

All About Us

All About Us is the newsletter of the Foundation for Nager and Miller Syndromes. We took our name from a story submitted by Monica Quinn in our Autumn 1991 issue. Monica captured our entire theme and purpose in three little words.

“Us” is all persons who live with our syndromes, the members of our families, our friends, our neighborhoods, the medical community and everyone we can reach. We offer information and communication for everyone interested in Nager and Miller syndromes.

As you read our newsletter, as you think about getting involved, as you contribute your features, reports, photographs, or your own experiences and poetry, think to yourself — “This is all about us.”

FNMS is an international support group dedicated to helping those affected by Nager and Miller syndromes. We serve as a clearinghouse of information and link families seeking support, hope, and advice. FNMS empowers families and guides them through the process of surviving to thriving.

FNMS is a parent-run volunteer organization with a 501 (c) (3) not-for-profit status.



Photo: Brad Kevelin

2013 FNMS family conference

Read all about this year’s wonderful conference from the vantage point of parent and founding FNMS director Margaret Hogan (p. 5). Our happy group is pictured above in the lobby of the Seattle Waterfront Marriott and enjoying some of the other sites Seattle had to offer below.



Castillo Family



Madsen/Jorde Family



Bueno Family

All About Us

www.fnms.net

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This foundation does not endorse hospitals, teams, products, or treatments. The information in this newsletter is provided to keep you informed of activities and progress internationally regarding Nager and Miller syndromes. Views expressed in this newsletter are not necessarily those of the Foundation for Nager and Miller Syndromes.

Director's Note: December 2013

Hello again! This year, as I struggled mightily to come up with something new and fresh to write, a hundred different ideas and tangents cluttered my mind to the point of total overload. What follows is my twelfth and final draft. I hope that it imparts most of what I wanted to convey.

As I procrastinate each year in writing this until the very last minute (and this year even beyond that) the holidays are always fast approaching. The time when I reflect upon the year gone by and all that I hoped to, but did not, accomplish. This year, instead, I am going to focus on my gratitude for each and every one of you. For if you are reading this you are an integral part of FNMS. Whether you are a donor, a volunteer, a vendor, a member family or a supporter of any of our member families, you are a part of the foundation. I feel truly blessed and grateful to have been involved with FNMS for the past 12 years and want you to know that I count each of you as a blessing in my life. Through your association with the foundation you have touched the lives of countless others — some you know and many that you may not know. As you peruse this issue, stop and look at the faces depicted in the photographs, these are just some of the people whose lives you have touched.

FNMS celebrated our 24th anniversary this past June by hosting our 9th Family Conference in my home town of Seattle, Washington. We were overjoyed to find that it was our most well attended conference to date. New member families from as far away as New Zealand and Germany were in attendance. It is always remarkable to see so many families gathered together from all over the world. They come to renew old friendships, meet and form new friendships, play together, and perhaps most importantly, learn from each other. This is always the ultimate goal of FNMS, bringing families together in a variety of ways to develop a strong support network and an even stronger bond. I often tell new members how important and life changing our conference will be for their child as well as their entire family. It wasn't until this year that I realized how each and every conference is life altering for me! It reminds me of what is really important in life, it renews my commitment to the organization and refreshes my spirit in a way that no other experience does.

So, thank you! Thank you all for the part that you play in building and sustaining FNMS!

DeDe Van Quill

Attend the "Taste of Morton Grove"



Support the foundation that supports FNMS each year by generously donating a portion of the proceeds from this event to FNMS.

Thursday, March 6, 2014
5:30 – 9:00 p.m.

White Eagle Banquets & Restaurant
6845 Milwaukee Ave., Niles, Illinois 60714

Contact Margaret Hogan (mepti40@aol.com) for information about tickets for this event.

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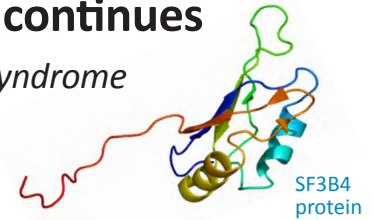
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Gene for Nager syndrome found ... but the search continues

SF3B4 is the major gene responsible for autosomal dominant Nager syndrome



Modern gene discovery methods help find Nager gene

The year 2012 signaled a major breakthrough in the search for the underlying genetic cause of Nager syndrome, which affects only a few hundred people worldwide. Researchers in the labs of Dr. Francois Bernier and Dr. Michael Bamshad teamed up to sequence DNA from 35 families (41 individual patients) with Nager syndrome. They found a total of 18 unique mutations in the gene, *SF3B4*, in 25 of those 41 individuals (61%) and published their findings in May 2012 (1).

