BULLETIN

CANADIAN GENETIC DISEASES NETWORK

IMPORTANT SCIENTIFIC ADVANCE AFFECTS RESEARCH ON BONE TUMORS IN CHILDREN

Vancouver: June 1, 1998: A team of Canadian Genetic Diseases Network scientists, led by Dr. Frank Tufaro at the University of British Columbia, has identified the structure and function of a tumor suppressor protein, EXT1, which is encoded by the gene responsible for Hereditary Multiple Exostoses (HME). HME is a disease which causes the formation of bone tumors in children.

The team has discovered that EXT1 encodes an enzyme in the endoplasmic reticulum which contributes to the biosynthesis of an important biological regulatory molecule, heparan sulfate. Hallmark changes in heparan sulfate have been found in a variety of cancerous tumors.

This advance is the first to report the function of the HME gene and represents a significant starting point for a molecular attack on the structure and function of this important gene family of tumor suppressors. HME constitutes 50% of benign bone tumors and 15% of all bone tumors, while EXT1 defects have also been implicated in other cancers. The benign growths associated with HME can degenerate into malignancies in connective cartilage tissues and bone joints.

The results of the study will be published in the June 1 issue of the prestigious scientific journal, *Nature Genetics.* .../2

Commented Dr. Tufaro from his laboratory in Vancouver, “It has been found that most HME cases have been attributed to mutations in this gene; however, the function of the gene and how mutations cause the disease has until now been unclear.” He added, “The discovery brings us one step closer to understanding the cause of devastating bone cancer.”

Dr. Michael Hayden, Scientific Director of the Canadian Genetic Diseases Network (CGDN) which is headquartered at the University of British Columbia, said “These new findings by Dr. Tufaro and his team is an exciting step forward in the research of the molecular basis of this disease. I am very pleased that CGDN and the NCE program played a key role in supporting the discovery.”

Since 1990, the Canadian Genetic Diseases Network has been committed to leading-edge research in human genetic disease and to creating strategic partnerships to commercialize discoveries. Fifty scientists and their teams, based in 18 universities, hospitals, and research centres across Canada, focus on the molecular and cellular causes of inherited disease. In 1997, CGDN was awarded a further seven years’ core funding by the federal Networks of Centres of Excellence Program via the Medical Research Council of Canada, to conduct an expanded research program “From Genes to Therapies.”

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