi came into this world by C-section on November 1, 2001. He was originally diagnosed with Myasthenia Gravis but at 21 months, we finally got the correct diagnosis of Myotubular Myopathy. Through a chance meeting, I met another family with an affected son and then found an Internet group of MTM mothers. All of the sudden I had community...a family. It was amazing to be able to talk to people that understood about our life, the ups and downs, the stress and celebrations. After going to the first MTM-CNM Family Conference, I knew that I had found "my people." My connections with different members of the MTM-CNM Community have grown over the years with the help of Facebook, and I consider them family.

In March 2015, Javad had a major setback. He had a massive seizure that caused cardiac arrest which resulted in essentially being in a coma-like state for almost six months. This event changed his ability to drive his wheelchair, move his arms, and do many of the things he had done previously. We continue on the slow road to recovery, but are hoping to get back to his old sassy self. He recently got a new wheelchair which has helped him be more mobile and we are waiting for a new speech board which should open his world. Javad starts high school in the fall and we are looking forward to his world opening up for him. Our family includes Javad, parents David and Shannon, older brothers Adam and Simon, Adam's wife, Sam, and sister Stesha. Javad is well loved by an amazing group of nurses, friends, and, of course, his family!



Stesha, Adam, Javad, David, Shannon, Simon

Our Families



(Front) Jasmin, Giovanni, Lupe
(Back) Joselyne, Chano

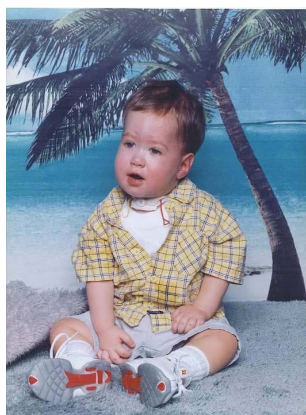
The Arteaga Family

We are the Arteaga family from Chicago, Illinois. The birth of our son Giovanni brought many new experiences into our lives. Some of them have been good and some have been very scary and challenging, but through it all Gio's strength keeps shining, and we are always fighting together with Gio. Through the MTM-CNM community, we are happy to have new friends and family. #togetherevenstronger

Gio is now three years old, and his current interests are in automobiles, especially the ones serving the community such as police cars, ambulances, fire trucks, school buses, and garbage trucks. He also loves monster trucks! At the MTM-CNM Family Conference, we are looking forward to learning more about the clinical trials and how this could help Giovanni and his friends with MTM.

The Biddle-Scott Family

Hi we are the Biddle-Scott family. We have had so many things going on over the past year!! Ashton's grandma Dawn earned her Angel wings June of 2016!! She is with our precious Angel boy Austen, who would be 15 next month. We also lost our family dog July of 2016!! This has been tough dealing with but we have been very blessed. In September of 2017, Ashton will become a big brother! We are expecting a little girl Addison Dawn! We are beyond excited! Ashton is getting ready to start the 7th grade. He went to MDA camp for the 6th year in a row and earned the butterfly award for trying the most new things.



Austen



Samantha, Ashton, Danny



Our Families

The Bowers Family

We are very excited that this year's Family Conference is in Nashville, our hometown since moving from Old Lyme, Connecticut 4 years ago. Although our son (and brother) Patrick passed away from MTM at the age of 8, almost 13 years ago, we continue to feel blessed to have had him in our lives and to remain a part of this amazing MTM/CNM Family. Patrick's love and gifts are felt every day in our lives and by those he touched. We know he has the same hope that we have, that at some point in the near future this disease will be treatable and will bring some well-deserved peace to those still in this fight.



Patrick



Sophie, Molly, Patrick, Carol

The Brassfield Family

We are from Oklahoma City, OK. I am a registered nurse entering my last year of my Family Nurse Practitioner program at the University of Oklahoma in the fall. I currently work for an infectious diseases practice as an infusion nurse and local hospital as a house supervisor. I have 3 awesome kiddos. Jaden is 13 and will be in 8th grade next year, Chason is 10 and will be a 5th grader, and Keegan is 9 and will be in 3rd grade. Lane would have been 16, he passed away just before his 9th birthday from complications of x-linked myotubular myopathy. He was silly, smart, stubborn, ornery and wonderful. He loved watching TV and his favorite movies were Cars and Fast and Furious. He kept us on our toes (ask about him taking his wheelchair for a swim). We couldn't take him anywhere without him running into someone that knew him. We were able to bring him to the first conference in 2009 and loved the experience and friendships made. I enjoy the memories that the sights and sounds of vents and suction machines and other essential equipment bring back at the conference. Hearing about the advancements in research, catching up with old friends, and listening to my heroes speak are some of the many reasons I look forward to every conference.



Lane



Keegan, Chason, Jaden, Rebekkah

Our Families



Colten, Travis, Kim, Evan

The Brown Family

Hi, we're the Brown family. Our son Evan was born on October 16, 2008. He came into this world blue and floppy. We were completely shocked and devastated, we weren't sure if he was going to survive. He was sent to Cincinnati Children's Hospital where he lived for six months. During his stay he was diagnosed with x-linked myotubular myopathy. After his diagnosis, Evan received a Trach and a G-tube and we started learning how to take care of him and his new equipment.

Evan came home on April 23, 2009. It was a huge adjustment to say the least. Our fourteen-year-old son, Colten has been the biggest help. He was only five when Evan was born, but he's his best friend and one of our best nurses. We've had our ups and downs over the last eight years but through it all we've gotten closer and stronger as a family.

Evan attends the local elementary school where he is a favorite among his peers. He often brings home notes and art work from the kids in his class. One of the highlights of this past year was organizing a 5k run/walk to raise money for the conference. We had a total of sixty-eight runners/walkers including the Bushey family and Myo the Traveling Turtle. It was a fun day, we had beautiful weather and it was an awesome way to bring the community together to raise money for a great cause.

The Browning Family

We are from St. Louis, Missouri and the proud parents of Jacob, a sweet, funny and determined 3 year old. Jacob was born on July 1, 2014 and after 3 1/2 months in the NICU, Jacob was able to come home on CPAP through a nasal cannula. It wasn't until 6 months of age that we had a diagnosis from genetic testing.

Jacob is a hard worker. He receives a great deal of physical therapy, occupational therapy and speech and language therapy each day. He LOVES to learn. We have always had a no limits attitude with Jacob and he constantly rises to our celebration of him with what he learns and can do. His favorite activities are playing with his animals, drawing, playing ball with the family (he always dictates who get the ball next), doing his language flashcards, and watching Sound of Music or Lion King. Jacob steals the hearts of everyone he meets with his sweet spirit and smiling eyes. It is so fun to watch him learn and express his wants and needs with his sweet and fun personality. He loves playing tricks on us and cracks himself up when he does so!

Jacob has a 6 year old brother Sam who will be going into first grade this year. Sam just adores his little brother and all he wants to do is constantly be around Jacob, and he tells everyone that he meets how adorable his 'Baby Jacob' is. He is Jacob's biggest fan and their bond is a beautiful thing to see.