Michael Bamshad, MD

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Francois Bernier, MD

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Calgary, Alberta, Canada;
Finding of Rare Disease Genes (FORGE) in Canada



Drs. Bamshad and Bernier used exome sequencing to identify *SF3B4* as the cause of this condition in most patients. Exome sequencing involves sequencing all of the protein-coding parts of a gene (i.e., the exons) that make proteins in our body — proteins that control development and growth and carry out important body functions. The researchers looked for variants or mutations in the same gene that showed up in more than one of the individuals with Nager syndrome who had their exome sequenced. By giving higher importance to mutations predicted to cause a greater disruption to protein function, the gene *SF3B4* emerged as the major candidate gene.

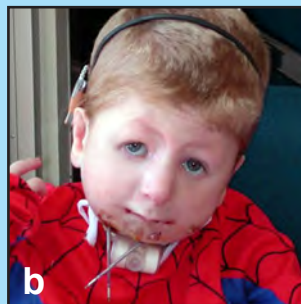
The sequencing of the *SF3B4* gene in additional individuals with Nager syndrome confirmed that *SF3B4* is the gene responsible for most cases of Nager syndrome. At least two recent follow-up studies to the original paper have confirmed

these results. Czeschik et al. (2) sequenced 12 Nager patients and found mutations in *SF3B4* in 7 of the 12 (58%), and Petit et al. (3) studied another 18 individuals with Nager from 14 families and found 9 mutations in *SF3B4*.

The researchers conclude that there must be more genes responsible for causing Nager syndrome, so new studies are underway that use exome and whole genome sequencing of individuals with Nager syndrome who did not have a mutation in *SF3B4* to see if they have mutations in a gene that works with *SF3B4* or otherwise. It was interesting that the clinical characteristics of individuals with Nager syndrome caused by *SF3B4* mutations were indistinguishable from Nager individuals without mutations in that gene.

One of the functions of the *SF3B4* gene is to encode a protein involved in splicing — a process that all cells use to link coding exons together before creating a protein. Splicing is really important for controlling how genes work and it is likely that mutations in *SF3B4* result in abnormal function of a specific gene or genes required during development. Further work will be required, however, before we truly understand why genetic changes in *SF3B4* result in Nager syndrome.

There is clearly a spectrum of severity in which the physical characteristics manifest in Nager syndrome. Some individuals may have a weak thumb that doesn't bend while another might be born with no thumb at all. Some might have a small jaw but their airway was managed at birth in such a way that they did not require a tracheostomy. Almost all Nager individuals have a degree of conductive hearing loss, but in addition to the loss of hearing, the degree of development in the inner and outer ear structures also varies. There may be clues to how this is determined in the Nager gene studies already published and could be an area explored in more depth in the future. Also, the fact that *SF3B4* does not seem to fully explain the cases of Nager syndrome may mean that the involvement of other mutations in the same or other genes may determine the severity of the physical characteristics of the syndrome.



Some of the physical characteristics of Nager syndrome and common surgeries: a) recessed lower jaw (micrognathia) and low-set ears, b) downward slanting eyes and need for multiple jaw distraction surgeries, c) pollicization surgery to remove underdeveloped thumb and create a functioning thumb from child's index finger, d) missing thumb (right hand), underdeveloped thumb (left hand), and crossed toes.

Comparing the genetics of Miller and Nager syndromes

On the surface, Miller and Nager syndromes are quite similar. Those affected by either one of these syndromes experience underdevelopment in the bones of the cheeks, ears, jaw, upper and lower limbs, in addition to other anomalies — some unique and some shared. For our families, these physical issues manifest in problems with breathing, eating, hearing, and in using their arms, legs, feet and hands for things that people who develop typically take for granted, such as feeding or bathing oneself, buttoning clothes, playing sports, riding a bicycle or driving a car, and numerous other activities. So although Nager and Miller are closely aligned when it comes to the physical characteristics attributed to each syndrome, how they develop at the genetic level appears to be very different.

In 2009, researchers from Dr. Bamshad's lab and others published a paper that described how exome sequencing could find the causative gene in single-gene (also called mendelian) diseases (4), and their proof of principle was the discovery of *DHODH* as the cause of Miller syndrome. Miller syndrome is inherited in an autosomal recessive manner. That means each parent has one copy of the mutated gene and each parent must pass down the mutated version of the gene to their offspring for the child to develop Miller syndrome. Copies of the mutated gene in the general population are rare, therefore it is rare for both parents to have a mutated copy of the gene. This explains why the occurrence of Miller is so rare. Nevertheless, when both parents carry a mutated copy of *DHODH*, the risk of having a child with Miller syndrome is 25% in each pregnancy.

