



MTM-CNM FAMILY CONFERENCE
CHICAGO, IL



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WELCOME

On behalf of the Conference Planning Team, welcome to the 2015 MTM-CNM Family Conference! We are so glad you are here! This is a unique opportunity to bring together families to support one another, to share information and resources to help better care for our loved ones affected by MTM or CNM, and to interact and collaborate with lead researchers and industry professionals in our field.

Following the 2013 Family Conference, it was clear to our planning team that the time had come in our journey to formalize the grassroots efforts of our team and develop a non-profit dedicated to the mission of connecting MTM-CNM families to resources, research and relationships. So in May 2014, we officially launched MTM-CNM Family Connection, Inc. We are excited about this new organization and look forward to continuing to serve our community through family conferences and beyond.

As we begin our 4th national conference, we celebrate with the theme: "Anchors & Oars: Strength for Today, Hope for Tomorrow". This theme illustrates our passion to help equip families with the connections and resources that will help us all stay strongly "anchored" 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 "grab an oar" and together move forward with hope: hope in celebrating each moment, hope towards treatments, and hope for carrying forward the legacies of our loved ones.

With scientific discoveries steadily progressing, and promising treatments on the horizon, this is a critical time to come together as one, to be organized in our next steps forward, and to share our journeys with the professionals who are working so hard on behalf of our loved ones and those who have gone before. By doing so, each and every one of you is helping to make this voyage through stormy seas a little smoother, anchoring us with hope, and bringing us closer to the peaceful harbor that we long for.

It is with deep gratitude that we would like to thank our corporate sponsors, family foundation sponsors, 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 truly special event. It has been a coming together of resources and gifts that have brought us here today and it is our hope that we all leave with a renewed commitment and resolve to pick up our oars and row together towards a bright and hope-filled future. May you all have the most wonderful weekend and leave with memories to last a lifetime!

With strength and hope,

MTM-CNM Family Connection, Inc.

Board of Directors & Conference Planning Team:

Erin Ward

President/Conference Director

Mark Ward

Treasurer/Planning Team

Marie Wood

Vice-President/Planning Team

Shannon Mashinchi

Clerk/Planning Team



AGENDA

2015 MTM-CNM FAMILY CONFERENCE AGENDA:

"ANCHORS & OARS: STRENGTH FOR TODAY, HOPE FOR TOMORROW"

GARDEN BALLROOM: ALL CONFERENCE SPEAKING PROGRAMS, COMMUNITY GATHERINGS, & MEALS
LINDBERGH MEETING ROOM: ALL KIDS' CLUB ACTIVITIES
EARHART MEETING ROOM: OUR MEMORIAL REFLECTION ROOM

FRIDAY, JULY 24TH

2:00PM – 4:00PM *"Meet your Cruise Crew!"* – Conference Check-in and Community Connections

5:00PM – 9:00 PM *"Setting Sail: Beginning our Journey Together"* – Opening Dinner, Introductions

SATURDAY, JULY 25TH

8:45AM – 9:00AM **ANNOUNCEMENTS AND WELCOME**

9:00AM – 9:40AM *"Getting our Bearings"* – An Introduction to Congenital Myopathies

Opening Remarks

Alan Beggs, PhD, Boston Children's Hospital

Diagnosis and Genetics of MTM/CNM

Johann Bohm, PhD, Institut Génétique Biologie Moléculaire Cellulaire

Natural History and Clinical Aspects of MTM/CNM

Jim Dowling, MD, PhD, (via video) and Hernan Gonorazky, MD, Hospital for Sick Children

9:40AM – 10:30AM *"Casting the Nets Wide"* – Several Promising Treatments

Therapeutic Approaches for Centronuclear Myopathies

Alan Beggs, PhD, Boston Children's Hospital

Inhibiting PI3 Kinase in MTM

Hernan Gonorazky, MD, Hospital for Sick Children

Interactions Between DNM2 and MTM

Johann Bohm, PhD, Institut Génétique Biologie Moléculaire Cellulaire

10:30AM – 10:40AM **BREAK**

AGENDA

2015 MTM-CNM FAMILY CONFERENCE AGENDA:

"ANCHORS & OARS: STRENGTH FOR TODAY, HOPE FOR TOMORROW"

SATURDAY, JULY 25TH (CONTINUED)

10:40AM – 12:00PM **"Full Speed Ahead"** – Progress on Gene Therapy

Introduction to Gene Therapy for MTM

Ana Buj Bello, PhD, Genethon

Recent Results With Gene Therapy in Animal Models

Martin "Casey" Childers, DO, PhD, and David Mack, PhD, University of Washington

Development of a Human Vector

Ana Buj Bello, PhD, Genethon

Pathways to Human Clinical Trials

Suyash Prasad, MD, Audentes Therapeutics

12:00PM – 1:00PM **"Catch of the Day"** Pickup Box Lunches

1:00PM – 1:45PM **"Take a Deep Breath"** – Respiratory Studies and Managing Breathing

Markus Renno, MD, Vanderbilt University/University of Florida

Barbara Smith, PhD, University of Florida

1:45PM – 4:45PM **"Shore Excursions"** – Afternoon Options

- **"Deep Dive"** (2:00PM – 3:30PM) Scientific & Medical Forum for Families and Professionals
– Bring your questions and ideas!
- **"Staying Afloat"** (2:00PM – 3:30PM) Hot Topics around daily life with MTM/CNM
– Families only, please
- **"Our True North"** Pay tribute to the memory of our Angels in the Reflection Room
– Connect with other families who have lost a loved one, do an activity together onsite or visit a local attraction together.
- **"All Aboard"** Enroll and participate in research studies with the various teams that are present
– Some studies require advanced registration
- **"Explorers"** Go offsite for some shopping or sightseeing adventures
- **"Make a Splash"** Leisure time at the hotel pool and kids' zero entry pool, and/or kids activity room

AGENDA

2015 MTM-CNM FAMILY CONFERENCE AGENDA: "ANCHORS & OARS: STRENGTH FOR TODAY, HOPE FOR TOMORROW"

SATURDAY, JULY 25TH (CONTINUED)

4:45PM **"We're gonna need a bigger lens!"** – Group Photo (wear your conference t-shirts!)

5:00PM – 9:00PM **"The Captain's Ball"** – Evening Program
An Update on Audentes
Matthew Patterson, Audentes Therapeutics

BUFFET DINNER

Variety Show showcasing the many gifts and talents within our community

SUNDAY, JULY 26TH

8:45AM – 9:15AM **"Radiance from the Lighthouse"**- OPTIONAL
A nondenominational Inspiration & Reflection Service, Earhart Room

9:30 AM – 12:00PM **"A Mighty Fleet"** – Important Work on Many Fronts
Molecular Diagnosis by Next Generation Sequencing
Soma Das, PhD, University of Chicago

RYR1-Related Myopathies
Michael Goldberg, MD, Allegheny General Hospital and RYR-1 Foundation

Beggs Laboratory, MTM Carrier Study
Casie Genetti, MS, Beggs Laboratory Boston Children's Hospital

CMDIR Studies for MTM: Genetic Testing, Event Study, Carrier Study
Sabine de Chastonay, PhD, Cure CMD/CMDIR

CMD Tissue Repository and Adapting canine pathology studies for gene therapy clinical trials
Mike Lawlor, MD, PhD, Medical College of Wisconsin

Patient Advocacy
Barbara Wuebbels, RN, Audentes Therapeutics

Reflections and Next Steps
MTM-CNM Family Connection Team

12:00PM – 2:00PM **"Pick Up Your Oars – and Row Together!"**
– Buffet Lunch and Closing Program

AGENDA

2015 MTM-CNM FAMILY CONFERENCE AGENDA:

"ANCHORS & OARS: STRENGTH FOR TODAY, HOPE FOR TOMORROW"

THROUGHOUT THE WEEKEND

- Participate in a physical assessment of motor function for XLMTM with the Nationwide Children's Hospital Team*
- Participate in the Observational Study of Respiratory Strength and Function with the team from University of Florida*
- Enroll in CMDIR studies, with Dr. Sabine de Chastonay
- Meet with Mike Lawlor to enroll in CMD Tissue Repository
- Meet with Genetic Counselor Casie Genetti, Beggs Lab Boston Children's Hospital
- Meet with Genetic Counselor Amy Knight Johnson, University of Chicago
- Meet with a respiratory specialist from Philips Respironics for product demos
- Visit the Reflection Room and pay tribute to the memory of our Angels
- Rest & Relax Poolside

***PLEASE PRE-ARRANGE A TIME WITH THE RESPECTIVE RESEARCH TEAM**

KIDS' CLUB ACTIVITIES

Although there will be volunteers helping in the Kid's activity room, nursing/respice care is not available for the Kids' Activities, caregivers should accompany children as needed.

SATURDAY, JULY 25TH

9:00AM – 12:00PM

Kid's Activities available will include Arts and Crafts, Wacky Photo Booth, Video Games, and Movies.

– Held in the Lindberg Meeting Room Saturday & Sunday

9:30AM – 11:30AM

"Marvelous Masks" Balloon Artist

"Our Superheroes - What's your super power?" – Kids will make their own superhero capes that represent their special gifts and talents.

"Celebration Hats" – Kids will work on making & decorating hats for the *"Parade of Hats & Capes"* that will take place during the variety show following dinner tonight.

12:00PM – 1:00PM LUNCH

Kids will join families for box lunches that can be enjoyed in the ballroom, poolside, or your own hotel room for some downtime.

1:45 PM – 4:45PM

Leisure Time with Families: Join a group outing to a local attraction, enjoy your own aquatic experience poolside with the family, or join us in the Kids room for a family-friendly movie and activities.

SUNDAY, JULY 26TH

9:00AM – 12:00PM

Arts & Crafts, Wacky Photo Booth, Movies, and video games

9:30AM – 11:30AM

"Marvelous Masks" Face Painting

"Fun with Foam: From Me to You" – Kids will make a special keepsake for their family as a 2015 Conference memory.

12:00PM – 2:00PM

Join families for closing lunch in the Garden Ballroom

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Matthew R. Patterson

President and Chief Executive Officer
Audentes Therapeutics

Mr. Patterson is the co-founder of Audentes and has served as President and Chief Executive Officer since the Company's inception in November 2012. Mr. Patterson has 20 years of experience in the research, development, and commercialization of innovative treatments for rare diseases and has held positions of senior management in both private and public biotechnology companies. Prior to Audentes he was an Entrepreneur-In-Residence with OrbiMed, the world's largest health-care dedicated investment firm. In this role Mr. Patterson identified and evaluated orphan drug investment opportunities. From 2004 to 2011, Mr. Patterson worked for Amicus Therapeutics, most recently serving as President and Acting Chief Executive Officer. Prior to his work at Amicus, from 1998 to 2004, he worked at BioMarin Pharmaceutical Inc., first serving as Vice President of Regulatory Affairs and later Vice President of Commercial Planning. From 1993 to 1998 Mr. Patterson worked at Genzyme Corporation in Manufacturing and Regulatory Affairs.

Mr. Patterson is a member of the Board of Directors of Gilda's Club of New York City, which provides social and emotional support for people living with cancer. Mr. Patterson received his B.A. in Biochemistry from Bowdoin College.



