MPS Research Gains $1 Million Increase in 2010

During the last several years, the Legislative Committee concentrated heavily on increasing research dollars through the National Institute of Health (NIH) for MPS and related diseases. With research funding by the NIH stagnant at $7 million for the last few years, we feared the loss of research jobs, and the delay of new treatments or therapies being brought to the marketplace for MPS and related diseases.

However, a $1 million increase was given due to the many excellent research opportunities available for funding. Much of this credit needs to go to President Obama and our MPS researchers for their superior grants and to our Policy with Partners members who advocated with Congress in Washington, DC, and at home. The NIH also is reflecting this $8 million amount for MPS research in 2011 and 2012.

Legislative Committee Travels to DC

May 16, 17 and 18 were busy days in Washington, DC, spent calling on members of Congress and their health liaisons to thank them for the $1 million increase in research funding while advocating for more research dollars in 2011 and 2012. At this time we also discussed the Society's priorities. It was very moving to once again have the resolution brought by Sen. Lindsey Graham (R-SC) declaring May 15, 2011, as National MPS Awareness Day. We thank Sen. Graham and his health liaisons, Colin Allen and Leigh Ellen Gray.

You will read more about our trip, National MPS Awareness Day and detailed talking points in the next issue of *Courage*. Thanks to board members Jeffrey Bardsley and Stephanie Bozarth, and MPS parent Amy Barkley for volunteering their time and expertise to help MPS families, and for accompanying Ernie and Debbie Dummann through the halls of Congress.

Legislative Committee:
Ernie Dummann, Chair
Debbie Dummann
Jeff Bardsley
Stephanie Bozarth
Chip Brady
Steve Holland
Austin Noll
MaryEllen Pendleton
Kim Whitecotton
Roger Chapin
Terri Klein
Kelly Rose
Laurie Turner
Barbara Wedehase

International Rare Disease Research Consortium Launched

In April, regulatory agency stakeholders, researchers, patient group representatives, members of the biopharmaceutical industry, and health professionals gathered for the second meeting and official launch of the International Rare Disease Research Consortium. Following a successful preparatory meeting in Iceland in October 2010, the second reunion picked up the pace with the endorsement by members to fulfill certain goals, including, notably, a commitment to the development of 200 new rare disease treatments by the year 2020 and the development of diagnostics for all rare disorders. Related challenges identified include the need to establish and provide access to harmonized data and samples, perform molecular and clinical characterization of rare diseases, boost translational, preclinical and clinical research, and streamline ethical and regulatory procedures.

Formed under the auspices of the U.S. NIH and the European Commission, the consortium will involve "an unprecedented cooperation at the international level" in order to fulfill the ambitious goals defined. As such, public and private stakeholders from all over the world are invited to join the fight to alleviate the suffering of rare disease patients and their loved ones. Participants included representatives from Canada and individual European countries (France, Germany, Italy, Spain and the UK, among others). Genetic Alliance President and CEO Sharon Terry, who participated in the three-day meeting, commented, "The energy generated by having concrete goals, with the commitment and leadership of major international funders throughout the entire meeting, was palpable. Now it is time to roll up our sleeves as a community, and focus our energy on getting the business plan in place to succeed."

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The loaded agenda began with an analysis of ongoing research efforts and future challenges, including a breakdown of country-specific activities. The conference then gave over to several breakout sessions, including Understanding of Pathophysiology of Rare Diseases (including genomic analysis by next generation sequencing, and animal and iPS cells models of rare diseases); Ontologies/ Disease Classification/Natural History; Biomarkers; Patient Registries and Biospecimen Repositories; Preclinical Research and Clinical Trials; and Communication/Publication/Information/PR/Data Policy. The meeting wound down with a report and conclusions from the breakout sessions.

Next Steps

A scientific and policy framework must be developed in order to "guide the research activities and foster collaboration among the stakeholders to systematically explore all the opportunities to accelerate the development of diagnostics and therapies for rare diseases." Funding and budget considerations also need addressing. It is anticipated that the governance of the project will be modeled after the Human Genome Project—and thus be open to research bodies from around the world. It is probable that a minimum financial contribution will be required by participants, although no figure has yet been determined. Participants also would have to share all relevant data, a process that would help harmonize terminology between countries.

