



A publication of ISMRD



**ISMRD wins \$10,000 in
Chase Community
Giving Program
See page 20 for details**


HIGHLIGHTS IN THIS EDITION	
From the President's Desk	Page 2
2012 International Scientific/Family conference and other reports	Page 3-6
Pernille Rolls ML III	Page 7
Ivan the Great - Galactosialidosis	Page 9
Life with a disability by Sylvia Webb continued from last newsletter	Page 10
Brian Reese – Alpha Mannosidosis – His Journey through Bone Marrow Transplant	Page 14
New Members	Page 17
Chase Community Grant - Fundraising - Donations	Page 20 -23

The International Advocates for Glycoprotein Storage Diseases

3921 Country Club Drive
Lakewood, CA 90712

info@ismrd.org
www.ismrd.org

501 (c) 3 nonprofit organization
FEIN #52-2164838

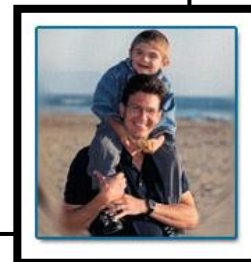


Our Mission:

ISMRD is the leading advocate for families worldwide affected by a Glycoprotein & Related Storage Disease. Through partnerships built with medicine, science and industry, we seek to detect and cure these diseases, and to enable a network of support and information.



From the President's Desk



*By Mark Stark
ISMRD*

I would like to say hello to all the ISMRD Network families and supporters. It has been a very exciting year for our organization, and it has gone by very fast. I was very humbled and honored at the beginning of the year to be asked to be President of ISMRD. I know I won't fill our outgoing President's shoes, but I will do my best to keep up his good work. This summer, we hosted a very successful and most ambitious event in South Carolina. By bringing together researchers and families from around the world; we were able to have a scientific conference, family conference, and natural history study with a tremendous level of participation. I am very pleased to report that the response to the conference was extremely positive, both the researchers and families had very high marks for the organization and content of this conference. I hope you enjoy reading about the conference in this issue of Pathways.

Of course this conference could never have happened without your support. As a very small organization, we are very dependent on our families to continually raise awareness of our children's extremely rare diseases, and to continually find creative ways to raise funds. Our board consists entirely of volunteers, we have no paid staff; and we work very hard to use the funds that are raised to make as many resources as possible available for our families. Of course, I am very fortunate that we have an incredibly resourceful and hardworking board; the amount of work they did for the conference is remarkable. Of course, I must also thank Dr. Sara Cathey; she was the principal researcher for the scientific conference, she was instrumental in our receiving funds from NINDS to support the conference, and she pulled together physicians and other researchers for the natural history study.

Looking forward, ISMRD has ambitious plans. We intend to fund research projects and help direct research funds towards early diagnosis, therapy, and ultimately a cure for these terribly debilitating disorders. We also intend to hold another scientific and family conference in about two years' time. I am pleased to report that through hard work by our board and families, we have been able to raise sufficient funds to commit to some research projects. We will be consulting with our scientific advisory board to select the most promising research studies that could be conducted with this money, and I look forward to keeping you informed of their progress. We will all need to continue to raise funds in any way possible for research. Research is very expensive, we have nine disorders to cover, and there are a number of promising research avenues towards our goal for cures for these diseases.

Finally, I would like to welcome new families to ISMRD. I know how difficult it is to find out that your child or loved one has a rare disease, and how alone and helpless that can make you feel. I hope you find the same sense of hope and family that my wife and I did when we first found ISMRD. Please reach out to us and ask for help, it is our most important work and greatest joy.

Mark Stark, President, ISMRD



2012 INTERNATIONAL CONFERENCE FOR GLYCOPROTEIN STORAGE DISEASES

The 2012 International Conference on Glycoprotein Storage Disorders was our most successful ever. Almost 100 patients, families and Professionals attended from all over the world, including Russia, Norway, Sicily and Australia. 30 medical professionals came from as far afield as Brazil, Belgium and Denmark to talk about the latest developments in research and treatments. There were also many speakers from the Charleston area who spoke on topics close to the hearts of families, including dental care, seizures, palliative care and post-secondary education.

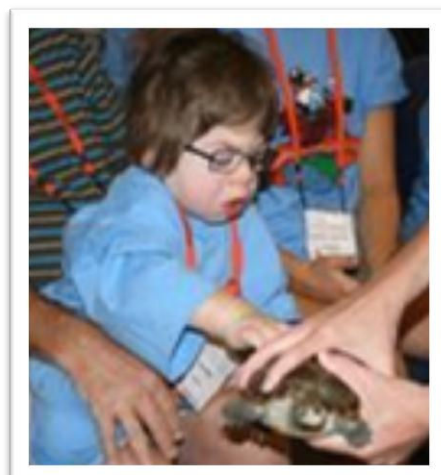
Conference fare included South Carolina specialties such as fried green tomatoes, she-crab soup and baked mac'n'cheese.



*Typical Low Country fare
featured at the conference*

While parents and carers networked and listened to speakers, their affected family members had their own busy program, including a fun day trip to the Charleston Aquarium and the Children's Museum.

Grace Webb (ML II/III) from
Australia pats a turtle at the
Charleston Aquarium



A Memorial Service was held for those who have passed away from a Glycoprotein Storage Disorder. The Charleston sky was filled with white balloons released to commemorate our lost loved ones.



ISMRD honours Jules Leroy for his lifetime of work for Mucopolidosis



Dr Jules Leroy, who discovered Mucopolidosis II, was awarded by Jenny Noble the inaugural ISMRD Lifetime Award, for his devotion throughout his career to work in the field of glycoprotein storage diseases.

Christopher Leonard (Fucosidosis) from the UK turned 21 during the conference.



Following are perspectives from others who attended the family conference and/or the Natural History Study extension. Our next edition will include accounts of the scientific conference from Drs Sara Cathey and Rosanne Taylor, a summary of the very positive results from our post-conference survey and additional family stories from the conference.

Carolyn Paisley-Dew
Board member, Australia



Dr Cathey and Andre Andrews at the South Carolina Conference

The following article was taken from the August 2012 edition of the Helix, the newsletter of the Greenwood Genetic Centre, where Dr Sara Cathey is based.

