

2019 MTM-CNM FAMILY CONFERENCE

July 19-21

St Louis



A DECADE Of Connections

MTM-CNM Family Conference



Welcome!

On behalf of MTM-CNM Family Connection, welcome to the 2019 MTM-CNM Family Conference!

As we begin our 6th Biennial MTM-CNM Family Conference, we celebrate a **Decade of Connections!** Ten years of building connections for our MTM-CNM Family and supporting our loved ones affected by myotubular and centronuclear myopathies. From the first conference in Houston in 2009, to Minneapolis for 2011 and 2013, to Chicago in 2015, to Nashville in 2017, and now our 2019 destination St. Louis, the miles traveled by families and medical professionals demonstrate the commitment of our community to come together and help make advancements for our rare disease community.

MTM-CNM Family Connection, Inc. was formally founded in 2014 out of the grassroots efforts of the family conferences and is a non-profit dedicated to the mission of connecting MTM-CNM families to **Resources, Research** and **Relationships**. We remain strong in our commitment to creating and cultivating a community of **Strength, Hope, and Family**. MTM-CNM Family Connection is grateful to have the opportunity to serve our community through our conferences and beyond in our work.

The theme for the 2019 MTM-CNM Family Conference is "**Through the Gateway: Expanding Possibilities**"! Inspired by the iconic Gateway Arch that marks St. Louis as the "Gateway to the West", we reflect on our own Gateway Moments and how our lives are changed as we enter a new world. These moments - getting a rare disease diagnosis, celebrating an unexpected accomplishment, mourning a loss, achieving a scientific breakthrough - can be powerful in shaping who we are as individuals and as a community. We don't really know what is on the other side of the gateway until we get there, though we may have heard stories from those who have gone ahead - perhaps sharing some hopes and dreams, perhaps some fears and worries. But we do know that life will be different, and the possibilities of what the future may bring are expanding! As an MTM-CNM Family, let's take a moment to look around this new landscape and step forward together...

It is with much gratitude that we would like to thank our sponsors: corporate sponsors, patient advocacy organizations, and 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 success.

It is our hope that you will find here a renewed commitment and resolve to move through the Gateway Moments together and look ahead towards the Expanding Possibilities for our loved ones living with MTM & CNM and our families. We hope you all have the most wonderful weekend and leave with **Connections** to last a lifetime!

MTM-CNM Family Connection, Inc.

Erin Ward, President
Marie Wood, Vice-President
Mark Ward, Treasurer

Agenda

2019 MTM-CNM FAMILY CONFERENCE "Through the Gateway: Expanding Possibilities" Hilton St. Louis Airport, St. Louis MO

THURSDAY, JULY 18TH

3:00PM - 4:00PM..... Orientation & Welcome Reception for 1st Time Families

5:30PM - 9:30PM..... Pre-Conference Community Dinner
(If you did not sign up for this during registration, and would like to attend, please contact info@mtm-cnm.org as soon as possible)

FRIDAY, JULY 19TH

2:00PM - 5:00PM..... Conference Sign-In and Community Connections Reception
Pickup welcome packets, enjoy light refreshments, and engage in some fun and informational activities

5:00PM - 9:00PM..... Opening Dinner and Evening Program
Community Social (cash bar), Welcome Address
Understanding Treatment and Trials: Robert Graham, MD, Boston Children's Hospital
Buffet Dinner, Professional and Family Introductions

9:00PM - Close Connections After-Hours
Networking, sharing, socializing, and learning together for all in the Constellation Lounge
9:30 Men's SALT group discussion with Levi Gershkowitz

SATURDAY, JULY 20TH

7:45AM - 8:15AM Morning Yoga (optional)
Welcome the new day with gentle movement, breathing, and mindfulness: Kristen Roberts

8:00AM - 8:45AM Breakfast

8:45AM - 9:00AM Announcements and Welcome

9:00AM - 10:30AM Scientific Session 1: Genetics and Basic Research
The View From Top of The Arch: Alan Beggs, PhD, Boston Children's Hospital
Genetics of Centronuclear Myopathies: Johann Böhm, PhD, IGBMC
Understanding YOUR Genetics: Jill Madden, PhD, MSc, CGC, Boston Children's Hospital
Centronuclear Myopathy Research at IGBMC: Johann Böhm, PhD, IGBMC

10:40AM - 12:00PM..... Scientific Session 2: Pre-clinical Therapy Development
Tamoxifen as a Therapy for MTM: Jonathan Volpatti, MSc, The Hospital for Sick Children
ASO as a Therapy for CNM: Belinda Cowling, PhD, Dynacure
UNITE-CNM Clinical Trial Update: Chris Freitag, MD, Dynacure

12:00PM - 12:45PM Lunch Break

Agenda

SATURDAY, JULY 20TH (continued)

12:45PM - 2:00PM **Scientific Session 3: Gene Therapy**

Late Rescue and Redosing in XLMTM Dogs: David Mack, PhD, University of Washington
Audentes Patient Advocacy: Kimberly Trant, Chelsea Karbocus, Audentes
Gene Therapy Clinical Trial Update: Sal Rico, MD, PhD, Audentes
Audentes Panel Discussion: Sal Rico, MD, PhD, Eric Mosbrooker, Kimberly Trant

2:00PM - 4:00PM..... **Caring For Individuals With MTM/CNM**

Part 1: Panel Presentations & Discussion: Respiratory, Neurology, Spinal Fusion for Neuro-muscular Scoliosis: Shared Decision-Making

Barbara Smith, PT, PhD, University of Florida
Robert Graham, MD, Boston Children's Hospital
Sarah Neuhaus, DO, National Institutes of Health
Lucia Bastianelli, RN, MSN, CPNP, Boston Children's Hospital

Part 2: Clinical Care Guidelines for Myotubular & Centronuclear Myopathy

Published Clinical Care Guidelines are important for promoting best practices amongst doctors and caregivers, especially for Rare Diseases. Families and medical professionals collaborate in a moderated open-forum to discuss further development of guidelines specific to MTM/CNM.

4:30PM..... **Group Photo**

Wear your conference t-shirt! Don't miss this chance to be a part of MTM-CNM History!

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

Community Social (cash bar)
Legislative Advocacy Panel
Marie Wood, MTM-CNM Family Connection
Lindsay Lykens, Parent Advocate
Max Bronstein, Audentes

Buffet Dinner, followed by the world-famous MTM-CNM Family Talent & Variety Show!

9:00PM - Close **Connections After-Hours**

Networking, sharing, socializing, and learning together for all in the Constellation Lounge

(continued next page)

KIDS ACTIVITIES

Kids of all ages are always welcome in the main conference sessions, but will also enjoy these fun activities in the "Kids Room" throughout the day on Saturday and Sunday morning:



Balloon Animals & Facepainting
Music Therapy Group Sessions and songwriting
Arts and Crafts
Video Games and Movies
Variety Show preparation
and more!