The study by Drs. Bernier and Bamshad showed that in children with Nager syndrome with unaffected parents the mutation in *SF3B4* was new and not inherited from each parent. Further, when a parent and their child were both affected, the causal variant in *SF3B4* was inherited from the affected parent. So for families with mutations in *SF3B4*, Nager syndrome is an autosomal dominant disorder. There are some papers in the

literature that suggest there might be an autosomal recessive form of Nager syndrome, but there is currently no way to prove or disprove this possibility. This means that for Nager families without an *SF3B4* mutation, the inheritance pattern cannot be determined with certainty.

Finally, in each of the studies involving Miller (4) and Nager (1), the researchers verified that mutations in *DHODH* did not occur in individuals with Nager syndrome, and that mutations in *SF3B4* were not seen in individuals with Miller syndrome.

What does the future hold?

The search for genes that explain *SF3B4* mutation-negative individuals with Nager syndrome is underway. As scientists learn more about the genetics of each of our syndromes, they also continue to learn about the lifelong challenges that patients and their families must face. What does the future hold? Technology has accelerated the pace of discovery to the point where the answer to that question lies in the very near future.

If you are interested in participating in the Nager genome study, please contact:

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Phone: 206-221-3849;

or

Francois Bernier, MD

Email: francois.bernier@albertahealthservices.ca

Phone: 403-955-7373.

References

1. Bernier FP, et al. *Am J Hum Genet* 2012; 9:925–933.
2. Czeschik JC, et al. *Hum Genet* 2011;132:885–898.
3. Petit F, et al. *Clin Genet* Sept. 4, 2013 (ePub ahead of print).
4. Ng SB, et al. *Nat Genet* 2010;42:30–35.

We remember long-time FNMS supporter

Trish Strickland

taken by angels
October 2, 2011



Cousins Trish Strickland and Margaret Hogan

Trish Strickland was my dear cousin and we were the same age growing up in the Chicago area. Trish was a very knowledgeable and discerning woman with a gentle spirit, who was smart, fun and kind. In 2006, I was thrilled to have Trish volunteer at the FNMS conference in San Francisco and interact with all our kids. Those kids made a great impression on Trish, and thereafter she chose FNMS a favorite among the other worthy charities she valued.

She was also a dedicated and valued employee of the Trust for Public Land (TPL) in San Francisco. I am very proud of Trish for her many accomplishments, especially in preserving much of the Northern California coastline. In doing so she helped create many beautiful hiking trails in that gorgeous area.

Trish bravely and privately fought cancer for 10 years and, after it returned, she made a point of putting FNMS in her will. FNMS's recent conference in Seattle in June 2013 was a great success — much in part to Trish's generosity.

Trish set an example of prioritizing what really matters in our short life and left this world a much better place.

FNMS 2013 in Seattle

— through my eyes —

by Margaret Hogan

UPON ARRIVAL IN SEATTLE ON THURSDAY, JUNE 30, my daughter Bridget, her boyfriend Erik, and I were picked up from the airport by FNMS director and parent volunteer DeDe Van Quill. This would be the 9th FNMS conference for Bridget and I that we've attended over the last 20 years. This much anticipated conference weekend was about to begin and I felt excited about what was to unfold in the Emerald City. As past director of FNMS, I recalled all the planning responsibilities, organizing of volunteers, coordinating activities, making the schedule of events and the numerous details that whirled around in my head before a conference. I wondered if DeDe might also have that same frame of mind while driving us to our hotel. She seemed confident and focused, and for good reason — everything was very well planned.

FNMS conferences have an energy unique to themselves and have been described by participants and volunteers to be like international 'family reunions' and even 'life altering'. The attendees already understand and have much in common with each other as parents, siblings, and grandparents of affected children or adults with Nager or Miller. For a pleasant change, there is no being stared at, being pointed at, or having to explain why you or your child look different when among our group. Looking different isn't just expected, it's welcomed.



Upon introductions, we feel connected by these two similar genetic conditions that have seriously impacted our lives. Here begins the sharing and learning from each other about the challenges and triumphs related to eating, hearing, seeing, speaking, walking, multiple reconstructive surgeries, and therapies. The importance of the psychological, emotional well being, and social aspects of living with Nager and Miller syndromes are topics of conversations.