The Greenwood Genetic Centre (GGC) and the family advocacy group ISMRD co-hosted the Third International Workshop on Glycoproteinoses on July 28-29 at the Crowne Plaza Hotel in Charleston. The glycoproteinoses are the group of ultra-rare lysosomal diseases that include alpha-mannosidosis,

betamannosidosis, aspartylglucosaminuria, fucosidosis, mucopolipidosis II, mucopolipidosis III, galactosialidosis, sialidosis, and Schindler disease.

Both a family conference and a scientific meeting were held, with some combined sessions. An example of the families who attended is the young couple who brought their not yet 3 year old daughter from Norway (Editor: Pernille Roll – read her family’s account below). She was diagnosed with ML III just two months prior to the conference. They arrived still shell-shocked from the diagnosis. By the end of the weekend they had met children and adults with the same diagnosis from around the world. They were able to interact with world class scientists and clinicians, experts in the field of glycoproteinoses. They returned to Norway with knowledge, new friends, and most importantly, with hope.

Another example is the 21 year old man with fucosidosis (Editor: Christopher Leonard) who travelled from the United Kingdom with his parents. He was diagnosed at the age of 6 months because he had an older affected brother, who died at 6 years of age. The parents learned of the work of a researcher from Australia, Dr. Rosanne Taylor, who had demonstrated that bone marrow transplantation was an effective treatment for canine fucosidosis. The parents were able to convince the UK National Health Service to treat their son with bone marrow transplant when he was 9 months old, making him the first person in the world to undergo BMT for fucosidosis. He walks, talks, and generally enjoys an excellent quality of life. He has outlived his brother by 15 years. Dr. Rosanne Taylor remains a leading fucosidosis researcher and presented her latest work during the scientific conference.

Other presentations at the scientific meeting covered the latest research on each glycoprotein storage disease, clinical considerations, transplant outcomes, and the hurdles to cross on the way to effective treatments for rare diseases. Topics addressed at the family conference included nutrition and feeding concerns, education issues, dental care, managing ataxia and seizures, and palliative care.

On July 27th and 30th GGC’s Charleston Office, located less than 2 miles from the meeting venue, held special glycoproteinoses longitudinal studies clinics. The comprehensive clinics included genetics, ophthalmology, cardiology, dentistry, psychology, orthopedics, and neurology evaluations. Thirty patients were seen in the two clinics (alpha-mannosidosis, 7 patients; mucopolipidosis III, eleven patients, mucopolipidosis II, two patients; intermediate ML II/III, six patients;

aspartylglucosaminuria, 2 patients; fucosidosis, 1 patient; galactosialidosis, 1 patient). The data collected will greatly expand knowledge of the natural history of these very rare disorders. Approximately 20 GGC faculty members and staff worked directly in the clinics in Charleston and dozens more GGC folks in Greenwood are involved in some way with the laboratory tests on all the samples collected. The conferences and clinics displayed the commitment and compassion of the Greenwood Genetic Center for an international audience. I am privileged and proud to be part of this organization that demonstrates that commitment and compassion every single day. Thank you, everyone at GGC, for your help.



The children and affected family members head off to the Charleston Aquarium on Saturday 28 July

On Saturday morning all our children, young adults and affected adults lined up for a photo shoot before heading off on their program. At 8.30am the bus arrived to whisk everyone away to the Aquarium, which sits right on Charleston's harbor. We split into two groups and the fun began. The 4D movie experience took us to the Arctic from the view of the sea life. We were misted with water, "things slithered around our legs" and punched our seat backs, and there was even a bit of a stinky fish smell!

Afterwards, we were given a tour of the Turtle Hospital, which has almost a dozen 'patients' in small rehab tanks. The whole group then toured the Aquarium proper; fish, reptiles, amphibians, and birds. One of the major highlights of the day was the classroom time. The wonderful Aquarium staff brought out turtles, lizards, snakes and even a small alligator for everyone to see and touch.

By mid-day stomachs were grumbling so it was time to stop and have a great deli lunch (some even ate two!!). The day wasn't over as we headed to the Children's Museum for a short visit. Most of the group enjoyed the hands-on experience there, while others were happy to sit and socialize with each other.

Soon it was time to return to the hotel. With Kevin, Carl and the driver Mike handling the loading/unloading duties, the tired but happy group was soon back at the hotel eager to tell their families about their exciting day's experience.

We mustn't forget all the wonderful volunteers who came to support our children and adults during their activities. Without this very generous support we would not have been able to provide such an exciting program. Thank you everyone who assisted us

Susan Kester, Children's Program co-ordinator

PERNILLE ROLL



Pernille Roll is 3 yrs old, was recently diagnosed with ML III and came from Norway with her family to attend the 2012 International Conference for Glycoprotein Storage Diseases. Here her father talks about their experience

After over nine months of studies and examinations due to changes found on the skeleton on our youngest daughter Pernille, we got the final diagnosis of what she is struggling with in March 2012. The outcome of the study was much more serious than any of us could imagine. The diagnosis of our little Pernilles disease was ML III.

Having digested this sad message we immediately started searching for information about the disease, information about the possible consequences of what the diagnosis will mean for Pernille and those closest around her. The search for expertise in this area was being done both in Norway and elsewhere in the world wide. With good help from close friends we started an intensive search on the net to define recourses and expertise in the area. We found Jenny - on the other side of the world.

The warm loving welcome from Jenny Noble felt good. We were not alone. There was an entire community out there who embraced us as a family. We got information, and quickly also an invitation to go to Charleston, US in July. Since there are no other person with ML III diagnosed in Norway - or Scandinavia for what we know, the knowledge in this field in the healthcare system and among doctors here is very poor. We decided soon to book our tickets. As a family we were very unsure of what that would meet us when we began our long journey to Charleston this summer.

It is not easy to describe the impressions and the feelings we were having after participated in our first ISMRD conference. It is all pretty new to us. We are still getting used to the idea and the fact that Pernille's future will be different, challenging, and also painful and hard for her. It was rough to face the future. It was hard to accept the facts. But we have no choice. The conference was a fantastic event. Pernille spent her days in childcare, apparently unaffected by the fact that all communication took place in a language she did not understand at all. She was exceptionally good taken care of. Mum & dad were being educated.



