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New model of muscular dystrophy provides insight into disease development

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Muscular dystrophy is a complicated set of genetic diseases in which genetic mutations affect the various proteins that contribute to a complex that is required for a structural bridge between muscle cells and the extracellular matrix (ECM) that provides the physical and chemical environment required for their development and function.

The affects of these genetic mutations in patients vary widely, even when the same gene is affected. In order to develop treatments for this disease, it is important to have an animal model that accurately reflects the course of the disease in humans.

In this issue of the Journal of Clinical Investigation, researchers at the University of Iowa report the development of a mouse model of Fukuyama's muscular dystrophy that copies the pathology seen in the human form of the disease.

By removing the gene fukutin from mouse embryos at various points during development, researchers led by Kevin Campbell were able to determine that fukutin disrupts important modifications of dystrophin that prevent the muscle cells from attaching to the ECM. Disruption of the gene earlier in development led to a more severe form of the disease, suggesting that fukutin is important for muscle maturation. Disruptions in later stages of development caused a less severe form of the disease.

In a companion piece, Elizabeth McNally of the University of Chicago discusses the implications of this disease model for the development of

new therapies to treat muscular dystrophy.

More information: Mouse fukutin deletion impairs dystroglycan processing and recapitulates muscular dystrophy, *Journal of Clinical Investigation*, 2012.
The attachment disorders of muscle: failure to carb-load, *Journal of Clinical Investigation*, 2012.

Provided by Journal of Clinical Investigation