We have been blessed with 3 steady nurses that help us care for Jacob. We continue to believe Jacob grows and improves on LOVE and everyone who meets him, falls in love with him (we call it the Jacob vortex)! We know Jacob's potential is limitless and we continue to trust God as we love and care for our sweet boy. (Psalm 139 - 'Jacob's Psalm')



Joanna, Sam, Jacob, Garrett

Our Families

The Bushey Family

Hello! We are the Bushey's, Matt, Natalie and twins Lilly and Cooper. We live in Springfield Ohio with our second graders. Cooper was diagnosed at 7 weeks with X-linked MTM, but it was evident at birth that something was wrong.

Being pregnant with twins I was always prepared for something to go awry, like having them early, not feeling a lot of movement etc., but having twice weekly ultrasounds and other testing we were told that there was "one in a million chance something would be wrong" and that both babies "looked perfectly healthy". Fast forward to 36 weeks when my water broke and craziness ensued. Like I said previously, when Cooper was born there was a panic that took over the room. All I heard was for someone to call the Neonatologist and not a baby crying. Then, in a blink of an eye someone went running with a bundle of blankets that was Coop. Five hours later he was finally stabilized and transferred to a higher acuity hospital. One more transfer, a major complication from a PICC line and 89 nights later we were finally all home together!

We have enjoyed traveling, going to baseball games and the zoo as a family. We certainly would not be where we are today without the love and support of this community. We feel incredibly lucky to be surrounded by so much love and support and we can't wait to get to know new families and visit with old friends!



Matt, Lilly, Cooper, Natalie



Jason, Briar, Stacey, Browning, Damon

The Cummins Family

Hello, We're the Cummins Family...Damon, Stacey, Jason, Briar, and Browning from Bessemer City, NC. We're excited to be at another MTM/CNM Conference after attending the first one in 2009! We look forward to making many memories with our "family"! We plan to walk away with closer bonds to each and every one of you!

Our Families

The Davies Family

We just recently moved to Phoenix, Arizona from Kansas City, Missouri. Ryan is a stay at home father. Kayla just started her nurse practitioner career at Banner MD Anderson. Makenzie is an active five year old who is the proud big sister of Grayson, two and a half years old. Makenzie is looking forward to starting kindergarten in August. Grayson continues to beat the odds and is a very active little boy.



Kayla, Makenzie, Ryan, Grayson

I was so excited when I found out that I was pregnant with my little boy. I had just lost a baby, so I was very anxious. I just had a feeling that something was terribly wrong. The doctors said everything was great. While I was pregnant with Makenzie, I felt every movement. This time was very different, I didn't feel him move at all. He was born two weeks early as a "floppy" baby. Immediately he was intubated, but was weaned to nasal CPAP within a day. We were transferred to Children's Mercy when Grayson was two days old. Grayson was found to have myotubular myopathy at four weeks old by a genetic test. Myotubular myopathy is just a part of Grayson's genetic problems. He also has a genetic condition, aniridia. It is a lack of iris and comes with a host of other eye complications, including legal blindness. Ryan and Makenzie also have the condition.

Grayson has been in and out of the hospital, but is doing fairly well at this time. We are looking forward to attending the conference and meet new and old MTM/CNM warriors and their families! Grayson and I attended the 2015 Family Conference and was overwhelmed with the support in the MTM/CNM Family.

The Ellis Family

Ayden Carter Ellis, better known as ACE, left to be with our Heavenly Father at just 17 months of age on May 12, 2014. During his time here: he astounded his doctors, physical therapists, and every one who knew him. His strength and determination to conquer MTM amazed us all! He will forever be remembered for his 'Happy' dance, his 'get it, get it, go's' and his Elvis dancing. We miss you, Baby Bear, but we know you will forever show us signs that you are watching over us! Love you always and forever!



Claire, Ayden, Jamus

Our Families

The Fontana Family

Hi, we're Scott, Staceyanne and Mason Fontana, and we live in northern California. Staceyanne was diagnosed with myopathy in her 20's, while Mason has x-linked MTM. Our journey has been an amazing adventure, filled with happiness, heartache, laughter, tears, joy and always love. From his birth, we have tried our best to fill Mason's life with as many experiences as possible. Some of our favorite memories are coast trips, Halloween costumes, Disneyland, exploring other states, Mason driving his chair anywhere he could get it, weekends in San Francisco, even flying him to Houston, TX for the first MTM/CNM Family Conference. Our absolute best experience was Mason's Wish, spending the day with the cast and then he starred in his own special episode of Yo Gabba Gabba. Mason doesn't tolerate his chair anymore, so we've now focused our adventures at home... movie nights, family games, lots of silliness and snuggles. Mason's favorite things are watching his shows (usually Gabba!), kicking his legs in his anti-gravity frame, and cuddling his beagles- but most of all, he loves playing with daddy and singing with mommy. This journey has been much longer than any doctor ever gave us hope of (what did they know?!), but that's how our family and perfect little man rolls... blessed with one miracle after another <3



Scott, Mason, Staceyanne

The Forbis Family

Hello everyone, we are the Forbis Family. We have 3 kids who are Logan (5 years old) and a set of twins Madison and Mason (2 years old). All three kids keep us busy. Logan has X linked MTM and has been our warrior from day one. Logan likes to play ball and his iPad but most of all he loves being around friends and Family. We as a family have great support back at home in North Carolina. We have been looking forward to the conference for a long time. At the conference we just want connect and meet other MTM families as well as learn about the most recent research/clinical trials.



Madison, Kathleen, Logan, Brian, Mason

Our Families

The Frase Family

In February 1995, our son Joshua was born. We were told he would not survive the day. They discharged us from the hospital when he was 24 days old with a NG tube, a suction machine, and apnea monitor. At 3 ½ months of age a muscle biopsy confirmed Myotubular Myopathy and at the time only 55 cases were known worldwide. They told us, “if he lives to see his first birthday, bring him back and we will re-evaluate him.”

It was the grace of God that Joshua survived so many life-threatening episodes because Paul and I were completely uneducated in how to take care of such a fragile little baby.

Before Joshua’s first birthday, Paul and I founded the Joshua Frase Foundation, which would raise money for research on a disorder that at that time, neither NORDS nor MDA had any information on. In fact, the only information we had access to was a packet of copied textbook pages on MTM sent to us by our neurologist. There were very few publications written at that time, some which were from the 1960’s. I thank God that we found Pam and Gary Scoggins who educated us on the care of our son. By 1997, our first team of researchers was established at Boston Children’s Hospital, Harvard Medical School. That began a two decade journey of working towards a cure for our precious MTM children. Through the years, we have expanded that partnership to Wake Forest Institute for Regenerative Medicine, the University of Washington, the University of Florida and Toronto Sickkids.

Paul retired from the NFL in 1999, and we set up shop in Ponte Vedra Beach, FL where we raised our two children (we had a healthy little girl, Isabella, when Joshua was 6 years old). Joshua and Isabella had a wonderful sibling relationship, and we tried to make life as normal as possible for both of them. Our precious son died on Christmas Eve 2010, just 40 days shy of his sixteenth birthday. He taught us so much about life. He possessed such courage, tenacity, and optimism in the battle for his life and never once did he question, “why me, God?” He inspired us to take risks and to never give up when the going got tough. His life inspired us to pursue our dreams with everything in us.

