Identification of the gene responsible for a new form of adult muscular dystrophy

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A study published in today's online edition the *American Journal of Human Genetics*, allowed the first identification of a new form of adult onset muscular dystrophy. The research team led by Dr. Bernard Brais, neurogeneticist at the Research Centre of the Centre hospitalier de l'Université de Montréal (CRCHUM) and associate professor, Université de Montréal, in collaboration with European collaborators, demonstrated that recessive ANO5 mutations will lead to abnormal membrane repair of muscle fibers.

The continuous stress induced by contractions of muscles lead to tears of its membrane that need to be rapidly repaired. "An understanding of how the loss of AN05 will lead to defective membrane repair will lead to better treatments of all muscular dystrophies were such abnormal process play a role. ", notes Dr. Brais.

In all the patients in the study, the researchers identified in all patients two recessive mutations inherited from both healthy parents. French Canadian cases were found to develop proximal limb girdle muscular dystrophy, usually in the thirties, while European cases developed a more distal non-dysferlin Miyoshi Myopathy (MMD3). One mutation appears to be more frequent in the Quebec population.

This is the first time Quebec researchers both describe a new form of muscular dystrophy and identify its causal gene.

Muscular dystrophy is the name for a group of neuromuscular disorders that are characterized by progressive weakness and wasting of the voluntary muscles that control body movement. As muscle tissue weakens and wastes away, it is replaced by fatty and connective tissue.

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