After coming home again and processed all of our impression, we are left with greater understanding about the disease(s) and we have learned a lot about ML III, both good and bad, just by participating these few days. We now understand and have more knowledge about what happens in Pernille's body, and how her development can and will be in the future and what challenges she and we together will face. Although each child with this disease is unique and the disease may and often do proceed differently from person to person, there is a lot of things we see is common. With the knowledge we now have, we can inform our doctors here at home about what will happen and in what areas they need to follow Pernille thoroughly on. This will help her further. As a direct consequence of Dr Cathys natural study and examinations by Dr Davids and Dr Leroy, Pernille now is scheduled to hip surgery in beginning of 2013, and she is testing for carpal syndrome this fall. We, and our doctors, will also be so fortunate to be supported in the future by the foremost expertise in the area by doctors attending the conference in Charleston.

Besides the knowledge and ability to meet with the premier expertise in the area we got throughout the conference to establish good friendships with other affected children and their families world wide, including the ML III families, which we put tremendous appreciating in. Being able to talk to others who have been in this position longer than us and who can provide advice and support is just overwhelming and fantastic. We would again like to thank you Jenny for inviting us to this conference to meet the rest of the ISMRD family. And thank you all for welcoming us in! We hope to keep in touch through email and Facebook, and look forward to see you all again at the next conference!



“IVAN THE GREAT”

Hello everyone my name is Ashley Pauls. The mother of an amazing 3 year old named Ivan. My little “Ivan The Great” was diagnosed in January 2011 with Galactosialidosis and am told it is EXTREMELY rare. I will never forget the day I found out his diagnosis and on top of that, that there is no cure or treatment for it. I felt like my heart was ripped right out from my chest. The whole world and how I perceived it changed instantly. I viewed the world and people in a whole new way.

This past year has been tough for us, as I am a single mom and trying my best for both Ivan and Melody. Melody is Ivan's big sister and does not have the disease. She is a great big sister to him. She understands all that is going on with him but I did not explain to her the end result in this disease.

Since Ivan was a baby I knew in my heart that something was not quite right with him. I continued to take him to doctors and insisted something still wasn't right. I couldn't put my finger on it but I just knew there was more to it than just little things here and there. I had a feeling that there was something bigger going on to because of all these different things. But never had I imagined it

would be something like this nor had I ever thought maybe just maybe it's something that will slowly end his life. Ivan is showing subtle symptoms but in time his symptoms will worsen. I just don't know how soon it will all happen. His internal organs will become enlarged; he will have back surgery in the near future because of the deformities and all his mental abilities he gains as he grows he will also lose as he grows. The scariest part is I am unsure about his future. I feel like we're going through this blindfolded because the doctors don't know much about the disease because Ivan is maybe one of three with this disease and probably the only one in the U.S. and I am aching to speak with anyone else going through this.

Ivan and I attended our first conference with ISMRD in South Carolina and it was unbelievable! Very informational and with all the other families there, we all could understand each other's pain, frustration and concerns. It felt more like a family and Ivan and I felt very comfortable there. Its more than an organizational support group, IT'S A FAMILY!! And I am glad to have gotten the chance to meet so many great, sincere doctors and so many beautiful children and their families.

What I have realized in the end through all of this: doctors, worrying, crying, praying and striving, I refuse to let this disease take a hold of our lives. It will not make our lives miserable or sad. It will not stop him or us from anything. And we will live each day filled with love, laughter and beautiful memories in our hearts. I cherish the most precious moments like when Ivan and his sister laugh so hard at each other they can't stop laughing or when he tries something difficult for the first time and triumphs it after a couple tries. He is a joy to our hearts and lives. I would not change a thing about my little man; I love him just the way he was born. He has made my life that much brighter and made me realize that there is nothing that important to live your life mad over, "Ivan The Great" has touched hearts and will continue to do so and make a change or difference in this world little or big. An idea, or vision always starts as a seed.....



Family Corner



Sylvia and family in 1999

LIFE WITH DISABILITY – IMPACT OF DIAGNOSIS

**By Sylvia Webb
Australia**

Sylvia Webb is an Australian parent with five children born with ML II/III and two without. She is a Christian and lives in Melbourne, Victoria with her husband Charles and her daughters Tegan and Grace. This article is the first in a series of subjects Sylvia has presented to Charles Sturt University Speech Pathology students over the past seven years to help them understand what it is like to be the parent of a child with a disability. Other subjects will be reproduced in forthcoming ISMRD newsletters. In this article, Sylvia describes what it was like for her family when her first two children were diagnosed with Mucopolysaccharidosis II.

News, whether good or bad, affects each of us differently. When a decision needs to be made to accommodate that news so that a person's life will be altered with the least amount of grief, a person will look at their experiences, beliefs, supports systems, etc and then make their decision. In my own life I have found that the same news at different times in my life did not necessarily result in the same decision.

I became a mum a couple of months after turning 24 years of age. I was filled with excitement and terror about being a mother. I knew that I could take care of a baby but I did not have the confidence that I would be a good 'mum'. And then it finally happened. At times, prior to this, I had experienced nagging feelings, for many reasons, that perhaps motherhood would escape me, especially since most of my sisters, who were all younger than me, were either pregnant or already mothers, a couple of them long before I had our first daughter.

Jade was born around three o'clock on a cold but sunny Friday afternoon in March 1982. Her birth had been induced after two blood tests had shown lowered hormone levels and I had not felt much movement over the previous two weeks. My labour and delivery were fairly standard although examination of the placenta after delivery showed a massive clot and evidence that only the outside ring of the placenta had been attached to the wall of the uterus. So we were lucky that Jade had survived to birth.

She was long and thin, almost scrawny, and of course I thought she was so beautiful. All I saw were her long fingers and pretty round face poking out of the bunny rug and crackling foil tissue paper that she was wrapped in to maintain her temperature. I imagined her growing up to be a piano player with those long fingers easily reaching across the keys.

