

NEWS RELEASE

FOR IMMEDIATE RELEASE

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

Montreal, Canada, January 21, 2010 – 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 ANO5 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

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.*

This study was supported by grants from the American Muscular Dystrophy (MDA) founded more than 50 years ago by Jerry Lewis, Muscular Dystrophy Campaign (United Kingdom) and the Jain Foundation.

*Muscular Dystrophy Canada

- 30 -

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About the study: from 12:00: www.cell.com/AJHG/ or call Nathalie Forgue

About the CRCHUM

With more than 350 researchers and 400 students, the CRCHUM is the University of Montreal's most important affiliated research centre. The CRCHUM is a hospital-based research centre focused on discovery, innovation and the non-invasive exploration of human beings and the transfer of results to patients and the population. Its research activities encompass 8 major themes: Cancer; Infection, immunity and inflammation; Cardio-metabolic diseases; Musculo-skeletal diseases; Neurosciences; Risks to health; Global health and Care systems and services.

About CHUM

The Centre hospitalier de l'Université de Montréal (CHUM) provides specialized and ultra-specialized services to a regional and supra-regional clientele. Within its more immediate coverage area, the CHUM also provides general and specialized hospital care and services. The CHUM uses an integrated network model to carry out its five-part mandate of care, teaching, research, the assessment of technologies and health care methodologies, and the promotion of health care. The CHUM has received accreditation from the Canadian Council on Health Services Accreditation, for the 2007-2010 period. The CHUM is a member of the vast network of excellence in health formed by the Université de Montréal and its affiliated hospitals. www.chumontreal.qc.ca

About the Université de Montréal

Rooted in Montreal and international in its vocation, the Université de Montréal is one of the largest French-speaking universities in the world. Founded in 1878, the Université de Montréal includes 13 faculties and two professional schools. With its affiliated schools, HEC Montréal and École Polytechnique, the Université de Montréal is the leading centre for higher learning and research in Québec and one of the most important in North America. The Université de Montréal brings together some 2,500 teachers and researchers and is home to over 60, 000 students. To learn more about the Université de Montréal : www.umontreal.ca