Paul and I still live in Ponte Vedra with our daughter Isabella. Our days are full of normal life stress, high school drama, our foundation work and carrying our son’s legacy with us in everything that we do. We would LOVE to meet you at this conference if we haven’t already met... and if we have, PLEASE come say hi!



Alison, Joshua, Isabella, Paul

The Fuller Family

My name is Linda Fuller and I live in Pachuta, MS with my husband and son who was diagnosed with MTM a few days after he was born. At that time, very little was known about MTM... there was no information what-so-ever available for parents, no home computer or Facebook... Things have really changed since that day 30 years ago! I thank the Lord for advancements and pray for our families, caretakers, and all those involved in research and support. So grateful to all who have made this conference possible!

Our Families

The Garland Family

We are the Garland Family from Greenwood, Indiana. Reese is 6 years old and getting ready to begin the 1st grade! He loves Hot-wheels, baseball, racing and bossing everyone around! Reese enjoys school, learning and seeing all his friends. This will be our 3rd conference and we cannot wait to reconnect with everyone.



Rob, Becky, Reese, Riley and JJ (the furry one)

The Gilson Family

The Gilson Family began in 1974 when we, Marshall and Trisha, high school sweethearts married. We were blessed with our first child, Stephanie in 1975. Stephanie was a very happy and content child and she soon was referred to as the “golden child” to all in our family. In 1980, son Ryan was born, a few weeks early. Ryan required the support of a ventilator and passed away at 11 days old. At the time we were to understand it was as a result of a brain hemorrhage. After genetic testing we were overjoyed to receive the go ahead to try for another child. In 1985 JT was born, pre-mature and not able to breathe on his own. We were devastated and struggled to understand how this could happen again. JT passed away just under 6 months old. The autopsy resulted in the diagnosis of X-Linked Myotubular Myopathy. In 2005, Stephanie received the news that she was a carrier. There was little information and support available in the 1980’s and for decades our family went through the motions of life, trying to heal. Our hopes in coming to the conference is to connect with the community, to gain insight into areas where may be able offer support, either in our home of Vancouver Canada and/or on a broader scope and to learning and participating in some of the research being done especially in the area of the female carriers, as we both have undiagnosed health issues.

The Gorenflo Family

This is one of our very first family photos, captured on the first day we were able to hold our son, a cherished memory. On December 5, 2016, Noah was born not breathing and a code blue was called. The flood of blue scrubs sprinting in and out of the operating room will be a memory that lives with us forever as a swirl of helplessness, confusion, and fear consumed our bodies. Noah spent his first four months in hospitals and it took a month until we received a diagnosis.

In spite of all that, we knew immediately that there is just something so special about Noah; he is a fighter, resilient, and just a happy baby.



Michelle, Noah, Joe

Being new parents, we have had a crash course in what that means; we had to learn to be advocates, decision makers, a voice for Noah! In our home, we are committed to maximizing Noah’s life and his life experiences and we share a united and guiding belief that Noah will be cured. It is our greatest honor and privilege to be Noah’s voices, advocates, and cheerleaders as we navigate each new experience.

Our Families



Jessica, Declan, Adam

The Hair/Marmon Family

Hi everyone! We're the Hair/Marmon family. Or, as we were nicknamed during our initial 19.5 month hospital stay, the "Declans." Yep, after being born a month early in March 2012, Declan spent 104 days in the NICU and an additional 16.5 months in rehab hospital (mostly due to insurance and waiver issues).

When we finally got Declan home at close to two years old, we ALL had to adjust to the change of living in the hospital bubble – where nothing is private – to living in the in-home nursing, therapy, medical world, where most things are still not private. But, how amazing to finally have our little guy home!

Declan was diagnosed with XLMTM at 7 months old, and has had to face many of the obstacles with which we're all familiar- trach, vent

support, g-tube, significant motor and developmental delays, as well as experiential delays from spending so much of his early life in the white, sterile walls of the hospital.

We've had ups and downs and ins and outs since we made it home from the hospital- bone breaks, gross and fine motor wins, surgery scares, being able to communicate via ASL and a speaking device, nursing issues, finally hearing his voice - but the one constant is always our silly, ornery, loving little boy.

Declan turned five in March. He just graduated from preschool and is preparing for kindergarten. He is *still* obsessed with Curious George, and has decided to start watching Jorge el Curioso- so apparently he's working on his fourth way to communicate. He loves his puppy Penny, the zoo, swinging, swimming, being destructive, drawing- especially giving himself and others tattoo sleeves, mom, dad, grandma and grandpa J and grandma and grandpa V. He is the light of our life, and we are so lucky to have him in it. We are so excited to be able to attend this year's conference, and are looking forward to all that the future holds for our kids!

The Hanna Family

Christian is 16 years old and attends Norman North High School where he is a member of the National Honor Society. Christian has a strong passion for music and is currently attending weekly classes and is learning to write music in hopes to one day pursue a career in this field. Along with music, Christian likes Netflix, You tube, going to the movies, and spending time with his sisters. Christian is truly a funny and unique spirit who is a blessing to our family. Through attending this conference we hope to gain insight and knowledge about MTM and the research being done to combat this disease. We want to be a more informed family on behalf of Christian, in order to create more positive outcomes for his life.



Christian

The Jackman Family

The Jackmans are from Banner, OK. Ronald is retired and likes to spend his time at the lake fishing. Dianne works as an AP & Inventory Manager for a local printing company. She enjoys sewing and working in the garden. They have three daughters Chrissy, Rebekkah & Ronda. Their son Ronnie Dale passed away shortly after his first birthday in 1975. He was born with x-linked myotubular myopathy. They have eleven grandchildren. Two of their grandsons have x-linked MTM, Christian who is 16 and lives in Norman, OK, and Lane who passed away in 2010. This is their first conference to attend.

Our Families



Jason, Kinsley, Jaxon, Ashley

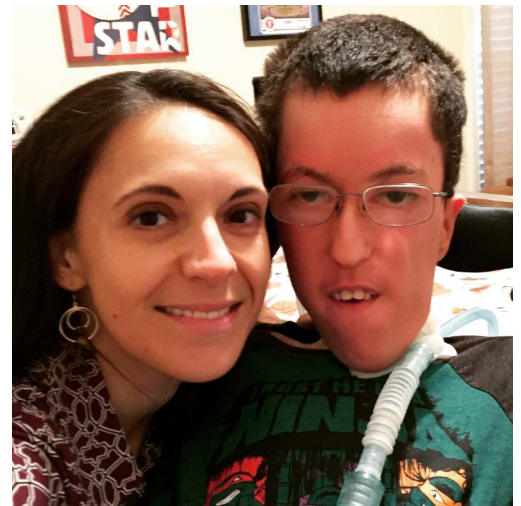
The Henson Family

We are the Henson's. Jason, Ashley, Jaxon, and Kinsley. We are from Inman, SC. Jaxon is 3 years old, and diagnosed with XLMTM. Some of Jaxon's favorite activities are playing ball, singing nursery rhymes, making crafts, blowing bubbles, and watching cartoons. Jaxon recently flew a kite on our last vacation and really enjoyed doing that as well! His favorite colors are orange and red and his favorite cartoons are Bubble Guppies and Team Umizoomi. Jaxon is such a bright, caring, and loving child! He is slowly warming up to his baby sister, Kinsley, and makes sure he gives her kisses each night before bed. He has such a fun and silly personality, that brings a smile to our faces and warms our hearts daily! Watching Jaxon grow and learn has taught us to look at the world differently, and for that we are very thankful. This will be our first time attending a conference, and we are very excited to learn more about treatment options.