I could never have imagined on the day of her birth or in the days to come how different her story would be.

Fast forward my life to April, 1984. Jade had turned two only a couple of weeks earlier and Tegan, our second daughter, was five months old. On the two-hour drive from our hometown of Lithgow to Sydney, I told Charlie that I suspected that whatever was wrong with Jade, Tegan had as well. I had observed even in those short five months that her development had been delayed.

Two days later, we were sitting in a small room in the Royal Alexandra Hospital for Children in Camperdown, Sydney. Two of the walls were panelled from the floor to half way up the wall and then glassed to the ceiling, so as Charlie and I sat there with the doctor I could see the children's beds in the ward outside. The doctor, who we had only met once previously, sat across from us with his back to a third wall with what seemed a very large desk for such a small room between him and us. The girls had had blood and urine samples taken and x-rays performed the day before and we were there to get the results.

Disbelief! Shattered! Confusion! We were a mixture of emotions but there were no tears or hysterics. How can this doctor be talking about our beautiful little girls? The words he's using, I'm sure they don't describe our daughters. It's one thing to finally have an answer to why Jade is not progressing, but to say that both girls are the same and use words like stunted growth, recurrent ear and chest infections, heart abnormalities, enlarged liver and spleen, hearing and sight problems, unstable joints, clawed hands, limited movements in all their joints, progressive delay in their physical and intellectual development and shortened life expectancy. Progressive degeneration; the words just reverberated over and over in my mind.

He gave us no title for what was wrong but told us that children with this disease don't usually walk, don't usually talk or have limited speech. They don't usually become toilet-trained, have many independent living skills such as feeding, etc. or go to a normal school. They remained very small, had organs affected in various ways, were often sick with recurrent ear and chest infections and both of the girls were affected. It all sounded a bit much. After I asked how long they would live, we were told that children with this disease were extremely lucky to reach their teen years and that they most often die from pneumonia or congestive heart failure.

Teens: that is ten to fifteen years. Hey, that's miles away. The girls are only two and a bit and five months old. Lots of things can happen in that time. I do remember being optimistic. But, I did not know if I could or even wanted to cope with this. I had known deep down that there was a problem and that possibly Tegan was affected as well, but he was describing a nightmare. We weren't devastated, that came later. Charlie sat quietly while I asked lots of questions. O.K. what options do we have? The doctor talked with us about having more children and told us about the options that were open to us.

There was having children of our own – well that was what we had always planned. Each pregnancy now carried a one-in-four risk that the baby would be born affected with the same disease as Jade and Tegan. There was artificial insemination by a donor. Mmmm, I don't know if I like that idea. I always thought that if you marry someone, you have their children. Charlie definitely did not like this option – having another man's baby. That really threatened his manhood.

There was adopting. Boy, I wonder how we'd go with that. An aunty and uncle had gone through adoption and had had to wait ten years before they were matched with a baby. I don't know if I could go through that process. Especially seeing I don't have any trouble getting pregnant myself. We could try fostering as an option. I wasn't sure that was such a good idea either. Often with fostering you end up with someone else's problems. We now had problems of our own to deal with. I didn't think that another person's social problems were what we wanted or needed in our lives on top of everything else.

And lastly, there was always institutionalisation. We were told that if we thought we wouldn't be able to cope with caring for the girls, and all that would entail, then the doctor would help with the process and we could have the girls placed in a home to be looked after by the state. Boy, were things going to be that bad? Hang on! Sorry, that's not really an option for us. The words were out before we even thought about it. Both of us felt the same. These are our daughters and our responsibility. Regardless of the outcome, we would handle it.

What about treatment? Lots of treatments had been tried but they had either failed or, are even now, in the very early stages of experimentation.

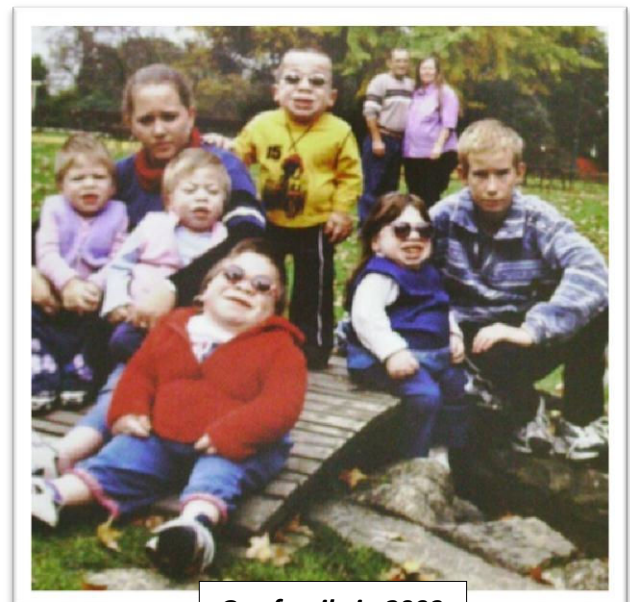
I remember asking about early intervention therapy and whether the doctor thought it would help. I didn't tell him I already had them enrolled in a Program. We were told that if we could teach the girls something that would lessen our work load then by all means go right ahead.

That was the end of the interview. Everything was matter-of-fact. Questions we asked were answered and we accepted what we were told. We had no reason to believe our life would be any different to what he told us.

The trip home was one of questions like, "What did you think about...", "What did he mean by...", "What about...". Most of these were Charlie asking me. Because I had eight years nursing experience, I *obviously* had all the answers.

We had two beautiful little girls, the apples of our eyes who now had a death sentence hanging over their heads (and ours). So what do we do now? Institutionalisation is out; experimental treatments are out; stimulation therapy is in; treatment of infections, etc, as they occur is in; hope is in; life is in. Quality of life, that is, because there sure as heck was not going to be much quantity.

