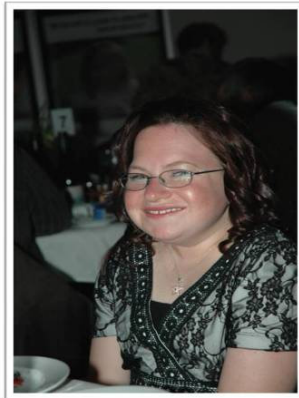




A publication of ISMRD



Sarah Noble's painting accepted for Genzyme Expression of Hope stage II
See page 4



Dr Emil Kakkis and Spencer Gates at ISMRD's Charity Auction Dinner - See page 3

HIGHLIGHTS IN THIS EDITION	
Message from the President	Page: 2
ISMRD Annual report adopted by Board of Directors for the 2008 year	Page: 3
ISMRD and GGC – A positive partnership for research	Page: 4
An Evening of Penguin Dreams – Charity/Auction/ Dinner May 29 2009	Page: 5
NIH Announces New Program to Develop Therapeutics for Rare and Neglected Diseases	Page: 13
A Snapshot of David Mackay	Page: 14
In Memory of Kelley Crompton	Page: 15

The International Advocates for Glycoprotein Storage Diseases

2922 Deerford Street
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 John Forman
President ISMRD*



Hello to the ISMRD network.

This second edition of Pathways for 2009 is an opportunity to give you all an update on progress for our organisation, and to let you know that things are now looking a little better for us since the financial and organisational difficulties we experienced early in the year. On the facing page is a copy of the annual report adopted by the board of ISMRD at our annual meeting held in May 2009. This provides a summary of the situation we were at then, as well as a record of our major achievements over the past years.

Back in May this year we faced significant challenges getting our organisation back on track. We were very low on cash and we needed to completely rearrange our operations as we no longer had an executive director to run ISMRD. This involved a lot of work, as I'm sure those of you who have moved home and state will recall the endless tasks of notifying changes, organising mail, accounting, banking arrangements, and more. For ISMRD the process was complicated by our board members being in so many different states and in different parts of the world.

Thanks to the tasks willingly taken on by our board members we have our address changes made, our committees reorganised, our audit completed, our licenses renewed, new accounting and banking arrangements in place, and all other administrative jobs organised. I want to give special acknowledgement of the work of board members Mark Stark, Andrea Gates and Jenny Noble who worked very efficiently as a team to organise many new financial and administrative arrangements for ISMRD. We can now get more focus on our mission of improving family contacts and information, building our networks with researchers and clinicians, and working towards treatments for our diseases.

In this newsletter you will see news of the fundraiser organised in California just a few months ago. Andrea Gates' fantastic organising efforts produced over \$25,000 for ISMRD. This event was an evening of fun and fundraising for ISMRD, but more importantly it got us out of the financial hole we were in and enabled us to face the future with more confidence. Thank you Andrea for your fantastic efforts for ISMRD. The big question is how can we ensure this effort is matched by similar efforts, large and small, to provide us more income in the future.

Other articles in this edition refer to our efforts to promote research. We have supported a second round of assessments for ML patients at the Greenwood center, we have plans to extend that study to Australia and New Zealand later this year, and we are working with researchers to develop a natural history study of Fucosidosis.

It is a tribute to our board that we have been able to maintain these efforts during a time of significant reorganisation. I am sure that the families, patients, friends and researchers in our wider network will be pleased to learn of these efforts and be appreciative of them, and be pleased to know the board of ISMRD is exploring possibilities for the next joint meeting of families and researchers to discuss our diseases.

I'm also pleased to see stories of the lives and achievements of our affected patients. Whether looking at their current work and achievements, or reflecting on those things at the time of their passing, these stories add the essential perspective of the very personal and human challenges and responses to glycoprotein diseases.

With my best wishes to you all,

John Forman
President, ISMRD

ISMRD Annual report adopted by Board of Directors for the 2008 year – including commentary to the date of our meeting 12 May 2009.

The most significant events during 2008 and in the following months up to May 2009, are a mixture of positive and negative.

- The financial and economic situation had a direct impact on our fundraising efforts and we were unable to secure sufficient funds to continue the role of executive director.
- Loss of the involvement of several board members led to continuing problems organising efficient administration for ISMRD, as well as problems with the operation of committees.

