BULLETIN

CANADIAN GENETIC DISEASES NETWORK

CANADIAN SCIENTIST HEADS DISCOVERY OF GENE LEADING TO INHERITED BLINDNESS

Toronto - November 14, 1997: An international team of scientists led by Dr. Roderick McInnes and Dr. Carol Freund, members of the Canadian Genetic Diseases Network who are located at The Hospital for Sick Children in Toronto, has identified a gene in which mutations cause an inherited retinal degenerative disease called cone-rod dystrophy. The disease develops over the course of many years and eventually leads to total blindness.

Mutations in the gene, called CRX, lead to the degeneration of the retina's light-sensing cells, the photoreceptors, which convert light energy into nerve impulses. The photoreceptor cells responsible for color perception and fine vision are called cones while rods are the cells governing vision in low light. The findings were made in collaboration with Canadian Genetic Diseases researchers Dr. Stephen Scherer and Dr. Lap-Chee Tsui, also at Hospital for Sick Children, and researchers in England and the United States, including Drs. Shomi Bhattacharya of University College, London, Constance Cepko of Harvard Medical School, and Samuel Jacobson of the University of Pennsylvania.

Results will be published in the November 14 issue of the scientific journal *Cell*.

Although cone-rod dystrophy is not common, Dr. McInnes explained that the identification of the gene has broader significance. "It's a major step along the road to understanding how photoreceptors develop and are maintained", he said. "We suspect that this gene may also be involved in other inherited diseases which lead to the degeneration of cones and rods, such as Retinitis Pigmentosa." He added that the CRX gene may be responsible for modifying the severity of other inherited diseases of the retina, explaining why two individuals in the same family and with the same disease may each experience very different rates of visual loss.

The current research builds on previous discoveries by Dr. McInnes and colleagues, the most recent in 1996, of a major regulatory gene called *Chx10* and its role in controling normal retina and eye development in mammals.

Dr. Michael Hayden, Scientific Director of the Canadian Genetic Diseases Network, headquartered in Vancouver, said "These new findings by Dr. McInnes and his team are an exciting advance in research into the molecular basis of inherited eye disease. The mutations in this disease may have relevance for people with different forms of blindness." He added " I am delighted that Network core facilities and Network funding played a key role in supporting this important work."

The Canadian Genetic Diseases Network is a nationwide Centre of Excellence committed to leading-edge research in human genetic disease, and to creating strategic partnerships to commercialize scientific discoveries. The Network's 50 scientists and their teams, based in 18 universities, hospitals, and research centres across Canada, focus on the molecular and cellular causes of inherited disease. The Network has recently been awarded a further seven years' core funding by the Networks of Centres of Excellence Program via the Medical Research Council of Canada, to conduct an expanded research program "From Genes to Therapies."

Information:
Carol Smith, Network Manager
Canadian Genetic Diseases Network
Tel. 604) 822-7217/822-7886
http://www.cgdn.generes