When we got back to Lithgow that day, we went to see each of Charlie's family members and told them about our trip and all that had transpired. Later, after Charlie had dropped me and the girls off at home, he went back and told his family that he believed that the doctor was lying, that there was nothing wrong with the girls and the doctor didn't know what he was talking about. I started to understand that we did things differently. We



Our family in 2003

processed things differently or at a different rate, which turned out to be a good thing at times because it meant that when I was down Charlie was able to support me and when he was down I could support him. We started to complement each other. A month after speaking with our doctor we received a phone call with the name of the disease that our Jade and Tegan were affected by - Mucopolysaccharidosis Type 2, also known as Leroy's I (Inclusion) Cell Disease.

In the medical literature which we started to receive in the following months I found out that children with this Syndrome grow to the size of nine month old babies, don't usually walk, have very limited speech, have many medical problems and often die by the age of 5-6 years of age. Also, the pictures with this literature showed features of the child becoming increasingly 'coarse', in crude language 'ugly'. My children were going to look 'ugly'.

My childhood was one of being raised in an immigrant family. Both my parents came from Holland in their late teens. I remember feeling 'different' at school. The other children poked fun at what we had on our sandwiches, the way our parents sounded when they spoke and although I went to a good catholic school and church, I was sometimes confused by some of the behaviour my mother in particular displayed during my upbringing. I remember walking down the street one day and her dragging us across the road because some intellectually disabled men were walking on the side that we were walking.

I have said in the past that people make their decisions based on their experiences, beliefs and their support networks and that as these things change throughout their lives then when faced with the same situation they will not necessarily make the same decision, particularly if things change. So it was with us.

Years before, I was faced with a pregnancy that had a high risk of producing a child with physical defects due to undergoing a pelvic procedure using radiation at a crucial stage of foetal development and I chose to terminate the pregnancy. The diagnosis of Jade and Tegan occurred after they were born and so our decision was mainly involving whether to keep them or place them in care.

When I was pregnant with our twins we were told that the prenatal diagnosis showed one twin who was unaffected and the other twin was possibly unaffected but this could not be guaranteed as the results were slightly elevated. Later, I wondered if it was explained this way so that I could carry on with the pregnancy with at least some hope.

After the twins were born my husband and I became 'born-again' Christians. We took the stand that if we were going to believe that God was all-loving and would watch over us and provide for us then it was all or nothing. So we chose not to have any of our subsequent five pregnancies tested.

As I look back over the past 35 years of child rearing and all that entails, I have no regrets. I believe that all my decisions were made based on the knowledge I had at the time, that they were 'educated' decisions. Some were harder than others. Some had lasting, perhaps negative effects. I own all of them.

Brian Reese

Alpha-Mannosidosis and Bone Marrow Transplant

**By Jean Reese
United States**

On January 2, 1997, my 3-year-old son, Brian Reese, received the devastating diagnosis of Alpha Mannosidosis. My husband and I were obviously shocked and saddened by this news. We were initially told that there was no cure, no treatment. Take your son home, love him, enjoy him, and keep him comfortable as best you can. There was little known about Alpha Mannosidosis at that time. We were told that children with this diagnosis have an unpredictable life expectancy. There is no way to know if a person will survive childhood or live into adulthood. The future that we (that ALL parents) dreamed of for our son was no longer possible. We didn't know where to turn.

Initially, my husband and I were barely able to function. We had to find a way to continue working and raising our 2 children; Erika age 6 and Brian age 3. We quickly became consumed with trying to find out as much as we could about Alpha Mannosidosis. The Internet was just beginning to find its way into people's homes in 1997. We were fortunate that I had Internet access through my position as an IT professional. I was able to connect with Paul Murphy and Dag Malm, the 2 people we credit with leading the charge to create a supportive community for families facing this diagnosis. Their mission was to increase awareness for the need to find a treatment and/or cure for people who suffer from Alpha Mannosidosis.

Through this connection we learned that research with gene therapy was in the preliminary phases. With a disorder with such a low incidence (1:500,000) the research dollars were just not flowing in fast enough to find a cure. We knew that any damage done to Brian's physical or mental capabilities could not be reversed. We felt like we had this enormous pressure to find a viable treatment as quickly as possible. At age 3 Brian was already showing signs of the disease: developmental delays, fine motor impairment as well as speech and language delays. Paul Murphy pointed us in the direction of Dr. Krivit at the U. of Minnesota hospitals in Minneapolis, MN. Dr. Krivit was a leader in the field of Bone Marrow Transplant research for children with genetic disorders.

Brian, my husband, and I made the 14-hour trip to visit with Dr. Krivit and Dr. Charlie Peters in Minneapolis. They were brutally honest with us, indicating the survival rate of 60% for non-related bone marrow transplants. Unfortunately, none of us were a suitable match for Brian. We turned to the National Marrow Donor Program and were blessed to find a donor. We were told that Brian's donor was a 42-year-old man, Paul, from the Boston area. We are eternally grateful for his selfless gift of life, as well as the American Red Cross and the numerous blood and platelet donors.

With the incredible support of a medical team at U of MN hospital and the aftercare here in Ohio from Brian's pediatrician, countless specialists and the staff at Nationwide Children's Hospital, Brian

survived the transplant journey. In a 13-month period he needed over 30 blood & platelet transfusions, spent over 70 nights in the hospital, made nearly 250 clinic/doctor visits and lived at the Minneapolis Ronald McDonald House for six months. He was in relative isolation and wore a mask any time he stepped outside the safety of our home. There were 3 different occasions when Brian was gravely ill from the transplant process and we were told to prepare ourselves that he might not survive. Somehow Brian pulled through each of these crisis situations, somehow we all did.

Brian does suffer some long-term complications from the BMT process. He has lens implants in both eyes to correct cataracts caused by the radiation and steroids. He has hearing loss caused by the toxicity in the drugs used to save his life when he battled pneumonia (twice) post-BMT.



Erika and Brian, July

When Brian's health stabilized, at age 5, he entered the world of public education. We were not familiar with how school districts are required to educate children with disabilities. We quickly found ourselves immersed in the IEP (Individual Education Plan) process. With the support of caring and knowledgeable academic professionals (and with the support of an advocate) we were able to secure the educational supports Brian needed to be able to access a free, public education. Our collective goal was to provide Brian with the supports he needs with the ultimate objective being for him to be as independent as possible. Brian is an outstanding student. He has the basics covered; never misses school and always completes his homework. School is hard for Brian. His disability makes the routine tasks of taking notes, studying for tests, and writing papers exceptionally challenging and time consuming. Brian has learned how and when to leverage the support of tutors and teacher assistance. We are so proud of his level of determination and never-quit attitude.