The Hollingsworth Family

Jacob Hollingsworth is 15 years old and lives just outside of Atlanta, GA, with his mom, Amanda, and part time with his dad, Dennis. Jacob is transitioning into high school this year and is looking forward to coursework in S.T.E.M. to prepare for his chosen career path in medical technology or surgery. Jacob enjoys attending Atlanta Braves baseball games, watching videos, going to museums, spending time outside, drawing on his iPad, and going to the movies. Jacob has recently published a Kindle book for kids, and he is currently working on two follow-up publications.

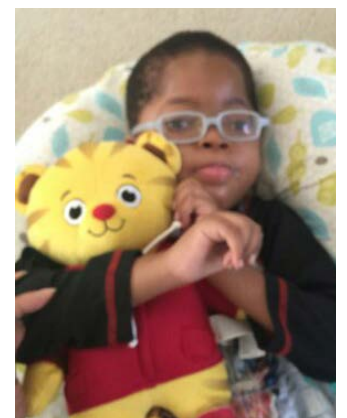
Jacob attended the first biannual MTM-CNM Family Conference in Houston in 2009. His experience in his words: "I loved meeting all my friends and just talking and talking." The Conference left a huge impression on Jacob and he has very much looked forward to returning to "see all of my friends again."



Amanda, Jacob

The Jones Family

The Jones family resides in South Carolina. Our son Matthew is 3 years old. Matthew enjoys book readings, listening to music, and playing with musical toys. Matthew's favorite TV shows are Dinosaur Train, Super Why, Floogals and Daniel Tiger's neighborhood.



Matthew

Our Families



The Lawton Family

Our son, Mitchell, was diagnosed with XLMTM at five months of age via muscle biopsy in 1990. At the time we did not know of anyone else with this diagnosis so we observed Mitchell and let him teach us what he needed to better his life. It was soon apparent that the biggest obstacle was respiratory, we sought help from pulmonologist and used our own medical knowledge to optimize his ability to prevent pneumonias. There was so little information in the early 90's but Mitchell was thriving and we felt optimistic. Here we are today, Mitchell is 26 years old and studying video animation at the University of Colorado on Denver's campus. It's been an extremely hard road and there were many, many times where we were fearful that he would not survive but Mitchell is the strongest person that we know. His

Charlie, Donna, Mitchell, Kyle

ability to live in the moment, make people laugh and feel loved inspires us everyday. Mitchell's younger brother, Kyle, is 22 years old and living in Hollywood, CA studying music. He's in the Bachelor's program at Musician's Institute with a primary focus on the electric guitar but he also plays drums, keyboard, writes and sings. He hopes to make a career in music either by playing, producing, teaching or all of the above. It will be fun to see where he takes his music. Charlie and I moved full-time to Las Vegas a year ago and we are loving life in the desert. Charlie retired from full-time work as an ICU physician and is working about a week a month around the country wherever his expertise is needed. He loves being able to only work when he wants, is enjoying the free time to play pickleball, golf, hike and is keeping his mind sharp by learning poker. I fully retired from nursing after having worked off and on for 33 years. I'm now able to work hard on my poker game with the poker playground in our backyard. Most everyone makes it to Las Vegas at one time or another and Charlie and I love to play tourist guides so if you ever find your way to our town please let us know. Would love to meet up! Being at the conference this year is very exciting as we look forward to hearing about the upcoming human trials for the gene therapy!



The Lewis Family

Caden came into the world May 14, 2002 and was quickly followed by his sister, Kate Elizabeth, 18 months later. At birth, Caden had a weak cry, didn't open his eyes, couldn't latch on and doctors described him as floppy. Within a few days he was able to take a bottle and also nurse. Concerns from the hospital nursery and pediatricians lifted and we settled into life with our new baby. Throughout his first few years his milestones were slightly delayed but still within a "normal" window in spite of his low muscle tone. At the age of four, our pediatrician referred us to a neurologist and he was diagnosed with X-Linked MTM through muscle biopsy. Caden is now 15, he is witty, talkative, loves to travel, watch movies, play video games, build Lego sets and play Minecraft. He is

Kate Elizabeth, Marty, Jessica, Caden

entering the 10th grade in high school! He works hard in spite of his weaknesses and because of that, he inspires more people than he may ever realize! Caden is a joy and we are blessed to be his parents. We are very excited about attending our second MTM-CNM Family Conference.

Our Families

The Lofton/Scott Family

Roy and I have a son, Corey. On Corey's 13th birthday we discovered that we were pregnant. My pregnancy was complicated by a lack of fetal movement and severe polyhydramnios. My doctor assumed that the polyhydramnios could be due to a cleft lip and/or palate and that the lack of fetal movement was due to the polyhydramnios. On June 9, 2003, the doctors decided that Cason was no longer benefitting from being in the womb and preformed a C-section. Immediately upon birth I was permitted to give Cason a quick kiss on the forehead before he was rushed to the NICU with respiratory failure and hypotonia. He was like a little rag doll, no muscle tone at all. Cason spent the next 30 days in the NICU and was discharged with an apnea monitor.

Cason spent only 5 days at home before being air lifted to Sacred Heart Children's Hospital in Pensacola with respiratory issues. The doctors performed a muscle biopsy, numerous blood test, MRI's, etc. in an attempt to diagnose Cason's medical complications. No results came back and we were told that Cason would probably not see his 1st birthday. We spent the next 2 ½ years in and out of the PICU with respiratory issues and Cason's muscle tone remained extremely low and we still had no diagnosis. Another muscle biopsy was performed and we were finally diagnosed with X-linked Myotubular Myopathy. Along with the muscle biopsy Cason was given a tracheostomy, a feeding tube was placed, and an orchiopexy for undescended testes.

When Cason was 5 years old tragedy struck. We lost Cason's dad, Roy, unexpectedly. Roy was a great dad that loved our boys with all of his heart and we miss him terribly. I hate that with all of the challenges Cason has he now has to fight them without his dad by his side. Somehow, Cason, Corey, and I have managed to stick together and get through one obstacle after another. Cason is now 14 years old and is such an amazing young man. He is so smart, creative, and imaginative. He is an amazing son, brother, and now uncle. I pray that a cure or treatment becomes available soon because he, along with his other MTM/CNM brothers and sisters, have so much to offer this world.



