

**For immediate release**

**First Demonstration of Muscle Restoration in an Animal Model  
of Duchenne Muscular Dystrophy**

*Research funded by Parent Project Muscular Dystrophy*

Using PTC124, a new type of drug that targets a specific genetic defect, **H. Lee Sweeney, PhD, the scientific director of Parent Project Muscular Dystrophy (PPMD)**, along with colleagues **at PTC Therapeutics, Inc. and the University of Massachusetts Medical School** has for the first time demonstrated restoration of muscle function in a mouse model of Duchenne muscular dystrophy (DMD). The research appears ahead of print in an advanced online publication of *Nature*, released on Sunday, April 22.

“This new class of treatment has the potential to help a large number of patients with different genetic diseases that have the same type of mutation,” said Dr. Sweeney, the senior author of the study and chair of the Department of Physiology at the University of Pennsylvania. This genetic flaw causes anywhere from 5 to 15 percent of the individual cases of most inherited diseases, including DMD, cystic fibrosis, and hemophilia.”

The new drug was developed by PTC Therapeutics, a biotech firm located in South Plainfield, NJ. It binds to the ribosome, a cellular component where the genetic code is translated into proteins, one amino acid at a time. The drug allows the ribosome to read through a mistake in the genetic code called a premature stop codon in order to properly make whole proteins.

In DMD, patients are missing dystrophin, a protein that helps keep muscle cells intact. About 15 percent of DMD patients do not make dystrophin because of the mutation. DMD eventually affects all voluntary muscles, as well as heart and breathing muscles.

## Muscle Restoration in Animal Model of DMD...2

PTC124 attaches to ribosomes in all cell types within the MD mouse model, overriding the mutation in the dystrophin gene that tells it to halt production of the protein. Instead of stopping, the full-length dystrophin protein is made. The drug enables enough protein to be made to correct defects in the muscle of the DMD mouse, and at the same time the drug does not prevent the ribosome from reading correct "stop" signals in the genetic code to make other necessary proteins.

"Enough dystrophin accumulated in the muscles of the MD mice so that we could no longer find defects in the muscles when we examined them," says Sweeney. "For all intents and purposes the disease was corrected by treatment with PTC124." The drug allowed dystrophin to be made in cells in which it was previously absent, to be delivered to the proper location at the cell membrane, and to induce restoration of muscle function in rodent muscles.

PTC124 is presently nearing the end of a Phase II multi-center clinical trial in DMD patients, of which Children's Hospital of Philadelphia is a major accruing site.

"This is a very big *first*," said **Kimberly Galberaith, Vice President of Parent Project Muscular Dystrophy**, which helped fund the research. "Twenty years ago, DMD was the first inherited disease to have its gene discovered. Now DMD is the first to be used as a model for restoration of muscle function through drug therapy. Clearly, the research dollars that go toward funding DMD research generate advances that are broad in scope and benefit families well beyond the DMD community."

Parent Project Muscular Dystrophy (PPMD) is the largest nonprofit organization in the United States focused entirely on Duchenne muscular dystrophy. PPMD funds critical research and provides families with up-to-date information and vital connections with other parents, supporters, and medical providers. For more information, visit [www.parentprojectmd.org](http://www.parentprojectmd.org).