Suyash Prasad, MD

Senior Vice President and Chief Medical Officer
Audentes Therapeutics

Dr. Suyash Prasad is a Pediatrician, Clinical Development Physician, and Translational Scientist, with a wide range of experience in drug development for infants and children with rare and severe disease. He is currently based in San Francisco in the role of Senior Vice President and Chief Medical Officer at Audentes Therapeutics, which is an organization that is dedicated to developing gene therapy approaches for treating infants and children with diseases that have no alternative options.

Suyash graduated in Medicine at the University of Newcastle-upon-Tyne, UK, where he received commendations for Pediatrics, Obstetrics and Gynecology, and Medical Ethics. His Pediatric training was completed at recognized centers of excellence in the UK and Australia before he moved to industry. He is a United Kingdom board certified physician and is a member of the Royal College of Physicians (MRCP), the Royal College of Pediatrics and Child Health (MRCPC), and a Fellow of the Faculty of Pharmaceutical Medicine (FFPM). Suyash has published in several scientific journals on aspects of Pediatric medicines development and is a past recipient of the Outstanding Contribution Award from the Faculty of Pharmaceutical Medicine of the UK Royal College of Physicians.

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Barbara Wuebbels, RN

Vice President, Patient Advocacy
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.



ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Alan H. Beggs, PhD

Sir Edwin and Lady Manton Professor of Pediatrics
Director of the Manton Center for Orphan Disease Research
Boston Children's Hospital & Harvard Medical School

Dr. Alan Beggs is internationally recognized as an expert in the genetics of congenital myopathies, most notably for his work identifying numerous genes for these and other genetic disorders of childhood. He received his A.B. degree in biology at Cornell University and his Ph.D. in human genetics at Johns Hopkins University. Dr. Beggs has been a standing and ad hoc member of numerous National Institutes of Health study sections and grant reviewer for the Muscular Dystrophy Association, March of Dimes and various international funding agencies. He is a member of the scientific advisory boards of A Foundation Building Strength, the Congenital Muscle Disease International Registry, and several biotechnology companies developing treatments for rare diseases, as well as on the Board of Directors of American MedChem, a nonprofit devoted to drug development for rare and neglected diseases of children. His laboratory has led studies to identify the genetic basis for a wide variety of rare conditions with a particular focus on skeletal muscle and its defects in centronuclear myopathies and related neuromuscular diseases. Current research is focused on utilizing genetic approaches in human patients and animal models to understand the pathophysiology of, and to develop treatments for, these devastating childhood disorders.



Casie Genetti, MS

Genetic Counselor
Beggs Lab, Boston Children's Hospital

Casie Genetti is a genetic counselor 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 this past June. She has been enjoying learning the ins and outs of her new position and is thrilled to have the opportunity to meet more members of the CNM/MTM 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.

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Sabine de Chastonay, PhD

Cure CMD, Congenital Muscle Disease International Registry

Dr. Sabine de Chastonay received a Bachelor of Science degree from the ETH in Zürich, Switzerland, and a Ph.D. in Molecular and Microbiology from the University of Bern, Switzerland. She has lived in the United States with her family since 1992, raising two children as a stay-at-home mom and volunteering for various community organizations. In 2010, Sabine began working as a Patient Contact Coordinator with Patientstar LLC, a company specializing in clinical trial recruitment. In 2012, Sabine joined Cure CMD as a Research Associate focusing on the Centronuclear Myopathies. Sabine is the principal investigator for the MTM Genetic Testing Study, sponsored by Valerion Therapeutics, the MTM Event Study sponsored by Cure CMD and Will-Cure, the MTM Manifesting Carrier Study, sponsored by the NIH and Will-Cure, and the RECENSUS Chart Review Study sponsored by Audentes Therapeutics. Sabine's goal is to work with advocacy organizations, biotech and pharmaceutical companies, and the scientific community to support clinical trial readiness for the MTM/CNM community.



ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Jim Dowling, MD, PhD

Hospital for Sick Children, Toronto, Canada

Dr. Dowling is a staff clinician in the Division of Neurology and a senior scientist in Genetics and Genome Biology at the Hospital for Sick Children in Toronto. He received his BS and MS from Yale University and his MD/PhD from University of Chicago. His PhD was performed in the laboratory of Elaine Fuchs. He did his child neurology training at Children's Hospital of Philadelphia and then a neuromuscular genetics fellowship at the University of Michigan. He did a postdoctoral fellowship at Michigan with Eva Feldman, and then became an assistant professor at the University of Michigan before moving to Toronto in 2013. Dr. Dowling's clinical expertise is in childhood neuromuscular disorders, and he is one of the leading authorities on congenital myopathies. His research examines disease pathogenesis and therapy development for these disorders. His laboratory has helped pioneer the use of the zebrafish as a muscle disease model, both for new genetic identification and for drug discovery. Recent research highlights include discovery of CCDC78 mutations as a cause of congenital myopathy and identification of N-acetylcysteine as a potential therapeutic for RYR1-related myopathies.



Hernan Gonorazky, MD

Hospital for Sick Children, Toronto, Canada

Dr. Hernan Gonorazky is an adult neuromuscular specialist working as a clinical and research fellow at Dr. Dowling laboratory at the Hospital for Sick Kids, Toronto. He is actually involved in different projects ranging from diagnostic to therapeutics aspects of early onset myopathies. Although Dowling laboratory uses different models with CNM, Dr. Gonorazky is focusing his research mainly in human cells. By differentiating skin cells (fibroblast) into muscle cells (myotubes) through different mechanism, we can now perform high-throughput screening to identify different mechanism involve in this group of disorders as well finding targets for potential treatment. In addition to research, Dr. Gonorazky has a profound interest on developing new noninvasive technologies for diagnostic and clinical follow up purposes, such as the implementation of muscle MRI. As an adult neurology with a keen interest in pediatrics neuromuscular disorders, he has acquired extensive experience in transitional care in a variety of disorders.

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Anna Buj Bello, PhD

Genethon, Paris, France

Having done a post-doctorate at the IGBMC, Illkirch, France, where she worked for several years on murine models of myotubular myopathy and was awarded with fellowships from EMBO and Marie Curie Actions, Dr. Anna Buj Bello joined the INSERM (National Institute of Health and Medical Research in France) in 2004 and works at Genethon since 2009. She currently heads one of Genethon's research teams, as well as a translational program investigating gene therapy for the treatment of myotubular myopathy. Recently, Dr. Anna Buj Bello was awarded the 2015 Outstanding New Investigator Award from the American Society of Gene & Cell Therapy.



Johann Böhm, PhD

Assistant Professor

Department of Translational Medicine

IGBMC (Institut Génétique Biologie Moléculaire Cellulaire) Strasbourg, France

The IGBMC is one of the leading European centres for biomedical research. Our institute is located close to Strasbourg, the capital of the European Union, and hosts 47 research teams working on rare and common human disorders. Our team focuses on congenital myopathies. These progressive muscle disorders affect children as well as adults in all populations, and represent a significant burden for the patients and their families. The different congenital myopathies are classified by their specific structural defects in skeletal muscle. Myotubular/centronuclear myopathy is characterized by the abnormal central position of the nuclei in the muscle fibers. The 16 team members are specialized in genetics, cell biology, pathology, gene therapy, or pharmacology, and our research activities encompass the identification of new genes, the generation and analysis of animal models, and the development of therapies. Within the past years, I have identified several genes associated with congenital myopathies, and I discovered and characterized two dog models for myotubular and centronuclear myopathy (published in PNAS 2010 and PLOS Genetics 2013). The Labrador retriever with the MTM1 mutation was later successfully tested for a gene therapy approach (Sci Transl Med. 2014). My presentation at the MTM-CNM Family Conference will address the genetic bases of myotubular/centronuclear myopathy, the importance of a precise molecular diagnosis, as well as the development of alternative therapies to efficiently treat MTM/CNM.

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Mike Lawlor, MD, 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 September of 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. Additional projects being pursued in the Lawlor laboratory are focused on 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.



ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Linda P Lowes, PT, PhD and Lindsay N Alfano, DPT, PCS

Nationwide Children's Hospital

Drs. Lowes and Alfano work at Nationwide Children's Hospital in the Center for Gene Therapy and perform clinical evaluations of children with neuromuscular disorders. They co-developed ACTIVE-mini (Abilities Captured Through Interactive Video Evaluation – mini) to measure movement in children with muscle weakness. Working in neuromuscular disease, Drs. Lowes and Alfano have experience with traditional assessments which are often fatiguing and not well-tolerated. They designed ACTIVE-mini to measure the movement ability of children lying on their back. This testing measures the spontaneous movement of a child over 2-minute trials without frequent position changes. The objective of their research is to further develop this tool for use in both clinics and clinical research trials to reduce the burden of testing of children.



Dr. Michael Goldberg and Dr. Morton Goldberg

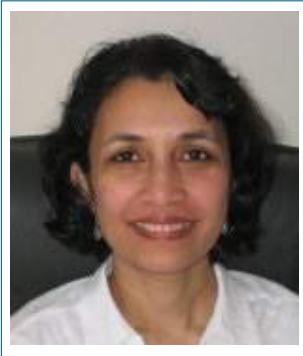
RYR1 Foundation

Michael F. Goldberg, MD, MPH, is the President of the RYR-1 Foundation. Mike is a neuroradiologist and Director of Neuroradiology at Allegheny General Hospital and in Pittsburgh, PA. He is an Assistant Professor at Temple University School of Medicine. He can be reached at mike@ryr1.org.

Morton F. Goldberg, MD, is an ophthalmologist specializing in retinal disease and genetics. He is the Joseph Green Professor of Ophthalmology at the Johns Hopkins School of Medicine and the Director Emeritus of the Wilmer Eye Institute. He is a member of the Institute of Medicine of the National Academies of Science of the United States.



ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Soma Das, PhD

Director
University of Chicago Genetic Services Laboratories

Soma Das is the Director of the University of Chicago Genetic Services Laboratories. Her laboratory was the first in the country to develop and perform molecular testing for congenital myopathies and she works with research groups to translate new gene discoveries into diagnostic testing for clinical use. Her laboratory uses cutting edge technology to develop the most comprehensive testing available for this group of disorders.



Amy Knight Johnson, MS

Genetic Counselor
University of Chicago Genetic Services Laboratories

Amy Knight Johnson is a certified genetic counselor, and graduated from the genetic counseling program at Northwestern University in 2010. She is currently the Senior Genetic Counselor for the University of Chicago Genetic Services Laboratories, an academic DNA diagnostic laboratory which focuses on testing for rare genetic diseases, including congenital myopathies. Amy is involved in a number of different roles within the laboratory, including interpretation of genetic test results, and development of new tests.



ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS

We are very excited that the team of researchers from the University of Florida is joining us again in 2015! 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 solutions for rare inherited diseases. Both of these initiatives are directed by Dr. Barry Byrne, MD, PhD. The UF team studies the potential interactive effects of regenerative therapies and rehabilitation to enhance neuromuscular function for individuals with inherited neuromuscular diseases.



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. Their approach is now being evaluated in a human clinical trial investigating the safety and efficacy of direct gene delivery to the diaphragm in Pompe disease. 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 in 2011 or 2013 will remember Dr. Barbara Smith, PT, PhD, who has collaborated with Dr. Childers with regard to measurements for pulmonary testing in the centronuclear myopathies, including XLMTM. Her ongoing research focuses on evaluation of respiratory motor control and the effects of therapies to facilitate recovery for patients with neuromuscular disease and breathing difficulties. She has worked with Dr. Byrne for several years in a clinical trial of intramuscular gene therapy to the diaphragm for children with Pompe disease. Dr. Smith has additional clinical and research experience with XLMTM, nemaline myopathy, and Duchenne muscular dystrophy, all of which is valuable for our community.