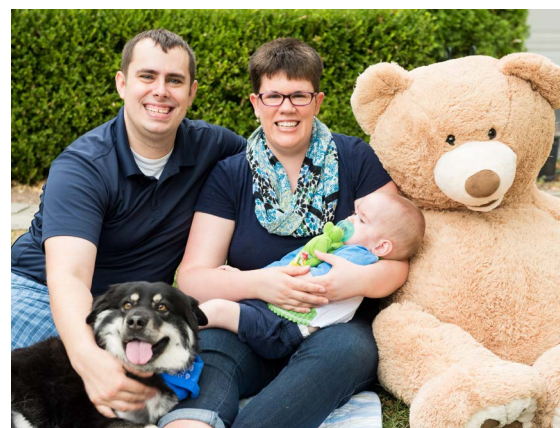
Dana, Corey, Cason, Roy

The Lykens Family

Hi Everyone! We are the Lykens Family: Craig, Lindsay, Happy (6), and Gil (almost 2), and we are from Ashburn, VA (about 25 miles outside of Washington, DC). Craig is an Assistant Manager for the Benchmarking Department at the National Business Group on Health in Washington DC, and Lindsay is the Corporate Office Manager for Command Security Corporation in Herndon, VA. We are excited that we will be joined by all of Gil's grandparents at the conference: Joe & Sherri Gillam, Tammy Lykens & Mary Jane McCall, as well as Gil's favorite nurse, Amy Truluck.

Gil is a playful and happy kid, who loves to laugh and be tickled, loves to listen to music and wiggle-dance, loves balance items on his feet, loves to fingerprint, and who very recently tried a tricycle for the first time (and he loves that as well!) Gil is starting to learn sign language, and knows about 30 signs right now. He gets very excited and happy when he sees other people using ASL, and thinks it is so neat when Mom and Dad sing and sign the ABCs with him.

We are so excited to meet you all at the conference!!

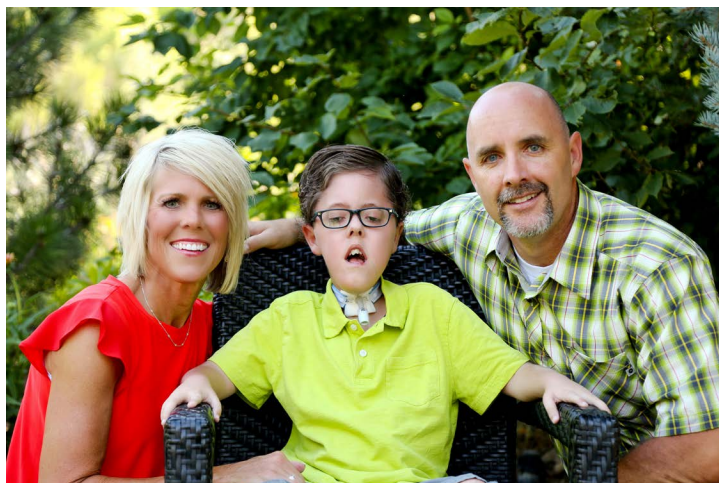


Happy, Craig, Lindsay, Gil

Our Families

The Maughan Family

When I (Eric) think of our family, what amazes me the most is how fast time passes. It seems like yesterday it was March 1990, when Debbie and I were married. 27 years later, we have a growing family; 4 children, 5 grandchildren, and a very busy life. I work from home as an independent sales representative. I enjoy anything outdoors and staying fit. Debbie is a



Debbie, Kash, Eric

making up stories and pretending to be a ninja. He dances, sings, and loves cuddling. He says he's going to be a professional You Tuber when he grows up, and I believe him. Our world is Kash, and we're fine with that.

marathon runner, just completing her first Boston Marathon in April 2017, and has Chicago and New York marathons on the list to be ran this year. On top of that, she is the one who holds our home together, ensuring that all is taken care of. She is an amazing woman!

Kash is the center of our universe. Everything revolves around him and his needs. He is the reason we do what we do, live the way we live, and have the perspective on life that we do. This perspective, living one day at a time, is what has kept us going. Kash gives us the strength and perspective we need. He is the most caring, loving, forgiving, and special boy that we have ever met. He is a grown man in a 10-year-old body. He understands his limitations and accepts them. But, he also dreams of walking and talking and being able to be just like everyone else. He loves computers, gaming,



Richard, Cooper, Calli, Reid

The Mayotte Family

We are most excited to connect in person with other families like ours and allow our oldest son, Cooper, an opportunity to meet other siblings of MTM Warriors. We hope to learn a great deal about all the research happening in our community!



Our Families

The McDermott Family

Greetings from the McDermott family: Daniel, Robin, Will (5) and Ryan (4). We live in sunny Phoenix, AZ. Ryan was diagnosed with CNM just before his first birthday. It took almost another year before we got the official XLMTM diagnosis through genetic testing. He has a few other medical challenges, but MTM is the most prominent.

Ryan is a very happy boy who loves toys that light up and make noise. He is especially fond of his drum set and switch adapted fans. He loves to dance and wiggle to music, play peek-a-boo, go swimming, and play with his family and friends. Will loves being a 'Big Brother.' We often see him showing Ryan fun things on the iPad, and reading books together. We all love doing things as a family and never let MTM stop us from living life to the fullest. We are grateful for all the support from our fellow MTM families. We have learned so much. With such a rare disease, we are often teaching the doctors and we would not be able to do that without this group. We are excited to see old friends and meet new families!



Will, Robin, Ryan, Daniel

The Miller Family



Kayla, Kyle

Hi, we're the Miller family, Jimmy, Darlene, and Kayla. We are from Lexington Park, Maryland. Sadly, our Kyle passed away on March 25, 2016. He was the crazy kid in the wheelchair that always liked to race everyone at the conference in 2015. I'm glad he got to be a part of that conference, as he talked about everyone and how much fun he had for months.

We are a busy family: Jimmy is a welder and I'm a Special Education Para Educator. Kayla is finishing up high school this coming year. Then off to college in the medical field. We enjoy trips to the beach and camping. Fishing and crabbing are still a big part of our lives (it was Kyle's favorite thing to do). We find ourselves talking about Kyle all the time. Like would Kyle do this or this is what Kyle would do. Even though he is no longer with us, he is always in our hearts.