Despite these organizational problems we have made some good progress in our research agenda:

- A very successful consensus development meeting was held in November 2008 to work on protocols for use of Bisphosphonate therapy in Mucopolysaccharidosis. A grant of NZ\$40,000 was received from the UK MPS Society to assist with this consensus development meeting.
- A grant of NZ\$60,000 was obtained by Jenny Noble from the AMP foundation to do work related to the consensus meeting, and to help fund the inclusion of Australian and New Zealand families in the natural history study.
- The relationship with the Greenwood Genetic Center has been maintained. Progress, though slow, is continuing with the natural history study. This includes support of US\$20,000 from the National MPS society directly to the Greenwood Center for this study, plus collaboration agreed to by Michael Beck and John Hopwood to extend this study to more countries.
- Connections we had established with researchers lead to a grant of US\$60,000 from the MPS Society to Richard and Jennifer Steet for work on a Zebrafish model of Mucopolysaccharidosis.
- A major grant application for US\$250,000 has been submitted to the NIH via the Lysosomal Diseases Network to seek funds for the natural history study. A decision is expected soon.
- Extensive background research has been done by John on newborn screening decision criteria in preparation for our advocacy aimed at getting Lysosomal diseases included in standard test panels. We are greatly assisted in this area through Barbara's role in newborn screening in the US, and our connections with the screening program in New Zealand. Close connection is maintained with the Genetic Alliance who have a researcher with particular responsibility for policy on this topic.
- ISMRD has maintained good links with a range of Lysosomal research groups and networks.
- Good connections have been established with ICORD, the international conference on rare diseases and orphan drugs. This group includes many regulators, industry, academics and officials from the US and EU, and its agenda includes a range of topics of close interest to us, such as orphan drugs regulations, research incentives, clinical trial requirements, etc.

The success we have had in making things happen since the start of ISMRD exactly 10 years ago should not be underestimated. In addition to items listed above, we have achieved significant support from the National Institutes of Health to hold two major scientific/family conferences in 2004 and 2007, with the combined value of NIH contribution for these two meetings likely to be well over US\$150,000.

However, most of the funds raised towards research and treatments for our diseases, do not pass through our books, and do not provide us with any overhead component. We are more likely to be a

catalyst for funds to go from funders to researchers, rather than have funds come directly to us. Securing administration funds for ISMRD has been a major difficulty for us. It is a significant fact that many donors do not wish to contribute to administration overheads, but another reality is that our ability to continue to advocate for our diseases and act as a catalyst for these research funds to flow, is severely compromised by our difficulties in getting enough money to secure our administration and advocacy functions. That was the major dilemma during the time we employed an Executive Director. Her work assisted this catalyst function well, but was unable to generate the essential administration funds to support her own role and our office functions.

A number of ISMRD families have run fundraisers on their own initiative to supplement ISMRD's own fundraising efforts, or made direct contributions to ISMRD. Over the past three years this has produced close to US\$40,000 on top of the US\$74,000 generated through the Walk/run events organised by Terri. All of these fundraising efforts have provided vital funds for us, and we are very grateful for them, but we have not been able to achieve these at a level sufficient to give us the organizational capacity we would like to have.

We must now revert to an all-volunteer operation and our busy lives and the economic problems make that more of a challenge than it was when we started 10 years ago. The key challenges for us continue to be: How to ensure efficient organisation of our accounts, receive mail, file returns, and maintain our database, fundraising activities, and our communications with families and other interested parties, so that our mission can be achieved.

John Forman
President, ISMRD



ISMRD Welcomes New Board Member

ISMRD is very pleased to welcome Pam Tobey to the board.

Pam is the mother to Autumn who is an MLIII adult. Pam and her husband, David, live in Arkansas and are parents also to another daughter, Michelle. She is a hairdresser and owner of her own salon.

Pam has conducted several fundraisers in her very small town....from baking and selling giant cookies to having gigantic yard sales. She is extremely passionate about finding funds for the ISMRD and dreams of the day when research is "rolling" and a cure found for Glycoprotein Storage Diseases.