With over 20 years of clinical practice and 8 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 and for identifying clinical trial outcomes that are feasible for patients who use ventilator support.

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Terry Sexton, ARNP

University of Florida

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. Thanks to Terry, UF's interdisciplinary neuromuscular clinic has grown exponentially in the past two years, which has been a lifeline for patients in the southeast. Although she has recently retired from her full-time work responsibilities, she remains affiliated with UF on a part-time basis, and would not miss the opportunity to reconnect with her friends again at the 2015 conference!



Lee Kugelmann, BA

University of Florida

UF feels grateful to welcome Lee Kugelmann to our team. Lee graduated from Emory University in 2014 and works with Drs. Bryne and Smith as a Clinical Research Coordinator. The UF team has appreciated Lee's enthusiasm, organization, and writing skills in her role as coordinator for the MTM respiratory study.



Markus Renno, MD

Vanderbilt University/University of Florida

Dr. Renno began a prestigious pediatric cardiology fellowship at Vanderbilt University in 2014, but remains a part of the UF group in its cardiopulmonary studies of MTM/CNM and other muscular dystrophies. As part of his ongoing involvement in the MTM respiratory study, Dr. Renno will provide medical oversight of the respiratory tests during the conference.

ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Mindy Cameron

Patient Advocate, University of Florida

Mindy's role on the UF team is liaison, advocate, and communicator-in-chief. She is the person to speak to if you have questions about the clinical and research activities at UF, including open research studies, advocacy and fundraising opportunities, speaking engagements, or visiting Gainesville.



Stephanie Salabarría, BS

Student Researcher, University of Florida

Stephanie has worked with Dr. Smith on her undergraduate honors research project for the past year, to evaluate the relationship between respiratory muscle strength and the coordination of breathing-related movements in the chest and abdomen in individuals with XLMTM. Stephanie's career goal is to become a pediatric physician.



ABOUT THE RESEARCHERS AND MEDICAL PROFESSIONALS



Dr. Martin "Casey" Childers, DO, PhD

University of Washington

Dr. Martin "Casey" Childers joins us from the University of Washington, Institute of Stem Cell and Regenerative Medicine. Dr. Childers is leading the exceptionally promising research into gene replacement therapy with Labrador retrievers who have MTM. This research is paving the way to human trials in the future. About his work, he says "I am passionate about finding answers for patients with devastating muscle diseases, particularly those with congenital myopathies and Duchenne muscular dystrophy. As a rehabilitation clinical specialist and basic scientist who investigates rare diseases, it is a privilege to help patients and families struggling with extraordinary burdens. Although we do not have many answers yet, efforts are now yielding new hope for our patients. This is an exciting time of discovery in muscle disease research."



Dr. David L. Mack, PhD

University of Washington

Dr. David Mack is an Assistant Professor in the Department of Rehabilitation Medicine as well as a faculty member of 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, which resulted directly from different phases of his professional training. The 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 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.

FAMILY AND 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.



CMDIR (www.cmdir.org)

The Congenital Muscle Disease International Registry (CMDIR) is a central hub for up to date information regarding clinical studies and trials for all types of Congenital Muscle Disease (CMD). Through the CMDIR, the CMD community has a home-a place to register with and without genetic confirmation of disease and a means by which to be contacted for clinical news. During the conference, seek out Dr. Sabine de Chastonay for more information.

CMD Tissue Repository (www.cmdir.org)

Dr. Mike Lawlor has led the establishment of a Congenital Muscle Disease Tissue Repository at the Children's Hospital of Wisconsin, which will allow the centralization of human tissue storage and distribution to scientists performing research on muscle diseases with the hope of improving the pace of research in our field. The CMD Tissue Repository is part of the triad of scientific resources that Cure CMD oversees, which also includes the CMDIR and the CMD BioBank at Coriell. Dr. Mike Lawlor will be at the conference to speak with families interested in enrolling in the CMD Tissue Repository.



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

Inspired by his experience at the 2014 Myotubular Trust Family Conference, Holger Fischer and his wife Jen Bilbao organized the first German Family Conference in June 2015. It was a great success! They founded the German CNM Association "Together Strong!" of which Holger serves as President. Their organization will aim to raise awareness, help support families, and raise funds for research. We are delighted Holger has travelled to be with us at our conference and please take a moment to meet him throughout the weekend.

FAMILY AND COMMUNITY ORGANIZATIONS



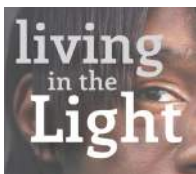
Information Point for Centronuclear and Myotubular Myopathy (www.centronuclear.org.uk)

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 14 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)

Created by Alison and Paul Frase in honor of their son Joshua, for over eighteen years the JFF has been raising funds to support research in hopes of finding a treatment and/or cure for Centronuclear Myopathies. 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. The JFF also host an International Family Registry/global map. Please take a moment to introduce yourself to Paul, Alison, and Josh's sister Isabella during the conference.



Living in the Light (www.frompatienttoperson.com)

Levi Gershkowitz is a photographer and writer creating compelling patient ethnography for education, advocacy and marketing—with a focus on rare diseases. Founded in 2012, Living in the Light is a patient advocacy initiative utilizing the potency of portrait photography to educate about the realities of rare diseases. The aim is to present a dignified perspective of individuals living with rare diseases, which conveys the unique wisdom and beauty they carry. We are thrilled to have Levi joining us this year to help capture some images of the 2015 MTM-CNM Family Conference.

FAMILY AND COMMUNITY ORGANIZATIONS

Myotubular Myopathy Resource Group



Myotubular Myopathy Resource Group (www.mtmrg.org)

The MTMRG was created by Pam and Gary Scoggin, and was one of the pioneers in connecting families and sharing information about MTM. We are grateful that they were the sponsoring organization of the first FamilyConferences by allowing conference funds to be raised under their nonprofit prior to the creation of MTM-CNM Family Connection, Inc. Please say hello to Pam at this year's conference!



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 groundbreaking research for our community. The Myotubular Trust offers scientists within the community competitive scientific grants. The Myotubular Trust also hosts European conferences for the MTM-CNM community. Our team, along with the community, learned so much from their conference held last year! The Myotubular Trust also manages the The Myotubular and Centronuclear Myopathy Patient Registry which is an international database specific to our condition. The registry has been developed 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). 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 the Foundation is to fill this much-needed void. We are very happy to have Drs. Michael and Morton Goldberg joining us for the 2015 conference!



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. You may visit the Will-Cure website to read more about the active and ongoing research they are funding within the community. Also be sure to say hello to Melanie, Dan, Juliet and of course Will at the conference.

CONFERENCE PLANNING TEAM



Erin Ward, Mom & Conference Director, Mark Ward, Dad & Planning Team Member, and Will Ward



The Ward Family

Our son Will was born in 2001 and spent the first few months of his life in the Children's Hospital Boston 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.

At the beginning for us, we had not heard of MTM before and even some of the doctors knew it only from textbooks, which were not exactly encouraging in their prognosis. Our family had an opportunity to experience "being at the right place at the right time" as we learned that one of the lead researchers in the world researching MTM was right at Children's Hospital Boston. Dr. Alan Beggs came to Will's bedside and shared vital information on this rare disease. An amazing fourteen years later, Will has just graduated from the eighth grade and will be attending our local High School in September. He loves the Boston Red Sox and he plays on a Challenger baseball team. Will enjoys music, books, movies, and likes to bowl. Will also has two very special dogs, Gracie & Simba, who come from a "family" of Labrador Retrievers who carry the same MTM gene.

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 in his lifetime there will be treatments and eventually a cure for MTM. We have learned the importance of helping to strengthen our community, coming together to support each other and celebrating our loved ones affected by myopathies. We hold onto the hope that anything is possible and together we can truly make a difference. With this mission in mind, in 2014 we helped to found MTM-CNM Family Connection, Inc., of which Erin serves in the volunteer role of President and Mark as Treasurer. We feel extremely blessed that we've had this opportunity to plan another conference for our community and hope that everyone has an amazing experience this weekend!

CONFERENCE PLANNING TEAM



Marie, Mom & Planning Team Member, David, Dad, Lucian, Blaise, Reid & Paley Wood, Brothers



The Wood Family

David, Reid, and I (Marie), along with “Granny” Sheri Peery attended the first MTM-CNM Family Conference in 2009. It was a life-changing experience, and I’ve been involved in the family conference planning team ever since. I currently serve on a volunteer basis as the Vice President of the MTM-CNM Family Connection and love my work. David is a philosopher with a PhD from Fordham University, who speaks and teaches at various venues and writes for journals and other publications. I attended the 2011 and 2013 family conferences on my own, but this year, the whole gang (including Granny) is here!

Since Reid was born in 2007 and after Paley joined our merry band in 2010, our lives have changed dramatically. In the early NICU days, Reid struggled to grow while on nasal cannula and didn’t do much better with CPAP trials, so he got a trach and g-tube at about two-months-old, which immediately resulted in rapid growth and strength. In the hospital, Reid was trialed without any vent support and appeared to be doing well until he had a major hypoxic event and had to be revived. This completely changed our approach of trying to make Reid fit somebody else’s mold, and we shifted to supporting Reid to be the person—body and soul—that he is.

Reid and Paley are both trached, vented, fed via g-tube and use wheelchairs. We’ve faced many challenges and difficulties because of Reid and Paley’s medical issues, however our family and faith have only grown stronger through it all; we are truly blessed. Lucian and Blaise are affectionate and playful brothers to Reid and Paley, who in turn bring out the gentler, nurturing side of their elder brothers. Joy and love are reciprocal impulses in our home.

Even though the littles need quite a few “accessories,” we see each one of the boys as perfect and adorable just the way he is. Helping Reid and Paley make progress in meeting milestones is a focus and we are proud of their achievements, but that is much less important to us than making moments meaningful. So, we are excited about what a treatment might mean for Reid and Paley and for every one of us in the future and we want to do all we can to help that cause, but tomorrow is not a guarantee, and we aren’t waiting for life to begin only after a cure. Life is happening right now, and a beautiful world is always before us: there are people to love, adventures to be had. We are embracing the present moment, simply grateful for each day, each smile, each hug and kiss.

CONFERENCE PLANNING TEAM

The Mashinchi Family

Javad Mashinchi came into this world by C-section on November 1, 2001. Everything was planned, grandparents outside and older sister waiting to be the first one to hold this new baby. Like a lamb, Javad entered the world, quiet as can be. As soon as the doctor announced we had a boy, I noticed that there was no sound, no crying, nothing. I kept asking why he wasn't crying and was told that everything was ok. The nurses whisked Javad off to the hospital's "Level 2 Nursery" but after 24 hours a decision was made to transfer him to the local children hospital where he would be admitted to the NICU. By the time I arrived on the Family Birth floor, the neonatologists had a variety of concerns for our son; generalized hypotonia, enlarged heart, respiratory issues. Luckily, a cardiologist was available and ruled out any heart issues (normal), but mentioned an enlarged thymus gland. They ran every test they could think of: Praeder-Willi, Myotonic Dystrophy, and a variety of others. All of these tests came out negative and left the doctors scratching their heads. We left the NICU six weeks later with no diagnosis and on to "live our lives." When Javad was a month old, he was given a diagnosis of Myasthenia Gravis. I learned everything there was to know about the disease and grew comfortable with the road that we were on.