Darlene, Jimmy, Kayla

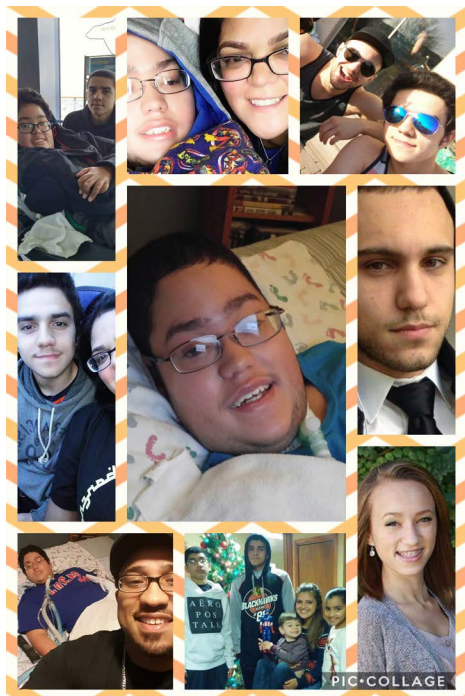
Our Families



Jayden, Brendon, Krysta, Camron

The Monk/Slemp Family

We are the Monk family: Brendon, Jayden, Camron and Krysta. We started our MTM fight on 11/20/14. Sadly, our little soldier Camron lost his fight on 6/2/17. But our family's fight has not ended. We work hard, we play hard, we live, we laugh, we love and we never take a moment for granted. Camron has always been and will always be our inspiration to get moving forward every day. We can't wait to meet you all. Much love, the Monk family.



The Munoz/Rocha Family

We are the Munoz/Rocha family! (Christi, AJ, Jason Jr. and Jason Sr., Julian and Destiny)

Jason was diagnosed with X-Linked MTM about 21 years ago, at 2 weeks old. Jason loves his computer and Pokémon! Jason is very transparent, and says what is on his mind! He has a very strong personality. (Sorry, he gets that from his Mama and Dad!). His strong will and determination has been Jason's motto his whole life! Jason excelled in all years of schooling, accepting two Presidential awards and being on honor roll many times.

Jason is starting to begin his journey into adulthood by furthering his studies with attending college. Jason has taken it upon himself to get into college doing things like other kids would do. He wants minimal help from parents! (This is really hard but makes us really happy)

Jason loves all his siblings, Julian, Destiny and AJ. He has a special bond with each. Jason is always supported by his grandparents (Nana and G-pa John), aunts (Angela, Lily and Shelley), and cousins (Sadie, Efrain, Gianna, Johnathan)! Jason makes us ALL stronger every day!

Our Families

The Orellana Family

We have three sons in our family, Dylan 26, Luciano 19, and Sebastian 14. This year we added a pug puppy named Draco, after the constellation - although Harry Potter is well-loved in this brood. We live a comotion-filled life in Northern California where we run our family business and the guys attend school. Sebastian is diagnosed with CNM from a Dynamin2 mutation. He is a self-avowed geek, avidly interested in computers and coding. In the Fall, he will attend a STEM academy for high school and pursue more of his interests there. Sebastian is also passionate about games, from chess to card to video to board games, and just now getting into D & D and Settlers of Catan. He loves to hang out with friends and his brothers, swim, and read as well.



Dylan, Jeanne, Sebastian, Luciano, Luis



Ashley, Melisa, Austin, Kelsey

The Saalwaechter Family

My name is Melisa Saalwaechter and this is my family. We are from Owensboro, KY. My daughter, Ashley is pictured on the left and she is 30 years old with a husband and 2 children. Kelsey, my other daughter, is 28 and at 5 days old was diagnosed with MTM from a muscle biopsy. Kelsey is a college graduate and works for a doctor's office. My son, Austin is 24 and is also a college graduate working for an electrical supply company. He hasn't been officially diagnosed, but has similar characteristics as Kelsey, so we assume he also has MTM. We visited a pediatric neurologist as the kiddos were growing up. The doctor always said my children have a mild case. Most people don't know they were born with a genetic disease because they both have done so well. We feel very fortunate. When Kelsey was born 28 years ago, I felt so alone and scared. Doctors knew very little about MTM and could give us very little information. Social media has helped us realize we are not alone and we are excited to be able to attend the conference this year! We are hoping to be led to a true diagnosis if it isn't MTM.

Our Families



Pam, Gary, John

The Scoggin Family

We're Pam, Gary & John Scoggin from Texas City, TX. We've been around the MTM community for a long time! John is 28 and holds a degree in Journalism/Communications from the University of Houston – Clear Lake. He stays busy writing online guides for video games. We have four dogs, including Pink, who came to us from the MTM dog colony.

The Sirmon Family

Benjamin Dean Sirmon was born February 16th, 2012 and remained in the NICU for three months. Once discharged, he was welcomed home by his two big sisters, Lennon and Isla. The month we attended our first conference, Benjamin was officially diagnosed with DNM2 Centronuclear Myopathy, a rare, more severe presentation of the disease. For three years, Benjamin traveled, enjoyed sporting events, loved the pool, danced along with his sisters, and made the world a better place. Benjamin, along with his friends and family, completed a fully accessible playground in South Louisiana before moving to North Louisiana where he quickly starting promoting and raising money for an even larger fully accessible playground. During this time, Benjamin welcomed with opened arms (eventually) a little brother, Rush.



Benjamin



Hillary, Kateri, KC, Isla, Rush, Lennon

One week after his third birthday, Benjamin passed away peacefully. We do not mourn the loss of our Superman, but celebrate his life, love and accomplishments. The following month, we found out we were expecting our fifth child. She has Benjamin's spirit and love of others. Our new little girl and the rest of our family will always have the most devoted and special guardian angel.

In June, The Smiles Park, another accessible playground we had worked so hard to promote, opened with Benjamin Bear in the center. Our family will continue to work towards accessibility for people with disabilities in Benjamin's honor.

Our Families

The Slaby Family

My name is Joe Slaby and I live in Terre Haute, Indiana. I was diagnosed with Centro Nuclear Myopathy when I was ten in the spring of 2001 (the Dynamin 2 genetic mutation was later diagnosed at Begg's Laboratory). Although there were symptoms at an early age, my progression didn't really truly begin until near the end of my middle school years where I began to experience greater mobility issues. I've been using a scooter full-time now for about three years whenever I leave my house. During that time I've been able to finish my college degree, work part-time during college, and eventually work full-time in an internship at a public accounting firm during my final semester. This past spring I graduated from Indiana State with a Bachelor's in accounting and I'm currently studying for the CPA Exam full-time. My folks have been beyond supportive of me every step of the way and I'm lucky to have them. In addition, the MTM-CNM Family Conferences have brought me close to a bunch of awesome people who I wouldn't have known otherwise. I'm beyond grateful to go to a place and connect with so many individuals every other year.



Andy, Xander, Jack, Carrie, Joe, Jack, Carol



Julie, Molly, Nathan, Trooper, Phil

The Swed Family

Hello! We—Julie, Phil, Nathan, Molly and our pup Trooper—are the Swed family. We hail from a small town in central Texas called Hutto. Nathan is 4 years old and his sister Molly is 8 months. It wasn't until Nathan was born that we realized we were in for a very atypical parenthood journey. With the help of Nathan's equipment, nurses, therapists, doctors, and Nathan's grandparents we are able support Nathan so that he can live a great life. He is full of joy and fun; we praise God every single day for the opportunity to love on him and his sister. He adores going to school, being a big brother, and playing trains. Being a part of the MTM community has made all the difference in our journey as parents; being connected to families who understand the specific battles we fight and celebrate our victories all the more because they understand what went into them is invaluable. We are so glad to be here and so excited for the opportunity to meet y'all!

