

JULY 26-28, 2013
BEST WESTERN PLUS AT MALL OF AMERICA, BLOOMINGTON, MN





Welcome

On behalf of the Conference Planning Team, welcome to the 2013 MTM-CNM Family Conference! We are so glad you are here! This is a unique opportunity to gather together families and professionals who all share a deep connection to Myotubular/Centronuclear Myopathy. It is a time for sharing and reflecting, for learning and teaching, for celebrating and inspiring.

The theme of our 2013 Conference is “The Road to Research, Resources, and Relationships”. This theme is inspired by the journey we are all traveling together. It encompasses the core elements of our Conference mission: 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 provide a unique opportunity for families to interact and collaborate with lead researchers in our field.

With promising therapeutic 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 researchers, clinicians, and 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 rocky road a little smoother and bringing us all closer to our destination.

It is with much gratitude that we would like to thank our sponsors: 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 success. It has been a challenging but inspiring journey, a coming together of resources and gifts that have brought us here today and it is our hope that you will leave with a renewed commitment and resolve to journey forward together. We hope you all have the most wonderful weekend and leave with memories to last a lifetime!

Warmly,

Erin Ward
Conference Director

Family Conference Agenda

FRIDAY, JULY 26

- 2:00PM – 4:00PM **“THE STARTING LINE”** CONFERENCE REGISTRATION AND COMMUNITY CONNECTIONS - REGENCY BALLROOM
- 5:00PM – 9:00 PM **“BEGINNING OUR JOURNEY TOGETHER”** OPENING DINNER - REGENCY BALLROOM

SATURDAY, JULY 27

- CONTINENTAL BREAKFAST** - HOTEL BREAKFAST ROOM
- 8:00AM – 8:45AM **YOGA FOR CAREGIVERS** (Optional) with Krista Hanson - REGENCY WEST MEETING ROOM
- 9:00AM – 9:15AM **OPENING ADDRESS** - REGENCY EAST MEETING ROOM
- “WELCOME” - DR. ALAN BEGGS (VIA VIDEO PRESENTATION)**
- DIRECTOR OF THE MANTON CENTER FOR ORPHAN DISEASE RESEARCH & THE BEGGS LABORATORY, BOSTON CHILDREN'S HOSPITAL
- 9:15AM – 12:00PM **“THERAPEUTIC TREATMENTS ON THE HORIZON: OUR HOPEFUL DESTINATION”**
- REGENCY EAST MEETING ROOM
- DEVELOPMENT OF GENE THERAPY FOR MYOTUBULAR MYOPATHY**
- ANNA BUJ-BELLO, PHD, GENETHON, FRANCE
- MARTIN “CASEY” CHILDERS, DO, PHD, UNIVERSITY OF WASHINGTON
- “PIT STOP” - SNACK BREAK**
- DEVELOPMENT OF ENZYME REPLACEMENT THERAPY FOR MYOTUBULAR MYOPATHY**
- MIKE LAWLOR, MD, PHD, MEDICAL COLLEGE OF WISCONSIN
- DEBORAH RAMSDELL, CEO AND HAL LANDY, MD, CMO, VALERION THERAPEUTICS
- 12:00PM – 1:00PM **“REFUEL”** Pickup Box Lunches
- 1:00PM – 3:00PM **AFTERNOON OPTIONS:**
- “NAVIGATING UNCHARTED TERRITORY”** Scientific & Medical Forum for Families and Professionals - REGENCY EAST MEETING ROOM
- “THE DAILY COMMUTE”** Family Exchange of ideas to overcome roadblocks, share our journeys, and tips for tuning up our daily routines - Breakout Suite TBD
- “MEMORY LANE”** Pay tribute to the memory of our Angels in the Reflection Room

SATURDAY, JULY 27 (CONTINUED)

“ALL ABOARD!” Enroll and participate in research studies with the various teams that are present

“SIDE TRIP” Outing to Mall of America, Nickelodeon Universe, Sea Life Aquarium

“STAYCATION” Leisure time at the hotel pool and water playland, and/or kids activity room

3:00PM-4:00PM **GROUP DISCUSSION** with Genetic Counselor Lindsay Swanson - Suite TBA
An opportunity to share and discuss questions about genetic, carrier-related, and family planning issues

4:45PM – **GROUP PHOTO** (wear your conference t-shirts!)

5:00PM – 9:00PM **“THIS IS HOW WE ROLL”** EVENING PROGRAM - REGENCY BALLROOM

A NEW DEVELOPMENT

- MATTHEW PATTERSON, CEO, AUDENTES THERAPEUTICS

BUFFET DINNER

VARIETY SHOW celebrating our gifts and talents and all that makes our community special

SUNDAY, JULY 28

CONTINENTAL BREAKFAST - HOTEL BREAKFAST ROOM

8:00AM – 8:45AM (Optional) **NONDENOMINATIONAL PRAYER AND REFLECTION SERVICE**
- REGENCY EAST MEETING ROOM

9:00AM – 12:00PM **“PATHWAYS TO CLINICAL TRIALS: OUR PREPARATION AND PARTICIPATION”** - REGENCY EAST MEETING ROOM

NATURAL HISTORY STUDY AND BIOMARKERS IN MTM/CNM

- JAMES DOWLING, MD, PHD, UNIVERSITY OF MICHIGAN

- LAURENT SERVAIS, MD, PHD, INSITUT OF MYOLOGY

RESPIRATORY MEASURES AND LESSONS FROM POMPE DISEASE

- BARRY BYRNE, MD, PHD, UNIVERSITY OF FLORIDA

- BARBARA SMITH, PT, PHD, UNIVERSITY OF FLORIDA

“PIT STOP” - SNACK BREAK

Family Conference Agenda

SUNDAY, JULY 28 (CONTINUED)

VIDEO PRESENTATION: MTM/CNM PATIENT REGISTRY

- ANNE LENNOX, MYOTUBULAR TRUST

GLOBAL MAPPING PROJECT

- ALISON FRASE, JOSHUA FRASE FOUNDATION

CONGENITAL MUSCLE DISEASE INTERNATIONAL REGISTRY STUDIES

- SABINE DE CHASTONAY, PHD, CMDIR

VIDEO PRESENTATION: CONGENITAL MUSCLE DISEASE BIOBANK

- TARA SCHMIDLEN, CORIELL INSTITUTE

CONGENITAL MUSCLE DISEASE TISSUE REPOSITORY

- MIKE LAWLOR, MD, PHD, MEDICAL COLLEGE OF WISCONSIN

12:00PM – 2:00PM

“ON THE ROAD AGAIN: CONTINUING OUR JOURNEY TOGETHER”

Buffet Lunch and Closing Program - REGENCY BALLROOM

KID'S ACTIVITIES - SATURDAY, JULY 27

9:00AM – 12:00PM

KID'S ACTIVITIES available will include Arts and Crafts, Wacky Photo Booth, Video Games, and Movies - REGENCY WEST MEETING ROOM

9:30AM – 11:30AM

“HAPPY FACES” Balloon Artist and Face Painting

“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.

1:00PM – 4:45PM

LEISURE TIME WITH FAMILIES Join a group outing to the Mall of America aquarium or Nickelodeon Universe, enjoy your own aquatic experience poolside with the family, or join us in the Kids room for “Finding Nemo” with sea-life inspired activities available.

KID'S ACTIVITIES - SUNDAY, JULY 28TH

9:00AM – 12:00PM

ARTS & CRAFTS Wacky Photo Booth, Movies, and video games

“FROM ME TO YOU” Kids will make a special keepsake for their family as a 2013 conference memory

“SOCK PUPPETS” Kids bring an ordinary tube sock to life by creating a monster, a monkey, an alien or whatever their imagination designs

12:00PM – 2:00PM

JOIN FAMILIES FOR CLOSING LUNCH - REGENCY EAST BALLROOM

OLDER TEENS AND YOUNG ADULTS:

As at past conferences, our older teens and young adults enjoy gathering together in a supportive community environment. Interested teens and young adults may wish to connect with Stesha Mashinchi, our teen & young adult group leader, upon arrival to discuss possible plans for the weekend including poolside gatherings and potential offsite visits to the Mall of America.

THROUGHOUT THE WEEKEND...