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

Agenda

SUNDAY, JULY 21ST

8:30AM - 8:45AM **Nondenominational Inspiration & Reflection Service (optional)**
Join us in the Reflection Room for uplifting verses, music, and fellowship

8:30AM - 9:15AM **Breakfast**

8:45AM - 9:15PM **Women's Coffee Chat**

9:15AM - 12:15PM **Gateway Moments & Expanding Possibilities**

X-Linked Carriers Discussion Panel

Jill Madden, PhD, MSc, CGC, Boston Children's Hospital

Cristina Liberati, MD, University of Florida

Emma Bliss, Remember The Girls

Emergency Preparedness

Robert Graham, MD, Boston Children's Hospital

Therapeutic Strategies: PT/OT/Music/Tech

Jenna Lammers, MSR, PT, University of Florida

Jaime Wilhite, MMT, MT-BC, Music Therapy St. Louis

Erin Ward, MTM-CNM Family Connection

Family Voices Panel: Overcoming Obstacles

12:15PM - 1:00PM **Lunch Buffet**

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.

5:00PM - 9:00PM **Post-Conference Community Dinner**

(If you did not sign up for this during registration, and would like to attend, please contact info@mtm-cnm.org as soon as possible)



Stay Connected!

While nothing quite compares with gathering in one place together at the MTM-CNM Family Conference, we are grateful to have so many ways to connect virtually online! Reach out, give us a Like, a Tweet, a Post, or a Share! We'd love to hear what you are up to, and to keep you posted on our activities. Stay connected, and help spread the word!



mtm-cnm.org



facebook.com/mtmcm (public page)
facebook.com/groups/mtmcmfamilyconference (private group)



linkedin.com/company/mtm-cnm-family-connection-inc



youtube.com/user/mtmfamconf



instagram.com/mtmcmfamily



[@mtmcmfamily](https://twitter.com/mtmcmfamily)



Special Guests

Jaime Wilhite, MMT, MT-BC & Tracie Sandheinrich, MA, MT-BC - Owners, Music Therapy St. Louis



Music Therapy St. Louis offers quality board certified music therapy services and consultation to patients, clients and families on both sides of the river of Missouri and Illinois. Music therapy is the clinical and evidenced-based use of music interventions to develop and enhance cognitive, social, emotional, physical and communication skills. Jaime has been a professional performing musician in the St. Louis area for 15 years. After a 10-year career in marketing she decided to take a leap of faith and switch professions to follow her passion using music to service others. Six years later, she was a practicing board certified music therapist

and attained a Master of Music Therapy at Maryville University.

Tracie grew up in a musical family, and graduated from Maryville University with a bachelor's of science degree in music therapy in 2008. She stated her own private contracting music therapy company in 2009, which eventually merged with Music Therapy St. Louis, where Tracie stands as co-owner and Director of Therapeutic Services.



www.musictherapystl.com

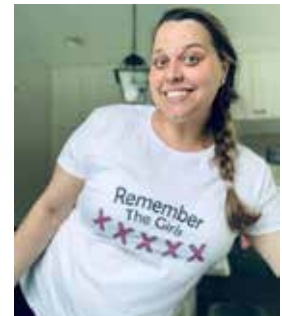
Emma Bliss - Vice-President, Remember The Girls



Emma Bliss found out that she was a genetic carrier for XLMTM the unfortunate way that many other women find out about their X-linked carrier status - by giving birth to a son with the affected disorder. Emma is a mama of two boys. Her second baby boy, Caleb was born in 2017 and diagnosed with Myotubular Myopathy at 4 months old in the NICU, where he spent his entire life earth-side. Caleb lived a beautiful and

peaceful but short 5 months and 1 day. From this loss and from the knowledge she now has, Emma has fostered a new purpose. Her mission is to raise awareness for genetic inheritance, more specifically X-linked disorders and to give back in the small way that she can.

Remember The Girls' mission is to raise awareness of the many issues facing female carriers of x-linked recessive genetic disorders; to provide a forum for x-linked females to share their stories, ask questions, provide and receive emotional support, and develop friendships; and to advocate for increased attention of the medical community to the physical and emotional issues of females who carry x-linked disorders.



www.rememberthegirls.org

Levi Gershkowitz - CEO, Living in the Light



Levi is a passionate writer, photographer and director with a love for people and the stories they carry. His approach to storytelling humanizes the experiences of people in life-limiting situations and through Living in the Light, leads a team of equally insightful individuals. We are thrilled to have Levi joining us again as Conference Photographer and also sharing his unique gifts of connecting people and facilitating dialog.

Founded by Levi in 2012, Living in the Light™ is a patient advocacy initiative utilizing the potency of fine art photography, compelling personal narratives, and engaging filmmaking to educate the biotech and medical community about the realities of rare diseases and the unprecedented effect they have on families and daily life.



www.frompatienttoperson.com

Research & Medical Professionals



Alan Beggs, PhD - Boston Children's Hospital

Alan Beggs is the Director of the Manton Center for Orphan Disease Research at Boston Children's Hospital and Sir Edwin & Lady Manton Professor of Pediatrics at Harvard Medical School. Following undergraduate studies at Cornell University, Dr. Beggs obtained his PhD in Human Genetics at Johns Hopkins University, with subsequent postdoctoral fellowship training in medical and molecular genetics at Johns Hopkins and Boston Children's hospitals. He has general expertise in laboratory and clinical applications of genetics to human disease, and since 1992 has directed an independent research program in the Division of Genetics and Genomics.



Boston Children's Hospital

Jill Madden, PhD, MSc, CGC - Boston Children's Hospital

Jill Madden is a genetic counselor and program manager specializing in the genetics of congenital myopathies as well as other rare and undiagnosed disorders. She earned her PhD in Genetics from Iowa State University in 2014 and continued her training as a postdoc at the University of Kansas Medical Center. After several years in the lab, Jill was craving the opportunity to work directly with patients and families, so she pursued a Master's degree in Genetic Counseling from the University of British Columbia in Vancouver, Canada. Jill began working with the Beggs Congenital Myopathy Research Program at Boston Children's Hospital in June 2018. She is thrilled to attend her first family conference and to have the opportunity to meet members of the MTM-CNM community! Jill coordinates the recruitment and enrollment of families for the Beggs Lab research study as well as the clinical studies that focus on identifying the genes and characterizing the symptoms associated with congenital myopathies.



Robert Graham, MD - Boston Children's Hospital

Dr. Robert J. Graham is a pediatric critical care physician with a special interest in acute and chronic care of children with technology dependence. Through clinical innovation, research, and teaching he seeks to extend critical care services beyond the intensive care unit (ICU) to optimize the outcomes for children and young adults with special healthcare needs as well as the experience of their families. In 2007, he developed a novel clinical program, the Critical Care, Anesthesia, and Perioperative Extension (CAPE) and Home Ventilation Program at Boston Children's Hospital, which provides home-visits, care coordination, and consultation for children with chronic respiratory failure. Dr. Graham's research includes health-services efforts investigating models of care, patient/parent-reported outcomes, and resource utilization, as well as clinical investigations focusing on patients with neuromuscular diseases.

Lucia C. Bastianelli, MSN, RN, CPNP - Boston Children's Hospital

Lucia Bastianelli is a pediatric nurse practitioner in the Cerebral Palsy Center at Children's Hospital specializing in perioperative care for children with complex neuromuscular conditions undergoing surgery for scoliosis and hip deformities. Lucia graduated from Regis College with a Masters in nursing and has over 20 years experience working with children with complex medical needs. In 2011, Lucia completed a Fellowship at Children's, named Leadership Education in Neurodevelopmental and related Disabilities (LEND) program at the Institute for Community Inclusion. Lucia is an active member of the American Academy of Cerebral Palsy and Developmental Medicine, (AAPDM). Lucia's previous experience includes clinical care and coordination for children with Duchenne Muscular Dystrophy at Tufts Medical Center. Lucia is a strong advocate for children with special needs and brings a unique perspective as a mother of young adult twins with Cerebral Palsy and complex medical needs. She is very excited to attend her first family conference and meet families and members of the MTM-CNM community.