Our Families

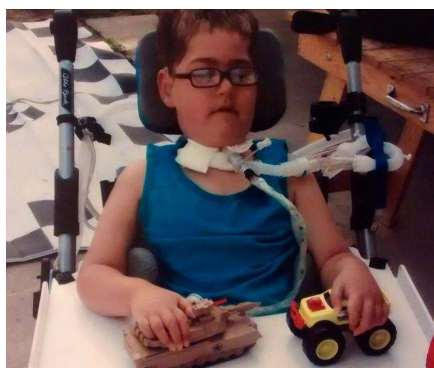
The Walker Family

On May 11, 2012 Jayden and Alexander came into this world at 27 weeks on the island of Hawaii. Jayden loved to watch super heroes on his iPad with daddy Johnny, while Alexander preferred a hot bath followed by a massage. Our sweet babies became angels on July 11, 2013. Our focus is to become as educated as possible and ultimately find a cure for mom Ashley. If we have not yet had a chance to connect please feel free to contact me by email anytime.

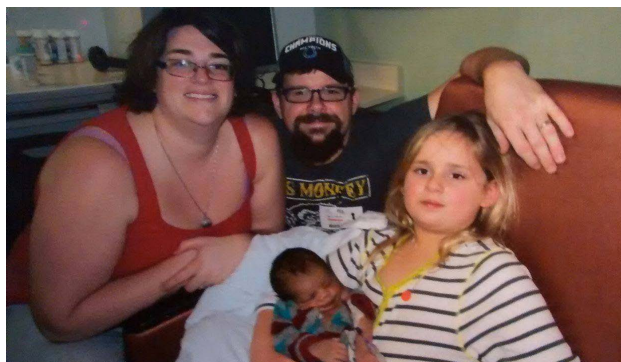


The Wilhelm Family

We are the Wilhelm family: Lindsey, Martin, Emily, Louie, and Joey. Our sweet Louie was born in Springfield, IL on June 17, 2011 with Myotubular Myopathy (MTM). All of us attended the 2013 MTM-CNM Family Conference, and we enjoyed meeting other families in person. Sadly, Louie passed away in August 2014 at the tender age of 3. His time here may have been short yet it was full of love. Louie is deeply missed and we hold his memories close to our hearts. The Illinois General Assembly passed a Resolution to make June 17, 2015 X-Linked Myotubular Myopathy Awareness Day in honor of Louie's birthday. This is just one way his light continues to shine in our lives and for all who knew him. Some time after Louie passed, we began dreaming of bringing another child into our family, and earlier this year that dream came true with our adopted son, Joey. We attended the 2015 conference and are happy to be back again.



Louie



Lindsey, Martin, Joey, Emily

Our Families

The Welter Family

We are the Welter family from Minnesota: Matt, Amy, Allie and Andy. This year Andy will be turning 16, seems like yesterday that he was a baby and we were just hoping he would make it to 1. Today Andy loves to travel, just recently we flew with him out to San Francisco. Andy had the time of his life, he loved flying and he did fabulous on the airplane. Some of Andy's interests are girls with short skirts on or anything that shows legs, yes he is a teenage boy. He also loves cats and dogs, he took an animal science class in high school and found his love of animals. Andy also likes to play on his surface (computer), and he loves to look up youtube videos of tunnels and hotels. He likes to plan his travel on what cool hotels he finds. Every year, he thinks we need to go on a family trip someplace.

Allie is going to be a junior in college this fall, at University of Wisconsin Eau Claire. Allie spent the past summer doing an internship with the Begg's Lab in Boston. What an opportunity for her, as her major in college is Biochemistry/Molecular Biology.

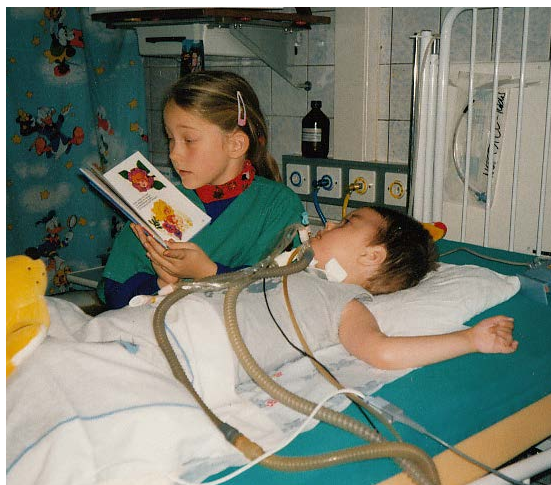
Matt and I (Amy) just had our 25th wedding anniversary and this will be our 5th MTM-CNM Family Conference. We have met some amazing families and can't wait to meet more this year in Nashville!



Matt, Andy, Amy, Allie

The Wüstner Family

My younger brother was affected by Myotubular Myopathy, he passed in 2009 at the age of 15, exceeding every prognosis doctors ever dared to make, enriching not only our life but the lives of everyone willing to get to know him. Marten was born in 1994 in a tiny community hospital and was immediately transferred to the University Hospital. When my mum was told that he is severely ill and might not live long enough for her to see him she told them "He is my son and I know he will not die." And that was it. Winning against the odds, surprising everybody with his strength, demonstrating his will to live and his way to celebrate life and making everybody smile became his attitude. Giving life a deeper meaning, making the ordinary extraordinary and teaching us that "impossible" is a non-existing entity. Our favorite brother-sister-activity was reading books, sadly we never made it through the Harry Potter series and of course teasing your big sister is always fun. He inspired me to become a doctor and to take responsibility as a work group leader in our German patient organization ZNM-together strong e.V.



Arlene, Marten



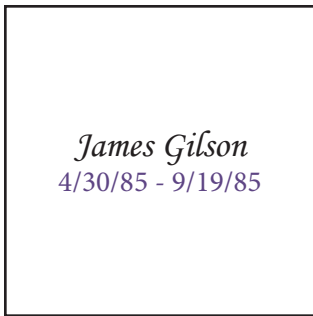
In Memoriam

We honor the legacy all those who have passed away from Myotubular Myopathy or Centronuclear Myopathy. The individuals shared here are beloved children or siblings of families attending this year's conference. We celebrate their memory as we gather together. Their spirits will always be with us. Please take some time throughout the weekend to visit our Memorial Reflection Room, a special place to honor, reflect, and celebrate the lives of all our loved ones who have passed away.