*Shannon, Mom & Conference Planning Team,
David, Dad, Adam, Brother, Stesha, Sister, Simon,
Brother and Javad*

When Javad was about 21 months old, our neurologist at Shriners Hospital told me that he felt Javad had been misdiagnosed and we should do a muscle biopsy. Five weeks later, I received a call with the diagnoses of Centronuclear - Myotubular Myopathy. Frantically looking on the Internet, I was stunned to learn that Javad had a disease that was so incredibly rare. I searched for information and found very little information that was recent. I felt so alone. I cried and wished that there was a way to get connected with others...I wondered if there were even any others?

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.

This last March Javad had a major health setback involving a seizure. This event changed his ability to drive his wheelchair, move his arms, and do many of the things he had done previously. We are now on the slow road to recovery, but are hoping to get back to his old sassy self. 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!

OUR FAMILIES



**Barbara, Mom, with
Phillip Baron**



**Dave Zulligen, dad, with
Phillip Baron**

The Baron-Zulligen Family

Until recently we were a very busy happy family of three. Dave is self-employed and was always available to Phillip (during our all too often drought in nursing) while Barbara worked a part-time job. Phillip was cognitively four years old so he was always playful, happy, and slightly mischievous. During his school years, Phillip's teachers and therapists were amazed at how much he loved school. Phillip was never able to walk and as he got older lost arm and hand strength gradually. He used a power wheelchair from the age of seven. He loved playing games on his PC and he became proficient at searching YouTube! When he was finished with school at the age of 22 he loved going on field trips with his nurse using the Pace bus. We loved going to Brookfield Zoo and many of Chicago's great museums.

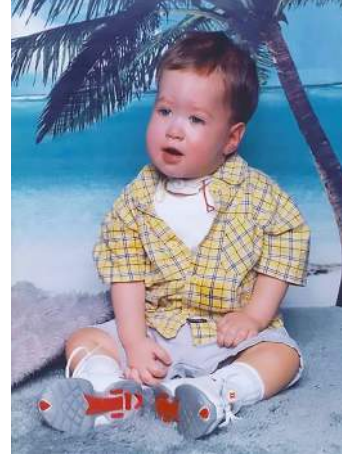
Every day Phillip matched his clothes with his many activities. We read books or watched movies, music according to his t-shirt. Phillip came by his OCD naturally. He obsessed about flags, penguins, elevators, bees and all holidays.

Phillip loved to have tastes of a variety of soups, dressings, sauces, and beverages. Whenever we have an eggnog or a spicy V8 we will lift our glass to Phillip.

OUR FAMILIES



Samantha, Mom, Danny, Dad
Ashton Paul



Austen O'Neal Scott



The Biddle-Scott Family

We are the Biddle-Scott family. We are from Logansport, IN. Danny and I have two children that are affected by MTM. Our first son Austen O'Neal was Born August 14, 2002 and earned his angel wings on February 6, 2004. He was an amazing little boy that changed my life for the better! He loved Winnie-the-Pooh and playing peek-a-boo! Our youngest son, Ashton Paul, was born in December 2004 will be in the 5th grade this year. He is full of personality and is extremely independent. Ashton loves to play games on the Xbox/PS4 and iPad. (Yes, he is slightly spoiled!) His favorite places to go are Walmart and Gamestop. He has recently been trying new foods such as barbecue chips and spicy mustard. My mother (Ashton's grandmother!) Dawn has been a huge help to me. Although she works full time at Walgreens, she is always willing to help out whenever and however she can. Also my oldest sister Shawnda (Ashton's "Aunt Dada") is attending this year's conference with us. They are both super excited to attend this conference with me!

OUR FAMILIES



Kristin and AJ, Parents of
Justin Bonny

The Bonny Family

We moved back to New Hampshire 2 years ago after living in Bolingbrook, IL for 11 years. Justin was born in the Chicagoland Area in 2003 and was initially diagnosed with MTM1 (recently we found out that his mutation occurred on the RYR1 gene). We were lost for a while in how to navigate our new normal but when Justin turned 3 we found the Yahoo groups for MTM and CNM and it changed our world, we had hope for Justin's future. We were lucky to be able to attend the family conference in 2009 and 2011 and were completely energized.

Justin loves the city and misses Chicago terribly but finds some comfort in our many trips to Boston. We now have a couple of seasons of adaptive skiing and power soccer under our belts which we all love and we have recently started kayaking. After several rough years we are finally settled and healthy!!! And looking forward to a great summer and a fabulous conference.



OUR FAMILIES



Rebekkah, Mom, Jaden and Keegan, Sisters, and Chason, Brother of Lane Garret Jackman



Lane Jackman



The Brassfield-Jackman Family

We are the Brassfield-Jackman family. We live in Oklahoma City, OK. My name is Rebekkah I'm a Registered Nurse working in an infectious disease office as an infusion nurse and part time as an intensive care nurse and for some reason decided I wasn't busy enough and started my pursuit of my Masters degree as a Family Nurse Practitioner this summer. I have four wonderful children. My oldest daughter is Jaden, she is the mother hen of the family. Chason is my eight-year-old son, that has the biggest heart. Keegan is my seven year old, and as her name is defined she is small and fiery. My oldest son Lane passed away in 2010 when he was almost 9 years old, talk about an ornery kid just ask the attendees of the first MTM-CNM family conference. (I also have a 14-year-old nephew, Christian Hanna, who has x-linked MTM.)

Our x-linked MTM story started in 2001 with the birth of Lane Garrett Jackman. He was your typical definition of MTM, floppy with poor respiratory effort. Lane lived an extraordinary life and we lost him unexpectedly in 2010. Lane overcame so many obstacles in his short life. He learned to drive a power wheelchair by the age of two and even tried driving into the pool by age six. He loved cars, going fast and cartoons. He loved being a brother, but probably enjoyed tattling more. He loved to sleep in on the weekends and hated getting up for school. He liked to play in the rain (although we were never sure if that was a good idea for his wheelchair.) He danced when he thought no one was looking. He was becoming quite the bowler. He was recognized anywhere took him. He was the best kid. We never knew how short his time would be, but we cherished every moment and miss him dearly.

OUR FAMILIES



**Kimberly, Mom, Travis, Dad, Colten, Brother
Evan Brown**

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 the next 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 twelve 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 six years with nursing issues and set backs in Evan's health, but through it all we've gotten closer and stronger as a family. Evan continues to grow and reach milestones little by little and has always been the sweetest, happiest little boy. Evan is a favorite at school and often brings home notes and art work from the kids in his class. One of the activities that we enjoy most as a family is going to the race track to watch Papaw's horses race.

OUR FAMILIES

The Browning Family



Joanna, Mom, Garrett, Dad, and Sam, Brother of Jacob Browning

God blessed us with Jacob on July 1, 2014. When Jacob was born he had difficulty swallowing and was not moving as expected. He immediately began his stay in the NICU, which lasted 3 1/2 months. Jacob was sent home on CPAP through a nasal cannula. A diagnosis was not discovered until January.

Jacob is a beautiful, sweet and determined boy who loves interacting with people. His spirit is filled with love and joy for life, which is infectious to all who encounter him. Jacob is a hard worker. He receives a great deal of physical therapy, occupational therapy and speech therapy each day. Jacob WANTS to move and it shows with how hard he works and the progress that he makes! Jacob claps his hands when we say 'YAY Jacob', he blows kisses, and he loves to play peek-a-boo with his daddy. Jacob's most favorite toy is his four-year-old brother Sam!

We are so fortunate for the nurses that help us care for Jacob. We truly believe Jacob grows and improves on LOVE and everyone who meets him, falls in love with him! We know Jacob's potential is limitless and we trust God as we walk through the everyday love and care of Jacob.
(Psalm 139 - 'Jacob's Psalm')



Rachel Bronstein

Buffalo Grove, IL

OUR FAMILIES



*Natalie, Mom, Matt, Dad, Lilly, Sister and
Cooper Bushey*

The Bushey Family



Hello! We are the Bushey's, Matt, Natalie and twins Lilly and Cooper. In about 3 days after we get back we are going to be moving to Springfield Ohio. The kids will be starting kindergarten (their first day is on their birthday) and Matt will be starting a new job as a Math Curriculum Coach, and I am a Labor and Delivery nurse. 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 acquity 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 certinally 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!

OUR FAMILIES



Randi, Mom, Jim, Dad, Peyton, Sister and Austin Cook

The Cook Family

Hello, We are the Cook Family. Randi (mom), Jim (dad), Peyton (6), and Austin (4). We live in Maple Shade, New Jersey. I was fortunate enough to attend the 2013 conference and am beyond excited that we will all be at the 2015 family conference along with Austin's great-grandmother, Nancy Wedin.

We have had only a small portion with our journey with MTM1 so far. When Austin was born on 10/28/2010, we were in and out of the Children's Hospital of Philadelphia for the first year and a half of his life. He had a muscle biopsy, g-tube, g-j tube, tracheotomy, chest tube, and was determined ventilator dependent. Since then he is now sprinting off the ventilator and breathing on room air during most days! We have got a power chair, that we are still learning how to drive, uses a stander daily, and a Kid Walk walker that he uses! Austin continues to amaze and surprise us!

Austin loves playing with cars, trucks, planes, and his iPad. We also love family camping trips with our pop-up camper. We also enjoy family outings to just about anywhere! Recently we have visited the Constitution Center (PA), the Circus, a day out with Thomas the Train, parks, fishing, and our extended relatives. This past year, we received our Make-A-Wish trip to Disney World. It was above and beyond amazing!

Peyton loves her brother unconditionally and is constantly looking after him. She loves spending time with him laying on the floor and hanging out. Peyton helps Austin in any way she can. We recently got an elevator lift on the front of our house, and she loves helping him ride it!

In November 2014, the newest member of our family arrived: Rosalind is a 3 year old lab-beagle mix from the MTM community in Seattle Washington. She is absolutely amazing and loveable. I am so proud that she could help us in some way come closer to a cure!

We can't wait to see everyone again and to meet all the new families, doctors, and researchers at the 2015 Family Conference!!!

OUR FAMILIES



Kayla, Mom, Ryan, Dad, Makenzie, Sister
Grayson Davies



The Davies Family

Meet the Davies Family! Ryan, Kayla, Makenzie and Grayson! We reside in Liberty, a suburb of Kansas City, Missouri. Ryan is a stay at home father. I am an oncology nurse and just recently graduated as an Acute Care Nurse Practitioner. Makenzie is an active three-year-old, who is the proud big sister of Grayson, seven months old.

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. We were finally discharged from the NICU after three and a half months. Unfortunately, he has had multiple admissions since his original discharge due to respiratory issues. On his five month birthday, he had respiratory failure and had a tracheostomy performed and ventilator support 24 hours a day. Since his surgery, he has thrived. He was finally discharged from the PICU the last week of June and is looking forward to this conference to meet other MTM warriors and their families!