Research & Medical Professionals

Sal Rico - Audentes Therapeutics



Sal Rico is Senior Vice President of Clinical Development at Audentes Therapeutics. He is a clinical pharmacologist with 15 years of clinical research experience in the pharmaceutical industry, contract research organizations and academia. 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.

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, and pharmaceutical medicine.

Kimberly Trant, RN - Audentes Therapeutics



Kimberly Trant is a Senior Director, and Head of the Patient Advocacy & Engagement department at Audentes Therapeutics. She began her career in healthcare as a pediatric and neonatal Registered Nurse (RN). Kimberly has spent the past 18 years in the pharmaceutical/biopharmaceutical industry in various roles

with increasing leadership where 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.

Eric Mosbrooker - Audentes Therapeutics



Eric B. Mosbrooker has served as Senior Vice President and Chief Commercial Officer (CCO) at Audentes Therapeutics since January 2019. He brings more than 15 years of healthcare leadership experience focused on launching and commercializing treatments for rare and orphan diseases. As CCO, Mr. Mos-

brooker is responsible for executing the global commercial strategy for the company's growing portfolio of gene therapy product candidates, and for building the commercial organization, including patient services, sales, marketing, and market access teams.

Mr. Mosbrooker joined Audentes from Origin Biosciences, where he served as Chief Operating Officer. Additionally, he has extensive experience in operations, management, healthcare, and technology consulting. Mr. Mosbrooker earned his B.S. in Industrial Engineering from the University of Wisconsin, Madison.

Chelsea Karbocus - Audentes Therapeutics



Chelsea Karbocus is Associate Director of Patient Advocacy & Engagement at Audentes Therapeutics. She joined Audentes Therapeutics in February of 2018.

Chelsea has spent the past 9 years in the pharmaceutical/biopharmaceutical industry partnering closely with patients, families and patient advocacy organizations. She is passionate about integrating the patient and family perspective into the work at Audentes and is driven to help bring innovative therapies to those living with rare conditions.

Chelsea began her career in healthcare advertising, in which she had the opportunity to work across numerous different therapeutic areas developing patient programs, engagement strategies and educational initiatives. In these roles, Chelsea sought to find creative ways to meet the needs of the patient community. She also had the opportunity to take part in launching numerous products and new indications.

Chelsea earned her B.S. in Communication from Boston University's College of Communications. In her free time, she volunteers at the Marine Mammal Center helping to rescue, rehabilitate and release marine mammals who are ill or abandoned.



Research & Medical Professionals

Max Bronstein - Audentes Therapeutics



Max G. Bronstein is the Senior Director of Health Policy & Corporate Affairs at Audentes Therapeutics. At Audentes, Max leads company interactions with state and federal policymakers and works with various coalitions to advance patient-focused policies for gene therapy and regenerative medicine.

In particular, Max is helping to lead legislative efforts to enable payers to engage in payment-over-time agreements for gene therapy products as well as ensure the strongest possible standards in the US for newborn screening of rare diseases.

Previously, Max was the Chief Advocacy & Science Policy Officer at the EveryLife Foundation for Rare Diseases, and has held various positions in the U.S. government, including at the National Science Foundation, the U.S. House Committee on Science & Technology, and the National Institutes of Health.

He holds a master's degree in public policy and a certificate in science and technology policy from the Ford School at the University of Michigan in addition to a BA in biology from Ithaca College, with a minor in writing. When not tormenting Members of Congress, he enjoys hiking around the San Francisco Bay Area and is an avid sailor, swimmer, and SCUBA diver.

Mark Meltz - Audentes Therapeutics



Mark Meltz is the Senior Vice President and General Counsel at Audentes Therapeutics and is responsible for leading the legal, compliance and corporate governance functions.

Prior to Audentes, Mark was the Executive Vice President and Chief Business Development

and Legal Officer at PaxVax, worked at Biogen Idec and was the Head of Legal for Novartis Vaccines for the Americas and North America regions.

Before joining the life sciences industry, Mark practiced law at the international law firm Bingham McCutchen LLP. Mark earned his Juris Doctor from Boston College Law School, Magna Cum Laude, and his Bachelor of Arts with Departmental Honors in Psychology from Yale University.

Katie Hogan - Audentes Therapeutics



Katie Hogan is Director of Patient Advocacy and Engagement at Audentes Therapeutics and is passionate about developing integrated advocacy and communications programs that positively impact the patient community. Previously, Katie served as Director, Corporate Communications at Audentes, and she brings more than

15 years of experience working on advocacy, communications, and public policy initiatives within the healthcare sector. Katie earned a Bachelor of Science (B.S.) in Communications from James Madison University in Virginia.



David Mack, PhD - University of Washington

Dr. David Mack is an Associate Professor in the Department of Rehabilitation Medicine and Bioengineering, as well as an Investigator in the Institute for Stem Cell and Regenerative Medicine at the University of Washington. He has a longstanding interest in how stem cells make cell fate decisions during embryonic development by coordinating their intrinsic genetic



program with cues from their surrounding microenvironment. The goal of the Mack laboratory is to apply their understanding of this basic question to the development of stem cell and gene therapy treatments for neuromuscular diseases. David's expertise is rooted at the intersection of genetics, developmental biology, cancer biology and biomaterials. The foundation is a Ph.D. in molecular genetics from the Indiana University School of Medicine. As a postdoctoral fellow with Dr. Gilbert Smith at the National Cancer Institute in Bethesda, he studied how 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 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.

Research & Medical Professionals

Belinda Cowling, PhD - Dynacure



Dr Belinda Cowling is Chief Scientific Officer at Dynacure. Her research has focused on understanding skeletal muscle function in congenital myopathies, and identifying novel potential therapies. She completed her PhD at Monash University, Australia, where she identified a novel regulator of skeletal muscle mass and strength, which is implicated in human disease and therapy. She completed a postdoc with Dr. Jocelyn Laporte, IGBMC, France, where she investigated the normal function of dynamin 2 in muscle, and how defects can cause centronuclear myopathy. In 2014 she became a tenured researcher for Inserm, where she identified down regulation of dynamin 2 as a novel therapeutic target for myotubular and centronuclear myopathies. In 2016 she co-founded the company Dynacure, and where she has been working closely with the research and development team. In 2018 Belinda joined Dynacure full time, and is focusing on translational research and supporting drug-candidate development. The lead program at Dynacure is focused on modulating the expression of dynamin 2 with antisense oligonucleotides for the treatment of centronuclear myopathies.

Chris Freitag, MD - Dynacure



Dr Freitag obtained his medical degree from Kiel University, Germany in 1994. After several years in different hospital posts, he started his career in the pharmaceutical industry at Roche in pharmacovigilance and medical affairs. At Shire, he held several positions in clinical development and medical affairs, covering the internal medicine portfolio, finally as VP, Global Clinical development and Innovation. In 2014 he joined BTG plc in the UK as SVP to head up Vascular Medicine and moved on to become responsible for global clinical development across the portfolio. He joined Debiopharm International SA in 2016 to lead the clinical R&D function managing international drug development across Phase I to III. He joined Dynacure as Chief Medical Officer where he is responsible for medical and regulatory strategy, including clinical development of the lead compound in CNM.