Patrick Bowers
7/25/96 - 11/5/04

Ayden Carter Ellis
12/27/12 - 5/12/14



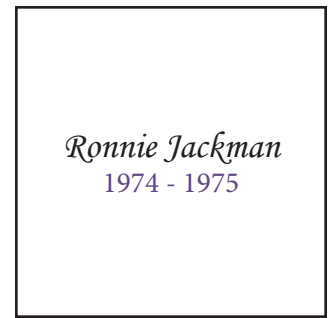
James Gilson
4/30/85 - 9/19/85



Ryan Gilson
7/16/80 - 7/27/80



Lane Jackman
10/16/01 - 9/9/10



Ronnie Jackman
1974 - 1975



Connor Miller
10/11/05 - 8/12/08

James Kyle Miller
12/15/01 - 3/25/16



In Memoriam

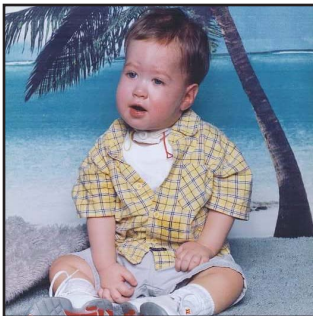
“They are not gone who live in the hearts they left behind”



Joshua Frase
2/2/95 - 12/24/10



Camron Monk
11/20/14 - 6/2/17



Austin Scott
8/14/02 - 2/6/04



Benjamin Sirmon
2/16/12 - 2/23/15



Alexander Walker
5/11/12 - 7/11/13



Jayden Walker
5/11/12 - 7/11/13



Louie Wilhelm
6/17/11 - 8/5/14



Marten Wüstner
6/17/11 - 8/5/14

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REEVE FOUNDATION
QUALITY OF LIFE
GRANTS PROGRAM

Family Fundraisers



The Brown Family organized an amazing "Run for Evan" 5K race



The Garland Family turned Rob's Slot Car Racing into a great Conference Fundraiser



Make a little magic...
Support #mtmcmfamily
www.mtm-cnm.org

The Ward Family "saved the beard" and celebrated "16 is Sweet" on Crowdrise



The Lykens Family "stood up for our boys" and raised funds with "Team Gilbug" t-shirts through Bonfire

Family Fundraisers

thirty-one™

Gifts, totes, bags sale by Calli Mayotte



Lemonade stand, bake sale, yard sale by the Harper Boys for Will Ward



Women's clothing sale by Joy Holmes for Reid and Paley Wood



Women's clothing sale by Shannon Mashinchi for Javad Maschinci



Scented products for the home sale by Heather Kirchmeyer for Reese Garland



younique

Makeup and skin care sale by Michelle Hill for Reese Garland

Family Fundraisers

Thank you to our Crowdrise team for "stepping up"
in support of MTM-CNM Family Connection

The Brown Family "Run for Evan"

Will Ward

Mark Ward "To Shave or Save"

The Garland Family

The Lykens Family "Team Gilbug"

The Hair-Marmon Family

Claire Hackett

Reid and Paley Wood

Amanda Hollingsworth

Christann Rocha

Javad Mashinchi

Stacey Cummins

Calli Mayotte

Ashley Henson

Natalie Bushey



crowdriseTM

If you don't give back no one will like you

Donors

With gratitude to our friends and family for giving from the heart:

Crystal Amos	Monica & Doug Hoepfner	Patti Reust
Michelle Anderson	Joy Holmes	Rose Family
Wendy Appel	Bill & Marianne Howe	David Rinaldi
Barghaan Family	Hughes Family	Christi Rocha
Gary Baumgard	Diane & Paul Huseth	Run for Evan
Naiem Bello	Margaret Klynstra	Penny Sage
Tammy Bennett	Kinnelon Education	Robert Schloss
Shirley Brown	Heather Kirchmeyer	Scully Family
Garret Browning	Emily Kmiecik	Matthew Seymour
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Couto Family	Lularoe Co.	Damian Siskavich
Mary Crauderueff	Lykens Family	Tim Stevens
Andrea DeBow	Eva Martin	Stansbury/Hanson Family
Barbara Deyermond	Javad Squad	Suarez Family
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Pamela East	Kate Maul	Beth Trifiletti
Simon Ellgas	Calli Mayotte	Angelica Santiago Townsend
Sheryl Forsberg	Susan McCarthy	Ellen & Doug Waddill
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Tina Cheng Faust	Lynn Mollica	Ward Family
Jen Bilbao & Holger Fischer	Pat & Tony Novotny	DKBJ Ward
Sheryl Forsberg	Tina Nguyen	Eileen & Leo Ward
Leslie Gaffney	Susy Ocampo	Ivy Ward
Garland Family	Jeff O'Connor	Kim Ward
Gerhardt Family	Katie O'Neil	Liz Ward
Joseph Gillam	Luis Orellana	Muttsy Ward
Morton & Myrna Goldberg	Candyce Payes	Will Ward
Ruth Gonya	Victoria Piantediosio	Welter Family
Hair-Marmon Family	Barbara Price	Weas Engineering
Jennifer Hampton	Russell & Leslie Price	West Family
Harper Boys	Nabeel Qureshi	Ryan West
Deb Harper & Family	Sharon Randall	Gretchen Wilson
Rose Ann Hickey	RARE Science RARE Bear Army	Wood Family
Michelle Hill	Julie Redmond	Reid & Paley Wood

MTM-CNM Family Conference

Reminder: Conference Policies for all attendees

As our conference grows, it is important for us to put conference policies in place to preserve the MTM-CNM Family Conference as a “sacred space” for individuals with MTM-CNM, families, and medical professionals that work directly with our community. All conference attendees and participants are expected to adhere to the following policies:

Confidentiality, Livestreaming and Social Media

For both the personal privacy of our families and to preserve the intellectual property of our researchers, livestreaming or audio/video recording of the conference presentations and sessions by conference attendees will not be permitted. MTM-CNM Family Connection will publish video recordings from the conference presentations and sessions at a later time depending on approval from the parties involved. At times, researchers or others may present information that is not ready for public disclosure, such as unpublished experimental data or future plans for research or clinical activities. Our community is privileged to share in these discussions at the Conference, but this information must remain confidential and may not be posted online or discussed in social media without express permission.

Photography and other Media

MTM-CNM Family Connection, Inc will have contracted photography and videography services to document the Conference weekend. Any use or republication of these photographs or videos in full or in part will require approval of MTM-CNM Family Connection, Inc. Conference attendees may take photos for personal use only; no other individual, organization, company, or entity is permitted to use Conference photos for promotional or organizational purposes without express written consent from MTM-CNM Family Connection. No other photography or videography services, print journalists or broadcast media are permitted, except those arranged through MTM-CNM Family Connection, Inc.

Competing Activities

The agenda of the MTM-CNM Family Conference includes a variety of structured and unstructured activities that are inclusive for all attendees, many of whom have gone through great lengths to attend the conference and meet with doctors, researchers, and families. No other individual, organization, company, or entity may create, organize, or otherwise arrange other activities targeting professional or family attendees of the Conference without express written consent from MTM-CNM Family Connection.

Solicitation

No individual, organization, company, or entity may engage in the selling of goods or services or solicitation of funds from conference attendees.



2009 MTM-CNM Family Conference
Houston, TX

2011 MTM-CNM Family Conference
Minneapolis, MN



MTM-CNM Family Conference

2013 MTM-CNM Family Conference



2015 MTM-CNM Family Conference Chicago, IL



See y'all next time... Get your boots on!



MTM-CNM Family Connection, Inc
www.mtm-cnm.org