OUR FAMILIES



Maggie, Mom and
Lincoln Avery DeLuna



The Deluna Family

Hello friends, we are the Deluna family! We are from Tampa, Florida and are so excited to be here representing our little man: Lincoln Avery Deluna. Lincoln is almost 19 months old and boy is he smart! His favorite activities are listening and signing to music, dancing, reading books, and playing with his medical equipment (although he has plenty of regular toys lol)! He is the light of our lives and our reason for existing. I am Maggie and my husband's name is Anthony. I am beyond grateful to have such an amazing, strong, and invested partner in life. He is my other half and my true love. As he said in his wedding vows-he is my knight, not in shiny, but "battle-tested armor." He is my rock. My incredible sister, Katie, lives with us to help with Lincoln. We honestly couldn't do it without her! She is my best friend and one of Lincoln's loudest cheerleaders. She has so graciously stayed back in Tampa to take care of Lincoln with the nurses so that we could be here at the conference. We are joined at the conference with my mother and father: Donna and Philip Germann, as well as my mother-in-law: Ronda Clark (my father-in-law, Ron, is in Kenya doing mission work!). Lincoln is blessed with the greatest set of grandparents who are there to help wipe our tears and cheer-on our successes. We are a "ride or die" crazy family and love one another fiercely. We know what loss feels like- we lost 3 infant boys to XLMTM almost 30 years ago. One of those boys was my brother and the other two were my cousins. We are so grateful for all of the research that has been done in these 30 years. The Beggs Lab, Dr. Childers, Audentes Therapeutics, Genethon, the incredible team of doctors at UF - Shand's Hospital, the online support groups, this beautiful family conference...we cannot "thank" enough. We are so thankful for you all. Lincoln's success is in part to you. It certainly takes a village. I am proud to say that Lincoln Avery is not only surviving but thriving!

OUR FAMILIES



Jennifer and Holger, Parents of
Emil Fischer



The Fischer Family

Hello, we are Jennifer Bilbao and Holger Fischer, the parents of our wonderful Emil, who was born on February 19, 2014 in Stuttgart, Germany with myotubular myopathy (MTM). After the diagnosis that Emil suffers from x-linked MTM. We were very sad and felt very lonely. However, only one day after this, Jen found the information about the Myotubular Trust. To our surprise, the Myotubular Trust was organizing a conference this very year in London. Holger registered and got even invited to take part in a focus group meeting about a gene therapy to heal MTM on the day before the actual conference.

How lucky we are! At one moment we thought we were all alone and by the next days we found out that there were others that would share their experience with us and that there was even substantial research going on to find a cure for that very rare disease! And this research was funded by the Myotubular Trust.

This conference inspired us to organize a similar one in Germany. In less than 6 months, we booked the hotel for the conference, arranged the children program, and contacted the families and experts in the field. The conference took place in June 5, 2015 and really surpassed our expectations. It was great to meet 24 families and to learn how to manage this condition and about therapy possibilities.

During the conference, we even founded the German CNM Association "Together Strong! (ZNM – Zusammen Stark! e.V.) to help the families in Germany to cope with CNM, to inform them about the condition and to support the running research at the moment through the Myotubular Trust. Take a look at our website: <http://www.znm-zusammenstark.org/>

P.S. If you want to know more about Emil and his daily life you can visit his blog: <http://emil-augustin.blogspot.de/>
He does not yet know English... it is in German and Spanish only but it has a lot of pictures.

OUR FAMILIES



Alison, Mom, Paul, Dad, Isabella, Sister and Joshua Frase



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 and a suction machine and told us “if he lives to see his first birthday, bring him back and they would re-evaluate him.” At 3 ½ months old a muscle biopsy confirmed Myotubular Myopathy, at the time only 50 cases were known worldwide. 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 only three publications written at that time, two 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 team of researchers was established at Boston Children’s Hospital and that began an almost nineteen-year 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 and the University of Florida.

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, 40 days shy of his sixteenth birthday. He taught us so much about life. He possessed such courage, tenacity, optimism in the battle for his life and never once did he question, “why me, God?” He inspired us to take risks, to never give up when the going got tough, and 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, middle 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!

Always for the children. Much Love, Alison, Paul, and Isabella

OUR FAMILIES



Sarah, Mom, Patrick, Dad, and
AJ Foye

The Foye Family

Wishing you all an enjoyable conference! Our teenage son has Titin-related Centronuclear Myopathy.



Kayla, Mom, Rob, Dad, Rilee, Sister and
Reese Garland

The Garland family

We are the Garland family from Indiana. Dad (Rob) is an automotive technician and Mom (Becky) works for the County Government. Our family is lucky to include Rilee who is 8 years old and getting ready to be in second grade and Reese who is almost 5 years old and getting ready to start his last year of preschool. Rilee would call herself an athlete who loves gymnastics, cheerleading, riding her bicycle, and playing ball with her little brother. Reese is the silly guy of the family! He enjoys music, dancing, and watching sports of any kind. Reese also likes to play games and watch Youtube on his iPad.

As a family we try to make everyday enjoyable whether we are doing something special like attending an Indianapolis Pacer game, or mundane like going to dinner or the grocery store. Becky and Rilee had the opportunity to attend the 2013 Conference and are super excited to share the experience with Rob and Reese this year! Go Colts!

OUR FAMILIES



Jessica, Mom, Adam, Dad, and
Declan Hair/Marmon



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.

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 for now almost two years!

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 saw Declan gain strength and momentum in the hospital (especially since his trach), but since he's been home, he's thrived! Our unmoving boy is doing amazing things. He is still working on head control, but can hold it up with no problem when in any of his chairs, his stander or sitting on mom or dad's lap. He can throw balls with dad, is king of pointing and can pick up the smallest speck of anything – an "OT's dream," as his therapist has called him. He has been driving a power chair with his head since he was 10 months old – one perk of living in a hospital, I guess! And, thankfully, he has had some great therapists at home who continuously push him (and us)!. Thanks to them, Declan has even taken steps in a gait trainer. And, he wants to so badly but that darn head control...

We also work on vent weaning daily, as well as on using the Passy Muir valve, so we have a busy little boy. Best of all, our wonderful little three year old is ALL OF THREE. He has the silliness, stubbornness, need for hugs and kisses, and temper to rival any three year old out there. He loves music, is obsessed with Curious George and would be happy just being around animals- especially dogs and horses. He is scheduled to start school in August, and we can't wait to see what happens once he gets around other kids his age!

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!

OUR FAMILIES



**Stesha with her Brother,
Javad Mashinchi**

Stesha Heselius-Mashinchi

My life with Javad has not been a normal one, but name one sibling who can say their life with their sibling is. It is OUR normal, but it's not the normal I expected from my life when I was little. I couldn't do the same things that my friends could do. I couldn't go to all the places my friends could. I knew from the day he was born, when I was 11 years old, that my life was never going to be the same. I could never go camping. I didn't invite many friends over. It was harder to make close friends because I had to REALLY trust them before I could let them REALLY know me and know about my brother. It wasn't that I wanted to hide him, it's that I could, I could pass for normal and not have to see the look on people's faces when they didn't know how to respond or started to feel pity. I didn't want pity. It was my life, it was what I knew, it wasn't pitiful, it didn't deserve their sympathy.

I don't want you to think for one second that I regret having my brother. I don't. I am stronger, more mindful, more loving than anyone I know. I have lived through more of a life, have experienced more, and had to see both a wider and more narrow scope than any other 25 year old that I have met. My life was just different. I've been suctioning since I was 12 years old. I can change a trach, change a vent circuit and sense illness from the smell of someone's breathe. I can carry on medical conversations with all my nursing friends. I have spent more holidays in the hospital on the hand holding side of the bed than I can count. My brother made me strong. My brother made me an ally. He changed my life, changed the way I thought and the way that functioned. My life as a sibling has been different than the relationships that my friends have with theirs, and there is nothing wrong with that.

We had the hard times. I had to grow up too fast when I was too young. But I got a family out of it, I got to be an advocate for other siblings. Sometimes people forget that just because we're "healthy" that we are living it too. We feel the missed play dates and the late pick-ups from soccer practice. We don't get to just tell people about what's going on at home when our siblings are sick. Adults understand, kids don't, and kids can be mean when they don't understand. The life of a sibling is different than the life of a parents because while parents lives stop (in a way) when our siblings are sick, ours don't, we just have to keep plugging through life and it can be hard. I don't regret for one moment that my life was different, I don't wish my life had been "normal" I just wish sometimes that I would have had the way to talk about it when I was younger. So here I am now, 25 years old, and I'm giving words and voice to the younger siblings that may not be able to express how they're feeling or feel guilty when they need to be needy. I'm here for them because I had to figure it out by myself and I don't want them to have to.

OUR FAMILIES

The Huseth Family



Christopher Huseth - Woodbury, MN



Andrew Huseth - Phoenix, AZ

Diane, Mom, Paul, Dad, Daniel and Benjamin, Brothers of Christopher and Andrew Huseth

Christopher (44 years old) was born in March of 1971 with classic symptoms of centronuclear (myotubular) myopathy. He was formally diagnosed at 9 months old upon a muscle biopsy at the time of a hospitalization with pneumonia. At this point, he also received a gastrostomy feeding tube. As one could imagine, very little was known to us, and we simply trekked forward with our lives.

Andrew (34 years old) was born in August of 1980 with the same symptoms as his older brother, and subsequently was diagnosed with centronuclear (myotubular) myopathy. During this time, we underwent further clinical analysis and testing. Evidence and findings of additional muscle biopsy procedures suggested the mother as a “carrier” and as owing to an x-linked recessive inheritance; at the same time, this was cautioned more as ‘research’ than an absolute diagnostic. In the late 1990s, both sons participated in the Dr. Herman research study, and a mutation in the MTM1 gene was not detected. As we’re aware, exponential strides and advancements have been made in the last 10-15 years, let alone the last 2-3 years. Through the generous support that is beyond our wildest imagination and words, Andrew received genetic testing and confirmation of a mutation in the RYR1 gene in November of 2013.

The development of motor skills and milestones came slowly for both boys, but none of it could have happened without receiving the abundant, much-needed support of family, friends, doctors, and early assistance development programs from their schools. Hospitalizations with pneumonia were common for both of them at young ages, but diminished as they grew stronger and older. Both of them were able to integrate and to excel both inside and outside the classroom. We are very blessed that both of them have thrived independently and have done well.

Today, Christopher makes his home in the St.Paul/Minneapolis area, works full-time for Comcast Communications and is an avid sports fan. Andrew makes his home in Phoenix, Arizona and works full-time as an urban planner in the private sector. Outside of work, he is a diehard Arizona State University football fan, and has been known to go skydiving on multiple occasions.

OUR FAMILIES



The Jarrell Family



*Krystal, Mom, Nathan, Dad, Connor and Griffin,
Brothers of Graham Jarrell*

Hello everyone! We are the Jarrell family. We live in Louisville, KY. Nathan and Krystal are small business owners and the proud parents of: Connor, who is 13 and going to be in 8th grade, Griffin, who is 11 and will be in the 6th grade and Graham, who is 2. Connor and Griffin both enjoy playing soccer and are very active. They completely adore their little brother and he loves them just as much! Graham absolutely lights up when he sees his brothers.

We were blessed with our precious Graham at 6:14 am on October 20, 2012. He was delivered via an emergency C-section due to an abruption. Graham was unresponsive and not breathing, but after a few minutes, the staff was able to resuscitate him. Because of this difficult delivery, Graham sustained white matter damage and a brain bleed, which led to hydrocephalus. Right after Krystal woke up from the anesthesia, she was allowed to see him for a few short minutes and then he was rushed to Kosair Children's Hospital NICU, where he would spend the next several months.