ISMRD and GGC - A positive partnership for research

By Dr Sara Cathey



ISMRD has a significant partnership with the Greenwood Genetic Center in promoting research through a natural history study for our diseases. Here's a commentary from Dr Sara Cathey on how this relationship developed and how it is making a positive difference:

In 2005, a family from Georgia traveled to Greenwood, South Carolina, so that their child affected with mucopolipidosis II could be evaluated by Dr. Jules Leroy. Dr. Leroy is a visiting Senior Scholar at the Greenwood Genetic Center. He had reported the first patient with ML II in the medical literature 40 years prior. With that visit an important relationship was established between ISMRD and GGC.

ISMRD advocates for families affected by the very rare glycoprotein storage diseases. Because these diseases are rare, families often feel like they are forgotten by medical science. Little information is widely available to patients or doctors. GGC is a leader in genetic services, committed to diagnosing genetic diseases, counseling families, researching the causes of birth defects and genetic diseases, and educating others. By working together, ISMRD and GGC are raising awareness of glycoprotein storage diseases. Since 2005, this partnership has led to

- better understanding of the usual course of disease with ML II and III
- extensive research of the gene changes that cause ML II and III
- reliable information about the diseases for families, physicians, and scientists
- international collaborations with researchers, all ultimately interested in effective therapies

ISMRD has hosted scientific meetings in the United States and New Zealand, fostering further research and discovery. GGC has hosted special ML Clinics attended by families from the US and Canada, with funding support from ISMRD. Around the world, numerous presentations and publications have made it impossible for medical science to “forget” these diseases.

The ML Project is going to expand. While the momentum continues and strengthens for ML research, the other diseases included in ISMRD’s mission are going to get the same attention by being included in an expanded natural history study.

There is strength in numbers. By unifying individuals with these rare diseases, ISMRD empowers families to tell the world to pay attention, learn, understand, and eventually, cure. Without ISMRD, this amazing progress would grind to a halt.

ISMRD is pleased to report such a positive comment from Dr Cathey and is delighted to reply with our own endorsement of the great interest and support shown by Dr Cathey and GGC.

An Evening of Penguin Dreams

Charity/Auction/Dinner, May 29 2009

*By Andrea Gates
Fundraising Chair*

**ISMRD’s first charity auction dinner was a success,
raising over \$25,000!!!**

The evening began with champagne, appetizers and a Jazz string quartet accompanied with snare drums played by Dr. Richard Blumenthal, Spencer Gates’ pediatrician. Soft Jazz played as people arrived, mingled and scoped out the auction items.

Our mascot, the penguin, was seen throughout the room. A set of 5 foot penguins stood at the entrance to the grand ballroom. Adorable ceramic penguins were on every auction table along with black and white balloons.

The dinner tables were set with black and white linens, vases filled with beautiful orange roses and of course penguins. Also on the tables were our thank you gifts. Since we are an international organization it seems fitting to have a set of crystal glass coasters etched with a globe of the world.



Dr. Emil Kakkis spoke before dinner and had the ear of everyone in the room. He spoke about glycoprotein storage diseases and the importance of research to find effective treatments and cures. Dr. Kakkis spoke of his new venture, “Kakkis Everylife Foundation- No Disease is Too Rare to Deserve Treatment” and that he would like to build a relationship with ISMRD families.

Guests were served a surf and turf dinner of Salmon Piccata and Roasted Tri-tip Chasseur. Dinner ended with a delicious fresh berry crepe topped with cream. Everything was wonderful from the salad, wine, dessert and everything in between.



Guests were entertained by the wonderful singing talent of Tony Odell. His voice was amazing and his choice of music was outstanding. Everyone enjoyed songs from Frank Sinatra, Johnny Cash, Jimmy Buffet, and Billy Joel just to name a few. A power point slideshow played throughout the evening. It was a touching way to remind everyone why we were there and who would benefit from our success.

Lew Cunnington was our auctioneer and emcee for the evening. He kept the audience’s attention throughout the night. Lew did a marvelous job with our live auction and raffle items. His personality and humor entertained guests until the very end of the night. We had well over a hundred auction items ranging from a weeklong vacation home on the beach, chartered fishing trip with Dan Hernandez,

collectables, art pieces, jewelry, a violin, to small items such as movie tickets. There was something for everyone.

All in all, it was a night to remember. This was truly a successful event in spite of our nation's economy. The donations were true acts of kindness and raised without any sponsor ships.

On a personal note, Spencer was thrilled to have three of his doctors, two of his therapists, his parish priest, family, his Principal, classmates, friends and neighbors attend. The room was filled with people who care about Spencer and his future. Spencer had no problem being the center of attention. Our family personally knew everyone in the room which made the evening very special to our family. We are truly blessed.