Here for the first time siblings can also speak comfortably with other siblings about their life experiences. There is always great purpose to and results from attending FNMS conferences. The educational component makes FNMS conferences worth attending, but the real breakthroughs occur when those who qualify (from experience) give support to those in need of support. Traveling away from the comfort and conveniences of their homes to stay at a hotel is very difficult for some of our families. For example, being away from food blenders for specially pureed foods is problematic, or traveling with cumbersome and heavy medical equipment makes travel almost impossible for some. It requires determination, flexibility and hope for those who travel despite these obstacles. These people know it's worth the hassle. For when we are face to face with each other, we profoundly inspire each other to do as my Dad used to say — 'to keep on keeping on'.



Some of the wonderful things I saw (or can remember) from this conference 5 months ago started happening right off the bat. DeDe and I gathered and alphabetized 27 thermal bags donated by Outrageous Sports, Inc., for attendees to receive at registration. Besides the conference schedules inside these bags there were many donated gifts: a white collapsible umbrella with the FNMS true-blue heart logo on them donated by Mike Hogan, the book ‘Eight Fingers Eight Toes’ donated by author Debbie Jorde, boxes of note cards of paintings that were originally painted and donated by artist Logan Madsen, and my favorite book, ‘Wonder’, donated by Random House Children’s Books. I saw a great outpouring of generosity from these donors in all these gifts.

As new families and veteran members came to registration, there were introductions, hugs and tears of joy that signaled that our conference was up and running now. First event was our ice cream social and DeDe selected the best flavors of Ben and Jerry’s ice cream and toppings for us to enjoy. Luckily we were allowed seconds — as in servings! Little by little everyone headed up to their rooms after their long travels. Some traveled as far away as New Zealand, Mexico, Australia, and Germany. However, David Gonzales, from Guadalajara, who is used to traveling this planet as a famous pianist, was not too tired to play piano in the hotel’s lounge area that evening. Ryan Van Quill, his son Jackson, and my daughter and her boyfriend were lucky enough to stay up and be there to hear David play. In respective order I see stamina, youth, and a couple of night owls there!



FRIDAY AFTER BREAKFAST we had some free time to explore Seattle. Later that day as a group we all boarded a ferry ride to Blake Island where we were treated to an authentic Native American tribal dinner and show. On the way over many took pictures of each other and I saw the magic of our adult members sitting with the some of the younger first time attendees. Also many long time conference attendees were mixing and catching up with each other. While the rest of us were socializing on board, Claudia Myall and Mike Hogan were competing for the Bravest Landlubber award! Once on shore we were brought mugs of cider with clams. After eating the clams, the shells were to be stepped on to add more crushed shells to the shell gravel path leading up to the lodge. I saw our kids having a blast being allowed to smash, break, jump and stomp on the clam shells that we adults kept



them well supplied with. The dinner was delicious and the cultural show kept everyone’s interest. The FNMS group shared the boat ride back with the public so we encountered some routine stares and questions from the curious about our group. After the boat ride back, we had the choice to either walk back to hotel or ride on the prearranged coach bus. I opted for the bus ride back and just before getting off back at the hotel our bus driver approached me with a question. He asked, ‘What kind of group are you?’ I replied, ‘A parent-run volunteer charity that helps families with these conditions’. To which he replied, ‘I’ve never seen a group like this before where everyone looks out for each other’. In that very short time he was with us, this young man knew he witnessed something special. His observation and comment deeply touched me. He helped me see FNMS through a stranger’s eyes but felt with his heart. Reminding me of the quote:

“IT IS ONLY WITH THE HEART THAT ONE CAN SEE RIGHTLY;
WHAT IS ESSENTIAL IS INVISIBLE TO THE EYE.”

Antoine de Saint-Exupery, Author of *The Little Prince*



ON SATURDAY MORNING, our educational speakers gave presentations to the adults while the kids were being entertained. In the past, speakers have included various therapists, specialty doctors, nurses and geneticists who lectured on issues specific to Nager and Miller syndromes. The morning's first presenter was Gay Lloyd Pinder, a mighty skilled speech therapist who blew us away with her technique and video history of treating Jackson Van Quill. She demonstrated how she patiently desensitized his oral aversion and eventually taught him to become a champion eater. It was thrilling to see how this dedicated professional accomplished something so monumental for Jackson that most of us have tried to do for our own children but have not had that fortunate an outcome.