At first, the doctors thought that Graham's muscle weakness and inability to come off of the ventilator was due to him getting too much anesthesia and the traumatic birth he sustained. However, at 3 months old, they did a muscle biopsy while he was in surgery to have a g-tube placed and it was from that biopsy we received his diagnosis of X-Linked Congenital Myotubular Myopathy. When we found out what it was and the prognosis, our hearts sank. We were determined to make every second with Graham count, because we didn't know how much time we would have with him. Krystal photographed him every day from day 1 until he was a year old. Graham has had a total of five surgeries (Ventricular Access Device placement, AV Shunt placement, G-tube placement, Tracheostomy, and a trach revision). He's had 4 bouts of pneumonia, once was double pneumonia, which caused his left lung to collapse. He has been the strongest person we have ever met. He's been through so much, but soldiers on with a beautiful smile. Graham has taught us so much and been such an inspiration to so many.

We were blessed to be able to bring Graham home May 5th of 2014. He has amazed all of us with his continuous strength improvement and growth in every area. He LOVES books and music. His favorite cartoons are Little Einsteins, Bubble Guppies and Mickey Mouse Clubhouse. We are currently working on getting him to hold his head up without support and communication. He's learned some sign language and shakes his head "yes" and "no" appropriately. He says "Mama" clearly, which Krystal absolutely loves! We are so very fortunate to have Graham in our lives and treasure every moment we have with this amazing, precious little boy!

OUR FAMILIES



Carrie, Mom, Ed, Dad, and Oliver, Brother of Elliot Koh



Elliot Koh

The Koh Family 

We are the Koh family from Denver Colorado: Ed, Carrie, Elliot, Oliver and our black lab Josie.

We became a family on May 18, 2012 when our sweet Elliot surprised us 6 weeks early! Our joy quickly turned to concern as he required extraordinary measures to breathe and was largely unresponsive. At two months of age, a muscle biopsy revealed that our little peanut had XLMTM.

After struggling with sadness, anger and fear, we found the strength to focus on making Elliot's life one full of love, laughter, and happiness. Everyday became a personal challenge: How do we make today a great day for Elliot! With that attitude and strength from family, caregivers and friends, we truly LIVED and LOVED with Elliot.

We lived in the NICU for Elliot's first 5 months. We were thrilled when we were finally able to get him HOME for good! We spent the last 2 ½ months of Elliot's life at home cherishing every moment. Sadly, on January 6th, 2013, God called Elliot home, and he peacefully left us to be free of his disease. Our little guy was a fighter from day one. In his short time with us, he demonstrated a level of courage, determination and strength that would make any parent proud.

The thought of experiencing joy again after losing our son was unfathomable. But God had a plan for us. He blessed us with a little brother for our angel Elliot. On July 22, 2014, our sweet, silly Oliver was born, kicking and screaming. Oliver brought joy back into our lives. We marvel at every milestone and thank Elliot everyday for giving us a perspective filled with awe, patience and gratefulness. Life has been a balance of living with the grief of losing Elliot and the joy of new life that Oliver has given us.

OUR FAMILIES



*Corey, Mom, Bruce, Dad, and Daniel,
Brother of Spencer Leibowitz*

The Leibowitz Family

Spencer was born February 12, 2001. He came a little earlier than expected, but we were thrilled. After spending a week in the hospital in which he was born he was transferred to CHOP (Children's Hospital of Philadelphia). After blood work and a muscle biopsy Spencer was diagnosed with X-linked Myotubular Myopathy. In July 2002 Spencer had a tracheostomy and has been ventilator dependent since. He remained at the hospital for seven weeks; his mother always by his side. At that point Spencer's personality really started to shine through.

At the age of five we got Spencer his first motorized wheelchair. After that there was no stopping him. Spencer started preschool at Easter Seals and then it was on to public school. We always felt that Spencer should be included with his same age peers in a regular school setting. In elementary school we saw tremendous growth academically, socially and physically. He has a core group of friends that he hangs out with. In March 2014 Spencer became a Bar Mitzvah (Jewish religious service where you read from the Torah). This was one of the proudest moments of his life. Spencer interests include Play Station 4, playing baseball and football and he is a big fan of NASCAR. Perhaps the most important aspect of Spencer's life is being a big brother to Daniel. Daniel is a rambunctious seven year old and the boys get along wonderfully.

Like many families we have been through a lot over the past 14 years, but it is only through the support of family, friends, doctors and nurses have we been able to get to this point in our lives. We consider ourselves truly blessed.

OUR FAMILIES



Jessica, Mom, Marty, Dad, and Kate Elizabeth, Sister of Caden Lewis



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 13, he is witty, talkative, loves to travel, watch movies, play video games, build Lego sets and play Minecraft. He is entering the 8th grade in middle 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 first MTM-CNM Family Conference.

OUR FAMILIES



Cason Lofton-Scott



Dana, Mom, Roy, Dad, and Corey, Brother of Cason Lofton-Scott

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 12 years old and is such an amazing young man. He is in the 5th grade (he had to start school 2 years late due to being so sick) and is a straight A student. 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.

OUR FAMILIES



Diana, Mom, Danin, Dad, Mikaela and Taylor, Sisters of Luke Manny



The Manny Family

We have always chose to live life to the fullest and try anything once! That's the Manny's mantra! From the day we took our Lukey home we have taken on that attitude. At first of course it was scary, a new world filled with new fears and new challenges like everyone one of us here have experienced. However, with Dad and Mom having fulltime careers and three children, two who were 16 months apart and a teenager, we adapted really quick. Being worried about germs and if Taylor grabbed a toy of Luke's without washing her hands quickly got thrown out the window.

Luke is the baby boy of the family. He is 7 going on 8, then there is Taylor our sassy little girl who is 8 going on 25, and our beautiful daughter Mikaela who is 20. We live in Rancho Cucamonga, California a southern California suburb that is about 40 miles east of Los Angeles. I work in Customer Operations Technology for a major natural gas utility company in California. Danin is a Maintenance Supervisor for a large plastic bottling manufacturer. Mikaela is in her third year of college at a local community college. She was studying to become a Physicians Assistance until she started her focus on Biotechnology.

She currently was accepted into an internship for a major biomedical company. She also owns her own health and wellness company called "Make Your Mark". Taylor is a sassy sister who adores her brother and her "bubby" is her best friend. She enjoys dance and musical theatre just like her mama. Often you find Taylor singing her heart out or bursting into song anywhere and everywhere. We have a cat who's name is Kujo, yes that's after the 1980's movie and a small Jack Russell Terrier/Corgie mix named Jax. Luke loves that dog more than his toys he's said.

Luke is 7 years old and is going into the 2nd grade. He attends a public school where he is in an orthopedically challenged classroom with other typical 2nd graders. He has been attending his school since he was 3 years old. He loves math and P.E. He is extremely smart for his age and attends a mainstream 3rd grade class for both Math and English. Luke loves playing video games on his ipad and is a Star Wars fanatic. That child knows more Star Wars facts than most adults. If you say the name of a character wrong, Luke is very quick to correct you. This year he played baseball for the first time in our local Challenger Little League. He was on his favorite team, "The Angels". In the fall he will be playing soccer. He loves playing soccer in his power chair in the street with his friends in our neighborhood. Luke loves going to his favorite store "Target" on the weekend, looking at all the new legos. We often go to Disneyland and he recently rode on his first rollercoaster, Radiator Springs in Cars Land. Although he said it was scary he was laughing the entire time. Although he gets scared at trying things that are foreign to him, we have shown Luke that he can adapt and try it. Many things like swimming he thought couldn't happen with his trach and vent, he now goes in the pool on a weekly basis and is my little fishy boy. We are so grateful to our members of this community who have gone before us, paving the way and showing us that LIFE IS POSSIBLE! It is my nature to research things and educate myself, when faced with something new or something challenging. When Luke was diagnosed with MTM we immediately took to the internet and jumped in with both feet. I have helped with the first MTM Conference and the one there after. We have been lucky to meet many families who we have bonded with over the years. I am lucky to call some of these mama's here my best friends. I attended the Parent Advisory Board for Audentes in November and was honored to share our experiences with them. We look forward to the up and coming possible treatments for our children. Life is GOOD always.

OUR FAMILIES



Robin, Mom, Daniel, Dad, and Will, Brother of
Ryan McDermott



The McDermott Family

Hi! We're the McDermotts: Daniel, Robin, three-year-old Will, and two-year-old Ryan. We live in Phoenix, AZ. We had no idea of the adventure that was in store for us when I was pregnant with Ryan. He got a g-tube and trach when he was 4 weeks old. After nearly two months in the NICU, we were allowed to take him home. Originally, we had been told that he would go home on hospice care. By the time he was discharged, that was thankfully no longer the case. He had two subsequent hospital stays after his initial discharge, and during the second visit when he was almost 5 months old, they tried putting him on a ventilator. This seemed to be the missing piece! He could breathe on his own, but he was expending every calorie he took in just to breathe. His growth took off, he was more awake and playful! He's been hospital free since October 2014. He was diagnosed with CNM, with a probably diagnosis of MTM just a week prior to his first birthday after a muscle biopsy. It took almost another year to get the official diagnosis of XLMTM through genetic testing. Ryan has defied the odds presented to us when he was born, and is stronger and happier than ever!

Ryan loves toys that make noise and light up, especially his drums and piano. He's learning about joysticks and how to activate his toys, thanks to Daniel who has done custom modifications to a lot of toys! He loves to dance and wiggle to music, play peek-a-boo, swimming, and playing with his family and friends. Will loves to show Ryan his toys, and how they work. He loves being a big brother and refuses to go to bed without giving Ryan a kiss each night. When they were younger, they would lie in Ryan's bed and watch movies together.

We are so thankful for this community, the support we've received and how much we've learned. With such a rare disease, more often than not we're teaching the doctors, and we wouldn't be able to do that without this group. We're so excited to meet everyone!

OUR FAMILIES



Darlene, Grandma, Jimmy, Grandpa, and Kayla, Sister of Kyle Miller

The Miller Family

Hi, we're the Miller family, Jimmy (husband), Darlene, Kayla (granddaughter) and Kyle (grandson). We are from Lexington Park, Maryland. We call Kyle our miracle baby. He spent the first 6 months of his life in a hospital. When he came home, we were told he would never be able to do anything and he more than likely wouldn't see his first birthday. Kyle is 13 years old now. As all of you know, our kids are amazing. He started proving the doctors wrong and continues to do so today.

We are a busy family: Jimmy is a Crane Operator and Welder and I'm a Special Education Para Educator. Kayla is starting high school in the fall and very busy with afterschool activities and sports. Kyle is going into the 7th grade. Kyle loves being outside. He enjoys family trips to the beach and camping. We love to crab and fish (Kyle's favorite). He enjoys boat rides and riding the 4 wheeler and his John Deere Gator. He loves being at the race track with his daddy. He loves to listen to music. He is all teenager. He just loves life. He is so excited to meet everyone, especially the other kids in wheelchairs.



OUR FAMILIES



Christi, Mom and AJ, Brother of
Jason Muñoz



The Muñoz Family

Jason Jr. was born bright and early on March 21, 1996 at 12:35am to Jason Sr. and Christi. This was the happiest day of our lives. This was also the scariest. Jason was not breathing at birth. Once Jason was stabilized, they brought him to us as he was ready to take the ride of his life--in a helicopter to Loyola Hospital at only 3 hours old.

