22nd Annual Family Conference in Washington, DC

This year's conference focused on advocacy and research. The family agenda offered unique insight for Society members on the role the federal government plays in shaping the public policy that affects our families. Families were given the opportunity to advocate by attending a brunch on Capitol Hill with members of Congress and their staff. The family conference was followed by a National Institute of Health (NIH)-sponsored scientific meeting that was attended by prominent researchers in the lysosomal storage disease (LSD) field from around the world.

Below is a summary by Denise Dengel of several presentations given March 5 during the family conference.

Lysosomal Storage Diseases: An Overview for the Non-Scientist

Dr. Kelley Moremen of the University of Georgia's Complex Carbohydrate Research Center gave a captivating talk describing (in layperson's terms) the basics of cell function, DNA and how defective genes result in LSDs.

Dr. Moreman explained that LSDs are caused by defects in the degradation of carbohydrates that result from a genetic alteration. The genetic alteration is inherited. This causes the creation of defective proteins (enzymes) that cannot do their job in chewing up carbohydrates in a specific part in the cell, the lysosomes.


Dr. Klane White and Denise Dengel with Paula Burg from Sen. Patty Murray's (D-WA) office

Dr. Moreman used an example of the human genome being an epic story (divided into 23 "big books" (chromosomes) that is read as sentences which are translated from genetic language to the language of proteins. Proteins (enzymes) do most of the work in the cells. If a lysosomal enzyme is defective, then the molecules that would normally be degraded will accumulate. This is the case in MPS disorders. The accumulation is progressive.

He compared normal lysosomal function to a recycling center. The materials are brought into the cell for recycling, broken down and then removed to make room for more material to be recycled. There is no room for storage. To explain LSDs Dr. Moreman used a landfill model. There is no breakdown of molecules; they accumulate. The accumulation, or having too many full lysosomes, compromises normal cell function. The defects in the system of the cell back up and hurt the overall cell function. The progressive onset causes disease symptoms.

This is a basic summary of the hour-long presentation. You may see Dr. Moreman's entire presentation on the Society's Web page, **www.mpssociety.org**, under Family Conference News.


The Sheaffer family with
U.S. Rep. Charlie Dent (R-PA)

Legislative Staffer and Lobbyist Panel Discussion

Les Sheaffer moderated a panel of Capitol Hill staff and lobbyists. He asked them several questions about working on Capitol Hill and the importance of patient advocacy groups, as well as some ideas on how to advocate.

The panel consisted of: Jill Canino, legislative assistant for health for Sen. Arlen Specter (R-PA); Judy Borger, former chief of staff to Congressman Jim Greenwood (R-PA) who currently works for Duane Morris, government affairs, LLC; and Jennifer Shevchek of Cavarocchi-Ruscio-Dennis Associates, also a former staffer in Sen. Specter's office.

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A sampling of the questions and answers:

What is your typical day like on Capitol Hill?

Judy: Loved that there was no typical day.

Jennifer: As a former staffer and now a lobbyist she is adjusting to not being on the Hill where information was brought to her. Now her job is to get the pulse of what's happening on the Hill for her clients.

Jill: There is no typical day. Her days are filled with meetings with advocacy groups. If the senator needs something at a moment's notice, her schedule can be in disarray.

Why is it important for patient advocacy groups to work with you?

Jill: It makes it personal. She has a chance to see pictures, hear stories and create relationships.

Jennifer: They want to hear from you, all politics are local. If you don't tell them about your group, who will? It's good to build relationships with your representatives.

Judy: No one can tell your story like you can.

In your opinion, do the efforts of patient advocacy groups make a difference?

Jennifer: She still has a picture of Brittany, Les Sheaffer's daughter, from their meeting four years ago. The picture put a face to MPS. She said don't forget to follow up.

Jill: Absolutely. Get out there and advocate! Remember, these are YOUR elected officials.

What advice do you have for families meeting with their elected officials?

Jennifer:

1) Be sure to make an appointment for a meeting, don't just show up. You can arrange meetings at the local level in the district offices. You do not need to come to DC to meet with them.
2) Be prepared to answer questions. Have facts available.
3) Keep it simple and brief — five to 10 minutes.
4) Listen to who you are meeting with carefully. If you do not know the answer to a question, let them know you will get back to them with the answer and then do it.
5) Be firm. For example, if you have a specific bill you want your representative to cosponsor, state the bill number and name and ask for their support.
6) Follow up within two weeks of the meeting send a thank you e-mail. Keep it to the point — review the meeting, answer any questions they may have had and thank them for their time.

Jill: Agreed that follow up is very important.

All three panelists stated and agreed that the MPS Society's Legislative Committee, beginning with Les' advocacy and continuing with Sissi and Kym, has laid the groundwork for good relationships on Capitol Hill. They are the best at doing this type of patient advocacy that they have seen. It is important as members of the MPS Society to work with the Legislative Committee on contacts you may make on Capitol Hill. What the Legislative Committee is doing is working; as a member you want to support their efforts.