While the adults were in the educational workshops, the children were supervised and kept busy with fun arts and crafts. Play clay was brought from Chicago and donated by Bridget's friend Erik. The first one I saw come out of the kid's room was Brittany Stevens wearing sunglasses, a feathered headband and strands of necklaces. What a sight to behold as Brittany proudly strutted around showing how she always keeps the fun in funky! Many kids also played games on iPads with the new kids they had just met during this weekend.



We tried something new and different this year in having two FNMS adult members, who were very young back when FNMS formed over 20 years ago, to serve as two of our presenters in the educational adult lectures. Without a doubt this was the conference highlight for me. Monica Quinn gave a very thoughtful and thorough PowerPoint presentation about her capability, responsibilities and independence in doing her job. Following Monica, Erica Perry took the microphone to speak to us on the hot topic of 'bullying'. Erica spoke of her experiences as an adult with Miller syndrome and gave great tips. Her talk was followed by a lively question and answer session. I don't believe anyone else is more qualified to educate us on these topics than those who live, learn and triumph over them daily. I was extremely proud of them both and moved to tears as our audience applauded and gave Monica and Erica well-deserved standing ovations. That was more than just a 'wow' moment for me. Having had the privilege of knowing these young ladies since they were very young, I saw they had grown into mature and confident contributing members of society. Their presentations were a sign that bodes well for FNMS's future. I felt such hope and excitement at the thought that our own kids could keep

FNMS going as a thriving global charity after we parents are gone. They had just proved that and more. Though it was caring parents who started FNMS, it's the next generation who will secure FNMS's future. During the weekend I saw many of the older members interested and caring for our new young members. It seemed to come to them so naturally.

After hearing the amazing presentations, Saturday afternoon's free time allowed everyone time to walk outside in the sunshine to the bustling Pike Street Market, stroll along the waterfront, see the original Starbucks, visit the Pacific Science Center, or see the fabulous Chihuly Garden and Glass exhibition. Mike and I had lunch with my sister Beth, volunteer emeritus, and her friend Mike Russell. This was Mike's first FNMS conference and over lunch he expressed how moved and impressed he was with all of our families. During Monica and Erica's presentation, I saw Mike sitting toward the back of the room and noticed he was captivated by what he was hearing. As a bonus, we did not need to hire security at the conference as both Beth and Mike are Chicago Police officers. While in Seattle it was FNMS for whom they were dedicated 'to serve and protect'.

ON SATURDAY EVENING, the crowd was gathering in the ballroom for the much anticipated dinner dance complete with a DJ. Parent volunteer Leslie Gaffney had scheduled to Skype with long-time member Melissa LeBaron (31 y.o.) via computer since she could not attend the conference. So before the dinner dance was in full swing some of us went to a quiet area of the lobby to take turns saying 'hello' and catching up with Melissa. I was happy this thoughtful prearranged meeting took place to help Melissa feel valued and still connected to all of us. After having a delicious buffet dinner, the DJ began playing music and Monica Quinn was the first one out on the dance floor. Old and young really let loose — let me tell you this group knows how to shake it! In this safe environment, free of judgment or staring, people could let down their guard and lose their inhibitions. We took a break from dancing to honor four birthdays with a cake and sang 'Happy Birthday' to Kelson Konowalchuk, MacKenzie Kehler, Sarah De Heer, and Heather Enter.





Mom's all the way! Above: Jackson and DeDe Van Quill, Stella and Heather Enter, Julia and Ursula Bednarek, and Bridget Ieronimo and Margaret Hogan. Right: David and Cristy Gonzales.



It was so much fun watching parents, brothers, sisters, aunts, uncles and grandparents celebrate the camaraderie out on the dance floor. Being musically talented, it was a natural attraction for David Gonzales to sit with and assist the DJ. Being a huge Beatle fan, I jumped when I heard Paul McCartney's 'Band on the Run' begin to play. But I was not the only Beatle fan on the dance floor, for it was Tinka Gaffney who requested this song and she played the air guitar with enthusiasm second to none! Celebrating through music and dance proved therapeutic for all. It helped distract from post-surgical pains, having feeding tubes, tracheotomies, body braces and chronic pain. This is just one of the benefits where I see the wonders of how our humble little support group that became an international charity can soothe one's soul. DeDe and I could stand back and see the fruits of our labors as directors. Before our eyes, we saw those of us who have experienced set backs, disappointments, failures, struggles and fears bravely choose to rise up as resilient and self sufficient individuals. We celebrated with everyone who had gathered in hope to forge ahead and go from surviving to thriving.