The early years of Jason's life had ups and downs with hospitalizations for pneumonia and RSV. This was a scary time but we persevered through this because our son was a living example of strength with a smile on his face. WE COULDN'T GIVE UP! We were on this merry go round of life with MTM. We kept trying to figure all this out and how to take care of him the best way. Our families were such a great support through these uncharted waters. Despite all these ups and downs, Jason was a happy little guy, always with a smile on his face and a shine in his eyes that would make someone fall in love with him the minute they met.

Jason's early school years of life were great as well. His teachers wanted to help him learn to his maximum capacity. When Jason was about 2 ½ years old, he was playing a game on his computer and decided to close the game and go on the internet (he signed in via the AOL dial-up!) and started "playing" on the internet! Hence the reason I call him my little genius. He has shown his strength and determination in so many ways other than with his muscles.

When Jason reached middle school, he excelled at everything that was handed to him. He needed a license to drive in the hallways due to his crazy power wheelchair driving! He received the presidential award and Honor Roll all three years. High School years mirrored Middle School. Jason did have a bout of bad health in his sophomore year of High School, he was pretty much home schooled for the whole year. Jason is a strong kid; when sitting up in a chair, he used to love to put his head back and pull it back up.

OUR FAMILIES



**Bobby, Dad and AJ, Brother of
Jason Muñoz**

The Muñoz Family (continued)

But one time he wasn't able to pull it back up--his trach became cut off, he stopped breathing, and had a heart attack. This was the most heart wrenching time in our lives. Jason was in a coma for 2 weeks; the unknown of what might happen was unbearable. We played his favorite show (Total Drama Island) on his DVD player and talked to him like he was awake every day. One day he woke up and wanted a part of the show to be played back again! This was another happy moment in our lives. He had a lot of rehab to get back to be able to do the littlest of things. From this Jason lost movement in his left side of his arm. We continue to move it and exercise it for him.

In Jason's Senior Year of High School, Jason's Math teacher let him know that he had the highest midterm score in the history of his teaching (of ~20 years); he received a perfect score! Jason graduated high school and this was the greatest accomplishment ever for him. This was a kid that wasn't supposed to be here, only given a year tops by the doctors. He surely showed everyone that they were wrong!

Jason is currently attending a transition program. They are helping learn how to get around town by public transportation and teaching him job skills that hopefully one day will help him get a job!! He is not too excited but his momma is! (Just kidding) They also have been helping him look into different area colleges. They teach him that he can be in this world doing all the things that everyone else can do--HE CAN DO IT TOO! He LOVES Pokémon, playing his Wii, playing his computer and watching movies. He has been very healthy the last 3 years or so. We just love him to pieces. We wouldn't change anything about him! He has made us who we are today. He has taught our family how to be strong, how to never give up and how to love. We all love him for that!

OUR FAMILIES



**Guadalupe Najera, Mom, Graciano Arteaga, Dad,
Joselyne and Jasmin, Sisters of
Giovanni Najera-Arteaga**

The Najera-Arteaga Family

We are the Najera-Arteaga family. Giovanni is our son with MTM and he has two sisters, Joselyne (age 7) and Jasmin (age 3). The road with MTM has not been easy, especially since Giovanni is the first one in the family with this condition. We have had difficult moments. Faith in God is what gives us strength to fight and carry on!



**Evelyn Najera and Sergio Gutierrez, Parents of
Mateo Najera-Gutierrez**

The Najera-Gutierrez Family

We are a local family who lives near Chicago, Illinois. Mateo has MTM.

OUR FAMILIES



Patricia Ocampo and Sergio Vazquez, Parents of
Emiliano Vazquez

The Ocampo-Vazquez Family



Hola! We are the Ocampo-Vazquez family and reside in Chicago. We are the proud parents to Emiliano, better known as “Emi” - our clever three-year-old son. Emi graced us with his presence two and a half months early, and upon his arrival, he was unresponsive with severe, low muscle tone. The physicians immediately went to work and got Emi to take some breaths. We were hospitalized at Northshore Evanston Hospital for two months. After several tests and still no diagnosis, we asked to be transferred to Children’s Memorial Hospital. On February 27, 2012, we were transferred to Children’s Memorial Hospital (now Ann and Robert H. Lurie Children’s Hospital). Soon after additional testing began, one of which was a muscle biopsy.

In early March, our Emi was diagnosed with X-Linked Centronuclear Myotubular Myopathy. Having a name to the disease brought us an odd sense of relief, although the fear began all over again once we started doing our own research on his condition. Shortly after his diagnosis, Emi received his very own trach and g-tube; Sergio and I received intense training on all his medical needs and equipment. We finally brought Emi home in August, after being hospitalized for the first eight and a half months of his young life.

Emiliano is a very curious little boy. Books and alpha letters are his favorite thing. He loves story time and enjoys “fanning” the pages with his little thumb. He also enjoys rides, whether they be on a car, his little tricycle, or in your arms. “Movement” is a big deal for him - and us. He continues to reach his own milestones. He brings us joy on a daily basis and continues to show us about life and “living in the moment”. We love our Emi unconditionally and could not be anymore proud of him. We continue to be hopeful for the next steps on clinical trials which will lead to a cure for this disease in the very near future. God speed.

OUR FAMILIES



Jeanne, Mom, Luis, Dad, Dylan and Luciano, Brothers of Sebastian Orellan



The Orellana Family

Our family is Luis (dad) Jeanne (mom) Dylan (brother) Luciano (brother) and Sebastian. Luis and Jeanne are self-employed and own a business in San Francisco. Sebastian has always been a deeply curious, bright and empathetic soul. Now entering seventh grade, he uses an iPad with various apps to help him mitigate a concentrated load of middle school work. Our goal for high school and college is to get him as independent as possible so he can achieve his dreams and live a fulfilling life. We know this conference will be a chance to connect to other families and learn more than ever and are thrilled to be joining.

Sebastian was born 5 ½ weeks early. The pregnancy had been atypical with third trimester maternal bleeding. In utero, he wasn't a huge mover/ kicker and was born floppy, with low Apgar scores, respiratory concerns and poor suction- the hospital simply said he was "hypotonic". All his childhood milestones were very late. We have had a multitude of mis-diagnoses from Myasthenia Gravis to MTM1. Recently, Sabine of CMDIR directed us to a new blood test study from the University of Chicago that identified certain gene mutations causing CNM. Excited, we took the test and shockingly it came back with a clear diagnosis of CNM caused by a mutation of the Dynamin 2 gene (and not what we'd thus far believed about MTM1). So much development was happening around MTM1, but very little around DNM2. At present, we know no one with the DNM2 diagnoses, and are hoping to meet others to help us into setting up a foundation to gather the affected people and start research. It's been a long journey, with hospitalization, surgeries, sleep studies, corrective orthodonture, and more, but we are hopeful and engaged.

OUR FAMILIES



Kathryn and Michael, Parents of Tommy Perez

The Perez Family

East Providence, Rhode Island



The Raddatz Family

Homer City, Illinois



Melody, Mom, Cliff, Dad, Micheal, Brother, Brittany, Sister, Cliff and Patrick Raddatz

OUR FAMILIES



**Carol, Grandma of
Nicholas Rocheford**



**Jaime, Mom, Bryan, Dad, and Abigail, Sister of
Nicholas Rocheford**



**Jaime, Mom and
Fleur**

The Rocheford Family

We are the Rocheford's. I am Jamie; my husband is Bryan; we have two beautiful children. Abigail, who goes by Abby, is six and is in first grade starting this year. Our beautiful son Nicholas is our MTM warrior! He was born in March of 2012, and we lost him in December of the same year. We had nine amazing months with him and we are so thankful for each of those days and the people Nicholas has brought into our lives! We are so thankful to my mother Carol LaDuke! When we needed her the most she was right there learning how to take care of Nicholas, so that we could live at home with him and not need help from outside. We are so thankful for her gift of help that we ask her to come to every conference with us. Our lab, Fleur, is the newest recent addition to our family. She came from an MTM research facility! She is the best grief support for myself.

This is our second conference and I wouldn't miss this for the world!



OUR FAMILIES



Pam and Gary, Parents of
John Scoggin



The Scoggin Family

The Scoggin family is doing great. Gary has retired and gone right back to work. Pam spends all her free time helping out at Church and chasing our three dogs, Salty, Pepper and Pink. Pink is a refugee from the MTM dog colony and doing really well as the newest member of the family.

John is now 26. He's survived adolescence, high school and college. He's now a graduate of the University of Houston-Clear Lake with a bachelor's degree in Communications. He's worked a little bit as a database developer and analyst for a local charity. But he spends most of his time today playing video games and writing an online guide for the video game Pikmin 3. In late August, for the third year in a row, John, Pam and Gary will be heading to Seattle for PaxPrime, the world's largest video game convention.

Medically, John is doing well. He has had a wide variety of surgeries – scoliosis (twice), gall bladder removal, spleen removal and a host of eye, ear and oral procedures. But overall he's quite healthy, going strong and anxiously awaiting news of progress in the MTM1 gene therapy.

OUR FAMILIES



Hillary, Mom, Kristopher, Dad, Lennon and Isla, Sisters and Rush, Brother of Benjamin Sirmon



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 the last 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.

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. Our family will continue to work towards accessibility for people with disabilities in Benjamin's honor.

One month after Benjamin gained his angel wings, we found out we were expecting our fifth child. Our new little girl and the rest of our family will always have the most devoted and special guardian angel.

The Sirmons

OUR FAMILIES

Joe Slaby



Joe Slaby

Hi, my name is Joe Slaby and I'm from Terre Haute, Indiana. I am 24 years old and was diagnosed with CNM in the spring of 2001. My family includes my dad Jack, mom Carol, brother Jack, and my three sisters Rachel, Christy, and Carrie. I also have four nephews and three nieces. They have had a definite impact on my life and I'm lucky to have their support.

Over the years I've learned to adjust and adapt to the lifestyle based on my progression. Although I can't walk up stairs anymore and have trouble getting out of chairs, I am still able to walk but predominantly use a scooter in my day-to-day activities. I strive to maintain the strength I have through daily stretching, light exercise, and a somewhat healthy diet.

I graduated with an associate's degree in accounting a couple of years ago and last year decided to go back to school at Indiana State University, my ultimate goal is to get enough credit hours so I can take the Certified Public Accountants Exam and become a CPA. In addition, I work part-time at the YMCA.

Attending the past three MTM/CNM Family Conferences and meeting the families who have shared their experiences with me has been an important part of my life. My family and I are looking forward to seeing everyone again along with getting to know new people!



OUR FAMILIES



Makayla and Emily, Daughters of Jackie Smith



The Smith Family

Hi! My name is Jackie and I was born with RYR1 Centronuclear Myopathy. I was born in 1979, when there was very little published or known about this disease. I was diagnosed at the age of 3 by muscle biopsy. My parents raised me to be very independent. They always encouraged me to try things and have fun in life. I have an awesome little brother who cares for me so amazingly. I have 2 daughters, Makayla is 14 and Emily is 10.

I love to read and write. I also have been learning all I can about this disease, and am spreading awareness of genetic muscle diseases.