Johann Böhm, PhD - IGBMC



I was born and raised in the rainy far north of Germany, studied the basics of genes and inheritance in the center of the country, and specialized in medical genetics in Freiburg, the most southern German city. To move further south and learn more about muscle disorders (and to have access to great French wine and cheese), I crossed the border in 2007, and I am since working on rare muscle disorders at the IGBMC in Strasbourg, France. Our team mainly focuses on centronuclear and myotubular myopathy, and we aim to identify the genetic causes of muscle disorders, understand what happens in the muscle fibers, and develop therapeutic approaches.



Since 2014 I am an associate professor, and in 2017 I was honored young (!) myologist of the year by the World Muscle Society. As a geneticist, I am in contact with many affected families in France, Europe, and abroad. Through DNA analyses, we solved many cases awaiting molecular diagnosis, and we thereby improved genetic counseling and disease management for the patients and families. And most importantly: knowing the genetic cause is the first step towards a therapy.

Jonathan Volpatti, MSc - The Hospital for Sick Children, Toronto



Jonathan is a researcher in Dr. James Dowling's lab specializing in phosphoinositide biology and drug discovery using model organisms including zebrafish, mice, and *C. elegans*. He received his BSc in Biology from Drexel University and completed his MSc in Molecular Genetics at the University of Toronto with Dr. Dowling. His work over the last five years has focused primarily on identifying



drug targets and therapies for MTM. He helped to identify PIK3C2B as a potential drug target for MTM and supported pre-clinical studies for tamoxifen as a drug therapy for MTM. He is looking to start his PhD training next year to continue working on disease pathogenesis and drug discovery for rare disorders. Jonathan is very excited to meet with everyone in the MTM-CNM community at his first family conference!

Research & Medical Professionals



Barbara K Smith, PhD, PT - University of Florida

After focusing on physical therapy clinical care for 13 years in the ICU, Barbara Smith transitioned into clinical research with an emphasis on the control of breathing in individuals with neuromuscular disorders. In addition, she studies the effects of both exercise and regenerative therapies on ventilatory function, particularly for individuals who use mechanical ventilation. Dr. Smith has both clinical and research experience with XLMTM, nemaline myopathy, Pompe disease, and Duchenne muscular dystrophy, and she collaborated with Dr. Childers' team on measurements of pulmonary function in the canine model of XLMTM. She is honored to participate in her 5th MTM-CNM Family Conference this year.



Cristina Liberati, MD - University of Florida

Dr. Liberati graduated from the Medical School of the University of Rome "Sapienza", Italy, in 2015. After a year of training in general medicine, she was incredibly excited to move to the USA and joined the UF research team in September 2016. During the past 3 years, Dr. Liberati has worked as medical supervisor and Clinical Research Coordinator with Dr. Smith and Dr. Byrne at UF on many different research studies for rare neuromuscular diseases. She and Dr. Smith have worked together to study the respiratory muscle responses to different therapeutic approaches (diaphragm pacing system, respiratory muscle strength training, etc.) in different medical conditions including XLMTM. This will be her second MTM conference, and she is very excited to return to the family conference and reconnect with old families and tie bonds with new ones!



Jenna Lammers, MSR, PT - University of Florida

Jenna received a BS in Biological Sciences from Clemson University in 1996, a Master of Science in Rehabilitation/Physical Therapy from The Medical University of South Carolina in 2000, and recently became a Certified Neonatal Therapist. She has worked in pediatrics for 19 years in a variety of settings: in-home early intervention, preschool, outpatient, acute care (general, PICU, CICU, BMTW, and NICU). Over the past 10 years she has been a clinical evaluator in over 30 drug and gene therapy trials for children with rare neurological diseases. Jenna recently took a new position at The University of Florida where she helps improve the continuity of care for children with rare neuromuscular diseases through their acute hospital admissions, outpatient neuromuscular clinic visits, and research trials.

Sarah Neuhaus, MD - National Institutes of Health

Dr. Sarah Neuhaus is a Clinical Research Fellow at the Neuro-muscular and Neurogenetic Disorders of Childhood Section, Neurogenetics Branch, National Institute of Neurological Disorders and Stroke, NIH. She completed her Child Neurology training at Duke University Medical Center and her Neuromuscular Fellowship at Johns Hopkins University. She now works with Dr. Carsten Bönnemann at the NIH, involved in diagnostics and therapeutics for rare pediatric neuromuscular disease. Her primary focus is on the XL-MTM Gene Therapy trial, ASPIRO, sponsored by Audentes Therapeutics.



MTM-CNM Family Connection

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:

- 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.



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



MTM-CNM Family Connection

The Ward Family



Erin, Will, Mark

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! Now eighteen, Will recently completed his Senior year, went to Prom, and graduated from our local High School with High Honors. Since the last conference, he had a dream-come-true moment meeting "Big Papi", his favorite Red Sox player. He attended his first stadium concert - his longtime music crush Taylor Swift. His favorite class was Biotechnology, where he had opportunities to present about new advancements directly affecting his life with Myotubular Myopathy. Will continues fundraising for the Beggs Lab Congenital Myopathy Fund through Boston Children's Hospital Marathon Patient Partner Program and leading his annual walk team. During transition planning with his school team, he shared he wants to help mom & dad with the conferences and be a patient advocate. 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 for all individuals with MTM/CNM. We believe 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. 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 hope that everyone has an amazing experience this weekend!

The Wood Family

The Conference is a tradition that the whole family looks forward to, and we each contribute in our own way! While Marie's primary job is "Mom CEO," she also passionately serves as the Vice President of the MTM-CNM Family Connection, which she cofounded alongside Erin and Mark Ward and Shannon Mashinchi. David, who is a philosophical and theological speaker and writer for his day job, will be helping out with video recordings again! Bill and Erlinda Robinson--Marie's parents-- came along to help at the Nashville Conference in 2017, and they had such a great time, they're back in 2019! Lucian (16) our resident history buff, and Blaise (14) who loves wrestling and chess, will be helping out in the kids' activity room! Reid (11) has had four surgeries in the past year related to xlMTM but that hasn't put a damper on his happy, extraverted personality one bit, so look out for his smiling face and give him some love! Paley (9), who like Reid also has xlMTM, survived a close call on May 27, 2019, during which Marie performed CPR to revive him while David and the kids came together as a team to get the situation in hand. Paley spent a week in the hospital before coming home the day before his 9th birthday. He is still in recovery from the ramifications of this traumatic event, but our sweet boy keeps doing his best every day to regain the mobility he had previously. Right after Nashville 2017, the Woods got a huge surprise with Marie's unexpected pregnancy, and in May 2018, "Baby Boy Wood #5" aka Kepler Caden Wood was born! Finally, the Wood Basketball Team is complete!



Paley, Lucian, Reid, Marie, Erlinda, Bill, David, Blaise

Our Families

The Bauer Family

We are the Bauer family (formerly Stewart). Alex is the oldest, age 25, with XLMTM and Levi (XLMTM) passed away on March 12, 2019 at age 24. Isabella is 17 and a manifesting carrier of MTM. I, Beth, am also a manifesting carrier of MTM and Stepdad, Scott. Alex and Levi were characterized as "severe XLMTM" at birth with APGARS of 0 at each measurement. However, they have defied the odds time and time again and both went to and lived at college, attending full regular curriculum. Despite many brushes with death, they made it past the predicted one year to live. This last year was difficult with Levi declining and no one able to help him or stop the decline. It was apparent he was passing away, but he did so with no fear, no pain and lots and lots of friends and family visiting in his last 6 weeks on earth. Many have told us what an inspiration Alex and Levi have been to them, and that is truly helpful dealing with our grief over losing Levi. He was a loving, caring, sweet son, brother and friend. Alex is extremely witty and loves to tell stories and chat.



