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Study shows a cause of retardation

Syndrome results from lack of protein for development, MDs say

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MONTREAL – One of the leading causes of mental retardation is the absence of a protein required for the proper development of neurons in the brain, according to a newly published study by a team of Quebec City researchers.

The underlying causes of fragile X syndrome were discovered by Dr. Edward Khandjian and Dr. François Rousseau of the Saint-François-d'Assise Hospital Research Centre, and published in the most recent issue of *Nature Genetics*.

Fragile X syndrome is the second leading cause of mental retardation after Down's syndrome. It affects about one in 2,000 children, leading to learning disabilities, hyperactivity

and autistic behaviour.

Although fragile X syndrome is a genetic defect, it shows up in many families with no previous history of mental retardation.

In an earlier study, the researchers found that one in 259 women in the general population are at risk of having a child with the disease.

While the gene involved in fragile X syndrome was discovered in 1991, Dr. Khandjian and Dr. Rousseau have only now discovered the role of the protein produced by the FMR1 gene, which is found in every tissue of the body, and concentrated in brain neurons.

The protein produced by the gene is essential to the proper functioning of ribosomes, the so-called protein factories of cells.

Because brain neurons are among the

cells that produce the largest volume of proteins, they are most sensitive to any breakdown of protein synthesis machinery. The researchers note that this is the reason mental retardation is the most visible result of the absence of FMR1 protein.

The discovery makes the fragile X syndrome the first hereditary disease of the ribosome, the most fundamental functional unit of the life of every cell.

The research by the two Quebec City doctors is supported by the Canadian Genetic Diseases Network, a consortium of 38 of Canada's geneticists. The network brings together researchers, hospitals and industrial partners.