OUR FAMILIES

The Swed Family



**Julie and Phil, Parents of
Nathan Swed**

Hello! We - Julie, Phil, Nathan, and our pup Trooper - are the Swed family. We hail from a small town north of Austin, Texas called Hutto. Nathan is currently 2 1/2 years old and he is a happy, silly, stinky toddler. It wasn't until Nathan was born (just a half-hour shy of Valentine's Day) that we realized we were in for a very atypical parenthood journey. None of the neonatologists knew what to make of Nathan's symptoms. It wasn't until after a muscle biopsy that was completed at the recommendation of a NICU doc (after they'd run out of other ideas of what to do) that we knew what direction Nathan's condition would take. With the help of Nathan's equipment, nurses, therapists, doctors, and Nathan's grandparents (Granny - who is here with us! Hi Granny! - Gramps, Meemaw, and Papaw) 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. He recently started preschool and loves to drive his wheelchair around looking for trouble. Nathan's most favorite thing in the world is to play trains. "Train" was his first word in sign language and watching model trains go around the room helped him gain head control. He also loves the color yellow, cats, construction vehicles, and laying on the floor next to his big lazy puppy dog. 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!

The Walker Family

San Diego, California



**Ashley, Mom of
Alexander and Jayden Walker**

OUR FAMILIES

The Walton Family



Emma and Conrad Walton

Hello everyone! This is Conrad and I am Emma, the lucky gal who gets to be Conrad's mom. We live together with our Yorkie, Maggie, in rural Pittsfield, Pennsylvania. Conrad's daddy, Scott, sadly passed away on September 30, 2013. We carry him with us in our hearts wherever we go.

Conrad is two years old and loves to sing and watch videos on his iPad. He is such a happy boy and like many two year olds - he has a mind of his own! Conrad is working hard with the help of his speech therapist, occupational therapist and teacher to get stronger and learn how to communicate. He enjoys clapping, playing with his train, and having stories read to him. Conrad is fortunate to have his grandparents visit him often when they are home for the summer. He likes it when grandma plays pat-a-cake with him and when grandpa plays "soccer" and "football."

I am a licensed professional counselor and work full time supervising a school-based behavioral health program for children. I am so thankful for the great team of nurses we have to make it possible for me to work, rest, and keep my sanity. Conrad is my greatest joy, but in my spare time I enjoy adding to my collection of Coach bags and Pandora jewelry. Recently I fulfilled a dream that Scott and I had of riding a motorcycle together by purchasing a Can Am Spyder. I hope that someday soon we find a cure for MTM so that Conrad can ride with me!

Conrad and I are blessed to be part of a wonderful church family who have taken good care of us these last two years. Conrad loves it when the weather is nice and we are able to walk to church together. He enjoys the music and seeing so many special friends who love him and pray for him every day. We are so excited to be here with you at the Family Conference and finally getting to meet you in person!



Scott, Conrad's Dad



OUR FAMILIES



The Welter Family



*Amy, Mom, Matt, Dad, and Allie, Sister of
Andy Welter*

In the winter of 2000, we found out we were going to have another baby. We were so excited because we already had our beautiful daughter, Allie, 5 years prior. The pregnancy had been going great, at 18 weeks we had an ultrasound done and they said we had a very active baby boy. About 4 weeks later we had another ultrasound because I had not felt the baby move yet. At that ultrasound they said he was kicking and moving but just not hard enough for me to feel him. Looking back now at all the ultrasound photos, in all of them, Andy is facing the same way, like he never moved. The doctors kept telling me everything was fine; our first baby was just very active. We ended up having a C-section and Andy came out extremely floppy and wasn't able to breathe long on his own.

It was a long road in the NICU. The doctors ran supposedly every test under the sun and they kept coming back with nothing. Finally, when Andy was about 2 weeks old, they did a muscle biopsy but that also came back with nothing. In September, we asked the doctors to just please put in a trach so we could go home and they did. We had to wait until November 1st, 2001, before we were able to take our sweet baby home.

Early on, Andy was off the vent all day and on at night. Andy had another muscle biopsy done a year later and still nothing was diagnosed. They did a MRI of his brain and found out that it was slowly deteriorating and they also did a nerve biopsy, which pointed towards neuroaxonal dystrophy. We were told we had maybe one year left with Andy and that he would become a vegetable. A miracle happened 6 months later, one of the MRI's showed that the brain deterioration had stopped! Even though we had no diagnosis we were relieved to know we no longer needed to worry about his brain. Andy was 5 when we switched neurologists and checked Andy for MTM. Sure enough, the test came back positive. I was also tested and found out I was a carrier.

Andy will be starting 8th grade in the fall. Andy loves the Hulk. He likes to pretend that the Hulk is going to smash things. Andy also loves to play video games. His favorite game is Mario Kart, and he doesn't like to lose. Every day he says or does something that just makes us so happy he is part of our life. He is a very mischievous boy with a great personality. Andy touches the hearts of so many people and we have learned so much from him. We love him very much and are grateful for what an incredible child he is.

OUR FAMILIES



Melanie, Mom, Brian, Dad, and Juliet, Sister of William Whiston

The Whiston Family



Dan and Melanie Whiston are the proud parents of William and Juliet. William entered the world four years ago and began our family's journey with MTM. William's medical story is similar to most. He was born with the classic MTM symptoms. Shortly after his first birthday, William was trached and placed on a ventilator. He recently transitioned from the LTV ventilator to the Trilogy.

William enjoys reading. A couple of his favorite books are *The Day the Crayons Quit* and *The Cat in the Hat*. He recently completed his first year of pre-school. We are pleased to report his first year was a huge success. He enjoys school and will begin his second year of pre-school in the fall. William's big sister Juliet (6) is William's best friend. The two are currently hooked on playing Xbox race car games together. Juliet will be going into the 1st grade this year. She is an energetic and fun girl who enjoys soccer, swimming and reading. She is William's biggest fan.

We live outside of Chicago in the charming town of Geneva, Illinois. William is fortunate to have grandparents that are active in his life. Grandma and Grandpa Whiston live in Geneva and Grandma and Grandpa Overholt live in central Illinois. William's Uncle Patrick and Aunt Megan also live just outside of Chicago. Our immediate family and our extended family are excited to have the ability to attend portions of the conference.

OUR FAMILIES



Lindsay, Mom, Martin, Dad, and Emily, Sister of Louie Wilhelm



The Wilhelm Family

We are the Wilhelm family: Lindsay (Mom), Martin (Dad), Emily (Sister), and Louie. Our sweet Louie was born in Springfield, IL on June 17, 2011 with Myotubular Myopathy (MTM). Lindsay, Martin, Emily, and Louie attended the 2013 MTM-CNM Family Conference and enjoyed meeting so many other families. Sadly, Louie passed away last August 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. We know his light continues to shine in our lives and for all who knew him.

OUR FAMILIES



*Stacey, Mom, Charles, Dad, Keyara and Amaiya,
Sisters, Kishawn, Kadrian and Kamryn Brothers of
Kamarion Willis*



The Willis Family

The Willis family is from Bristol, IN! Kamarion, affectionately called “Kmoney,” is a fun, bright seven-year-old boy. He has two sisters, Keyara (6) and Amaiya (6), and three brothers Kishawn (14), Kadrian (4), and Kamryn (4).

Kmoney was born March 22, 2008, presenting at birth with hypotonia and breathing issues. Several tests were run, but they all came back negative, so on April 7 he was taken by helicopter to Riley Children’s Hospital in Indy (three and a half hours away). He had more tests--still no explanation for the hypotonia. He had his trach & g-tube surgery on April 30, and on May 15, he was finally sent home on trach collar with supplemental O2. December of 2008, at pulmonary checkup, one of the doctors was horrified at the way he was breathing, finally confirming our worries. Kmoney was admitted to trial with CPAP, which made his blood gases worse, and then he was put on a ventilator. He spent his first Christmas in the hospital, returned home shortly before New Year’s. Even with the support of the ventilator, it took 3-4 months for him to get down in the 40 bpm range, because he had gotten so used to breathing that rapidly! We spent 22 months on the vent, giving him constant PT at home to increase strength. In August 2010 we passed a sleep study and were able to come off the vent completely on October 15, 2010! In December 2010 a muscle biopsy was done; the results received February 2011 indicated XLMTM. Kmoney is a true blessing to our family and a jokester who keeps us smiling.

TRIBUTE TO OUR ANGELS

We honor on all those who have passed away from Myotubular Myopathy or Centronuclear Myopathy. The individuals shared here are beloved children 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 (In the Earhart Room), a special place to honor, reflect, and celebrate the lives of all our loved ones who have passed away.



Phillip Baron
01/04/90 to 06/16/15



Joshua Miles Frase
02/02/95 to 12/24/10



Adam Hoyle
1987 to 1988



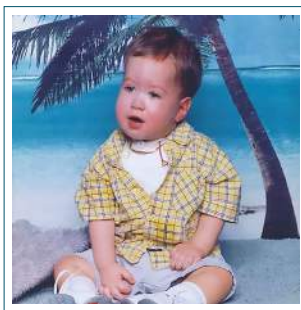
Lane Garret Jackman-Brassfield
10/16/01 to 09/09/10



Elliot Charles Koh
05/18/12 to 01/06/13



Patrick Connor Miller
10/2005 to 08/12/08



Austen O'Neil Scott
08/14/02 to 02/06/04



Cliffy Raddatz
12/27/91 to 01/29/01

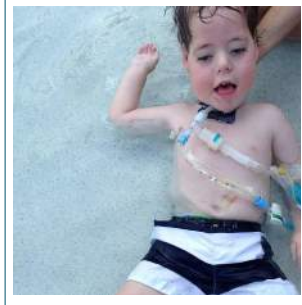


Nicholas Rocheford
03/18/12 to 12/17/12

TRIBUTE TO OUR ANGELS



Matteo Serafano
02/25/08 to 09/27/13



Benjamin Dean Sirmon
02/16/12 to 02/23/15



Alexander Edward Walker
05/11/12 to 07/11/13



Jayden Donnell Walker
05/11/12 to 07/11/13



Louie Wilhelm
06/17/11 to 08/05/14



ANGELS NOT PICTURED

- *Joshua Hoyle* 1989
- *Layton Daniel Deluna* 2014



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- **Genetic Testing**
- *CMDIR*
- **MTM Phone-based Event Study**
- *CMDIR & Jim Dowling*
- **Biomarker Study - Jim Dowling**

For more details, see our poster at the
MTM-CNM Family Conference

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conference theme!*

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*Thank you for
raising funds!*

*Becky Garland
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and family*



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MEMORIES AND MEMORIES IN THE MAKING



2009 MTM/CNM Family Conference Group Photo



MEMORIES AND MEMORIES IN THE MAKING



2011 MTM/CNM Family Conference Group Photo



MEMORIES AND MEMORIES IN THE MAKING

2011 MTM/CNM Family Conference Photos



MEMORIES AND MEMORIES IN THE MAKING

2011 MTM/CNM Family Conference Photos



MEMORIES AND MEMORIES IN THE MAKING



2013 MTM/CNM Family Conference Group Photo



MEMORIES AND MEMORIES IN THE MAKING

2013 MTM/CNM Family Conference Photos



MEMORIES AND MEMORIES IN THE MAKING

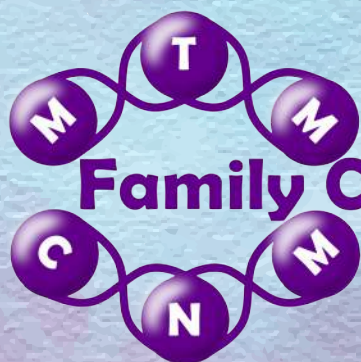
2013 MTM/CNM Family Conference Photos



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