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The Clark family with J.K. Robinson
(Sen. John Warner's (R-VA) office)

by
Sissi Langford,
chair, Committee on Federal Legislation
and
Kym Wigglesworth,
co-chair, Committee on Federal Legislation

The Wigglesworth family with Mary Beth Stanton
(Sen. Herb Kohl's (D-WI) office)

The Dummann's with Gerald Hood
(U.S. Rep. Don Young's (R-AK) office)

Erik and Sara Fletcher

Marqueta and Kevin Johnson with Sen. Richard Burr (R-NC)

Jessie Brenton and Adam and Joan Cohen speak with a congressional staffer

Texas families meeting with Emily Dillard from Sen. John Cornyn's (R-TX) office

Individuals Disability and Education Act (IDEA) Update

Dr. Alexa Posney, director for the Office of Special Education Programs, U.S. Department of Education, gave a timeline and history of how the educational system has changed over the years from denying those with disabilities to including them in the educational system. She pointed out that 30 years ago one million kids with disabilities did not go to public school. She stated that systems change — in the 1970s there was a de-institutionalization and by the year 2000 there was inclusion.

She pointed out that in today's educational system children with degenerative disorders are seen as a success when they can maintain their current level of functioning. In the past the goals were generally set for improvement. Dr. Posney credited the work of groups like the MPS Society in successfully advocating for inclusion of language specific to children with degenerative disorders in the reauthorization of IDEA.

Dr. Posney emphasized the importance of being involved in your child's individual education plan (IEP) and setting goals that fit your child's particular needs. She also spoke on the importance of having each person on your child's care team involved in the IEP, and that you have a right to have them all sit down and meet together for the IEP meeting.

Capitol Hill Brunch

On March 6, families traveled to the Russell Senate Office Building to attend a brunch with members of Congress and their staff. This was an extraordinary opportunity for our families to interact with elected officials. By all measures this event was a success. Staff members from 32 congressional offices attended in addition to Sen. Richard Burr (R-NC), Sen. Johnny Isakson (R-GA) and Rep. Charlie Dent (R-PA).

As many of you are aware, members of the Legislative Committee have been meeting with congressional members and staffers for more than six years to advance public policy that will improve the lives of individuals and families affected by MPS. During these years we've established working relationships with many of these offices. We always bring along pictures of our own children and share pictures of other individuals with MPS. Many staffers keep pictures of our kids in their offices. The pictures help to humanize these disorders for people who have never met anyone affected by MPS. Many of these staffers attended the brunch. Personally meeting the families and spending time with the kids made a profound impact. We have further solidified the existing relationships with many of these congressional offices and have many new contacts thanks to Society members inviting their congressional delegations. Thanks to all who participated in this amazing event.

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Legislative Committee

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NIH-Sponsored Scientific Meeting

This landmark meeting entitled, “Towards Clinical Progress in the Mucopolysaccharidoses” brought together researchers from around the world to collaborate, stimulate research and guide future actions in addressing MPS and related diseases. This meeting was the result of years of collaboration between the NIH, specifically the National Institute of Neurological Disorders and Stroke (NINDS) and the MPS Society’s Legislative Committee. We were honored to have Dr. Story Landis, NINDS director, open the meeting.

The workshop objective for this two-day event was to identify and address impediments to effective therapies, and to develop a strategy to determine the minimum standard protocol for clinical application of effective therapies for MPS and other LSDs.

Researchers who participated in this meeting included many who have been working on effective therapies for MPS for many years. There also were young researchers in attendance, many of whom were mentored or inspired by the seasoned researchers, who added new and innovative ideas to the meeting. Information on current research was presented, some of which has yet to be published. Some of the goals for individual sessions included: addressing the hurdles researchers face in moving from animal to humans in clinical trials; identifying the limitations and need for improvements in traditional therapeutic approaches for MPS; addressing the practical hurdles in moving current therapeutic strategies into the clinic; is gene therapy the most promising therapeutic approach to cure MPS and other LSDs?; and early intervention and meaningful clinical outcomes — what are the issues and hurdles to effective therapy for MPS? (e.g. newborn screening, genotype/phenotype correlations, objective end-points, combination therapies, regulatory issues, etc.).

One session was devoted to identifying novel approaches and evaluating strategies directed toward the central nervous system and blood brain barrier. Each of the speakers in this session presented research that is being funded through the NIH/LSD Research Consortium (LSDRC) program announcement (PAS-04-120) which resulted from NINDS and the LSDRC collaboration.

The format of the meeting lent itself to collaboration and discussion. Most of the sessions started with brief presentations from researchers and included time for questions and discussion. There also were blocks of time devoted to panel discussions. This resulted in open debate and the free exchange of ideas. Although this meeting was designed for scientists, parents were welcome and were able to ask questions and participate in discussions. Three members of the Society participated in a parent panel discussion called *Parents with CNS involved children (a day in the life)*. Larry Kirch, Kris Klenke and Les Sheaffer gave a glimpse of their lives with Allison, Kraig and Brittany. This prompted numerous questions from and a lengthy discussion with the researchers.

This meeting also can be counted as a great success and would not have been possible without the support of the NIH, specifically NINDS, the National Institute of Arthritis and Musculoskeletal and Skin Disease, and the Office of Rare Diseases. The following people were responsible for organizing the meeting: Mark Haskins, V.M.D., Ph.D. (University of Pennsylvania); Sissi Langford (National MPS Society); Catherine McKeon, Ph.D. (National Institute of Diabetes and Digestive and Kidney Diseases); Mark S. Sands, Ph.D. (Washington University in St. Louis); and Danilo Tagle, Ph.D. (NINDS). These very busy people each dedicated hours of time organizing and ensuring the success of this meeting.


Dr. Mark Sands with a Congressional Staffer


Jenny and Gordon Wingate, Cheryl Guidry and a Congressional Staffer


The Keith family with Lindsey Anna from Sen. Mitch McConnell’s (R-KY) office


Ron and Kristin Mattheson with Brad Conway from Sen. John Sununu’s (R-NH) office