- Participate in the Natural History Study with the team from University of Michigan*
- Participate in the Observational Study of Respiratory Strength and Function with the team from University of Florida*
- Enroll in CMDIR studies, including the MTM Event Study with Dr. Sabine de Chastonay
- Meet with Genetic Counselor Lindsay Swanson, Beggs Lab Children’s Hospital Boston
- Visit the Reflection Room and pay tribute to the memory of our Angels
- Take Family Portraits with SergioV (www.sergiovphotography.com)
- Participate in self-care activities available throughout the weekend including yoga and massage
- Rest & Relax Poolside

*Please pre-arrange a time with the respective research team.



ALTHOUGH THERE WILL BE VOLUNTEERS HELPING IN THE KID’S ACTIVITY ROOM, NURSING/RESPITE CARE IS NOT AVAILABLE FOR THE KID’S ACTIVITIES, CAREGIVERS SHOULD ACCOMPANY CHILDREN AS NEEDED.

Meet the Research Teams...

RESEARCHERS



Anna Buj Bello, PhD
Genethon, Paris, FRANCE

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Having done a post-doctorate at the IGBMC, Illkirch, France, where **ANNA BUJ BELLO** 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. How wonderful to have such excellent international collaborators participating in our conference!



Martin "Casey" Childers, DO, PhD
University of Washington

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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." We look forward to the opportunity to meet with Dr. Childers again and hear about the latest and greatest in gene replacement therapy and the hope of treating those with Myotubular Myopathy!



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 performed the pathological analyses for a number of preclinical trial studies for animal models of X-linked myotubular myopathy that are currently being performed worldwide, including anti-myostatin therapy, gene therapy, and protein replacement therapy. He has also recently begun evaluating myostatin inhibition in murine models of nemaline myopathy.

In the spring of 2013, Dr. Lawlor's laboratory became the site of the Congenital Muscle Disease Tissue Repository, which is meant to provide a central place for the donation and distribution of patient tissues. It is our hope that such a central resource for tissue storage and distribution will improve the pace of research in our field.



Laurent Servais, MD, PhD
Institut of Myology, Paris, France

Collaborating with the Dowling Lab Team is **LAURENT SERVAIS**, MD, PhD is a child neurologist working in the Institut of Myology, in Paris, France. He is the head of the Clinical Trials and Databases service. Together with Genethon (France), Valerion (US) and Dr. Jim Dowling (University of Michigan), he is preparing a natural history study using innovative methods to longitudinally measure strength and function of patients with XMTM in Europe and in US.

Meet the Research Teams...

RESEARCHERS



Lindsay Swanson, MS, CGC
Research Genetic Counselor
Beggs Lab, Boston Children's Hospital

LINDSAY SWANSON is a genetic counselor specializing in the genetics of congenital myopathies, including centronuclear and myotubular myopathy (CNM/MTM). She received her Master's degree in Genetic Counseling from the University of Minnesota in April 2012 and began with the Beggs Congenital Myopathy Research Program at Boston Children's Hospital in July 2012. During the past year, she has worked with many families with congenital myopathies from around the world and has greatly enjoyed getting to know members of the CNM/MTM community. Lindsay coordinates the recruitment and enrollment of families into the research program and develops and conducts clinical studies to identify symptoms and genes associated with the congenital myopathies.



Sabine de Chastonay, BA, PhD
CMDIR

SABINE DE CHASTONAY received a bachelor's 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 US with her family since 1992 and raised two children as a stay-at-home mom involved in various community organizations. From 2010 to 2012, she worked as a Patient Contact Coordinator with Patientstar LLC, a company specialized in patient recruiting and site selection surveys for clinical trials. In April 2012, she joined the Congenital Muscle Disease International Registry (CMDIR) as a volunteer Research Associate with a focus on Myotubular and Centronuclear Myopathy. Sabine will lead the MTM Genetic Testing Study, sponsored by Valerion Therapeutics, and is the research coordinator for the recently launched MTM Event Study. Her goal is to work with existing organizations and the scientific community to support getting the MTM/CNM community into clinical trials.

Meet the Research Teams...

UNIVERSITY OF *Michigan* TEAM

Returning to the conference this year is the team of researchers from the University of Michigan! The team is led by Dr. James Dowling. His research focuses on better understanding congenital myopathies, in particular Myotubular and Centronuclear myopathies, as well as finding new treatments for these conditions.

The team will be inviting patients and families diagnosed with MTM/CNM to continue or begin participation in the Natural History study. The purpose of the study is to better understand how the symptoms of MTM/CNM change over time and find targets for new treatments. The team is recruiting living patients as well as those who have passed away. Participation includes filling out a survey, drawing out the family tree, and collecting medical records. This year, the team is adding two new components to the study. The first component will be performed in collaboration with Dr. Laurent Servais. Dr. Servais will be using new tools to test muscle strength in patients with MTM. The second component is the collection of blood for biomarkers in patients with MTM. Biomarkers are markers in the blood that help researchers track disease progression and test the effectiveness of a medication. These new components are not required for participation in the study, but highly encouraged.

If you are interested in learning more about the Natural History study, please contact Kim Amburgey by email kamburg@med.umich.edu or by phone (734) 647-9224. You can also check out their website at http://sitemaker.umich.edu/dowling.lab/cnm_mtm_rm_natural_history_study. For those who have already enrolled in the study, you will be contacted directly by the team for instructions for continued participation.



James Dowling, MD, PhD

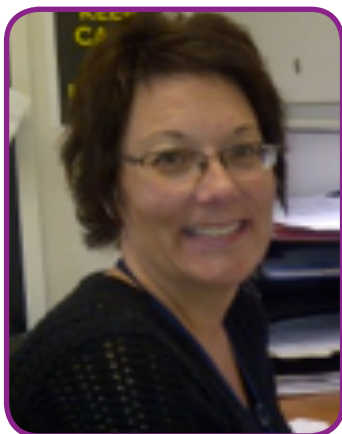
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DR. JIM DOWLING is a pediatric neuromuscular specialist and researcher at the University of Michigan. His laboratory is dedicated to learning more about CNM/MTM. The Dowling laboratory uses zebrafish and mouse models of CNM to better understand how and why these diseases occur, identify new genetic causes and develop new therapies. One potential therapy to come out of his lab is a medication called Mestinon. Using CNM animal models, he identified problems with the communication between the nerve and muscle; Mestinon helps to improve this communication which in turn may lessen fatigue and possibly improve muscle movement in patients. In addition to research, Dr. Dowling is also the co-director of the Pediatric Neuromuscular Clinic, a Muscular Dystrophy Association (MDA) sponsored clinic. In this clinic, patients with a variety of nerve and muscle conditions including muscular dystrophies, congenital myopathies, and inherited and acquired neuropathies are evaluated and treated.



Kimberly Amburgey, MS, CGC

KIMBERLY AMBURGEY is a genetic counselor in the Pediatric Neuromuscular Clinic at the University of Michigan. In the clinic, she arranges genetic testing for patients with congenital myopathies, helps them navigate the diagnostic odyssey, and helps patients and families understand the implications of positive test results. Her clinical research focuses on congenital myopathies, including CNM/MTM. She has been involved in studying the prevalence of congenital myopathies in southeastern Michigan, the first prevalence study done in the United States. At the CNM/MTM family conference, Kimberly will be recruiting for the natural history study. This study will track the progression of disease over time and help to identify potential targets for future clinical trials.



Kimberly Creamer

KIMBERLY CREAMER is the clinic coordinator for the Pediatric Neuromuscular Clinic at the University of Michigan. Her role in the clinic is to coordinate care and act as the main point of contact for patients and their families. In addition to her clinical roles, she is also involved in several clinical research projects. Similar to the last CNM/MTM family conference, she will help enroll participants in the Natural History study

Meet the Research Teams...

UNIVERSITY OF *Florida* TEAM

We are very excited to have a team of researchers from the University of Florida joining us this year! The University of Florida team is led by Dr. Barry Byrne, MD, PhD, and the team studies the potential of gene and cell therapy to enhance muscle regeneration for individuals with inherited neuromuscular diseases. At the conference, the U of FL Team will have the opportunity to connect with interested patients and families who may qualify for their “Observational Study of Respiratory Strength and Function in X-Linked Myotubular Myopathy.” The purpose of the study is to gain a greater understanding of the year to year changes in individuals with Myotubular Myopathy.



Barry Byrne, MD, PhD

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DR. BYRNE'S laboratory and research is focused on understanding the pathophysiology of inherited cardio-skeletal myopathies and therapeutic strategies using gene therapy. Specifically, his lab has developed novel gene therapies for neuromuscular diseases such as Pompe disease. They have used AAV vectors to achieve sustained correction of the gene deficiency in Pompe disease models, leading to restoration of contractile and metabolic function of striated muscle and motor nerves. Their approach is now being evaluated in a human clinical trial investigating the safety and efficacy of direct gene delivery to the diaphragm. 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 X-linked MTM.