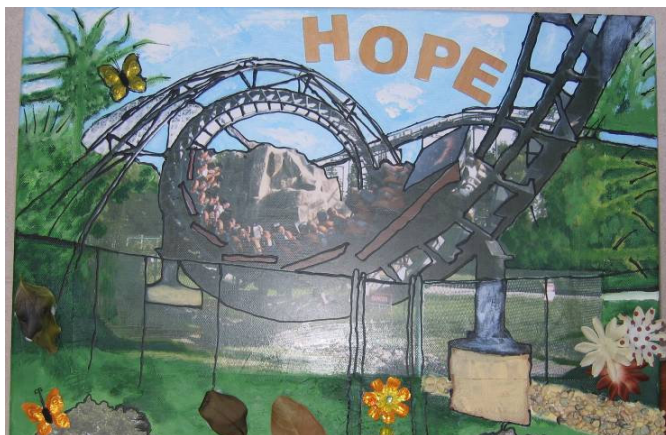
If two people along with some help from family and friends can host a successful dinner, ask yourself what you can do. The possibilities are endless.

Many thanks to those who contributed, both near and far to the success of ISMRD's first Charity Auction Dinner.

Andrea Gates
Fundraising Chair



Early this year Sarah Noble submitted a painting to Genzyme's Expression of hope stage II. ISMRD is very proud to announce that the painting was accepted and will be one of their featured works of Art.



Sarah was also asked to write a little bit about her painting and what it meant to her. Below is what was submitted.

My name is Sarah Noble. My art displays my life living with Mucopolidosis Type III. It is like living on a rollercoaster. Some days I feel wonderful and others I am not so well. Last year I had major cervical fusion and had many highs and lows while I recovered. Although there are no therapies for ML I live with hope that one day there will be a treatment for ML.

Art is my passion and a way to escape this terrible disease. I have a great sense of humor and get along with most people. I love to watch DVD's with friends and chat with friends on-line. My goal in life is to live every day as it comes and help other kids/adults like myself. I would also like to say thanks for giving me the opportunity to paint for the expressions of Hope stage II.



Rare Disease Day 2009: Patient Care, a Public Affair!

The first Rare Disease Day sponsored by EURORDIS (European Organisation for Rare Diseases) was held last year in Europe on February 29, 2008. February 29th was chosen since it is a rare day and it is symbolic of rare diseases.

Building on the success of last year, Rare Disease Day 2009, spread beyond Europe seeing new partners from the United States, Canada, Latin America, Taiwan and China joining in the action, with momentum building in Australia and New Zealand.

This year was the first time Rare Disease Day was observed in the U.S.A. The National Organization for Rare Disorders mobilized more than 200 patient organisations generating great enthusiasm. Two government offices that are instrumental in rare diseases and orphan products in the U.S.A, the NIH Office of Rare Diseases and the FDA publicly rallied their support.

To read more about rare disease day go to.

www.eurordis.org

http://rarediseases.info.nih.gov/rarediseases_day.aspx



A Return Visit to the Greenwood Genetic Center in South Carolina

By Denise Crompton

Three years ago, a number of MLII and MLIII families gathered at the Greenwood Genetic Center in South Carolina to meet with the doctors who are researching these diseases. ISMRD members had worked diligently to obtain funding for the project. In July 2009, many of us made a return visit to meet with Dr. Sara Cathey and Dr. Jules Leroy, as well as some guest doctors who donated their time to conduct testing. They were able to provide reports that patients could take back home to assist their personal physicians with their care.

We met at a wonderful new building, The South Carolina Treatment Center for Genetic Diseases, which had been dedicated only a week before our arrival. It was a treat to have a tour of the new facility, where families were able to avail themselves of the expertise of geneticists, psychologists, orthopedists, and ophthalmologists.

While there, my husband, Bob, and I learned that great strides are being made in researching both MLII and MLIII. And we were able to witness considerable maturation in some of the children since

the last time we saw them. Much of the time that we were together in groups, we were laughing. Kids are kids! With friends and grandparents along to help with the little ones, we became a crowd when all together... a loud and very noticeable group! The folks in the restaurants that our group invaded were full of questions. I can't tell you how much Bob and I enjoyed watching the affected children and their siblings interact. It is so important for them to be with others who understand their needs. Besides the meetings with the doctors, it was great to hear the parents sharing their experiences.

