



The latest in research and development

MDA-sponsored scientists, working with hospitals and private-sector researchers, are constantly working to develop possible cures and treatments for muscular dystrophy. More than 80 percent of all money contributed to MDA goes toward research and treatment.

The following are only some of the kinds of scientific research and studies that are currently underway. More can be discovered at mda.org/publications/resdev.html.

“Read-through” drug goes to the next testing phase—The drug PTC124, which causes cells to “read through” a specific type of genetic error that forms a molecular stop signal and affects some 15 percent of children with Duchenne muscular dystrophy (DMD), begins undergoing testing at a higher dose, after earlier tests showed it was safe and well tolerated at a lower dose level. MDA’s donation of \$1.5 million to PTC Therapeutics helped move the drug through testing.

300 “antisense” compounds developed for possible use in DMD—An MDA-funded team in Australia developed some 300 “antisense” compounds that can coax muscle cells to skip over errors in the dystrophin gene and produce functional dystrophin protein molecules. Dystrophin is needed but missing in DMD. One such compound is already being tested in boys with the disease.

Toxic neighboring cells identified in ALS-affected nervous system—MDA-supported researchers have found that nervous system cells called glia secrete an unknown toxic compound that kills neighboring motor neurons, the muscle-controlling nerve cells affected in amyotrophic lateral sclerosis (ALS). They say transplanting stem cells that become good glia into people with ALS may be beneficial.

Tricostatin helps mice make protein needed in SMA—MDA-backed scientists find that the compound trichostatin (TSA) increases levels of a needed but deficient protein in the cells of mice with spinal muscular atrophy. TSA belongs to a family of potential new medications known as HDAC inhibitors, which cause cells to interpret genetics instructions as “open” and ready to be used, rather than “closed” and unavailable for use. These results provide a basis for testing HDAC inhibitors in people with SMA.

Researchers identify new type of muscle stem cell—MDA-supported researchers in Italy announced they’ve identified a new type of muscle stem cell that they believe is highly promising for treatment of muscular dystrophies. These new stem cells, called “pericyte-derived,” are located around small blood vessels in muscle tissue. When injected into mice with DMD, they matured into muscle fibers and improved the ability to use the muscles.

Largest ever ALS drug search begins—MDA and the ALS Therapy Development Institute launched the largest drug discovery project in amyotrophic lateral sclerosis in history. The three-year, \$36 million endeavor will attempt to identify biochemical targets and find drugs that work on them.

Two anti-scarring drugs show promise in mice with DMD—An MDA research grantee is among the scientists who announced that two drugs, losartan and pirfenidone, have shown promise in reducing scar formation (fibrosis) in mice affected by DMD. Scar formation resulting from excess deposits of connective tissue is a major factor in muscle damage in DMD and other muscle diseases.

Gene therapy trial for Duchenne dystrophy has begun—Scientists and physicians have launched the first U.S. human gene therapy trial directed at DMD, with the support of a \$1.6 million grant from MDA. The first of six boys with DMD receives an injection of genes for dystrophin, the missing protein in DMD, in one arm and a placebo in the other. The scientists will later measure dystrophin production and monitor the effects of the gene transfer on the children.

While these summaries show that MDA is advancing against the effects of these diseases, it clearly illustrates that the battle has a long way to go. We know so much more than we did about muscular dystrophy, and as techniques and knowledge increase, we get closer to the cure.

I want to personally thank each of you for the work and support you have given these families. Next month, I hope to print the summaries of each branch and announce the winners of each category. We are tabulating the numbers with MDA, and the total amount raised will be a number for which we can all be proud. ☒