Lee Ann Lawson, BSN, MSN

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LEE ANN LAWSON'S career focus has been to improve the care of children with severe pediatric diseases. During the past 5 years, she developed extensive experience coordinating large trials with interdisciplinary teams and facilitated the IRB and IND approval process for a current clinical trial of diaphragm gene therapy for children with Pompe disease. Ms. Lawson has collaborated with Dr. Byrne and Dr. Smith on previous studies involving respiratory muscle testing and home visitation in ventilator-dependent children with Pompe disease.



Barbara Smith, PT, PhD

Those of you who attended in 2011 will remember **DR. BARBARA SMITH**, PT, PhD, who served as a consultant to Dr. Childers with regard to pulmonary testing in ventilated patients. She has continued studying the effects of respiratory motor training, in order to facilitate recovery for patients with neuromuscular disease and impaired breathing. Her work includes respiratory muscle testing in ventilator-dependent children with Pompe disease as part of a clinical trial of intramuscular gene therapy to the diaphragm. Dr. Smith has additional experience with XLMTM, nemaline myopathy, and Duchenne muscular dystrophy, all of which is most valuable for our community.

With 18 years of clinical practice and 5 years of full-time research experience, her specialty in respiratory muscle assessment will be an asset for collecting and interpreting the preclinical respiratory data and preparing translational clinical trial outcomes that are feasible for patients with neuromuscular diseases.

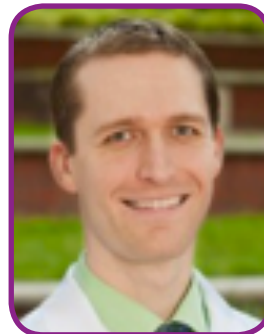
ADDITIONAL TEAM MEMBERS FROM THE *University of Florida* ASSISTING THROUGHOUT THE WEEKEND INCLUDE:



Terry Sexton, ARNP
Clinical Program
Coordinator



Danny Martin, PhD
Professor of Physical
Therapy



Markus Renno, MD
Pediatric Fellow



Manuela Corti, PhD, PT
Postdoctoral Research
Assistant.

Corporate & Community Collaborators

AUDENTES *Therapeutics*



Matt Patterson
President & CEO Audentes Therapeutics

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MATT PATTERSON is the President and CEO of Audentes Therapeutics, a new biotechnology company based in San Francisco, CA. Audentes' goal is to develop and gain approval for new medicines for rare muscle diseases using gene therapy technology. Matt has nearly 20 years of experience working for biotechnology companies focused on the research, development, and commercialization of medicines to treat rare diseases.

VALERION *Therapeutics*



Deborah Ramsdell
President & CEO Valerion Therapeutics

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DEB RAMSDELL has 25+ years as a drug development executive, primarily in therapeutic biologics. For the past 5 years, has worked on orphan diseases at Lotus Tissue Repair and Enobia Pharma. Currently, Deb is President/CEO of Valerion Therapeutics, an early stage biotech company with a platform delivery technology with a focus on rare genetic muscle diseases.

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VALERION *Therapeutics* (CONTINUED)



Hal Landy
MD, Chief Medical Officer,
Valerion Therapeutics

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DR. HAL LANDY is a board certified Pediatric Endocrinologist with 25+ years of clinical research experience in academics and industry and over 20 publications, primarily in rare orphan diseases. He previously held VP positions at Serone, Genzyme & Repliden with numerous successful drug approvals in growth, wasting and lysosomal disorders in the US, EU, Canada and Japan. Most recently the CMO and VP, Medical Affairs Enobia Pharma prior to the acquisition by Alexion Pharma and Medical Consultant to Lotus Tissue Repair.



Kristen Cunningham, Regulator Affairs
Consultant, Valerion

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KRISTEN CUNNINGHAM has worked in the biotech/ pharmaceutical industry for 20 years in Drug Development, mainly in the areas of Clinical and Regulatory Affairs. Prior to joining Valerion Therapeutics, Kristen worked for Enobia Pharma in Cambridge, MA (acquired by Alexion Pharmaceuticals). At Enobia, she had the opportunity to work on an Orphan Disease Clinical Program treating patients with Hypophosphatasia (HPP). Prior to Enobia, Kristen spent 5 years at Cubist Pharmaceuticals as a clinical consultant working on anti-infective studies. Kristen joined the Valerion Team in July 2012.



Corporate & Community Collaborators

PHILIPS *Respironics*

A global leader in sleep and respiratory therapy equipment, Philips Respironics provides meaningful innovation for individuals with neuromuscular disease, such as the Trilogy ventilator, CPAP and BiPAP machines, CoughAssist, and nebulizers. A local respiratory specialist from Philips will be available during a portion of the conference to provide information and demonstrate and answer questions about their equipment.

PASSY - *Muir*

Invented by a patient named David Muir, the Passy-Muir® Tracheostomy & Ventilator Swallowing and Speaking Valve is a simple medical device used by tracheostomy and ventilator patients. When placed on the hub of the tracheostomy tube or in-line with the ventilator circuit, the Passy-Muir Valve redirects air flow through the vocal folds, mouth and nose enabling voice and improved communication. See their information at our resource table, and enter the raffle to win a Toby Tracheasaurus plush toy!

AFM *Productions*

The French Muscular Dystrophy Association (AFM) is composed of patients and their families who are affected by neuromuscular diseases. There are about 200 neuromuscular diseases against which the AFM fights. Through their non-profit laboratory Genethon, the AFM has provided significant support for the development of gene therapy vectors for myotubular myopathy, and they are working on a documentary following Dr. Anna Buj Bello's work in this field.

MTM/CNM PATIENT *Registry*

The Myotubular and Centronuclear Myopathy Patient Registry is an international database specific to our condition. It is managed from the UK and operated by Myotubular Trust. The registry has been developed in partnership with TREAT-NMD and with a number of leading neuromuscular researchers. Anne Lennox of the Myotubular Trust will be sharing an overview via a video presentation.

CMDIR

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 *Biobank*



The Congenital Muscle Disease Biobank was established in August 2010 to facilitate research on the diagnosis, treatment and prevention of congenital muscle diseases. Many of the samples in the CMD biobank have come directly from patients and families who learned about the biobank through participation in Cure CMD, the Congenital Muscle Disease International Registry, A Foundation for Building Strength or the Joshua Frase Foundation. Tara Schmidlen from the Coriell Institute, which houses the Biobank, will be sharing an overview via a video presentation.

CMD TISSUE *Repository*



Dr. Mike Lawlor has recently 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. 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.

JOSHUA FRASE *Foundation*



Created by Alison and Paul Frase in honor of their son Joshua, for over sixteen 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. Please take a moment to introduce yourself to Paul and Alison during the conference.

WHERE THERE'S A WILL *There's A Cure*



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. Say hello to Melanie, Dan, Juliet and of course Will at the conference.

Meet the Conference Planning Team...

THE *Ward* FAMILY



Erin Ward, Conference Director **Mark Ward**, Planning Team Member,
Will Ward ~ Methuen, MASSACHUSETTS

Our son William 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, William came home with a tracheostomy and G-tube on July 3rd, which we will forever celebrate as “William’s Independence Day”! William has thrived at home and faces each day with incredible resilience and his strong will. William uses assistive technology to communicate and he is also a power- wheelchair user.

At the beginning for us in 2001, 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 William’s bedside and shared vital information and resources on this rare disease.

An amazing twelve years later, Will is now entering seventh grade and has been fully integrated in our neighborhood school since preschool. He loves his friends, the Boston Red Sox, and he plays on a Challenger baseball team. Will enjoys music, plays an electronic guitar and adaptive bongos. He also completed Cub Scouts, likes to bowl, and made his first communion. 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 William just as he is, 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. We feel extremely blessed that we’ve had this opportunity to plan this conference for our community and hope that everyone has an amazing experience this weekend!

THE *Wood* FAMILY



Marie Wood (Mom), *Planning Team Member* **David Wood** (Dad),
Lucian (Brother), **Blaise** (Brother), **Reid** and **Paley Wood**

G.K. Chesterton once wrote that every person “is a great might-not-have-been” and in that spirit, we give thanks for every day with Reid and Paley as a day that might-not-have-been. Reid and Paley have x-linked MTM and they utilize trachs, ventilators, and g-tubes for their basic life needs. Although Reid and Paley are not as physically strong as their peers, the people in their lives recognize the incredible strength of their spirits. Reid is simply “joy” personified, who captivates hearts with his 50 million gigawatt smile, silly pranks, and merry, infectious laughter! He is deeply creative and loves music, painting, and drawing. Our little Paley is sweetness itself, endearing himself to all with his patience, tenderness, curiosity, and happily humming ways. His favorite activities are singing and story time!

