Medical Xpress

Discovery one step closer to treatment for Duchenne muscular dystrophy

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(Medical Xpress)—A University of Alberta researcher has pinpointed a mutation that brings the medical community another step closer to treating those who suffer from a fatal type of muscular dystrophy.

Toshifumi Yokota, a muscular dystrophy researcher in the Department of Medical Genetics, led a team of researchers that discovered a mutation that results in dramatic regrowth of the dystrophin, which is a protein that essentially acts like a support beam to keep muscles strong.

Yokota found that lab models with the mutation possessed many unique muscle fibres that are somehow involved with the regrowth of the important protein that is virtually non-existent in those suffering from Duchenne muscular dystrophy. This disorder is an incurable and fatal neuromuscular condition that causes muscles responsible for movement to weaken, which leads to difficulty walking or breathing.

"If we can find the mechanism that causes the dystrophin protein to regrow, it would be a drug target for the treatment of Duchenne muscular dystrophy," said Yokota, who holds Muscular Dystrophy Canada's Friends of Garrett Cumming Research Chair and the H.M. Toupin Neurological Science Chair. "Our discovery is very promising."

Yokota noted the increased dystrophin protein didn't result in muscle regeneration and the team is continuing their research to find out why. "We are now working on the next steps: what causes these muscle fibres to increase in number?"

The first co-author on the study, published in the peer-reviewed journal *PLOS ONE*, was Yokota's undergraduate summer student, Merryl Rodrigues.

Garrett Cumming, for whom Yokota's Muscular Dystrophy Canada chair is named, added, "This is very exciting news and hopefully these findings will lead to a viable treatment and ultimately a cure."

Provided by University of Alberta

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