Sports are where we see his enthusiasm. Brian has played all sports (with a passion for soccer) since age 5. He is an avid snow skier and loves to wakeboard. However, he does struggle with physical limitations due to his Alpha Mannosidosis and the chemo/radiation he received during the BMT. As kids his age hit puberty and grew bigger and stronger, Brian leveled off. Despite 9 years of daily growth hormone injections, Brian has reached an adult height of 5' 0". We know that this much growth is a success for a child with a lysosomal storage disorder. Brian sees this in a negative light; just another way he is different from his friends. We empathize with him. The teenage years are difficult for many people, and he is no exception. We pray that he will see these differences in a more positive light when he is an adult.

Fast forward to September, 2012... Brian is fully immersed in his senior year of high school. He continues to enroll in general education curriculum, with an interest in business classes. He is a member of his high school soccer team. Despite having to endure painful joints (which are likely

due to his genetic disorder), he dutifully ices his back, knees, and ankles after every practice and he happily returns to the playing field the next day.

Brian doesn't want to miss out on anything in life. He has watched as his sister has had her ups-and-downs as a college student. He wants to have that opportunity. As his friends are beginning their college search, so is he. My husband and I are spending time sitting in the Admissions Office on college visits with Brian. We had never allowed ourselves to dream of a college experience for him. Brian proved us wrong! None of us know what his potential college life will look like. We will take this one step at a time. Knowing Brian, he will find a way to make this work. He continues to be an inspiration to us.

We feel exceptionally blessed to have the unconditional support of our family and friends. They were part of this journey, propping us up when we needed it and helping us celebrate the many milestones along the way. At a time when the world felt like it was closing in, there were many new paths laid out before us. We were introduced to people with indescribable medical intelligence, patience and passion for helping people like Brian.

This experience has changed all of us in ways we never imagined. Erika, Brian's sister, is pursuing a degree in the medical field. She is interested in working in pediatric hematology /oncology. I made a career change from business to education. I am an Intervention Specialist and work with children with special needs. My professional life has helped us understand Brian and support him in ways we did not know were possible. My husband is a math teacher and he has also been an active part of Brian's academic success.

Receiving a diagnosis of a life-threatening genetic disorder has had a significant impact on our family's life. All families who face this same situation understand how overwhelming the feelings of helplessness and fear can be. Fortunately, the ISMRD has evolved into an organization that is leading the charge to support research, support families, as well as increase public and professional awareness for MPS and related disorders. We respect and appreciate the dedication of the leaders and members of this group who continue to advocate for these children in need.



**Brian playing soccer
15 years later,
September 2012**

NEW MEMBERS

We warmly welcome to our Penguin Family

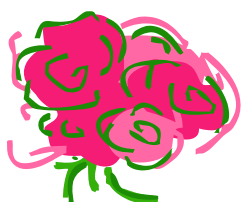
- Fahad Alhosani and his son Khalifa (2) who has Fucosidosis. They live in the United Arab Emirates
- Sherrie Armour, whose children Dakota and Savahna have ML III (USA)
- Antonella Di Gori whose son Julian Biasi (one year old) has ML II (Australia)
- Marta Diaz, whose children Pedro and Javier have Alpha Mannosidosis (USA)
- Erin Ginn, mother of Eryana Nicole Jones who had Sialidosis (USA)
- Smith Lambert and Sanjare Ferdinand. Smith has Late on-set Sialidosis type I (USA)
- Truls and Birthe Roll and their daughter Pernille (3) who has ML III (Norway)
- Julia Taravella and her children Daniel and Alexander who have Aspartylglucosaminuria (USA)
- Adina Trestianu and her two year old daughter Bianca who has- ML II
- Sadia Zaidi and Talha Zaidi who has Alpha Mannosidosis (USA)
- Juanita Van Dam and her children Damian and Jesse-Rose, who have ML III (Australia)
- Lori Smith and Katelyn who has ML III (USA)
- Parker Meador, Mark Barham, Heather Anna, Majorie Claire, Vivi Roll



Some of our Penguin children and young adults have recently had surgery or are on the waiting list for surgery

Your Penguin family are thinking of you and Praying for a good outcome

- Katelyn Smith
- Sarah Noble - a successful shoulder replacement
- Lonnie Tice - open heart surgery on 8 October
- Sean (Kester) - knee surgery and awaiting a second operation
- Sarah Burgess (ML III), UK - knee replacement
- Lori Smith - hand surgery
- Skylar Thomas - back surgery in October
- Callie Nagle - hip surgery in October
- Sean (Parks Hoffman) - Knee Surgery
- Meg Rust – Carpal Tunnel surgery on both hands



**The ISMRD extends
its deepest sympathy
to the family of Etko,
who passed away in
July this year**

Research Corner



Workshop on Natural History Studies of Rare Diseases: Meeting the Needs of Drug Development

From Genzyme Rare Community newsletter, September 2012

If there is one common denominator that binds the rare disease community together, it's the importance of having a natural history of a rare disease. Having a solid understanding of the dynamics behind a particular rare disease can foster an interest in research culminating in the development of a therapy.

On May 16 and 17, 2012 many individuals representing the diverse sectors of the rare disease community gathered at the National Institutes of Health Campus in Bethesda, Maryland to learn and discuss how to facilitate a collaborative process for developing rare disease natural histories. Knowledgeable speakers from academia, research institutions, patient advocacy organizations, NIH/FDA, and industry shared their expertise and opinion on what a framework for natural history collaboration could, should and would look like. Their presentations sparked thought provoking questions and discussion. A link to the full agenda and presentations speaker slides has been provided and can be found here:

https://events-support.com/Documents/Agenda_NHS_Final.pdf.

One participant remarked how gratifying it was to hear that all the speakers were "in sync" with each other on the necessity for all stakeholders to get behind a natural history movement. This attendee was especially pleased that the opinions and concerns of the rare disease patient community were well represented and articulated in a rare disease patient advocate.