OUR FAREWELL BREAKFAST ON SUNDAY wrapped up a truly extraordinary weekend. In the past we have even had children cry when it was time to say goodbye to their new friends who 'look just like me'. After most left to catch their flights or begin their long drives back home a few of us lingered in the room afterward. Someone had a beach ball and we began tossing it up in the air and eventually passing it from person to person. Paris Van Quill was by far the most athletic who could keep the ball in play and then a spontaneous game of volleyball began in around

our breakfast tables and chairs. Debbie Jorde had quietly attracted my daughter Bridget over to her for a nice, long chair massage. Wisely, Debbie's son Logan (33 y.o.), and daughter Heather (37 y.o.), had been strategically positioned to protect the person receiving their chair massage from being hit with our frequently errant beach ball. All ages and abilities tried their best to keep the ball in play. Best Play of the Day goes to Logan Madsen who kept giving the ball to Raymond Whiteman (7 y.o.) to kick serve the ball back into play. Best Player Award in the Over-40 League goes to Greg Myall. Congratulations, Greg!



My heartfelt thanks to DeDe, her husband Ryan, daughters Peyton and Paris and son Jackson who gave tremendously of themselves and ran a most successful conference weekend. DeDe continues to volunteer her precious time as FNMS's Director. On behalf of FNMS I thank DeDe for all the work she does for FNMS while still living a full life as a wife, mom to 3 and being a full-time real estate agent. Brava! Brava! DeDe's sister Alexa Larsen easily earned the award for being the Best Sister Volunteer Organizer at this conference. No easy job, but she made it look easy and kept us all together. Great thought had been put into every detail of this conference. If you missed it, I strongly urge you to try to attend the next FNMS conference.

Speaking of the next conference, rumor has it that Leslie Gaffney will be hosting it in Boston in 2015. First, my sincere thanks to Leslie who donates her skills and so much time to professionally create the FNMS newsletter. Leslie knows that our newsletter is such an important tool for informing our families and our donor/supporters. Leslie lives a busy life as wife, mom to Tinka (10 y.o.) and working full time. Thanks to Leslie for her dedication and sacrificing her sleep to edit these newsletters into the wee hours of the night.

I pray we all see each other again and even more families in Boston. As we know, Boston was the place where the famous historical words were said, 'The British are coming, the British are coming'. Bostonians best prepare because 'FNMS is coming'! I wish love and peace to each of you in the New Year. Lastly, many thanks and blessings to all our donors whose continued support helps us to help each other.



First and lasting impressions

— of the 2013 FNMS conference —

We are the [de Heer family](#) and traveled from New Zealand for our first family conference in Seattle. It was a really special time for us for several reasons, and the most wonderful opportunity for Theo (6 y.o.) to finally meet some other children with Nager Syndrome.

Theo was born in June 2007, and from about July 2007 I have been lucky enough to be in contact with three families that I met through FNMS. The Perry's here in New Zealand have been the most wonderful contact for us and we are lucky to have developed a really close relationship with Erica in the last 6 years. We have also been in regular e-mail contact with Tinka's family, Saul's family and Anna's family (not able to be in Seattle), my lifeline with all things 'Nager' in the last 6 years. For us to finally get the chance to meet these wonderful families was a dream come true for me. It was like meeting a friend who we hadn't seen for a long time. Theo and Codie are both still a bit young to really appreciate the value of this, however, for me it was a gift more precious than gold. It was sad that some other families were not able to make it, but I know that I will treasure the time when we can finally meet just as much, and that the new families we were able to connect with was just as amazing. This can feel like such a lonely journey for us, being geographically quite isolated here in New Zealand, however, technology has made the world a smaller place. I also really valued having my extended family welcomed into the FNMS family as it was a special opportunity for them to come with us.

My only regret is that we didn't have longer to enjoy the company and share our experiences. However, I know that in 2 years time we will get another opportunity to reconnect with these wonderful people, and perhaps welcome more into this special community we are fortunate to be a part of. Thank you FNMS for organizing this most wonderful conference.

[Monica Quinn](#): This summer, my family and I got to go to Seattle for our FNMS 2013 Family Conference. We had so much fun that it was hard to see it end! I just love coming to these get togethers because I can spend time with my best friends and not worry about being "disabled" for a few days!

My favorite part was when my parents and I went to the Space Needle. I felt like I was standing on "top of the world" when I looked at the city! I also liked making my presentation about my former job and how it intertwined with having special needs. I almost cried when I got a standing ovation. It was special making FNMS history!