We’ve had a few close calls, but Reid and Paley continuously refresh and inspire us in the midst of difficulties. Lucian and Blaise--marvelous, hilarious characters in their own rights--adore their little brothers and love to make them smile! As parents in a situation both precarious and wonderful, David and I have had to surrender to the knowledge that ultimately these lives are not in our hands, yet we are determined to do everything we can to actualize our sons’ greatest potentials, praying and trusting in God as we do.

We also are profoundly aware of the need for support in the journey with MTM and CNM. After attending the first family conference in 2009, we were thrilled to meet so many wonderful people and to form ties of friendship that have helped sustain us through uncertain times. I (Marie) found the conference mission close to my heart’s desire to serve the community. One of the ways I have had the privilege to do so is as a member of the planning team since 2009.

Meet the Conference Planning Team...

THE *Mashinchi* FAMILY



Shannon Mashinchi (Mom), Conference Planning Team
David Mashinchi (Dad), Adam (Brother), Stesha (Sister),
Simon (Brother) and Javad

Javad Mashinchi came into this world by C-section on November 1, 2001, quiet as a lamb. Right away I noticed that there was no sound, no crying, nothing. In spite of being told that everything was ok, I knew in my heart that it wasn't. The nurses whisked Javad off to the NICU. The neonatologists had a variety of concerns for our son; generalized hypotonia, enlarged heart, respiratory issues. They ran every genetic test they could think of: Praeder-Willi, Myotonic Dystrophy, etc. These tests came out negative, but eventually at one month old, Javad 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.

When Javad was about 21 months old, our neurologist suspected that Javad had been misdiagnosed and suggested a muscle biopsy. The biopsy provided Javad with the new diagnosis of Centronuclear/Myotubular Myopathy. I was stunned to learn that Javad had a disease so incredibly rare. I searched for information and found very little that was recent. I felt so alone. I cried and wished that there was a way to get connected with others, wondering if there were even others?

Through a chance meeting, I met another family and then an internet group. Suddenly, I had community! These (mostly) women, whom I had never met in person, became closer to me than people that I saw every day. They knew how I felt, knew my daily struggles with caring for a medically-fragile child. They gave me encouragement, advice, and listened to my worries. One day, another MTM mom and I conceived of the idea of a family conference. This dream first came to fruition in Houston, TX in 2009 and I am honored to be a part of its continuation.

Javad is now 11 years old and attends fifth grade at our local elementary school. He is deeply loved by his brothers, Adam and Simon, and by his sister, Stesha, who assists the conference planning team with organizing activities for young adults and teens at the conference. Javad has grown and thrived, partly because of our incredible love and passion to give him the best life possible, but also due to the support of other incredible families that I have met who have children with MTM. The bonds of our community are priceless.

THE *Sanders* FAMILY



Chrissie Sanders (Mom), *Past Conference Director 2011*,
John Sanders (Dad), **Chelsea**(sister), **Jace** (brother),
Joey (5/29/07- 2/28/09), **Chloe**(sister) and **Jacob**(brother)

Our beautiful boy, Joseph, was born on May 29, 2007, in Las Vegas, Nevada, weighing 7 pounds 4 ounces and measuring 21 inches long. We knew something was wrong when he didn't cry, move, or open his eyes.

Joseph was whisked off to the NICU where the nurses began to work on him. We were told he was suffering from "wimpy white boy" syndrome and would be in our room in a few hours. Those few hours grew to a few days.

Eventually we were able to take our precious angel out of the hospital and begin our life together at home. Joseph began occupational and physical therapy at eight weeks of age. He began to thrive and grow, although he had many struggles and underwent 14 hospitalizations in his 20 months of life.

At one year old, a muscle biopsy revealed that Joseph had Myotubular Myopathy (later testing showed that it was x-linked and probably a sporadic mutation). Life became so sweet for us around that time as Joseph became stronger. He could control his head, roll over, and demonstrated increasing control of his arms and legs. Joseph was a quick study in sign language! He mastered, "eat," "more," "bye-bye," "please," "thank-you," "drink," and "monkey."

In early February 2009, Joseph was hospitalized with RSV and pneumonia. Our champ fought it off in a few days, but then in February 26, 2009, Joseph became sick again and had to be hospitalized. In less than 48 hours of admission, Joseph suffered from a massive hepatic hemorrhage, later discovered to have been caused by a rare liver disease called Peliosis Hepatitis. Doctors explained that nothing more could be done. John and I stayed beside Joseph, kissing his forehead as he ran into the arms of Jesus.

In June of 2010, John and I welcomed boy/girl twins, Jacob and Chloe. Their older brother Jace will be 13 yrs old this year. As our family continues on, not a day goes by that we don't talk or think about Joey. John and I have been active in the community because we believe in the support of friends going through this same experience.

Conference Volunteers

THE *Welter* FAMILY



Amy Welter (Mom), **Matt Welter** (Dad), **Allie** (Sister), & **Andy**
~ Farmington, MINNESOTA

A special thanks to the Welter family, our hometown family, for helping the conference planning team with local logistics, kids activity room staff, and their ongoing support of the conference in both 2011 and this year. Thank you!

*I*n 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 your 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.

...CONTINUED

Andy will be starting 6th grade in the fall. Andy loves to play outside, play on his IPAD and play baseball. 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.

THE *Majeau* FAMILY



Debbie (Mom), **Joël** (Dad), & **Ashtyn** (daughter)
~ St. Georges, Manitoba, CANADA

We'd like to thank Debbie for her willingness to support the conference in anyway needed and the time she has given to help coordinate the kids' activities for the conference. Thank you!

We live in St. Georges, Manitoba, Canada...a small town about an hour and a half from the capital of Manitoba which is Winnipeg (pretty much right smack dab in the middle of the country). Joël owns his own carpentry business and Debbie works for a family and community resource center in addition to a couple other jobs. Our daughter Ashtyn was born on March 17, 2012 and is the light of our life.

We became a part of the MTM-CNM community when our nephew was born in 2009 with XLMTM. He was such an important part of our life and we were so embraced by the MTM-CNM families that they have all become an extension of ours. We couldn't imagine life without all of our "family" members and are honored to be a part of your lives.

Conference Volunteers

THE *Hanson-Stansbury* FAMILY



**Krista Hanson (Mom), Burke Stansbury (Dad)
& Lucas Hanson**
~ Seattle, WASHINGTON



We are so excited to have Krista volunteer this year to offer a session of "Yoga for Caregivers" on Saturday morning. We'd also like to thank Krista and Burke for helping to facilitate our parent exchange of helpful resources and daily living ideas on Saturday afternoon. Thank you!

*L*ucas just finished his first year of preschool, and he will turn four this August. Lucas is a huge animal lover, and he especially loves dogs, birds, dinosaurs, and sea creatures. He has also loved music since he was a baby -- he goes to a weekly "music together" class, his favorite part of school is singing at circle time, and he loves dancing to rock music with his Daddy. And Lucas's new passion this spring is baseball. He's already been to two Mariners games.

Lucas has x-linked MTM, so his entry into the world was marked by the intense challenges that many MTM families go through. He was born 2 months early, he was immediately intubated, and we spent the next three months in the NICU in Washington DC. Though we didn't get a diagnosis for six months, it was clear from the beginning that Lucas's life would include significant health challenges. So of course now, nearly four years later, we are incredibly proud of everything that Lucas can do. And we're thrilled when we look back and see how far we've come as a family, and how mobile we can be with all this equipment!

Last year we moved to the Pacific Northwest, to be closer to our families and to the incredible lakes, trees, and mountains in our corner of the country. Krista and Burke are both involved in progressive politics, and because of our experience as Lucas's parents we are learning about the history of the disability rights movement, including many amazing parents who came before us. We benefit from many of their victories, including Early Intervention, the right to access to school for all children, curb cuts, ramps, and elevators at baseball stadiums. Burke works for a national grassroots organization that works to strengthen programs like Social Security and Medicare, and Krista teaches yoga.

... THE *Vazquez-Ocampo* FAMILY ...