Certainly the discussion surrounding the need of rare disease natural history should and will continue. Garnering reliable information on how this vital topic pertains to each rare disease organization is necessary so that a constructive and meaningful discussion can be had among the entire rare disease community.

Sponsors of this meeting were the NIH Office of Rare Disease Research (ORDR), the National Center for Advancing Translational Sciences (NCATS), National Institutes of Neurology Disorders and Stroke (NINDS), National Institutes of Health Clinical Center, Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), the FDA, the FDA Office of the Commissioner, FDA Office of Orphan Product Development (OOPD), and the FDA Center for Drug Evaluation and Research (CDER).

DONATIONS AND FUNDRAISING



CHASE COMMUNITY GIVING GRANTS \$10,000 TO ISMRD

ISMRD has received a grant of \$10,000 from Chase to use towards our mission of advocating for glycoprotein storage diseases.

This year, Chase provided \$5 million through its Chase Community Giving program to the charities who could raise the most votes. This money was divided among 196 recipients, including ISMRD.

ISMRD received double the number of votes this year that it received last year, up from 700 to 1400. This resulted in the grant to us of \$10,000! THANK YOU CHASE!!!

Chase Bank has awarded over US\$28 million in grants to over 8000 charities in the USA, including all 50 states, plus Washington D.C. and Puerto Rico.

Chase Community Giving was introduced in 2009 as a way of letting the public determine where the funds go. It's a program that lets fans of Chase Community Giving, and Chase customers, vote to help determine where Chase donates millions of dollars.

See http://www.facebook.com/ChaseCommunityGiving/app_162065369655 for more information

Thank you to each and every one of you that voted for ISMRD and encouraged their family, friends and network to vote for us. TOGETHER WE DID IT!





Dakota's Mom Julie Fuller and Anna James

Rock 4 Dakotah is a wonderful event that I had the good fortune of being able to attend with my husband Bret and my daughter Anna (Mucopolipidosis III) this past September. This yearly event is a fundraiser that is held every September in remembrance of a wonderful young man called Dakotah Smith, who passed away in April 2006. Dakotah suffered from Mucopolipidosis. Dakotah's mom, Julie and her family and friends put hours and hours into a wonderful event that celebrates Dakotah's life and that donates much needed funds to organizations such as ISMRD.

We arrived in Mechanicsburg, PA on the evening of Saturday, September 21st after a long 12 hour drive. Julie had taken care of our hotel reservations for us and had arranged a welcome basket for Anna that just about made her day! The event was held at Appalachian Harley Davidson in a wonderful facility they had at the back of the dealership. The music started at 10am, and was a mix of everything from Veggie tales to some ZZ top! Local bands, such as the well followed Emily's Toy Box donated their time to perform on stage. Vendors such as Avon, and a wonderful local woodworker were selling their wares, contributing some of their profit to the fundraiser. Raffle tickets were selling like crazy and some great deals were to be found at the silent auction. Anna won a 5 pound bar of Hershey chocolate!! We are still debating how to eat it all!

I was amazed and very touched to see just how many people loved Dakotah and his mom, Julie. I know how much hard work went in to making this fundraiser happen, having worked on several myself in the past. Dakotah was looking down on us and obviously took care of the weather for us. The forecast was for gloomy skies and possible rain, but we had gorgeous sunshine. All in all, it was a wonderful time for friends and family to connect and remember a very special little boy. I don't think there was a dry eye in the house as we all joined together to sing together on what would have been Dakotah's fourteenth birthday.

September 22nd, 2012 will be a day I will remember with great fondness. I'll look forward to September 2013 as I do my best to work out getting to Pennsylvania once again to celebrate a very special young man.

Jackie James
ISMRD Board Member
St Louis, Missouri



ISMRD Honor Roll

**Thank you to everyone who provided funds for the ISMRD
by donating, fundraising or grant writing**

<i>Daniel & Katherine Adams</i>	<i>Genzyme Corporation</i>	<i>Carolyn Paisley-Dew</i>
<i>Brian Alters M.D.</i>	<i>Esther & Gordon Gow</i>	<i>Kim Parks</i>
<i>José & Maria Alvarez</i>	<i>Charlette & Ernest Hatch</i>	<i>Frank Parlato</i>
<i>Sherriee Amour</i>	<i>Joy Hayes</i>	<i>Erin Patterson</i>
<i>Elizabeth Arcuri</i>	<i>Adriane Hollon</i>	<i>Karen Patterson</i>
<i>Whigham Avery & PA Winesett</i>	<i>Bill & Dona Holzman</i>	<i>John Payne</i>
<i>Nancy Baker</i>	<i>Carolyn Holzman</i>	<i>Judy Rompf</i>
<i>Linda Barham</i>	<i>William Holzman</i>	<i>Robert Royston</i>
<i>Kim Bedell M.D.</i>	<i>Ruby Howard</i>	<i>Scott & Greta Salhus</i>
<i>Dan Bindman & Norma Edwards</i>	<i>Jackie James</i>	<i>SGL Carbon LLC</i>
<i>Dennis & Dianna Bisnar</i>	<i>Thomas Johnson</i>	<i>Patrick & Kim Silva</i>
<i>Marilynn Bodnar</i>	<i>Susan & Carl Kester</i>	<i>Linda & Chuck Sloan</i>
<i>Patti Bowman</i>	<i>Mary Kimmet</i>	<i>Mark Stark</i>
<i>Harry Budiono</i>	<i>Betty King</i>	<i>Tania & Elie Suares</i>
<i>Shuangping Chen</i>	<i>Terri Klein</i>	<i>Ben & Annette Sy</i>
<i>Cristina & Francis Coffey</i>	<i>Ned & Elizabeth Kotick</i>	<i>Brian & Alice Takamiyashiro</i>
<i>Linda Comins</i>	<i>Thomas & Linda Kovessy</i>	<i>William & Sandra Thomas</i>
<i>Penny Connell</i>	<i>Stan & Carol Kulesza</i>	<i>David & Pam Tobey</i>
<i>Donna & Stanley Cravens</i>	<i>Geraldine Kunstadter</i>	<i>John & Denise Trauger</i>
<i>Allan CUILTY</i>	<i>Nicholas & Barbara Link</i>	<i>Anne Turbridy</i>
<i>Mary Anne CUILTY</i>	<i>Kiley & Tammy London</i>	<i>Ultragenyx Pharmaceutical Inc</i>
<i>Kathy & Brad Davis</i>	<i>Kristen & Steve Luna</i>	<i>Thien Van & Kim Phi</i>