We definitely rocked all night at the dance to fabulous music. Peyton and I wouldn't stop until the lights went out! We are truly a family because we support each other about our daily challenges and reach out when times are tough. It's fulfilling to know that FNMS is a terrific organization to count on and I hope it's around for years to come! I miss everyone, but I can't wait to see you all in Boston at FNMS 2015!



[de Heer family](#)
"I liked the dancing on the last night. I liked meeting all of the children and playing with them."

[Theo de Heer](#)

[Tracy and Peyton Smith](#): This year was our first conference and I am so glad we attended. Peyton had such a great time, but it was really special to myself and my mother-in-law. It is so rare we can meet other families whose children have Nager Syndrome. The conference allowed us to see families and their children in different stages that we have not yet arrived at and provided so much hope for Peyton's future. For Peyton it is the first opportunity to build relationships with other children that I have no doubt will continue into her adulthood and will provide her the support she will need. As her parent I can give her all the love in the world, but no one else will understand exactly what she is going through except the other children with her syndrome. I am so glad she will have those wonderful examples to look up to.



[Dancing fools Erica, Theo, Saul and Peyton tearing up the dance floor on the final night of the FNMS conference.](#)

[Ursula and Julia Bednarek](#): First, Julia was happy that Stella could also come from Germany. Second, she likes that the meeting always takes place in a different city and looks forward to exploring the different things to do there.

The language barrier is unfortunate and limits Julia's interaction with other children. For me it is also a shame that I do not speak English. I would have been so happy to and wanted to talk with all of you about everything.

[Lara Beranek and family](#): Dave and I loved talking to the other parents who have had similar experiences with their kids. He and I were both really struck by the different experiences had by people in different countries or even in other areas of the United States. It was eye opening to hear of the things some have endured. We found it very special to be part of a group where these people, who have dealt with people and places that were not accepting of them, could feel comfortable and accepted. Going to an FNMS conference is like going to a family reunion — we all just start up where we left off. And if we haven't met yet, we will be friends by the end of the weekend! Getting to meet Sarah de Heer and the rest of the crew, whom I had only known through Facebook, was a very special moment. My only wish is that we'd had a week together. There was just not enough time to visit with everyone enough!

Mira really enjoyed getting to see some of the younger kids at the conference. She was very excited to see Raymond again and learned that Peyton was tons of fun in a tiny package. Mira loved the boat trip to Blake Island. Mira also said she really loved the kids' activities while the grown-ups listened to speakers. She said they had a great time!!

Saul was super excited to see his girl, Erica Perry, and to hang out with Raymond. He loved sharing electronic devices with Raymond and Peyton. His all-time favorite part of the weekend was obviously the dance! Although it took him a song or two to get his groove on, once he was dancing, he didn't want to stop!

Silas says he was very excited to get to go to a new state and to see the city of Seattle. He really enjoyed hanging out with other teens from around the country and state. His favorite parts were building stuff with clay and perfecting throwing room key cards around the conference center.



[Julia with Sherman, Carla and Maurice Perry](#)



the event. We often have volunteers and spectators make donations the day of the event as well. A couple of years ago we added a virtual race. People complete the three legs of the triathlon (1/4 mile swim, 20K bike, 5K run) on their own and submit their times to us. For us this was a way for people who are unable to travel to Marathon, Wisconsin, to still participate and support FNMS. Yes, that means YOU can participate from wherever you live! You can even lie about your times because we have no way of checking ...

Each year, we have raised between \$4,000 and \$6,500 for FNMS. Having organized this event for so many years it has become

Many of you know us and our story, but for those of you that don't, I'll recap. My name is Lara and my husband is Dave. We have four kids. Our youngest son, Saul, was born with Nager syndrome in 2004. As all of you who have children with these syndromes know, hearing that diagnosis was kind of a life changer! Thankfully we found FNMS, because the information we received from them helped us immensely. The packet of information and resources that came in the mail was not only useful in educating ourselves, but also the doctors and specialists caring for Saul in those first months. We live in a small town in central Wisconsin. Of the many doctors we saw, only the geneticist had ever seen or heard of Nager syndrome, and that was a case she'd seen in 1980. FNMS was an amazing source of support to our family back then and continues to be today.

