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BULLETIN

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CANADIAN GENETIC DISEASES NETWORK

January 2, 1996

TWO CANADIAN RESEARCHERS MAKE MAJOR ADVANCE IN UNDERSTANDING CAUSE OF FRAGILE X SYNDROME

Canadian researchers Dr. Edward W. Khandjian and Dr. François Rousseau of the Unité de Recherche en Génétique Humaine et Moléculaire of the Saint-François-d'Assise Hospital Research Centre in Québec City, have announced a major advance in understanding the role of ribosomes in causing Fragile X syndrome of inherited mental retardation. Dr. Rousseau is a member of the Canadian Genetic Diseases Network.

Fragile X syndrome is the most common specific cause of mental retardation in children after Down's syndrome. Until now, the function of the FMR1 gene which is inactivated in the disease, has remained unknown. The Québec City team is the first to shed light on the normal function of the gene and to make a significant contribution in determining the role of the protein produced by the gene. The absence of this protein causes Fragile X syndrome.

Drs. Khandjian and Rousseau have succeeded in determining that the product of the FMR1 gene is a component of ribosomes, the protein factories of the cell where synthesis of all proteins takes place. This advance makes Fragile X syndrome the first known hereditary disease of the ribosome, the fundamental functional unit of the life of every cell.

The results of their research is published in the January 1 issue of the scientific journal *Nature Genetics.*

The association of the FMR1 protein with ribosomes shows that this protein has a role to play in the protein factories of the nervous system as well as all other cells in the body. Dr. Rousseau commented,"The absence of this protein in individuals affected with the most frequent form of inherited mental retardation is likely to stem from a suboptimal function of the protein factories in neurons." Neurons are cells which produce the largest amount of proteins and are potentially the most sensitive to any alteration in protein synthesis machinery.

Dr. Rousseau added, "This discovery opens a new major avenue for research in nervous system disorders as it shows that defects in one of the several ribosomes-associated proteins can cause mental retardation, autistic behavior or hyperactivity which are all symptoms of Fragile X syndrome."

Fragile X syndrome is a hereditary disease which typically affects one boy in 1500 and one girl in 2500. Several cases of the disease have appeared in families with no prior familial history of mental retardation. In November, Drs. Khandjian and Rousseau reported a high prevalence of Fragile X unaffected carriers (1 in 259 women) in the general population. These women are at risk of having a child with the disease.

Dr. Michael Hayden, Scientific Director of the Canadian Genetic Diseases Network which has been the principal supporter of the research, said "The discoveries made by the Québec team have made significant advances in the understanding of this devastating disease." The Canadian Genetic Diseases Network is a consortium of 38 of Canada's leading geneticists who are linked with 12 universities, 9 hospitals, and 10 core facilities across the country. The Network performs leading-edge research on common genetically transmitted diseases and works with industry partners on methods of detection and treatment of the diseases. Seed funding for Network research projects is provided by the federal Networks of Centres of Excellence Program via the Medical Research Council of Canada.

For further information contact:
Carol Smith, Network Manager
Canadian Genetic Diseases Network
Tel. 604-822-7217/7886