Patricia Ocampo (Mom), **Sergio Vazquez** (Dad)
& **Emiliano Vazquez**
~ Chicago, ILLINOIS

We are very grateful to have both Patricia and Sergio sharing their gifts and talents with us this year. Thank you Patricia for helping design this year's conference program, and thank you Sergio for utilizing your photography skills as our conference photographer this year. We appreciate you both and your willingness to share your passions with the conference!

When Sergio and I found out we were expecting, like all families, we were ecstatic. The thought of having a baby was a bit scary but put an automatic goofy smile on our face. On December of 2011, our son Emiliano was born. He thought it would be a good idea to grace us with his presence 2 1/2 months early, upon his arrival he was unresponsive and very floppy; the docs immediately went to work and got Emiliano to take some breaths. We were hospitalized at Northshore Hospital for 2 months. After many, many tests and still no diagnosis, we asked to be transferred to Children's Memorial Hospital.

On February 27th we were transferred to Children's Memorial Hospital, now 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. Knowing what Emiliano was fighting brought us an odd sense of relief, although the fear began all over again once we began our research on his condition.

Shortly after Emi received his very own trach and g-tube, and Sergio and I received intense training on all his medical needs, supply and equipment.

We finally brought Emi home on August 2011, 8 1/2 months later. Emi is now a year and seven months young, he continues to strive and reach his own milestones, he is a very curious little boy. He loves musical instruments - especially the drums, he also loves to read or should I say "flip the pages" on books and is a big wiggle worm. He brings us joy on a daily basis and has shown us so much about life, he taught us to be patient, strong, and to believe in miracles, he also taught us to live and enjoy life at the moment.

We are super proud of Emiliano and love him unconditionally, and we are hopeful for progress to be made in finding a cure very, very soon. God speed.

Attendees

THE *Bauer* FAMILY

*H*i, we are the Bauer Family! Alex is 19 and lives at and attends Northwestern University in St. Paul, MN majoring in creative writing and journalism, with a Bible minor. Isabella is almost 12 and will be in 7th grade. Levi is 18 and will attend Bethel University in St. Paul in the fall majoring in accounting with plans to become a CPA. Amanda is 19 and attends St. Olaf College in MN majoring in Economics and Math. Beth is the mom and is a nationally known healer and Myofascial Release Therapist. Max is 16 and will be in 11th grade. Jay is the Director of Engineering at a farm equipment company. Scott is 21 and recently graduated with a degree in IT and is now employed full-time in his profession.



From L to R, Front row: **Alex, Isabella** (Sister), **Levi**.
Back row: **Amanda** (Sister), **Beth** (Mom), **Max** (Brother),
Jay (Dad), **Scott** (Brother) ~ Apple Valley, MINNESOTA

THE *Biddle-Scott* FAMILY

*W*e are the Biddle-Scott family and we drove nearly 550 miles one way to get here! Scott and I had two children that were affected by MTM. Our first son Austen O'Neal Scott lived from August 2002 until February 2004. He was the light of my life and loved Winnie the Pooh. Our second son, Ashton Paul, was born in 2004 and will be in the 3rd grade this year. Last year he earned all A's and is extremely independent. Ashton loves to play games on the Xbox and iPad. His favorite places to go are to Walmart and to McDonalds to get French fries. Recently, he has fallen in love with Goldfish snacks.

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. She is super excited to attend this conference with me!



Samantha (Mom), **Scott** (Dad), **Austen** (8/14/02 - 2/6/04),
Ashton, & **Dawn** (Grandmother) ~ Logansport, INDIANA

RACHEL *Bronstein*

*M*y name is Rachel Bronstein and I am 27 years old with RYR1 CNM. I hold a bachelors degree in Family Social Services and a masters in Rehabilitation Counseling from Northern Illinois University.

Rachel Bronstein
~ Buffalo Grove, ILLINOIS

I currently am a freelance counselor, but hope to obtain employment with an agency in the future. CNM has impacted my life by helping me to realize that I have limitations and am not like everyone else. It has caused me to realize who my true friends are and how many people truly care about me.

THE *Bushey* FAMILY



Natalie (Mom), **Matt** (Dad), **Lillian** (Sister), & **Cooper**
~ Xenia, OHIO

*H*ello, we are the Busheys: Matt, Natalie, Lilly and Cooper! We live just outside of a town called Xenia, Ohio. Matt teaches 8th grade math, and I just finished nursing school and have taken a job as a Labor and Delivery nurse. Lilly, two minutes older than Cooper, is a bona fide princess, and Cooper is our superhero, who loves music!

During my entire pregnancy, we had absolutely no idea that anything was wrong. The doctor kept preparing us for Lilly to be our "sick baby," but little did anyone know that it was going to be Cooper to give us a run for our money! After coding multiple times at birth and being nearly taken out by a PICC line, he always has come back fighting, which is why our friends in the NICU dubbed him, "Cooperman the Superman." After our initial diagnosis, we quickly searched for some sort of literature on MTM and came across the group on Facebook. We could not have been more thankful for the way we were received and the support that was given. And now, whenever we have a question about something, we ask our MTM family before the doctors, because more often than not they have a better answer! Thanks for all that you do!

Attendees

THE *Cook* FAMILY

*H*ello, we are the Cook Family: Randi, James (a tire pressman), four-year-old Peyton, and two-year-old Austin. We live in Maple Shade, NJ. Austin was diagnosed with Myotubular Myopathy when he was about 7 months old. While most of his life was spent at Children's Hospital of Philadelphia, we are happy to say that he has now been home for over a year! At one and a half years old, Austin received a trach, and he is vent dependent, although we are now sprinting him off the vent! Peyton is such a great big sister, and she helps out so much. She is involved with almost every aspect of his care.

Austin's favorite activities are playing on the iPad, family time, walks, camping in our pop-up camper, watching tv, hanging out with Peyton, and playing with his trucks and other toys. We are very excited to meet everyone!!



James (Dad), **Randi** (Mom), **Peyton** (Sister), and **Austin**
~ Maple Shade, NEW JERSEY

THE *Frase* FAMILY

*I*n 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 that “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 by the grace of God Joshua survived so many life-threatening episodes since Paul and I were completely uneducated in how to take care of such a fragile little baby.

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Alison (Mom), **Paul** (Dad), **Joshua** (2/2/95-12/24/12)
& **Isabella** (Sister) ~ Ponte Vedra Beach, FLORIDA

Before Joshua's first birthday, Paul and I founded the Joshua Frase Foundation to 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 was a packet of copied textbook pages on MTM sent to us by our neurologist. 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 a sixteen-year journey of working towards a cure for our precious children. We have expanded that partnership to Wake Forest Institute for Regenerative Medicine and the University of Washington.

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 a few months 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 we do. We would LOVE to meet you at this conference if we haven't already met... and if we have, come say hi!

THE *Garland* FAMILY



Becky (Mom), Rob (Dad), Reese, & Riley (Sister)
~ Greenwood, INDIANA

*M*Meet the Garland Family! We are Becky, Reese, Rilee, and Rob. We hail from Greenwood, Indiana, which is one of those fly over states in the Midwest. The two cuties in the middle are two and five. When Rilee was only two years old we told her she was going to be a big sister. She was so excited! Of course she wanted a little sister, but we were blessed with a boy. Reese came into the world on August 26, 2010 via emergency c-section 4 weeks earlier than we planned. He had immediate respiratory failure and was intubated shortly after birth. He was transported the next morning Riley Children's Hospital in Indianapolis, Indiana. He spent 4 months in the ICU there and was considered the "big boy" of the NICU!

He finally came home December 14, 2010 with far more accessories than the average newborn. We have since been relishing in our simple life as the parents of two amazing little people who hold our hearts in their little hands. We are proud parents who, like most other parents, think our kids are the cutest, brightest kids in the world. We love to spend time together as a family and are known for throwing the best cookouts in the neighborhood!



Attendees

CONNIE *Guinn*

Elijah Stephen Guinn was born at 6:18 pm on June 21, 2001, in Wichita Falls, Texas. When he was born, a hush fell over the room --that's when we knew something was wrong. I was later told that our beautiful 7 lb, 3 oz, 22-inch long son tumbled into the world like a rag doll. My sweet baby was only with me for 23 short hours, a time spent being lovingly held by his family before departing on the evening of June 22, 2001. Although Elijah was never formally diagnosed, he presented with all the symptoms of classic x-linked MTM.