Levi, Beth Isabella, Scott, Alex

The Bliss Family



Robert, Wyatt, Emma, Caleb

We are the Bliss family from New England! Robert, Emma, Wyatt & Caleb Bliss. Our Caleb came into this world early and silently in November 2017. At 34 weeks, it was unexpected after a perfect pregnancy and zero complications. Caleb had many issues upon his birth and we were unsure what was connected to what. After 4 months in the NICU, genetic testing came back with the formal diagnosis and everything pieced together - Myotubular Myopathy (XLMTM). With zero family history, it was quite a surprise.

Caleb lived 5 months and 1 day in the NICU at CT Children's Medical Center, where he was held and loved his entire beautiful life. Since his birth and his passing from this earth, we have found peace, healing and gratitude for the time Caleb gave us. Caleb will forever be our greatest blessing and what he taught us will forever be the turning point in our lives. Caleb has an older brother, Wyatt who he adored. Every time Wyatt was able to visit, Caleb light up through his twinkling eyes, fleeting smile and unwillingness to close his eyes when Wyatt was around. It was an in-

describable bond albeit the surroundings and confusion of NICU life. Caleb will always be remembered for his bright blue eyes that spoke the words he was unable to express, for his cute little "butt chin" dimple and for his calming, laid-back personality. Caleb loved his late night daddy time where his dad would read him Ferdinand the Bull over and over and over again. He loved bath time and getting his head scrubbed. He brought so much love and joy to those around him and we are forever grateful to the NICU staff that cared deeply for him.

Emma has found a passion in the genetics world of medicine, advocating for X-linked carriers through the "Remember The Girls" organization. Having been through all stages of the single gene disorder process - with Caleb, in pregnancy and through her PGD IVF journey, there are many challenges, ethics and decisions to face. It can be a difficult path to navigate as a carrier but Emma's wish is to bring hope to those struggling and provide resources to those looking for others experiences.

Our Families

The Browning Family



Joanna, Sam, Jacob, Garrett

We are the Browning Family and live in St. Louis, Missouri. Jacob was born on July 1, 2014 and was diagnosed with MTM at the age of 6 months. Jacob spent his short but meaningful 3 1/2 years of life placing an undeniable mark on our hearts and this world. Jacob was the most sweet, funny and determined boy who loved to learn and always had his teachers and therapists in awe. He stole the hearts of everyone he met with his sweet spirit and smiling eyes. Although we lost Jacob on November 16, 2017, Jacob lives BIG in our hearts and is with us always. He transformed us as individuals, parents, and as a family and we honor and celebrate him in everything we do.

Jacob has an 8 year old brother Sam who will be going into third grade this year. Sam is Jacob's biggest fan and their relationship was the most beautiful thing to see. We are also expecting a baby girl in October 2019.



Sam and Jacob

The Bushey Family

Hello! We are the Bushey's, Matt, Natalie and twins Lilly and Cooper. We live in Springfield Ohio with our almost 10 year olds! Cooper was diagnosed at 7 weeks with X-linked MTM, but it was evident at birth that something was wrong. Since that diagnosis we have learned how to live the best life with it and make the most of everything we do! We enjoy traveling, and have even made it across the country from Ohio to California three times! Disney is always a fan favorite and Cooper really likes going to baseball games.

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!



Lilly, Natalie, Cooper, Matt

The Charette Family

Toronto, Canada

Our Families

The Davies Family

We are blessed to have had three years with our warrior, Grayson. Grayson passed away on January 24, 2018 due to respiratory complications. Our family now lives in Phoenix, Arizona; originally from Kansas City. Ryan is a stay at home father. Kayla works as a nurse practitioner at Banner MD Anderson. Makenzie is an active seven year old who is the proud big sister of Grayson. She misses her brother very much. Makenzie is looking forward to starting second grade this year. She is doing well in school despite being visually impaired. Ryan and Makenzie have another genetic condition called aniridia. It is a lack of iris and comes with a host of other eye complications, including legal blindness. We are looking forward to attending the conference and meeting 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. I was also able to attend the 2017 Conference and am excited to bring the rest of the family this year.



Grayson



Kayla, Makenzi, Ryan

The Davis Family

The Davises live here in the St. Louis area since 2008. Luke and Christy have been married 22 years and are the proud parents of Joshua (XLMTM, 21 years old) and Lindsay (18 years old), and their Jordan recieved his angel wings at 19 months in 2008.



Christy, Luke, Lindsay, Joshua



Jordan

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 1/2 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 reevaluate 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, Medical College of Wisconsin 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, preparation for Isabella’s transition to college this fall at liberty University, 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

I, Linda Fuller, and my husband Phil live in Pachuta, MS. I am blessed with 2 sons, Mark and Matt Snowden. Matt was diagnosed with MTM when he was 13 days old. At that time, very little was known about MTM...there was no information whatsoever available for parents, no DNA testing, no home computer or facebook. We enrolled in Dr. Beggs’ research when Matt was about 9. Matt is now 32 years old and enjoys playing video games and watching movies. We are so blessed with the love and support of family and friends. I thank the Lord for all the advancements that have been made and pray for all our MTM/CNM warriors, their families, caretakers, and all those involved in research and support. So grateful for the MTM-CNM Family website and to all involved in making this conference possible!



Matt, Linda, Phil

Our Families



Jessica, Declan, Adam

The Hair/Marmon Family

Did you know there were THOUSANDS of types of dinosaurs that used to roam the Earth? We didn't either. At least not until some little boy developed an obsession with all things dinosaurs last year- just in time for Christmas presents he actually enjoyed opening! And because Declan must know all the things, we also now know what all dinosaur names MEAN. After all, his favorite game is "look it up" for every. single. thing. :)

But life would be pretty boring without this inquisitive little kid who has come so far from spending the first 19.5 months of his life in the hospital (mostly due to red tape). Because of him we've learned new ways to communicate, how to advocate, how to appreciate the little things and how to make every second matter.

Declan turned seven in March! He just finished first grade, and attended camp for kids with trachs and vents this summer. In addition to dinosaurs, he loves animals, the zoo, swinging, being destructive,

school, music, dancing and his family. Mom (Jessica) and dad (Adam) work- sometimes traveling quite a bit- and mostly try to keep it together. Oh, and look up dinosaur name meanings... 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

Greetings from the Heartland!!!!

My name is Christian. When one thinks of the United States, you would normally call to mind destinations such as California, New York City, D.C., the Grand Canyon, Disney World, and other places of notoriety this country has to offer. Well, that "ain't" us! We're from small town Oklahoma, home of the Great Plains, waving wheat fields, and the University of Oklahoma (GO SOONERS)! Our family, like many within the MTM-CNM family, have seen our fair share of trials and tribulations associated with this disease. I hope that scientific research will continue improving the quality of life for our small community and disabled persons globally.

