# Heart-function protein may help muscular dystrophy patients live longer

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A Rutgers-led team may have found the key to preventing Duchenne muscular dystrophy (DMD)-related heart disease, the leading cause of death in patients living with the disease.

The study, published in the *Journal of Clinical Investigation (JCI)*, examined the role of Connexin-43 (Cx43), a protein that regulates heart function. The researchers found that Cx43 was dysfunctional in both human and mouse DMD hearts, so they modified the Cx43 protein in the hopes of alleviating heart disease. The researchers discovered that altering the Cx43 protein through a process called phosphorylation protected DMD mice against irregular heart beat and late-stage failure.

"For many DMD patients, the heart muscles gradually break down, leading to death. Our findings may help give hope to millions of patients," said study co-author Diego Fraidenraich, an assistant professor of Cell Biology and Molecular Medicine at Rutgers New Jersey Medical School.

"Medical advances have managed to slow down the disease progression in most muscles in the body, but there are yet to be any discoveries that target or prevent deterioration of the DMD heart, which remains the number one killer among these patients," said co-author Eric Himelman, a Ph.D. candidate at Rutgers New Jersey Medical School. "Therapies based on our finding may help prolong the lives of muscular dystrophy and other heart disease patients."

DMD, a genetic disorder characterized by progressive muscle degeneration, is the most common type of muscular dystrophy, affecting about one in 5,000 males and typically beginning at about age 4. The average life expectancy is 26.

These findings were simultaneously published together in JCI insight with another Rutgers-led study that examined Cx43 activity in the heart

Next steps include developing drugs that directly target Cx43 in DMD hearts, with a goal of potentially introducing clinical trials using Cx43 modification as a therapy for DMD patients.

The study was funded by the National Institutes of Health, the Muscular Dystrophy Association and the American Heart Association, and includes an interdisciplinary team of investigators with complementary expertise from Rutgers University, Fred Hutchinson Cancer Research Center, New York University and Baylor College of Medicine.

**More information:** Eric Himelman et al. Prevention of Connexin43 remodeling protects against duchenne muscular dystrophy cardiomyopathy, *Journal of Clinical Investigation* (2020). DOI: 10.1172/JCI128190