



## Thanks for a job well done!

**W**hen President Rolando and I decided to present the idea of a national bowlathon to increase our contribution to the Muscular Dystrophy Association, we had high hopes. Those hopes were met when we raised more than \$340,000 during the first bowlathon. Our goal this year is to top that amount and raise at least \$500,000.

But that will be done only if as many branches as possible turned out last month. I hope to report in the next *Postal Record* that we exceeded our goal. If you had an event, send in the report from the packet your branch received. If you didn't get the reporting form, please go to NALC's web page and click on "MDA" under the "Community Services" tab to find the reporting form to send to headquarters. We want to make sure your branch gets credit for the work you've done.

**To give some insight to how your money is used in research endeavors,** I have listed just a few of the many projects that are currently ongoing to help find a cure:

**Two DNA changes needed to cause FSHD symptoms—**MDA-supported scientists uncover a previously missing piece of the puzzle posed by facioscapulohumeral muscular dystrophy. Not only does this disease require the presence of a contracted area of DNA on chromosome 4 (a previously recognized factor), but it also requires a "permissive" DNA signal on the same chromosome, which allows toxic proteins to last long enough to cause muscle damage. The study is expected to provide new therapeutic targets.

**DMD gene repair strategy takes a big step forward—**An MDA-supported research group reports that a new generation of molecules can help cells permanently repair errors in the dystrophin gene, fixing the underlying cause of Duchenne muscular dystrophy. In experiments on cells, the new molecules stimulated more than 10 times the DNA repair levels of previous molecules, providing a "proof of concept" for gene repair as a therapy for DMD.

**Synthetic enzyme approved for late-onset Pompe disease—**Lumizyme, an enzyme manufactured by Genzyme Corp., becomes commercially available for the treatment of late-onset acid maltase deficiency (Pompe disease) in individuals 8 and older. MDA-supported basic research played a role in the development of both Lumizyme and Myozyme, Genzyme's enzyme replacement drug for infants and very young children with Pompe.

**Therapeutic strategy in MTM opens muscle fibers—**MDA-supported researchers report a new molecular strategy

designed to transport the needed myotubularin enzyme (a type of protein) into muscle fibers in myotubularin-deficient mice and perhaps eventually in humans with X-linked myotubular myopathy.

**More evidence implicates immune system in ALS—**Investigators at the ALS Therapy Development Institute find that disrupting an immune system pathway called CD40L delays disease onset and extends survival in mice with a disease mimicking human amyotrophic lateral sclerosis. The experimental drug candidate, a blocking protein, or "antibody," called ALSTDI-00846, prevents interaction between two key CD40L pathway components, blocking a signal to the body to launch an immune system attack.

**ALS clinical trial delivers drug to central nervous system—**Human testing begins of ISIS-SOD1-Rx, an experimental "antisense" drug developed with MDA support, in people with the SOD1-related form of familial amyotrophic lateral sclerosis. The phase 1 clinical trial is the first in which a drug designed to block production of the toxic SOD1 protein is administered directly into the central nervous system of humans.

**Toxic clumps not the only molecular cause of OPMD—**An MDA-supported study team finds that a loss of function of the PABPN1 protein likely is a contributing factor, along with the formation of potentially toxic protein clumps in muscle cells, in oculopharyngeal muscular dystrophy—a finding that may lead to new therapeutic strategies.

**New muscle stem cell found—**MDA-supported scientists in France identify a previously unknown muscle stem cell in the spaces between muscle fibers in mice. Called "PICs," the cells may play an important role in muscle regeneration and repair and could have implications for treatment of muscular dystrophies.

**Gene therapy rescues mice with SMA—**A research team reports "unprecedented" improvement in newborn SMA-affected mice that received gene therapy via intravenous injection. Newborn mice that received the treatment demonstrate near-normal motor function and brain-to-muscle signaling, as well as a dramatic increase in survival. The team utilized key findings derived from previous MDA-supported studies in their investigation.

**As you can see, MDA is working hard during a difficult time** in our economy to deliver help and hope to MDA families. Thanks from the bottom of my heart for all you do! ☐