On a personal note, we were pleased that we were able to give the doctors the 14-page autopsy report of our daughter Kelley, who died on March 17, 2009, at the age of 45. Dr. Leroy said that Kelley will be teaching scientists for many years to come, because we also had tissue sent to the genetic center for study. That was as Kelley wished. It is our hope that, in the future, there will be more treatments available not only for those affected by MLII and MLIII, but also for those affected by the other diseases under the ISMRD umbrella.

Both Drs. Cathey and Leroy stressed to us the importance of all of us to keep the ball moving along the path toward answers for our rare diseases. Although all families are dealing with their own family situations, we do need to have a strong cohesive group to continue this necessary work. The families with the other rare diseases covered by ISMRD are in the same situation now that we were in not too many years ago. Instead of being satisfied with where we are now, we need to reach out to others who may be looking for us. Research is important, but keeping this organization running is also important. We need to help families attend conferences and to make the connections they need in order to obtain necessary help. Everybody donates to one charity or another. It might be helpful to ask those we know to remember ISMRD in the future.

ISMRD is thrilled to be partners with the Greenwood Genetic Centre for the Natural History study project. We are very proud of the significant fundraising achieved by our families and the GGC for the Mucopolidiosis study. We again look forward to working in partnership with the GGC for the extension of the Natural History Study which will include the other Glycoprotein diseases. When this project commences we will again need significant funds to support families to attend the GGC, or to send Dr Cathey to the Families. This is the largest project ISMRD has undertaken and we look forward to the continued partnership with the GGC and our ISMRD families.



The ML Penguins return to Greenwood

**Dr Sara Cathey to travel to New Zealand and Australia
In November for extension of
Natural History Study for the Glycoproteinoses**

The International Society for Mannosidosis and Related Diseases (ISMRD) has been instrumental in the establishment of a natural history study of the Glycoproteinoses. As a result, Dr Sara Cathey from the Greenwood Genetic Centre in South Carolina, USA, will visit New Zealand and Australia in the middle two weeks in November to visit participants with one of the Glycoproteinoses:

Alpha-mannosidosis, Aspartylglucosaminuria, beta-mannosidosis, Fucosidosis, Galactosialidosis, Schindler disease, Sialidosis, Mucopolipidosis II, Mucopolipidosis III

This natural history study will advance scientific knowledge available about the Glycoproteinoses disorders for researchers, newly-diagnosed families and their physicians; will assist with early diagnosis and symptom management; and is a necessary step towards a cure.

There will be two parts to the study: sample analysis and collection of clinical information.

Blood, urine and/or possibly skin samples will be taken. Clinical information will be obtained by record review, telephone interviews, and direct examination where possible.

Any information that may be beneficial to the patients will be provided to them or their families; a greater understanding of glycoprotein storage disorders may be gained through their participation. Databases on each of the glycoproteinoses will be established in order that information on these disorders and access to biological specimens may be readily shared with clinicians and scientists. The compiled clinical data will be a resource for families, medical care providers, and researchers.

This study builds on the Mucopolipidosis Project which the Greenwood Genetic Center, with assistance from the ISMRD, began in 2005. The information gained from that study has already had important outcomes for families and practitioners.

If you have an affected family member, or know of anyone with one of the Glycoproteinoses, please pass this information on to them. The Glycoproteinoses are rare, and we need as many participants as possible in the study.

ISMRD is very excited at being able to include New Zealand and Australia in what will be the world's largest Natural History for Glycoprotein Storage diseases. This is an amazing opportunity to contribute your information to the Natural History Study.

To take part in the study, please email Jenny Noble jenny.noble@xtra.co.nz

FUCOSIDOSIS NATURAL HISTORY STUDY GENE MUTATION ANALYSES TAKING PLACE NOW

CALL FOR PARTICIPANTS

As part of the Natural History Study for the Glycoproteinoses, samples are currently being collected from people anywhere in the world who have Fucosidosis, for analysis at Prof John Hopwood's laboratory in Adelaide, Australia.

If you have an affected family member, or know of anyone with Fucosidosis, please pass this information on to them. Fucosidosis is one of the rarest lysosomal storage disorders, and we need as many participants as possible in the study.