Benjamin Elisha Guinn was born at 12:50 pm on September 12, 2002, in Houston, Texas. After a muscle biopsy, and DNA testing, he was diagnosed with x-linked MTM. He used a g-tube button to eat, had a trach, used a ventilator to help him breathe, used a wheelchair for mobility, and used a speaking device for communication. He was a wonderfully sweet little boy with a great big smile to share with everyone. He was both fragile and resilient, and had a great disposition. My sweet boy was with me for almost 8 years before passing away unexpectedly on the morning of June 12, 2010.

I am working through the grieving process, June is an especially hard month of course, and it hurts not being able to hold Elijah and Benjamin or to watch them learn and grow. Benjamin attended the 2009 conference and there are memories that make it difficult to attend the conference, but life goes on, and there is a lot of work still to be done.



Past Conference Director, 2009
Connie (Mom), **LeeAnn** (Sister) **Elijah** (6/21/01 - 6/22/01)
& **Benjamin** (9/12/02 - 6/12/10)
~ Wichita Falls, TEXAS

THE *Huseth* FAMILY



Diane (Mom), **Paul** (Dad), **Christopher**, **Daniel** (Brother), **Benjamin** (Brother), & **Andrew Woodbury**, Minnesota
~ Phoenix, ARIZONA

Christopher (42 years old) was born in March of 1971 with classic symptoms of the disease. He was formally diagnosed with Myotubular Myopathy at 9 months old upon a muscle biopsy and testing at the time of a hospitalization with pneumonia. At this point, he also received a gastrostomy feeding tube. As one would assume, very little information was available to us regarding the disease in those days, and we simply trekked forward.

Andrew (32 years old) was born in August of 1980 with the same classic symptoms and subsequently also was diagnosed with Myotubular Myopathy. During this time, we underwent genetic testing and counseling. Evidence and findings to us suggested that of a recessive-type gene as the origination. Many years later, both sons participated in the Dr. Herman research and study, and it was determined that neither son has the x-linked form of Myotubular Myopathy.

The development of motor skills and milestones came slowly for both sons, 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 very well.

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

Attendees

THE *Koh* FAMILY

We are the Koh family from Denver Colorado: Ed, Carrie, Elliot 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. Elliot was soon transferred to Colorado Children's Hospital. A muscle biopsy revealed that our little peanut had a rare disease known as MTM.

After struggling with sadness, anger and fear, we found the strength as a couple 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 developed his "bucket list" and quickly started marking off the items. He went outside, laid in the grass, felt sand between his toes, cuddled with as many people as possible, dressed up for Halloween, had a birthday party, got baptized, finger painted with his cousins, met Santa and many more wonderful things.

Although we dreamed of taking Elliot home, no patient had ever been discharged from the NICU on 4L of heated high flow oxygen. But through the work of respiratory therapist "angels" and a supportive home health care company, we were able to get Elliot HOME for good!

We spent the last 2 ½ months of Elliot's life at home cherishing every moment. Small moments like lying together in bed, all with his O2 and suction equipment in tow, were a dream! We invented, modified and rigged ways to improve life for our special needs child. We celebrated the holidays with Elliot in good health. Those 2 ½ months will be the best days of our lives. We were a happy family.

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. Every little movement he made was a milestone, every smile was cause for celebration. We look forward to the day that we can all be together again with our little man.



Ed (Dad), Carrie (Mom), and Elliot (5/18/12 - 1/6/13)
~ Denver, COLORADO

THE *Mortenson* FAMILY



Marty (Dad) & Mitchell

THE *Rocheford* FAMILY



Jamie (Mom), Bryan (Dad), Abby (Sister), Carol (Grandmother) & Nicholas (3/18/12 - 12/17/12)
~ Princeton, MINNESOTA

REGINA *Mulligan*



~ Bemidji, MINNESOTA

We are the Rocheford family, my name is Jamie, my husband's name is Bryan. We have two amazing children the beautiful girl that you will grow to love at this conference is Miss Abigail, she goes by Abby though. Our beautiful little MTM warrior's name is Nicholas. We lost our warrior last December 17, 2012 only 9 months from the day we received him. Bryan and I are grateful for our support system. No matter what we had to learn in order to have our son at home, my Mother Carol Laduke was always right there, learning with us. We are so thankful for her help that we asked her to join us for the conference.

Attendees

THE *Serano* FAMILY

*M*atteo Serafano Matteo was born on February 25, 2008 and hails from Huntington Beach, California, where he lives at home with his parents, Don and Nancy. His recent milestones include attending pre-school where he was named student of the year! He is learning to use his communication device, Nova Chat 10 and also learning how to drive his new power chair.



Nancy (Mom), **Donald** (Dad), & **Matteo**
~ Huntington Beach, CALIFORNIA

THE *Sirmon* FAMILY

*O*n February 16, 2012, our family was blessed with Benjamin Dean Sirmon. His two older sisters Lennon (4) and Isla (3), could not have been more excited. After three months in the NICU, Benjamin came home with g-tube, trach, and full time vent, and was diagnosed by muscle biopsy with Myotubular/Centronuclear Myopathy. At this time, all genetic tests have returned negative, but we continue to wait on two last tests.

The Sirmons are a very busy family living in Luling, LA. Dad (KC) is an orthopedic surgery resident in New Orleans and Mom (Hillary) is a pediatric physical therapist turned busy stay at home mom. Lennon and Isla are busy, energetic, loving, and spunky little girls who always want to include their little brother in fashion shows, camp outs in the living room, and dance time in the afternoons. Benjamin is an amazing kid that loves life, he is always happy, and though he has little movement, can change lives.



Hillary (Mom), **KC** (Dad), **Lennon** (Sister), **Isla** (Sister),
& **Benjamin**
~ New Orleans, LOUISIANA

THE *Slaby* FAMILY



Jack (Dad), **Carol** (Mom), **Jack** (Brother), & **Joe**
~ Terre Haute, INDIANA

*H*i, my name is Joe Slaby and I'm from Terre Haute, Indiana. I am 22 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. They have had a definite impact on my life and have been very supportive.

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. At times, I use a scooter for longer distances. I strive to maintain the strength I have through daily stretching, light exercise, and a somewhat healthy diet.

Recently, I graduated college with an associate's degree in accounting and am taking some time off so that I can focus on starting a power wheelchair soccer team in Terre Haute along with working on a couple of other side projects.

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Attending the past two MTM/CNM Family Conferences and meeting the doctors and 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 these same families along with getting to know new ones.

THE *Whiston* FAMILY

*T*he Whiston Family is very excited about attending their first MTM-CNM Family Conference. In attendance are Dan, Melanie, Juliet, William and Dan's Parents Brian and Debbie Whiston. William has X-linked MTM and just celebrated his 2nd Birthday. We, like many others have definitely had our share of struggles over the past two years. However, we are happy to report that we have now been able to take our family on multiple mini-adventures including going to zoos, festivals and a baseball game! Dan, Melanie, Brian and Debbie are all board members of Where There's A Will There's A Cure. We are honored to meet our MTM-CNM Family. Thank you for the amazing support you have given us.



Daniel (Dad), **Melanie** (Mom), **Juliet**(Sister), and **William**
Brian & Debbie Whiston (Grandparents)
~ Geneva, ILLINOIS

Attendees

THE *Wilhelm* FAMILY

*H*i there! We're the Wilhelm family, Martin, Lindsey, Emily, and Louie. Martin and I have been married for a wonderful eleven years and have our two beautiful children to show for it. Emily is four and Louie is two. Louie was diagnosed with x-linked MTM at three months old through a muscle biopsy. Emily is loving swimming lessons right now and Louie loves to be in the water too! Emily also enjoys painting and drawing. Louis enjoys getting caught up in cartoons and loves playing with everyone. We are so excited to be part of the 2013 conference!



Lindsay (Mom), **Martin** (Dad), **Emily** (Sister) & **Louie**
~ Dallas City, ILLINOIS

THE *Vandermaazen/Bierstaker* FAMILY

*O*n November 30, 1993, our son, William Michael Bierstaker, "Willie" was born. We knew immediately something was very wrong. He was very weak and barely had the energy to take a breath. In a matter of hours he was whisked away to the NICU at another hospital. In the course of his first year, he had numerous surgeries including the placement of a g-tube, and tracheotomy. We got so familiar with the hospital it was like a 2nd home. Willie's muscles remained very weak, but when he was feeling well, he had an amazing smile and an engaging personality.

He had pneumonia several times but surprised us all and pulled through. We decided to keep him on a ventilator 24/7. This was a turning point for him: instead of using all his energy to breathe, he could use it for other things. He was happy most of the time, alert and wiggled around.

