

New research increases understanding of Duchenne muscular dystrophy

13 October 2016

A new paper, co-written by faculty at Binghamton University, State University of New York, increases the understanding of Duchenne muscular dystrophy (DMD)—one of the most common lethal genetic disorders—and points to potential therapeutic approaches.

"The findings suggest that the immune system has an important role in the [muscle disease](#) of Duchenne muscular dystrophy," said Eric Hoffman, professor of [pharmaceutical sciences](#) and associate dean for research at Binghamton University's School of Pharmacy and Pharmaceutical Sciences.

DMD, the most common of many types of muscular dystrophy, generally occurs in young boys, where they first show signs of [muscle weakness](#) in early school years. The disease then gradually destroys [muscle tissue](#), so that boys lose the ability to walk in their early-mid teens, and then later succumb to respiratory or cardiac failure. All patients with DMD have gene mutations in the dystrophin gene so that they do not make dystrophin protein in muscle tissue. The dystrophin gene is the largest gene in the human genome; lack of dystrophin protein was discovered as the cause of DMD by Hoffman in 1987.

While all DMD patients share the same genetic and protein problem in muscle, they often show differences in the severity of their disease. Some are particularly severe (losing ambulation at just 8 or 9 years of age), whereas others are much less severe (walking until their 20s).

Hoffman and his colleagues sought to determine why different DMD patients responded differently to the same loss of the [dystrophin protein](#) in their muscle. After studying hundreds of DMD patients, they confirmed that the gene CD40, an important modulator of the immune system, held a polymorphism that modified the severity of Duchenne muscular dystrophy. Polymorphisms are

subtle differences in genes that dictate the color of hair, height, skin color and many other aspects that make each person unique.

"Slight differences in the immune system lead to different reactions regarding the muscular dystrophy. The immune system needs to be balanced: too much or too little is a bad thing in any immune response. This balance may be shifted in muscular dystrophy due to the CD40 polymorphism," said Hoffman.

"If a genetic polymorphism of CD40 leads to milder disease in DMD, then it follows that drugs targeting CD40 may also improve patient symptoms," added Hoffman. "This research opens new therapeutic avenues to try to develop therapies for DMD."

The paper, "Association Study of Exon Variants in the NF- κ B and TGF β Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy," was published in *The American Journal of Human Genetics*.

More information: *American Journal of Human Genetics*, [dx.doi.org/10.1016/j.ajhg.2016.08.023](https://doi.org/10.1016/j.ajhg.2016.08.023)

Provided by Binghamton University

APA citation: New research increases understanding of Duchenne muscular dystrophy (2016, October 13) retrieved 11 October 2022 from <https://medicalxpress.com/news/2016-10-duchenne-muscular-dystrophy.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.