I love music, TV, traveling, writing, spending time with family, and just life altogether! This year, I graduated high school in May and look forward to beginning my first semester of college in the fall. Obviously, attending conference with my MTM family will be the highlight of my summer before I embark on this new stage of life and I cannot wait to make new memories with y'all!!!! Look out, St. Louis; the Hannas are coming!!!!



Dylan, Lauren, Christian, Chris, Madison

Our Families

The Hanson/Stansbury Family



Ida, Burke, Lucas, Krista

Ida, Lucas, Krista and Burke live in Seattle, Washington. Lucas will be a fifth grader in the fall, and he is passionate about rock music and technology (which he calls “my beloved screens.”) Lucas has x-linked MTM. Ida is four and loves art and making up stories, and she is incredibly adept at noticing what is and isn’t wheelchair accessible. Burke and Krista are both involved in social justice work, including grassroots organizing around the NW and support for families of children with disabilities and chronic illness around Seattle. We love finding ways of getting our whole family to places that we couldn’t have imagined going when Lucas was a baby – up a gondola in the Redwoods, out onto ocean beaches, and hiking in the mountains on accessible trails. And we also find joy in the smaller victories: making up a game like name-that-tune or rock-bands-A-to-Z so that Lucas will also have fun when we go out for pizza as a family.

The Ice Family

I live on a family farm with my parents Bernard & Sherry Ice near my brother Jimmy, his wife, Jessica, and niece and nephew, Grace and Ryan. Born in July, 1970, a floppy baby, I was initially diagnosed with the classification of Myotonic Atrophy, Type I Smallness with Central Nuclei, at NIH in 1974. I walked at age 4 and was able to speak and start 1st grade on time. I was weaker than my classmates and would be susceptible to falls. I learned to get myself up like an inch worm, with my head being the last to come up. At age 12, I had an acute pneumonia which necessitated a five month stay in Louisville Children’s Hospital and ultimately a left lower lobectomy. I came home ventilator dependent and in a wheelchair. I tried to wean off the vent during daytime hours, but was largely unsuccessful. I eventually started ambu bagging myself during the day and using mechanical ventilation while sleeping. For 25 years I stayed out of hospitals until normal respiratory bugs started getting antibiotic resistant. I’ve had several “debugging” stays in ICU’s since. In 2015, I was admitted with the same course in mind but complications following an abdominal surgery left me unresponsive for 10 days and I have been fully ventilator dependent since then and have my first power wheelchair.

During this recovery I was encouraged to rediscover my underlying muscle disease. This led me to University of Louisville Neurologist Dr. Martin Brown. Using my old NIH records and exam I was given targeted genetic tests. In October of 2017 I had a new name and a starting point, Myotubular Myopathy.



Gary, Bernard, John, Sherry, Jimmy, Grace

Our Families

The Jackman Family

We are from Oklahoma City, OK. I am a nurse practitioner working for an infusion center. I have 3 awesome kiddos. Jaden is 15 and will be a Sophomore next year, Chason is 12 and will be a 7th grader, and Keegan is 11 and will be in 5th grade. Lane would have been 18, 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.



Chason, Jaden, Rebekkah, Keegan

Ronald and Dianne Jackman



Dianne, Ronald

They are from Banner, OK. Ronald is retired and likes to spend his time at the lake fishing. Dianne recently retired and 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 second conference to attend.



Our Families

The Kalejaiye Family

Our names are Segun and Ronke Kalejaiye. We are originally from Nigeria, but we currently reside in Durham, NC. We are blessed with 3 adorable children (Praise, Precious and Prince). Praise is 6 years old, Precious is 4 and Prince is 10 months old. Prince was born on Sept. 9, 2018 at Duke Hospital NC and was diagnosed with XLMTM in Nov. of 2018. Prince had a very rough start to life, as we were at the NICU for 4 months. We are thankful that he made it out of the hospital alive despite all he went through. Prince's condition came to us as a shock, as we had a healthy pregnancy. We have never heard about the CNM-MTM condition until Prince was diagnosed. Our little Miracle has helped us to discover our hidden strengths and abilities as a parent. We have had to stretch ourselves more than we could have ever imagined in the past 10 months. Prince loves to smile and dance, his favorite song is 'Baby Shark'. Praise and Precious are very helpful and understanding of Prince's condition. They enjoy spending time with him, and take turns to read him a book daily.



Ronke, Segun, Prince, Praise, Precious



The Lawton Family

Mitchell was born on September 6, 1990. His Apgar scores were 1 & 2. He was placed on a ventilator but extubated after 24 hours. During those first few months Mitchell continued to grow but his cry could hardly be heard and he had to be bottle fed because his ability to suck was so weak. At the age of five months Mitchell was diagnosed with MTM via muscle biopsy. There were many challenging times with Mitchell's health, many hospitalizations, many broken bones; but he was always able to persevere. Mitchell is now 28 years old and thriving at the University of Colorado-Denver studying Video Animation. He enjoys gaming, board games and spending time with friends and family.

Mitchell

The Lewis Family

We are the Lewis Family from Madison, Mississippi. Caden is 17 years old and going to be a Senior in high school this year! He enjoys hunting, traveling, movies, and video games. This will be our 3rd conference and we look forward to seeing everyone.



Jessica, Caden, Kate Elizabeth, Marty

Our Families

The Lykens Family

Hi Everyone! We are the Lykens Family: Craig (34), Lindsay (34), and Gil (almost 4), and we are from Ashburn, VA (about 25 miles outside of Washington, DC). Gil just finished his first year of Pre-K and we are excited to see what next year will bring. He loves to watch videos on youtube kids, especially songs about counting, and enjoys counting along on his fingers! Gil loves to zoom around the house in his manual wheelchair, and likes to explore and go outside. Gil has been making great progress on his "Good Boy Chart" this summer, and has already earned trips to the local farm/petting zoo, Chuck E. Cheese, the Clemyjontri accessible playground, and to Target to pick out a new toy! We expect that he will earn lots more fun adventures this summer. While Lindsay and her mother (Sherri) are at the Family Conference this year, Craig and Gil will be making mischief at home with Gil's other Grandma, Tammy, Lindsay and Sherri are excited to see you in St. Louis!



Craig, Gil, Lindsay

The Marik Family

We live in El Reno, Ok. I'm a Physical Therapist Assistant working at Mercy in the Home Health department and the mom to 4 wonderful children. Lauren is 26 and is an RN currently working at the Heart Hospital in OKC, she was recently accepted into Nurse Practitioner school. Madison is 22 and just graduated with her Bachelors in Psychology and is currently working in a daycare. Christian is 18 and just graduated high school and will be attending OU in the fall and loves doing genealogy. Brooklyn is 13 and is going into the 8th grade and plays competitive soccer. This is Brooklyn and I's first conference. Lauren and Christian were at the last conference. We are excited to be here and to hear what research has been done to find a cure.



Madison, Brooklyn, Christian, Chrissy, Lauren, Dylan

Our Families

The Mashinchi Family



Javad, Shannon

Javad Mashinchi is a funny, sweet and loving seventeen-year-old young man. In 2015, Javad had a major health setback that changed his overall trajectory. Prior to this, Javad had been attending school with his peers in a much different world. The seizure took his ability to voluntarily move and the stamina to be in his wheelchair for any length.

Javad attends school at home with amazing instructors that come to the house every day. He has been working hard to regain movement and working towards being in his chair for extended periods of time. Javad will be a junior this upcoming school year and our goal is for Javad to attend school for at least a half day. We continue on the slow road to recovery and are seeing more and more of his sassy self emerging every day.