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Lisa Beth (Mom), **Michael** (Dad), **Elisabeth** (Daughter),
and **Willie** (11/30/93-8/8/02)
~ Appleton, WISCONSIN

So we took him home with all of his heavy and cumbersome equipment. But we were determined to find ways to help him do as many “normal” childhood things as possible.

Life was incredibly busy and very difficult at times. We cried gallons of tears, but felt incredibly privileged to have the opportunity to care for Willie. We learned to appreciate the simplest of things.

We were blessed with family and many friends who helped and supported us and who prayed for us when things got rough. We always had in the backs of our minds how very fragile Willie’s health was and we knew that any cold could turn into pneumonia. We spent hours upon hours praying for a miracle, only to finally realize that each and every day is a miracle. God answered our many prayers by giving us strength to face our challenges and faith to recognize His Hand in our daily lives. Willie left this world and entered eternal life on August 8, 2002 at 5:50 am at the age of 8.

It’s been more than 10 years since Willie passed away; years that have been filled with deep, deep sorrow. Losing a child has been described as one of the most painful experiences ever. But time does make the grief easier to manage, and the one thing that has been most important is the knowledge that Willie is at home with Jesus. He is free of all of his physical limitations, and one day we will all be together again.

Our daughter, Elisabeth is now in college, Mike and I have moved ahead with our careers. We would not trade the 8 years we had Willie with us for anything. He taught us what is truly important in life. He could speak only a few words, but his life spoke volumes; he couldn’t walk, but he took us places we never dreamed we’d go; his life on this earth was only 8 years long, but he changed our lives and the lives of so many others in countless ways.



Tribute to our Angels

In Loving Memory

We reflect on all of the children who have passed away from Myotubular Myopathy or Centronuclear Myopathy. For the children named below and for those we do not know, we celebrate their memory as we gather together for our 2013 MTM-CNM Family Conference. Their spirits will always be with us.