The Study will enable families to contribute to a better understanding of the progression and effects of Fucosidosis. Because it is so rare, little or no information is available about Fucosidosis, and compiling information about its course from infancy to adulthood is vital.

This stage of the study will include a gene mutation analysis, which can be conducted on already-harvested fibroblast cultures (skin samples which may already have been taken for diagnostic purposes and which may still be in storage) and where possible, urine samples.

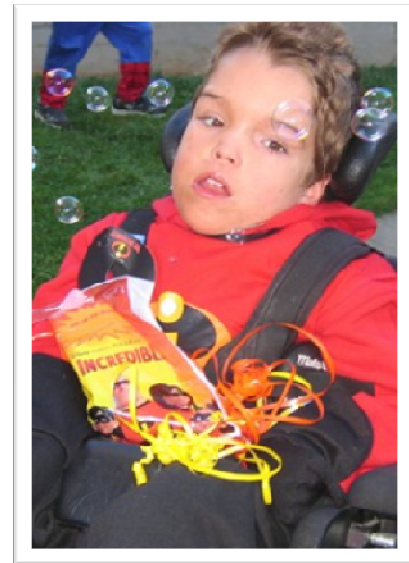
Fucoside storage will be measured from these samples using mass spectroscopy. This biochemical information will later be put together with any clinical information that may be available.

This is an amazing opportunity to contribute to the information available concerning Fucosidosis.

To take part in the study, please email

ISMARD www.ismrd.org or

Professor Hopwood at john.hopwood@adelaide.edu.au



*Matthew Paisley-Dew, from Australia,
had Fucosidosis
He is seen here at his 10th birthday party*

Dr. Emil Kakkis Sets up EveryLife Foundation

Emil Kakkis, M.D., Ph.D.

Kakkis EveryLife Foundation, President



"No disease is too rare to deserve treatment"

Dr. Kakkis is best known for his work over the last 18 years to develop novel treatments for neglected rare disorders. He began his work in a research bungalow at Harbor-UCLA working with minimal funding and support to develop an enzyme replacement therapy (Aldurazyme®) for the rare disorder MPS I. The struggle to get the therapy translated from a successful canine model to patients succeeded due to the critical financial support of a new patient organization formed by Mark and Jeanne Dant for their son Ryan, called the Ryan Foundation.

Kakkis' collaboration with the Ryan Foundation in the early development of Aldurazyme was highlighted in a 60 Minutes II segment aired in April 2001 ("Saving Ryan"), and Reader's Digest article in May 2001. Aldurazyme development was later supported by BioMarin™ and eventually their partner Genzyme™ leading to FDA approval in 2003. Dr. Kakkis has learnt a great deal from the Aldurazyme development story and the critical lessons that can guide the changes needed in the development process to accelerate new innovation in treatments for rare disorders.

During his tenure at BioMarin, Dr. Kakkis guided the development and approval of two more treatments for rare disorders, MPS VI and PKU and has contributed to the initiation of 7 other treatment programs for rare disorders, three of which are now in clinical development. Dr. Kakkis has left his position as Chief Medical Officer of BioMarin to pursue changes in the drug development and regulatory system. His focus will be on improving the diagnosis and treatment of rare disorders; specifically the process by which treatments for rare disorders are tested and approved.

Dr. Kakkis graduated from Pomona College, magna cum laude and received the Vaile Prize in Biology for his thesis research in 1982. He received a combined MD and PhD degrees from the UCLA Medical Scientist Program in 1989 and received the Bogen prize for his research on c-myc oncogene regulation. He completed a Pediatrics residency at Harbor-UCLA Medical Center in Torrance, CA and completed his fellowship training there in the UCLA Intercampus Medical Genetics Training Program in 1993. He became an assistant professor of Pediatrics at Harbor-UCLA Medical Center from 1993-1998 where he initiated the enzyme therapy program for MPS I. He is board certified in both Pediatrics and Medical Genetics. He joined BioMarin in 1998 and held various positions including Chief Medical Officer from 2006 to 2009. He received the Lifetime Achievement Award from the National MPS Society for his work on Aldurazyme. He has authored numerous scientific articles on MPS I, immune tolerance during enzyme therapy, intrathecal enzyme therapy and studies on treatments for MPS VI and PKU. *To read more go to: <http://www.curetheprocess.org/>*

NIH Announces New Program to Develop Therapeutics for Rare and Neglected Diseases

ISMRD is thrilled to see this new program that is being approved by the NIH. We have worked with many other not for profits to promote improvements to policy and research and we welcome this initiative. Here is the press release from the NIH

Bethesda, Md., Wed., May 20, 2009 — The National Institute of Health is launching the first integrated, drug development pipeline to produce new treatments for rare and neglected diseases. The \$24 million program jumpstarts a trans-NIH initiative called the Therapeutics for Rare and Neglected Diseases program, or TRND.