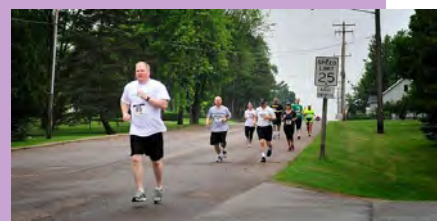
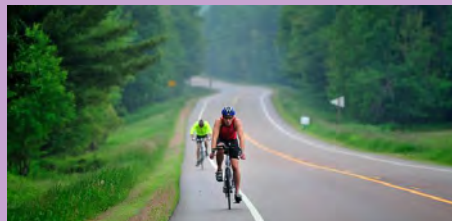
After Saul was home and through the worst of his surgeries, I wanted to give something back to FNMS. I wanted to help ensure that FNMS could continue to support families who receive the news that their child has one of these very rare syndromes. When Saul was two, we decided to have a bike ride followed by a big party. We encouraged guests to donate to FNMS in lieu of bringing gifts to Saul. We were so excited to have raised \$2,500 that day — we were hooked! It was the beginning of an annual fundraiser we organize in our little town of 2,000 residents.

This past June, we held our 8th Annual Ultra-Mini Triathlon (yes, we have evolved!). We have two triathlon events, one very short one and one sprint length. We added a 5K run to our list of races in 2013 at the request of people in the community who are not triathletes. All participants pay a race fee, plus we ask local businesses to be sponsors of

almost routine. Dave and I each have a list of tasks to complete and a general idea of a deadline for each. We have family and friends that volunteer their time. We get a large group of volunteers signed up to help on the day of the event. On the day of the race, we are on top of the world! The excitement, the emotion, the amazing athletes, the love from our community — it is such a special day for us. Part of our routine on race day is to reflect on the day and cry. Really! We usually have family and a few close friends over and we sit around our dining room talking about the day. Undoubtedly, somebody is overcome by the wonder of the day and tears fall. The feeling of doing good and giving back is completely amazing!

As a mother of four kids, one with Nager syndrome, I totally understand being busy. I know that organizing an event like this may seem impossible. I am here to tell you it is possible!! The reason our event has grown each year is because we aren't doing this alone. The very first year was an eye-opener for us. People wanted to help. They brought money. They even asked us to do it again! It was a lot simpler than you might think. Remember that part where I said doing good feels amazing? If you start something good, like a fundraiser for FNMS, people will want to be a part of it. They get a little of that amazing feeling from taking part. It is not difficult to encourage people to participate in something that makes them feel good.

A fundraiser can take many different forms. Our triathlon is a rather big event. We have also taken advantage of other opportunities to raise smaller amounts of money for FNMS. We have sold concessions at a basketball game or track meet when no school group was available to do it. We raised about \$300. We found a local company willing



You can't spell TRIUMPH without some TRI at the beginning and a big UMPH at the end!

to lend us a dunk tank each spring at no cost. We set it up at our kids' school one day in May and any kid that brings \$2 can throw three balls. Teachers, principals, and students volunteer to sit in it, so kids are really excited to throw some balls! This one raises around \$600.

I encourage you to try a small fundraiser. Get permission to set up a stand at a local event or in a mall or busy area. Sell baked goods, crafts, jewelry, whatever you can think of that works for you. Write a letter to local businesses explaining why FNMS is important to you and ask for a small donation. Talk to your school's student council and see if they would be willing to do something to help raise money. Our school just had a dance last night with the proceeds going to a local charity. Have a big birthday party and ask for donations instead of gifts. Find someone who sells catalog products (like Pampered Chef or Lia Sophia) and ask if they would consider doing a party where the profits go to FNMS. The possibilities are endless. What ideas do you have?



The money donated to FNMS supports the foundation in many ways. This newsletter is one way. Paying for the hosting and management of the website is another. Getting packets of information out to new families is yet another. The big one is the family conference that takes place every couple years. My family has attended three of the FNMS

Family Conferences. If you have never gone I strongly encourage you to find a way to attend the next one. It is an experience like no other! Many of us may never meet another individual with Nager or Miller syndrome without an organized event like the conference. We learn new things to help our kids, network with other families, and make friends with folks from around the world. To me, the most satisfying part of the conference is the atmosphere of acceptance. Seeing the kids feel accepted and comfortable enough to "just be kids" is priceless.

To find out more about our event visit www.ultramini.org or like us on Facebook

Marathon Ultra-mini Triathlon Donors

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(December 2011 through December 2013)

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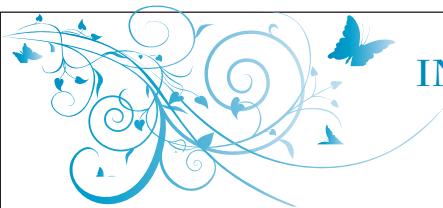
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