Jonathan Balmer

3/11/02 - 4/7/02



John Balmer

12/1/09 - 4/8/13



Logan Tyler Beasley

5/5/97 - 7/7/03



Thijs Marijn Bergijk

3/17/06 - 4/21/06



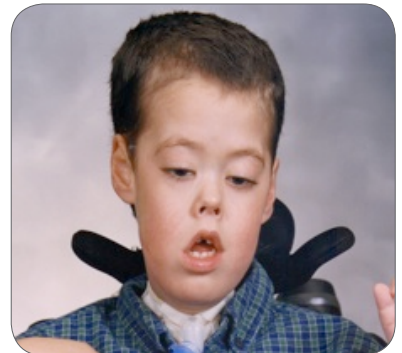
Jack Adam Blunsdon

1/11/06 - 1/11/07



Patrick Michael Bowers

7/25/96 - 11/5/04



Trent Burley

3/18/99 – 10/8/04



Kevin Campos

7/4/03 – 7/2/04



Gabriel Cole Currin

5/8/03 – 8/22/12



*Andre Colton
Narro-Boggs*

6/25/05 – 7/5/10



Anthony Ray Cox

12/25/88 – 1/30/89



Scott Crane

7/25/87 – 6/11/11



Samuel Cuthbertson

10/21/03 – 12/12/03



Daniel

2/2/06 – 10/23/07



Dillon James Daniels

6/22/91 – 7/11/91



Tribute to our Angels

Jordan Davis

4/5/07 - 11/23/08



*Raymond Chester Devore
"RC"*

1/21/05 - 11/22/11



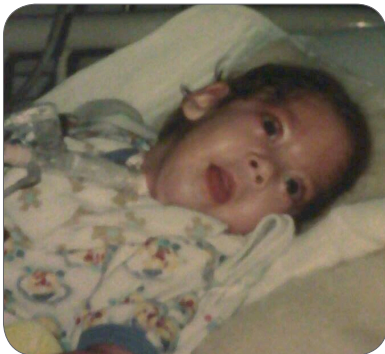
Joshua Miles Frase

2/2/95 - 12/24/10



Ethan Trey Gibson

4/7/01 - 3/24/02



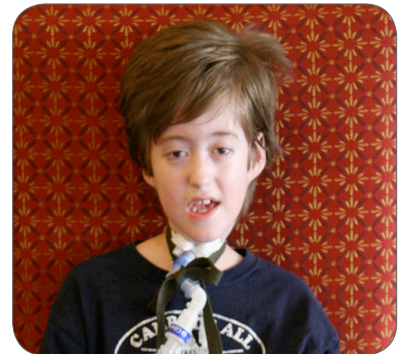
Kaden James Grant-Buck

4/19/06 - 2/13/08



Benjamin Elisha Guinn

9/12/02 - 6/12/10



Elijah Stephen Guinn

6/21/01 - 6/22/01



Maddux Achilles Haggard

2/4/05 - 2/10/05



Louis Thomas Hart

10/31/08 - 1/2/10



Marlon Louis Hart Knowles

5/18/05 – 8/28/05



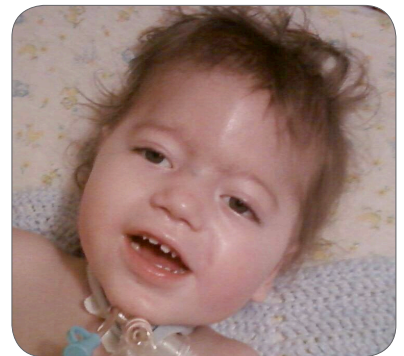
Carter Dean Hershey

10/30/03 – 11/5/03



*Kenneth Ray Houston, Jr.
"Scooter"*

4/23/10 – 7/17/12



Kaylee Rae Howeth

11/29/05 – 5/31/12



Sol Xavier Humphreys

5/22/08 – 6/20/08



Patrick William Huss

9/17/99 – 2/3/05



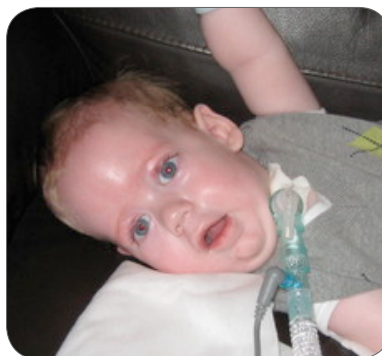
*Lane Garrett
Jackman-Brassfield*

10/16/01 – 9/9/10



Nathaniel Jenkins

3/26/08 – 3/18/09



Zachary Josephson

2/15/89 – 2/6/96



Tribute to our Angels

Paul Kahn

11/6/45 - 1/1/10



Broderick Dillon Knapp

9/30/95 - 4/01/02



Ingo Thomas Kuippenburg

8/23/04 - 2/9/05



Elliot Koh

5/18/12 - 1/6/13



Samuel William Kowalski

9/5/05 - 6/3/07



Benjamin Laager

10/23/05 - 1/7/06



Tom Lennox

7/21/03 - 5/29/07



Nathan Loughlin

10/28/00 - 11/14/00



Sean Loughlin

10/7/04 - 11/06/04



Alex Loughlin

12/25/08 - 12/25/08



Killian Loughlin

4/1/11 - 4/25/11



Vitus Hollaender Maansen

11/5/08 - 6/14/10



Padraig Martin Maguire

7/10/87 - 10/19/87



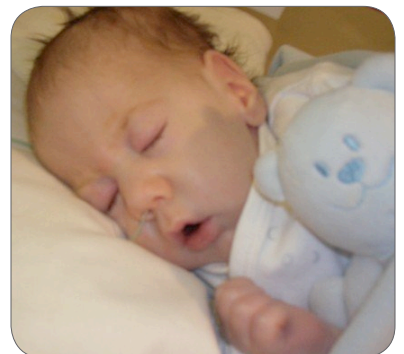
Donn Joseph Maguire

7/17/88 - 3/15/89



Archie Mcinerney

8/26/05 - 2/4/06



Michael Keith McLaughlin

4/22/09 - 6/17/09



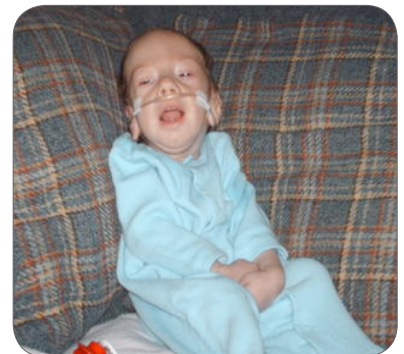
Matthew McRill

9/5/04 - 10/8/04



Patrick Connor Miller

10/05 - 8/12/08



Tribute to our Angels

*Liam Thomas Merriam
Muendel*

11/6/45 - 1/1/10



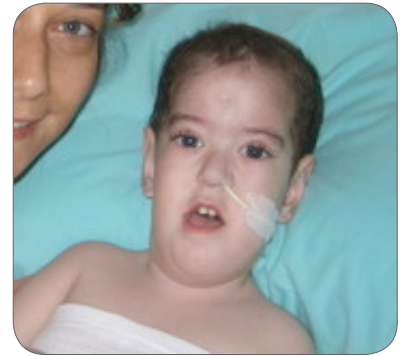
Austen O'Neal Scott

8/14/02 - 2/6/04



Iker Paltura

4/07 - 11/23/08



Taxon Pepworth-Lehmann

4/18/08 - 6/17/08



Taxon Persons

6/25/10 - 10/24/11



Rachel Allie Pierce

8/13/99 - 3/5/04



Nicholas Rocheford

3/18/12 - 12/17/12



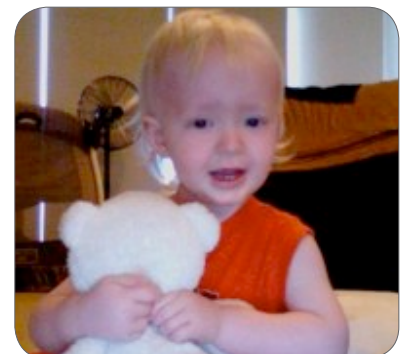
Aaron Ryan

9/10/07 - 3/14/11



Joseph Daniel Sanders

5/29/07 - 2/28/09



Owen Michael Servo

6/26/10 - 9/12/11



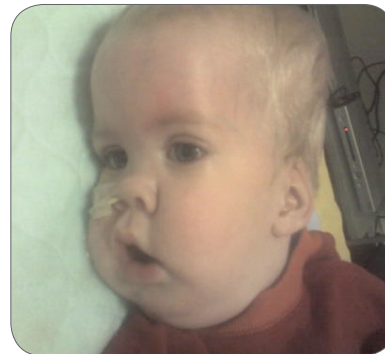
Joshua Seville

4/3/00 - 7/13/11



James Smith

9/16/05 - 8/29/06



*Cameron Alburtus
Maria Stok*

1/19/06 - 8/11/06



Justin A. Tineo

4/7/03 - 7/26/07



James Franklin Watson

2/13/81 - 6/25/82



Kaleb Anthony Williams

10/06/07 - 5/17/08



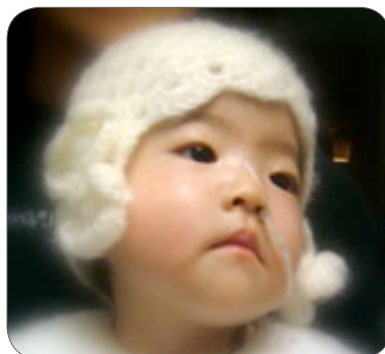
Matthew Wright

5/28/01 - 8/27/04



Evelyn Qing Zeng

12/25/04 - 12/12/07



Tribute to our Angels

Always in our Thoughts, Forever in our Hearts...

(ANGELS NOT PICTURED)

<i>Andrew</i>	2/16/86 - 5/24/86
<i>Nakai Angel Beasley</i>	11/25/80 - 11/23/81
<i>Willie Bierstaker</i>	11/30/93 - 8/8/02
<i>Jason Bubniak</i>	8/8/03 - 12/9/03
<i>Andrew Ronald DeSchampe</i>	8/26/01 - 9/16/01
<i>Alex Fallon</i>	5/4/05 - 6/17/07
<i>Robert Andrew Hann</i>	2/22/87 - 2/6/12
<i>Patrick Stewart Hann</i>	2/24/95 - 2/27/97
<i>Ryan Cedric Hokse</i>	5/5/06 - 6/4/06
<i>Miles Daniel Josephson</i>	4/4/88 - 4/4/88
<i>Lenzie Keith</i>	10/3/76 - 7/4/81
<i>Joseph William Means</i>	4/11/66 - 4/11/66
<i>Nathaniel</i>	7/27/00 - 10/26/00
<i>Cliff Bolter Raddatz</i>	date not available
<i>Roman</i>	9/29/10 - 1/24/11
<i>Michael Andrew Russell Steckler</i>	12/18/91 - 10/30/95
<i>Shannon Miles Sanders</i>	6/25/79 - 6/25/79
<i>Chad Tetreault</i>	3/25/88 - 8/20/02
<i>Matthew Tyler Thomason</i>	1/12/93 - 5/17/93
<i>Joshua Whistler</i>	4/2/05 - 1/17/06



"In Loving Memory"

*I*n Loving Memory
of our MTM-CNM Angels,
including those whose names
are unknown to us
but held in the hearts of those who loved them.

May you always feel
your child's love surround you
and know their loving legacy
lives on in our community.

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

All our love,
2013 MTM-CNM
Family Conference



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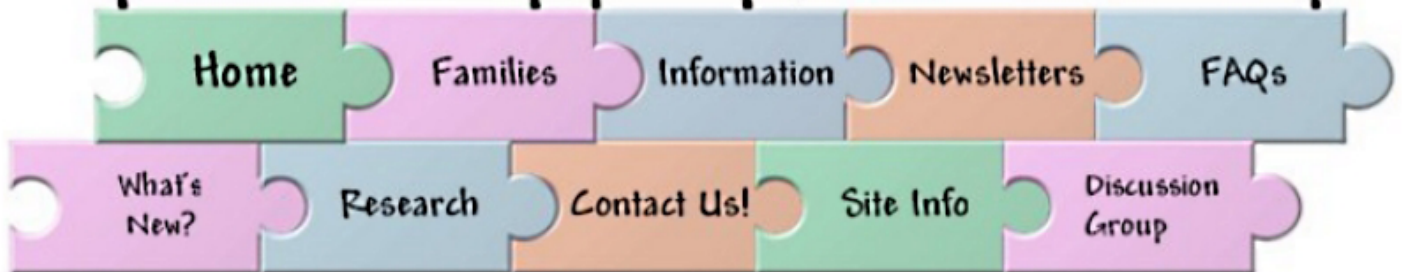
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Thank You to the Scoggin Family for being pioneers in the MTM/CNM Community, and for their sponsorship of the 2013 MTM-CNM Family Conference through their non-profit organization, the MTMRG.



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"The truth of the matter is nobody is perfectly normal. We all have various types of characteristics, but one thing is for sure none of us are the same and none of us are completely different."

- Joshua Frase
www.joshuafrase.org



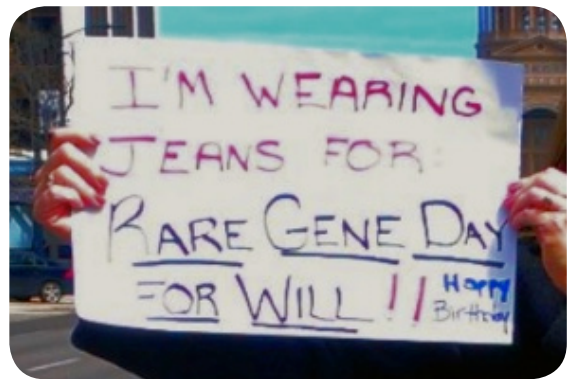
Many thanks to the Sanders Family for their annual Fireworks Booth, in loving memory of their son Joey

Conference Donors



Thanks to all who have helped to support the Family Conference by purchasing items from our Zazzle Store!

And many thanks to our family, friends and members of the Comprehensive Grammar School who participated in our "Jeans for Rare Genes" day!



Thank You to the Wood Family for their generous donation!



Thanks to the
Slaby Family
for their Rocking
fundraisers this year
with their letter-writing
campaign and yard sale!



Thank you to the
Hanson/Stansbury family
for their generous donation!



In appreciation of your
generosity, Majeau Family!



Thank you to Sergio of
SERGIOV PHOTOGRAPHY
for donating his time and
helping us make lasting
memories.

Conference Donors

With great appreciation to all our 2013 MTM/CNM Family
..... Conference Donors

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Doug & Ellen Waddill

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Dan & Kim Ward

Kim & Ivy Ward

Leo & Eileen Ward

The Whiston Family

The Wood Family

Jim Zinkan

Memories from 2009 & 2011



2009 MTM/CNM Family Conference Group Photo



2011 MTM/CNM Family Conference Group Photo

Memories from 2011