Court Okays Federal Funds for Embryonic Stem Cell Research

A federal appeals court has sided with the Obama administration, overturning a judge's ruling last summer that NIH funding of embryonic stem cell research violates a law against destroying human embryos. A panel of three judges on the U.S. Court of Appeals for the District of Columbia Circuit —who were all appointed by Republican presidents—ruled 2-1 in April 2011 to strike down the injunction issued by federal judge Royce Lamberth that barred the NIH from paying for research on embryonic stem cells (ESC).

Lamberth ruled that ESC research violates a federal law known as the Dickey-Wicker amendment that bars use of government money to destroy human embryos.

After the prospect of losing funding put the ESC research community into a panic, the same court that issued the ruling ordered that the NIH could continue to fund ESC research until a formal decision was made on Lamberth's injunction.

The Obama administration argued that millions of dollars worth of research would be put in peril if the injunction was allowed. Soon after he came to office, President Obama lifted a ban on stem cell research imposed by President George W. Bush and expanded federal funding for ESC research.

The NIH came up with ethics guidelines in 2009 to govern ESC research that stated federally funded embryonic research can only involve leftover embryos from in vitro fertilization procedures with the parents' consent, and that there be a "clear separation" between the decision to create embryos for reproductive purposes and for research purposes.

But critics say that ESC research destroys human life and is not something the government should be paying for.

The case was originally brought by two researchers who work with adult stem cells, which are found in various tissues throughout the body, who argued the NIH's guidelines violate the Dickey-Wicker amendment, and Lamberth agreed.

"Responsible stem cell research has the potential to treat some of our most devastating diseases and conditions and offers hope to families across the country and around the world," said White House

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spokesman Nick Pappas. “Today’s ruling is a victory for our scientists and patients around the world who stand to benefit from the groundbreaking medical research they’re pursuing.”

NIH Director Francis Collins, MD, PhD, said he was happy and relieved with the ruling. “I am delighted and relieved to learn of the decision of the Court of Appeals,” he said in a prepared statement. “This is a momentous day—not only for science, but for the hopes of thousands of patients and their families who are relying on NIH-funded scientists to pursue life-saving discoveries and therapies that could come from stem cell research.”

NORD Gala

On March 17, 2011, the National Organization for Rare Disorders (NORD) honored two members of the U.S. Congress for improving the lives of people with rare diseases. The awards were presented at the annual NORD Partners in Progress Celebration held at the Andrew W. Mellon Auditorium in Washington, DC. Each year, NORD celebrates pioneering achievements of individuals, organizations, and companies in public policy, patient advocacy, medical research and product development.

Actress Patricia Richardson of TV’s Home Improvement, Strong Medicine and The West Wing emceed the event with more than 500 guests from patient organizations, government agencies including the NIH and Food and Drug Administration, academic research centers, and pharmaceutical companies in attendance.

Among others honored were the Honorable Joseph Crowley (NY-7) and the Honorable Leonard Lance (NJ-7) who received the National Health Leadership Award. Representatives Crowley and Lance are the co-chairs in the U.S. House of Representatives of the new Rare Disease Congressional Caucus.

The caucus was formed to bring Congressional attention to the nearly 7,000 known rare diseases that currently have no approved therapies; ensure sufficient funding for research and orphan product development; explore ways to incentivize companies to create new drugs, biologics and humanitarian use devices; and provide an opportunity for members of Congress, families and advocacy groups to exchange ideas and policy concerns. Rare and neglected diseases affect nearly 30 million Americans.

“The Rare Disease Congressional Caucus will provide an important forum in Washington for the exchange of ideas and information related to rare diseases,” Rep. Lance said. “Congressman Joe Crowley and I are committed to working together in a bipartisan capacity with like-minded members, policy advocates and families across the nation to increase awareness and education of rare diseases.”

Stephanie Bozarth, Jeffrey Bardsley, and Ernie and Debbie Dummann were guests of BioMarin Pharmaceutical and would like to thank them for their hospitality.