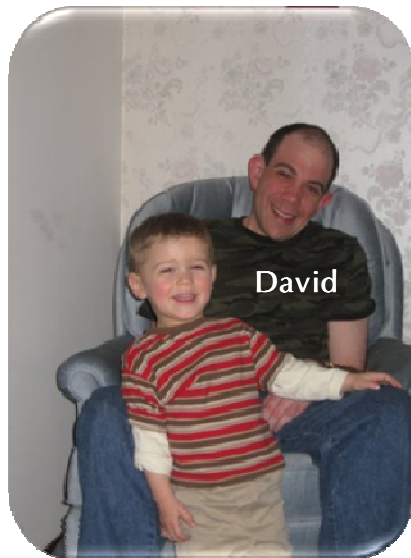
The program, is unusual because TRND creates a drug development pipeline within the NIH and is specifically intended to stimulate research collaborations with academic scientists working on rare illnesses. The NIH Office of Rare Diseases Research (ORDR) will oversee the program, and TRND's laboratory operations will be administered by the National Human Genome Research Institute (NHGRI), which also operates the NIH Chemical Genomics Center (NCGC), a principal collaborator in TRND. Other NIH components will also participate in the initiative.

A rare disease is one that affects fewer than 200,000 Americans. NIH estimates that, in total, more than 6,800 rare diseases afflict more than 25 million Americans. However, effective pharmacologic treatments exist for only about 200 of these illnesses. Many neglected diseases also lack treatments. Unlike rare diseases, however, neglected diseases may be quite common in some parts of the world, especially in developing countries where people cannot afford expensive treatments. Private companies seldom pursue new therapies for these types of illnesses because of high costs and failure rates and the low likelihood of recovering investments or making a profit.

"NIH is eager to begin the work to find solutions for millions of our fellow citizens faced with rare or neglected illnesses," said NIH Acting Director Raynard S. Kington, M.D., Ph.D. "The federal government may be the only institution that can take the financial risks needed to jumpstart the development of treatments for these diseases, and NIH clearly has the scientific capability to do the work."

A Snapshot of David Mackay

By Colin Mackay



David Andrew Mackay will turn 30 on July 4th of this year. He enjoys his job and has an active social life and an incredible circle of friends. It's not unusual for David to be two towns away and run into people he knows. He is on a first-name basis with our town's Mayor and loves to network and meet people everywhere he goes. He was diagnosed with Alpha-Mannosidosis at the age of two. David has always enjoyed life and has tried his best to participate in the activities his siblings or peers took part in. For many years he was part of the 9th Whitby Scouts as a Beaver, Cub, Scout and Venturer and he really enjoyed fundraising events and weekend camps away from home.

He has been a member of the Sunrise Youth Group for 15 years and has been to many dances, social events and camps – both in tents and in cabins - with that organization. David has been volunteering one night per week at a local nursing home, helping with the social activities by pushing wheelchair bound residents to the auditorium and back to their rooms, also serving snacks to the residents during the entertainment. David was recently honored by the Province of Ontario with an award for 15 years of volunteering at a special awards dinner banquet.

Seven years ago David decided he wanted a job that pays money. He entered a government training program and they found him a job which he has now held at Swiss Chalet, a local restaurant, as a dishwasher/cleaner for more than 6 years. The staff and management have been extremely supportive and say that he is one of their best and most conscientious employees. Despite being unable to read, write or handle money, he has learned by repetitive training and sticking to a regular schedule. David believes that the more dishes he washes each day, the more money he earns on his pay cheque. David lost 40 pounds during the first year on the job and has kept his weight down at a healthy level since then. The doctors feel that this job is excellent in order to keep him mobile and we hope he can manage the tasks for many years to come.