David & Leslie Dewinter

M & M Durham

Erik & Susan Duthe

Christina Duthie

Charles & Sharon Emerson

Shelby & Jan Essman

Charles & Cynthia Ewers

Loughridge Farm

Kathleen Gann

Gates Family Trust

Kevin & Andrea Gates

David Gayl

Colin Mackay

Marina Oil Services

Laurie & Stan Martin

René & Susana Martinez

James & Mary Martorana

Grace & Paul McAllister

Mark & Karen McEniry

Gregory & Sherry Michael

Gerald Middleton

Robert & Paulette Montelone

Jerry Montoya

Howard Vincent O'Connell

Vantage Credit Union

Connie & Sergio Villasenor

Robert and Melody Voltz

Raymond & Helen Wells

Ed White

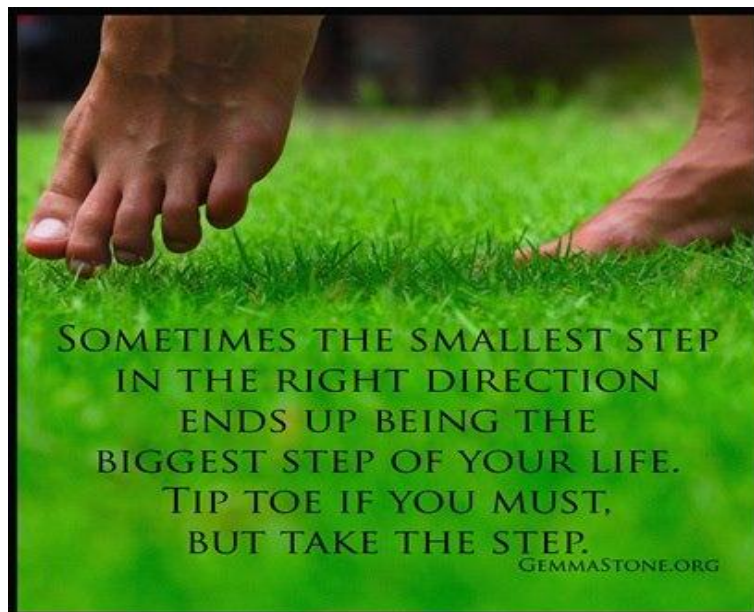
Michael & Carla Whitt

Jason & Magdalyn Williams

Caroline Williams

Carol Winger

Jamie & Ronni Young



ISMRD are the International Advocates for the following disorders: *Alpha Mannosidosis, Aspartylglucosaminuria, Beta Mannosidosis, Fucosidosis, Galactosialidosis, Mucopolipidosis II (I-Cell Disease), Mucopolipidosis III (Pseudo-Hurler Polydystrophy), Schindler Diseases and Sialidosis*



Contacting ISMRD

ISMRD Board of Directors

President: Mark Stark
Vice President, Administration: Jenny Noble
Vice President, Fundraising: Pam Tobey
Vice President, Research: John Forman

Directors

United States: Jackie James | Andrea Gates | Susan Kester |
Melissa Rust | Tish Adkins
Australia: Carolyn Paisley-Dew

Founded in March 1999

Postal Details 3921 Country Club Drive
Lakewood,
CA 90712,
United States

E-mail: info@ismrd.org
Website: www.ismrd.org | FEIN: 52-2164838

ISMRD'S SUNSHINE CARE Committee



ISMRD has a group of parent volunteers called the "**Sunshine Committee**". Our purpose is to coordinate support for families in need. The type of support varies on the circumstance -- from birthday and weddings, an illness or death in the family, or a family experiencing surgery or a medical crisis. In any case, we provide a little "sunshine" for the family by providing flowers, encouraging messages via email, cards or a phone call -- whatever we think the family would find most helpful. In order to help others, our group relies on the support of all families because, in essence, we are all part of the ISMRD "Sunshine Committee".

If you are in need of assistance or know someone in our Penguin community who is in need, **please contact Susan Kester**. She will coordinate with the appropriate parties to determine how we can best help.



Feedback Form | Donation

ISMRD would like to hear from you! Send us your feedback, your request for further information or make a donation. Just fill out the appropriate boxes below, cut out this page and then return.

Donations: contributions to ISMRD are tax-deductible in many countries. Consult your nation's local or central tax-collection agency. A copy of our current financial statement is available upon request by contacting ISMRD at our address at **3921 Country Club Drive, Lakewood, CA 90712, USA**. Documents and information submitted to the State of Maryland are available from the Office of the Secretary of State or the State Licensing Department. Please contact us for further information.

Tell us how you can help! We would like to hear from you and offer you a part in our vision to link families, support research, develop therapies and find cures.

- Send us names and e-mail addresses of family, friends, and professionals who would be interested in receiving our newsletter or who want to know more about our mission.
- Tell us what you can help us with
 - Fundraising
 - Publicity and communication
 - Do you have any other ideas or other ways that you can help ISMRD?

Name: _____

E-Mail: _____

Please help our Cause

ISMRD is a 501(c) charitable organisation based in the United States serving a global constituency. We provide our services, which include our newsletter, website, outreach activities and support of research, without requesting monthly dues or any other financial restrictions. We gratefully accept donations that will enable us to continue toward our goal of a future free of the tragic consequences of Glycoprotein Storage Diseases.

Yes I would like to Contribute the following (check one)

\$100
 \$75
 \$50
 \$25

Please Make Your cheque payable to **ISMRD** Thank you

Please give us your name & how to contact

Name: _____

Street: _____

Street 2: _____

City/State/ Province: _____

Country/Postal: _____

E-mail: _____

