

Myotonic Muscular Dystrophy Research at UVA

What is Myotonic Muscular Dystrophy (MMD)?

MMD is a genetic disease that causes the progressive wasting of muscle, and damages the heart, brain, and other organs and tissues. No existing treatments stop or reverse this debilitating and life-threatening disease.

MMD is the result of a mutation that causes excessive “repeats” of defective RNA sequences. Cruelly, the disease gets progressively worse as it is passed from generation to generation. It is rarely diagnosed until a child either dies in infancy or is severely debilitated.

The University of Virginia’s Mani Mahadevan, M.D., is recognized as one of the world’s

“I have a sense of hope that I didn’t have five years ago,” says Mahadevan. “We can realistically look at developing treatments in the next five to 10 years.”

foremost experts on MMD. In 1992, he was a member of the research team that discovered the gene mutation that causes MMD, and he has dedicated his life to finding a cure for the disease.

Mahadevan and his UVA research team were able to reverse MMD in animal models by “turning off” the defective RNA. Remarkably, their experiments showed that the disease not only ceased in its progression but that previously damaged muscles and organs actually healed.

Mahadevan has a three-pronged approach to attacking MMD:

- **Silencing RNA.** Mahadevan and his team are searching for ways to replicate in people the extraordinary results they saw in an animal model. This could effectively “cure” MMD.



- **Therapeutic Proteins.** Patients with MMD do not adequately produce certain beneficial proteins because of clogged cell nuclei. Effective protein replacement therapies, while not curing the disease itself, is a very promising avenue for alleviating the symptoms of MMD.
- **Drug Therapy.** There are vast catalogs of pharmacologic compounds that may help patients with MMD. Some have been approved for use in humans for other diseases, and others are not yet approved. Mahadevan has established strategic partnerships in order to expedite the discovery of promising drug therapies for MMD patients.

Mahadevan and his team intend to find ways to improve the lives of children living with MMD today and find the cures of tomorrow.

If you want to join the effort through making a tax-deductible contribution, please contact Scott Karr at 800-297-0102 or skarr@virginia.edu.