David enjoys going to local sports bars with friends, having a beer and watching his favorite sports teams: the Toronto Maple Leafs (ice hockey) and the Toronto Blue Jays (baseball). David lives in Whitby, Ontario, Canada, with his parents and is fortunate to have his sister Jennifer, brother-in-law Michael, nephew Camden (2), niece Melody (6 months) and brother Derek living nearby. The whole family gets together frequently for meals, or outings. Uncle David really enjoys spending time with the little ones.

To tell both sides of David's story, he is also like a typical guy – leaving a mess in the kitchen, clothes on the floor in his bedroom, and he can be very stubborn and unhappy when he doesn't get his way. He tires more quickly now after a long day or too much walking and gets headaches in noisy or busy environments.

David loves food, travel and people, and is excited to the point he can barely sleep before a vacation, so it's often best just to surprise him. He finds joy in life, and so do the people that know him.

In Memory of Kelley Crompton July 4 1963 – March 17, 2009

On March 17, 2009, when our daughter Kelley was 45 years old, her lungs could no longer function, so God took her into His loving arms. It was most appropriate for Kelley (my maiden name) to gain her angel wings on St. Patrick's Day!

My husband, Bob, and I never liked the fact that our daughter had the rare disease of ML III, but we knew that we would all be pretty miserable if we had not decided to accept it. We prayed for guidance from God throughout Kelley's life, and He always led us to the right people for the help she needed.



We had a beautiful celebration of her life with family and many friends. The cards and wonderful letters we received have made us realize just how many lives she touched... in a very positive way. We were fortunate to have her as long as we did.

Kelley had always made stuffed animals for all of her nieces and nephews, so our thoughtful daughter-in-law, Linda, chose to add a "Kelley touch" to the reception after the Mass. Since there are seven children in that family, they had collected many of them through the years. Linda let them pick out their favorites, and brought the rest to the reception. She arranged them on a table with a note, inviting everyone to take one along in memory of Kelley. It was quite a sight seeing a bunch of adults hugging their stuffed animals.

Kelley's Journey on earth has ended, but her spirit lives on. I now know that my book, "Kelley's Journey" was a prelude to the last six years of her life, and the many wonderful people we have met along the way. So much happened during the past six years, the story will not really be complete until I write about that, as well as the wonderful tributes people have made in Kelley's honor. There will be a revised and expanded "Kelley's Journey!"

In her quiet way, Kelley touched many lives, teaching all of us about acceptance, patience and fortitude. She showed us that it is possible to be pleasant even when in pain or gasping for breath. She taught all those around her that it makes no sense to feel sorry for yourself or complain about your situation. We are all here for a finite amount of time. Some of our children gained their angel wings at a very young age and some have been trying to hold out for the record. They all have had a profound effect on many people. God has used them to help people learn some of life's valuable lessons. The age at which one dies is not as important as how that person lived the life they were given. Kelley believed that, if you can adopt a good attitude, when you play the hand that has been dealt to you, and if you ask God for help along the way, you will have the strength to face the challenges of life, whatever they be.

The years have taught me that, although we no longer can enjoy someone's physical presence, the spiritual presence of those who have taught us much about living life in harmony with God's will... that spiritual presence will always be available to help us through any trials we may face. The time came when Kelley's spirit needed to be released from the body that so restricted her. There were many things that she never understood, but I have never known anyone who had as much acceptance of God's will as she did. Bob and I are doing okay, because we know that we did everything we could for her, and ultimately God is in charge.

Shortly after she died, when I was going through some of the books in her room, a little piece of paper fell out of one that I picked up. She had cut it out of the church bulletin in March of 2004, and was using it as a bookmark. The poem was "AN IRISH BLESSING!" Yes, so fitting that she left us on St. Patrick's Day!

Denise Crompton, July 2009



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: John Forman | **VP & Treasurer:** Mark Stark

Secretary: Jenny Noble

Directors

United States: Barbara Burton, MD | Andrea Gates | Pam Tobey

Australia: Carolyn Paisley-Dew

Canada: Bill Simpson

Founded in March 1999

Postal Details 2922 Deerford Street
Lakewood,
CA 90712,
United States

E-mail: info@ismrd.org

Website: www.ismrd.org | FEIN: 52-2164838



'It was only a little pee, why the 30 minute timeout?'

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. **2922 Deerford Street, 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

other \$ _____

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: _____