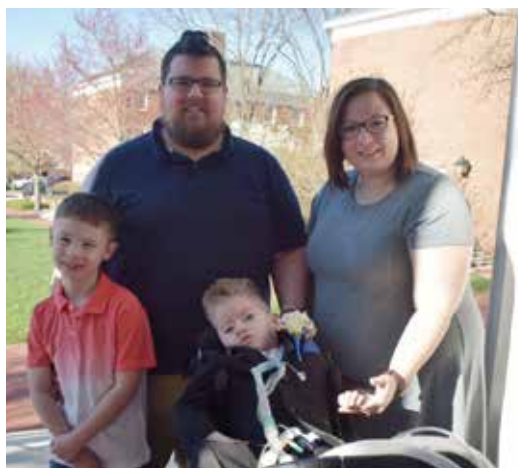
Javad currently lives with his dad, David, and his brother, Simon. His mom, Shannon, lives less than six minutes away and hangs out with Javad all the time. His sister, Stesha, lives in Chicago and his brother, Adam, lives in Auburn, California with his wife, Sam, and their son, Graham. The family works together to provide Javad with all the love, care, and support he needs to continue to live a healthy and happy life.



Arienna, Amanda, Alex, Ashley, Mary

The Mathews Family

Hello everyone! We are from Stroudsburg Pa and are excited to attend our 1st MTM/CNM family conference. Alex is 14yrs old has been diagnosed with unspecified Myotubular Myopathy. He is pictured with his sister Arienna 9yrs old his Mom Amanda his Aunt Ashley & his grandmother Mary.



Cooper, Richard, Reid, Calli

The Mayotte Family

Richard, Calli, Cooper, and Reid are from Crawfordsville, IN. Reid is 4 years old and thriving now that his medical interventions are finally stabilized. You can find the family at various sports complexes supporting Cooper's athletic endeavors. Reid is enjoying preschool and socializing with his friends. We are excited to be back at the MTM-CNM Family Conference this year to spend time with our MTM family whom we have missed so much!

Our Families

The McDermott Family

Greetings from the McDermott family: Daniel, Robin, Will (7) and Ryan (6). 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 happy boy who loves LED pinwheels, YouTube (in any language, he particularly likes Spanish), and balloons. He loves to dance and wiggle to music, go swimming, drive his wheelchair (he's still in training), and play with his family and friends. Will loves being a 'Big Brother.' He loves to help with therapy, play catch with his brother, and build plastic cup towers for Ryan to knock down. We 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!



Ryan, Daniel, Will, Robin

The Munoz/Rocha Family



Christi, Jason Jr, AJ, Jason Sr

Hello, we are the Munoz/Rocha family. Our family includes Christi Rocha, Jason Munoz Sr., Jason Jr., AJ, Julian, and Destiny!

Jason always has a huge support system. He is loved by his Nana Mary and Grandma Sally. His Grandpa John. His aunts, Lily, Angela and Dee. His Uncles Jay and Josh. Also his cousins Sadie, Efrain, Gianna, Jonathan, Jaxson, Matt, Rachel, and Caleb.

Jason was born on March 21, 1996 and was diagnosed with XLMTM. Jason is strong willed and determined and that has been his life motto!

Update on kids: Jason, 23, health wise, he is doing great! Jason is registered and he will start taking a Criminal Justice classes at the College of DuPage starting in August. He is excited about that! He has been enjoying his life (after high School), he has been keeping himself busy with trips to the mall. He still LOVES Pokémon (that will never change). His brother AJ, 18, is leaving for college soon, August 10th. He received a football scholarship from McKendree University.

“Really all of my family and myself are doing great. We are all just trying to live our lives to the fullest” (Jason, July 2019).

Our Families

The Orellana Family

The Orellana family is Luis & Jeanne, our three sons Dylan, Luciano & Sebastian, and Draco, a pug who has no idea he is not a housecat. Sebastian is heading into Junior year in 2019 and we all looking forward to seeing everyone again at the MTM CNM 2019 gathering! I couldn't find a good shot with all 5 so the 3 sons will do. 😊



Sebastian, Luciano, Dylan



The Payne Family

We are the Payne family, Greg and Stephanie, from Atlanta! We have been married for 2 years and consider ourselves to be a fun loving family that enjoys music, traveling and cooking new recipes. We are dog lovers and our goldendoodle puppy, Buster, is the center of our world! We are blessed to have the love and support of our family and friends, in particular Stephanie's mom and sister who have been with us every step of the way. Stephanie was originally diagnosed with LGMD but recently received a new diagnosis from the Mayo Clinic as a manifesting carrier of XLMTM. The past few months have been spent digesting Stephanie's new diagnosis and trying to educate ourselves on the best ways to manage her condition and symptoms. It's been challenging, overwhelming and scary at times, however, we are adjusting to the realities of MTM and remain full of fight and hope. This is our first time attending the conference, and we are so excited to engage with the members of this brave community. We look forward to making new friends, learning more about clinical trials and gaining access to new resources to help us navigate the challenges of MTM.



Stephanie, Greg

Our Families

The Rennie-Roberts Family

Hi there ! We are the Rennie-Roberts family from Ile Perrot Quebec, Canada. Our James is ten years old now and loves to sing and take pictures when he is not cuddling with his two dogs. James's dad works near home as a technician for a major data center and Mom is a part-time yoga teacher. Gaelan was loved from the moment we knew we were expecting. He arrived on March 30th 2007 and stayed only a short time with us before he got his wings on April 13th 2007. We miss him every-



Gaelan

day and know that where he is he is happy and at peace.



David, James, Kristen

The Rocheford Family

Bryan, Jamie, Abby, and Nicholas. Grandma Carol. We live in Minnesota just north of the twin cities. Abby is ten and sometimes very shy, loves music, you tube videos and can talk your ear off about L.O.L dolls. Nicholas was born March 18, 2012 and diagnosed with X-linked MTM about a month later. We were lucky enough to enjoy nine months and on December 17, 2012 we lost our warrior. Nicholas had a smile that would light up a room and cheerful disposition no matter what was going on around him. Grandma Carol has been a treasure to this family and we would not think of leaving her home. Grandma Carol was a crucial part for us to be able to bring Nicholas home and still be able to work.



Abby, Jamie, Nicholas, Bryan

Our Families

The Scoggin Family



John

We're Pam, Gary & John Scoggin from Texas City, TX, on Galveston Bay, south of Houston. We've been around the MTM community for a long time! John is 30 and holds a degree in Journalism/Communications from the University of Houston – Clear Lake. He stays busy writing online guides for video games. So far his guides have had over 420,000 pageviews. His guides have won a couple of awards for their usefulness and completeness. The picture shows John in his “nest” with his computer, dual monitors and video game console. Check out his work at <https://gamefaqs.gamespot.com/community/TheDogfather/contributions>.

We have four dogs, including Pink, who came to us from the MTM dog colony. We stay busy with John as well as community and church activities.

The Scott-Biddle Family

Hi, we are the Scott-Biddle family! We are from Indiana. We are excited to be able to make it this year!! We have 3 children. Austen is our angel baby. He would be 16 in August. Ashton is 14 and getting ready to go in to the 9th grade. He loves video games and roller coasters! Addison will be 2 in September. She is full of energy and loves Mickey mouse!



Danny, Ashton, Samantha, Addison



Austen

The Slaby Family

My name is Joe Slaby and I live in Terre Haute, Indiana. I was My name is Joe Slaby and I live in Fishers, Indiana. I was diagnosed with Centro Nuclear Myopathy when I was ten years old in 2001. The Dynamin 2 genetic mutation was later diagnosed at Beggs Laboratory. Although there were symptoms at an early age, my progression didn't truly begin until the end of my middle school years when I experienced mobility issues. After falling more frequently, having greater difficulty walking and standing up from chairs, I began using a scooter which I have done ever since. During that time I've gone from working part-time at the YMCA to graduating college and passing the CPA Exam. I currently work full-time as a fund accountant outside of Indianapolis. My family has always been my greatest supporters and provided stability over the years to allow me to develop into the person I am today. In addition, the MTM/CNM Family Conferences have brought me close to many amazing people who helped me to accept myself. I'm forever grateful to go to a place and connect with so many individuals every other year.



Joe

Our Families



Phil, Nathan, Molly, Julie

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 6 years old and his sister Molly is 2. 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 (who are here with us in St. Louis! Hi, y'all!) 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 Pokémon. 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; we are so excited for the opportunity to meet new friends and visit with old friends!

The Utama Family

Dominic and Heather have 3 little kids. Our children are Marcus, age 5, Mary, age 4, and Mimi, age 3. Our oldest child, Marcus has hypoxic brain damage and needs a ventilator. Nobody in our family has MTM-CNM, and we really appreciate being able to join with other families at the conference though Marcus' diagnosis is different. Marcus stopped breathing when he was still a newborn, and ever since, he's needed a lot of medical interventions. We live in St Louis. There are great resources here, but meeting Marcus' needs can be confusing for sure. We look forward to meeting other families!



Mary, Heather, Marcus, Dominic, Mimi

The Walker Family

My name is Ashley Walker. I am married and currently reside in Carlsbad, California. My whole life, from the time Dr.'s started diagnosing me, my parents were told that I had a mild form of limb-girdle muscular dystrophy. They were told this even through a muscle and nerve biopsy. Dr.'s would tell my parents that I would never menstruate and that I would be wheel chair bound by the time I turned 30 years old. Well to their surprise, On May 11, 2012 at 23 years old, I gave birth to Identical twins! While my husband and I were stationed on the island of Hawaii, we welcomed our sons Jayden and Alexander into the world at just 27 weeks. Our boys immediately needed assistance with feeding and breathing. When our twins turned 9 months old, all 3 of us received genetic testing and were given the diagnosis of x-linked MTM. We made it our mission to learn how to care for and ultimately bring our babies home from the PICU. Our twin boys passed away when they were just 14 months old. My favorite memory of Jayden is watching super heroes with his daddy and how Alexander loved having a warm bath and snuggles with his mom.

We feel immensely blessed to have had them for as long as we did. This is going to be our 3rd conference!! I am constantly overwhelmed and grateful for the support of our MTM family, which is exactly what keeps me coming back! I look forward to seeing old friends and making new friends while I attend! It really helps meeting other females affected by MTM and connecting to learn more about gene therapy.



Our Families

The Whiston Family

We are so excited to be attending our third conference. This conference is especially exciting as William is turning 8 years old on the day we arrive! We are looking forward to meeting new families, reconnecting with our MTM friends from all over the country, and possibly playing a few rounds of Super Smash Brothers or Mario Kart. Our family lives in a suburb outside of Chicago. If you ask William he might sing Go Cubs Go for you! Our two kids, William and Juliet (10), are both fans of movies, iPads, and ice cream. Instead of dwelling on our MTM journey through hospitals and "medical misadventures" our family is focused on having fun and seeing the world. We are excited to visit friends and family in St. Louis and look forward to touring Missouri and Kansas as part of this trip.



Melanie, William, Daniel, Juliet

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 in early 2017 that dream came true with our adopted son, Joey. We attended the 2015 and 2017 conferences, and are happy to be back again.



Louie



Martin, Emily, Lindsey, Joey

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.



Levi Bauer
11/17/94 - 3/12/19



Caleb Bliss
11/6/17 - 4/7/18



Jacob Browning
7/1/14 - 11/16/17



Grayson Davies
12/6/14 - 1/24/18



Jordan Davis
7/5/07 - 11/23/08



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



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



Ronnie Jackman
1974 - 1975

In Memoriam

“Always Remembered, Forever in our Hearts”



Gaelen Rennie
3/30/07 - 4/13/07

Nicholas Rocheford
3/18/12 - 12/17/12



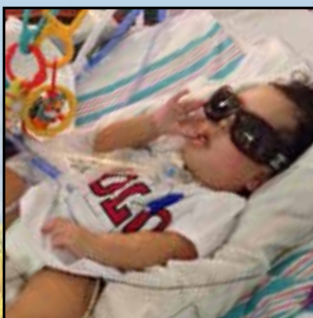
Austin Scott
8/14/02 - 2/6/04

Daniel Villareal
12/21/73 - 12/24/73



Edwardo Villareal
10/21/75 - 12/2/75

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



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

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



Connections Continue!



MTM-CNM Family Connection Inc is honored to have been awarded a 2019 RARE Patient Impact Grant from Global Genes. This grant enabled us to in turn provide travel grants to help families to attend the Conference.

Our team will be at the Global Genes Patient Advocacy Summit 2019 in San Diego, CA, September 18-20. MTM-CNM Family Connection will again host a family gathering at noon on Saturday September 21, in San Diego, as we did following the 2017 Summit in Irvine, CA. Hope you can join us!



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.

Solicitation

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

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 some of these community leaders throughout the weekend or connect with them following the conference via their respective websites.

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



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



Joshua Frase Foundation (www.joshuafrase.org)



Myotubular Trust (www.myotubulartrust.org)



Fundacja Oswoic Miopatie (www.oswoicmiopatie.com)



RYR-1 Foundation (www.ryr1.org)



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



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Our sponsors are generous in their financial support, and unwavering in their dedication to families and individuals affected by rare diseases.

Thank You.

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Fundraisers & Donors

Huge thanks to these superstars for leading fundraising activities on Crowdrise and Facebook:

Kimberly Brown
Caity Crosby
The Garland Family
Levi Gershkowitz
Christian Hanna
Shannon Mashinchi

Calli Mayotte
Christi Rocha
Joe Slaby
The Ward Family
Amy Welter
The Wood Family

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

Atlas MedStaff
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The Barghaan Family
Kelly Berger
Debbie Biasini
The Browning Family
The Burgess Family
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Catherine Schaeffer
Millie Sellin
Megan Shea
The Shirk Family
Joe Slaby
Joe Slaby
Melanie Spring
Burke Stansbury
Evelein Van Londersele
Bille & Kaes Van't Hof
Eileen & Leo Ward
Dan & Kim Ward
Kim & Ivy Ward
The Ward Family
The Welter Family
Jason Wheeler
Marie Wood
The Wood Family

Gateway Moments....



2009 MTM-CNM Family Conference
Houston, TX



2011 MTM-CNM Family Conference
Minneapolis, MN



2013 MTM-CNM Family Conference
Minneapolis, MN



...a DECADE of Connections...

2015 MTM-CNM Family Conference Chicago, IL



Photo: Sergio Vazquez



MTM-CNM FAMILY CONFERENCE 2017



NASHVILLE TN



www.mtm-cnm.org

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...Here's to Expanding Possibilities!



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